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Identification of common genetic risk variants for autism spectrum disorder

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Part I

Supplementary Notes

Sample description and data processing

1.1 The iPSYCH ASD sample

1.1.1 The iPSYCH sample

The iPSYCH ASD sample is a population based case-cohort sample extracted from a baseline cohort consisting of all children born in Denmark between May 1st 1981 and December 31st 2005[1]. Eligible were singletons born to a known mother and resident in Denmark on their one-year birthday. Cases were identified from the Danish Psychiatric Central Research Register (DPCRR)[2], which includes data on all individuals treated in Denmark at psychiatric hospitals (from 1969 onwards) as well as at outpatient psychiatric clinics (from 1995 onwards). Cases were identified with schizophrenia, bipolar affective disorder, affective disorder, ASD and ADHD up until 2012. The controls constitute a random sample from the set of eligible subjects.

1.1.2 The iPSYCH ASD GWAS sample

The ASD GWAS is based on a register update. Cases were selected from the iPSYCH sample as those diagnosed with ASD in 2013 or earlier by a psychiatrist according to ICD10, including diagnoses of childhood autism (ICD10 code F84.0), atypical autism (F84.1), Asperger's syndrome (F84.5), other pervasive developmental disorders (F84.8), and pervasive developmental disorder, unspecified (F84.9). As controls we selected from the random iPSYCH control cohort children that did not have an ASD diagnosis by 2013.

For descriptive statistics and numbers describing the data flow for the steps described below see Table 1.

1.1.3 Genotyping and calling

The samples were linked using the unique personal identification number to the Danish Newborn Screening Biobank (DNSB) at Statens Serum Institute (SSI), where DNA was extracted from Guthrie cards and whole genome amplified in triplicates as described previously[3, 4]. Genotyping was performed at the Broad Institute of Harvard and MIT (Cambridge, MA, USA) using the PsychChip array from Illumina (CA, San Diego, USA) according to the instructions of the manufacturer. Genotyping was carried out on the full iPSYCH sample in 23 waves and so was the subsequent data processing.

Genotype calling of markers with minor allele frequency (MAF) > 0.01 was performed by merging callsets from GenCall[5] and Birdseed[6] while less frequent variants were called with zCall[7]. Details on the merging procedure for variants with MAF > 0.01 are described elsewhere (<https://sites.google.com/a/broadinstitute.org/ricopili/utilities/merge-calling-algorithms> and [1]). Prior to the subsequent QC and imputation SNPs were excluded when they were on either of two lists: a) a global blacklist comprising SNPs for which genotyping failed in 4 cohorts genotyped at the Broad as part of the PsychChip project (Psychiatric Genomics Consortium) with Illumina's PsychChip and/or b) a local blacklist of SNPs for which the MAF in the GenCall and Birdseed callsets were substantially different ($\Delta_{\text{MAF}} > 5\%$) prior to the merging of variants.

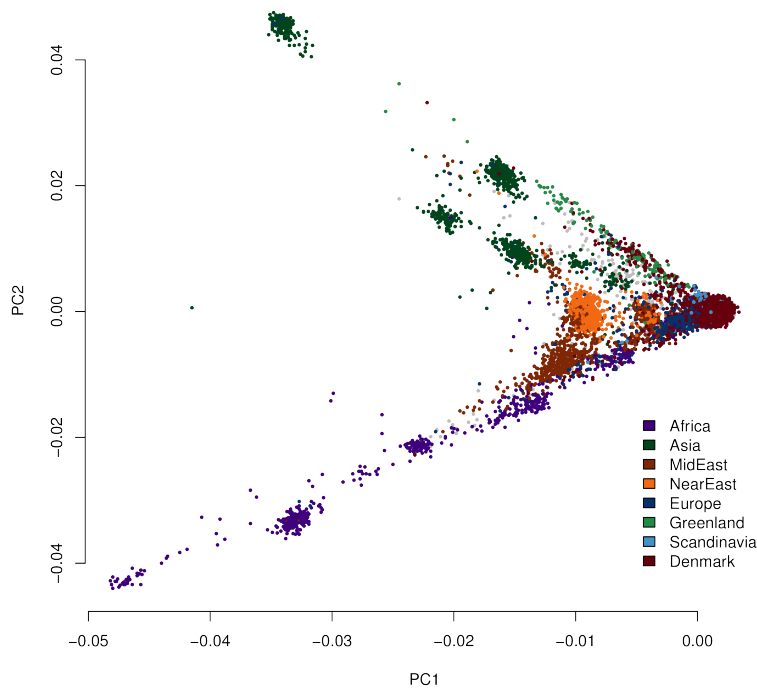
1.1.4 QC and Imputation

Ricopili[8], the pipeline developed by the Psychiatric Genomics Consortium (PGC) Statistical Analysis Group was used for quality control, imputation, principle component analysis (PCA) and primary association analysis. The data was processed separately in the 23 genotyping batches. Details for ricopili are described elsewhere (<https://sites.google.com/a/broadinstitute.org/ricopili/home>). In brief, the quality control (QC) step included manipulations of the data ensuring proper subsequent processing as well as filtering of SNPs and samples using a broad variety of different measures. Before subsequent imputation the data was (strand) aligned with the respective reference sample. Phasing was achieved using SHAPEIT v2[9] and imputation done by IMPUTE2[10, 11] with haplotypes from the 1000 Genomes Project, phase 3[12, 13] (1kGP3) as reference. Post imputation filtering of SNPs was carried out using measures for accuracy of imputation (INFO scores > 0.1) and MAF (> 0.5%).

1.1.5 PCA

After excluding regions of high LD[15], the genotypes were pruned down to a set of roughly 30k markers by pruning in a sliding window fashion using PLINK 1.9[16, 17]. This was done with an r^2 limit of 0.075, window size 1000 and step size 100. Using PLINK's identity by state analysis pairs of subjects were identified with $\hat{\pi} > 0.2$ and one subject of each such pair excluded at random (with a preference for keeping cases). PCA was carried out using smartPCA[18, 19]. A sub-sample of European ancestry was selected as an ellipsoid in the space of PC1-3 centred and scaled using the mean and 8 standard deviation of the sub-sample consisting of those whose parents and grandparents were all known from the registries to have been born in Denmark (n=31 500).

Principal component (PC) one and two of the initial PCA are shown in Figure 1 annotated by parental origin.



Supplementary Note, Figure 1: PCA plot showing the first to principal components of the initial PCA on the 39 542 unrelated individuals coloured by parental origin (if at least one parent was born outside of Europe that dictates the colour, red indicates all parents and grandparents were born in Denmark.)

A secondary PCA was run to provide covariates for the association analysis. These covariates were tested for association to the outcome (adjusting for index variables for the waves). In the association analysis we included PC1–4 plus any PC above that which was nominally significantly associated to the outcome.

1.2 The PGC ASD sample

In brief, five cohorts provided genotypes to the sample (n denoting the number of trios for which genotypes were available): The Geschwind Autism Center of Excellence (ACE; n = 391), the Autism Genome Project[20] (AGP; n = 2272), the Autism Genetic Resource Exchange[21, 22] (AGRE; n = 974), the NIMH Repository https://www.nimhgenetics.org/available_data/autism/, the Montreal[23]/Boston Collection (MONBOS; n = 1396, and the Simons Simplex Collection (SSC; n = 2231). The trios were analysed as cases and pseudo controls. A detailed description of the sample is available on the PGC web site: https://www.med.unc.edu/pgc/files/resultfiles/PGCASDEuro_Mar2015.readme.pdf and even more details are provided in[24].

The contributing samples were all processed for quality control, imputation, principle component analysis (PCA) and association analysis in the Ricopili pipeline[8]. In particular SHAPEIT[9] was used for phasing and imputation was done by IMPUTE2[10, 11] employing the 1000 Genomes Project, phase 3[13, 14] (1kGP3) as reference. Trio samples were imputed as a case-pseudo-controls design.

The CEU subset was chosen using a Euclidean distance measure weighted by the variance explain for each of the first 3 PCs. Individuals more distant than 10 standard deviations from the combined CEU and TSI HapMap reference populations were excluded.

1.3 The follow-up samples

1.3.1 deCODE

The Icelandic sample has been described previously[25]. Briefly, patients were ascertained through the State Diagnostic Counselling Center and the Department of Child and Adolescent Psychiatry, and were diagnosed based on ICD-10 criteria. Controls were recruited through ongoing projects at deCODE. Chip-typing, long-range phasing, imputation and association analysis were carried out as described previously[26].

Patients and controls from Georgia, Serbia and Ukraine were recruited by mental health clinics in their respective countries. Patients were diagnosed using ICD-10 criteria[27]. Samples were genotyped using Illumina arrays, phased with EAGLE v2, and imputed using IMPUTE v2 based on the 1000-genomes Phase3 reference data set.

All studies were approved by relevant ethics and data protection agencies, and written, informed consent was obtained from individuals providing blood samples.

1.3.2 Finnish autism case-control study

The families for the Finnish autism dataset were recruited via Finnish university and central hospitals, mainly Helsinki University Hospital, Jyväskylä Central Hospital and Kuopio University Hospital. The control samples originate from Finnish population cohorts[28]. Two groups of families were included 1) families with at least one child diagnosed with autism, including families with additional siblings diagnosed with other ASDs or 2) families with no individuals diagnosed with strict autism but at least two individuals with a diagnosis of Asperger Syndrome[29–32].

Diagnostic evaluations were made by a multidisciplinary group of clinicians at the neurological department of hospitals. Data were collected from extensive diagnostic examinations including neurological examinations, assessment of developmental history as well as psychological and neuropsychological examinations. Final diagnoses were based on ICD-10 and DSM-IV diagnostic nomenclatures. Families with known associated medical conditions or chromosomal abnormalities such as fragile X syndrome were excluded from the study. All families were Finnish except for one family where the father was of Turkish origin. Subsequently, the ADI-R was administered to autism families willing to continue to participate in the study. In a subset of the Finnish autism families a 96% concordance rate was observed between ICD-10 and ADI-R diagnosis of autism[33] making the Finnish autism families clinically comparable to international family sets used for genetic studies.

One individual was included from each family in the case-control datasets, choosing the most severely affected individual (i.e. autism before Asperger Syndrome). If several individuals in the family had the same diagnosis, the individual with highest genotyping success rate was included. Genotyping was performed in multiple stages

using Illumina HumanHap300/550, HumanHapCNV370 and Human610 arrays. All genotypes were called using Illumina BeadStudio.

1.3.3 PAGES

Data for this project were available from three sources, controls obtained from the Swedish Schizophrenia study and limited to the samples used in the Gaugler et al.[34]; ASD cases and controls genotyped at MSSM; and controls genotyped at MSSM. These samples were genotyped on different platforms and at different times. For details see Table 1. Genotypes for autosomal SNP were imputed up to the 1000G Phase 3 v5 reference panel using the Michigan Imputation Server in a separate job for each of the three sources.

Supplementary Note, Table 1: Available data from each of the three sources (207,822 SNP were genotyped on all three platforms.

Source	Cases	Controls	SNPs
Gaugler study (G)		2580	491,543
Nov 2016 (N)	1075	235	678,539
Jan 2017 (J)		1365	884,875
All	1075	4180	1,279,969

Removing controls appearing in more than one sample retaining index cases in a handful of full-sib pairs and parent-offspring resulted in 2477, 235, and 1355 controls from G, N, and J, respectively. In addition 1069 cases remained from N.

Genetic ancestry was determined using dacGem in GemTools[35, 36] based on 11,402 SNPs. These SNPs were selected from the set of 207,822 SNP with the conditions that they are autosomal, have a minor allele frequency > 0.05 , and pass pruning using `-indep-pairwise` with settings `window=50`, `overlap=5`, and $r^2 < 0.01$. The settings for dacGem were to use 500 random individuals in the base and to no longer sub-divide clusters after the initial round. dacGem identified 2 significant dimensions of ancestry and broke the data into 5 clusters.

For the replication we limited ourselves to the 622 and 3841 Swedish ancestry cases and controls, respectively. This reflects the exclusion of 304 PAGES samples that potentially were included in the combined iPSYCH-PGC-Aut analysis. Data were analysed using a model including the 2 significant eigenvectors obtained from GemTools.

1.3.4 BUPGEN

The study was part of the national Norwegian BUPGEN network, recruiting patients from Norwegian health services specializing in assessment of ASD and other neurodevelopmental disorders. The current sample comprised 102 children with ASD recruited between 2013 and May 2016 and assessed at age 6-18 years, with childhood autism (28%), atypical autism (8%), Asperger syndrome (37%) and unspecified pervasive developmental disorder(27%). 656 Controls were collected from the population of South-East Norway.

The children were assessed by a team of experienced clinicians (clinical psychologists and child psychiatrist). Diagnostic conclusions were best-estimate clinical diagnoses derived from tests, interview results and observations. All diagnoses were based on the International Statistical Classification of Diseases and Related Health Problems 10th Revision (ICD-10) (27). Criteria and autistic symptoms were evaluated using Autism Diagnostic Interview-Revised (ADI-R) (28) and/or Autism Diagnostic Observation Schedule (ADOS) (29). In addition, the assessment included a full medical and developmental history, physical examination, and IQ assessment.

Samples were genome-wide genotyped with Human Omni Express-24 v.1.1 at deCODE Genetics (Iceland). Quality control of genotypes, imputation, principal component analysis and primary association analyses were performed using the bioinformatics pipeline Ricopili[8] applying standard settings except where noted. Subjects and SNPs were included in the analyses based on the standard quality control settings in the Ricopili pipeline, with one adjustment: The SNP call rate filter was set to 0.96 instead of 0.98, as a stringent SNP call rate filter would exclude a large number of SNPs. Imputation was performed using the standard settings of the Ricopili pipeline, using haplotypes from the 1000 Genomes Project, phase 3 (1KGP3)[13, 14]. The data set included samples from three

separate batches; one of the batches contained only 2 cases and was excluded from the analysis after the imputation was performed. After these quality control steps, the study contained 194 cases and 3802 controls.

Prior to performing principal component analysis, a set of regions with high LD[15] was removed, and the number of controls was randomly reduced to four times the number of cases. The analysis was performed using Ricopili with the following settings: `ldsubgr 8000` (using all individuals), `nwind 1000` (the number of SNPs in window for pruning), `stepw 100` (window step size), `r2 0.075` (limit for LD pruning), `noproject` (perform smartpca on complete dataset). Removal of genetic outliers based on the principal components was performed in two steps. In both steps outliers were defined as samples that were outside of a 4-dimensional ellipsoid with center based on the first 4 principal components of the samples, and size based on the variance in the first 4 principal components among the samples. In the first step, an external set of homogenous Danish samples from the iPSYCH data set was included in the analysis. This set of homogenous Danish samples was assigned as defining the center and variance of the ellipsoid, and samples with a distance larger than 8 standard deviations in from the center in the 4-dimensional space were removed. This step excluded samples that were highly genetically heterogenous from Scandinavians. The second step was performed with the remaining samples, and without including the set of homogenous Danish samples. Genetic outliers were defined as having a distance larger than 4 standard deviations from the center. After removal of genetic outliers, the study contained 164 cases and 3449 controls. A final PCA was performed with these samples and the PCs used in the subsequent association analysis.

Association analysis was performed using standard Ricopili settings. The number of controls was randomly reduced to four times the number of cases. As the criterion for which principal components to include, we included the first four plus any of principal components 5–20 which had a significant correlation with case/control status. This resulted in principal components 1–4 and 8 being used as covariates in the association analysis.

Statistical Analyses

2.1 GWAS

2.1.1 SNP-wise analysis

Association analyses were done by applying PLINK 1.9 to the imputed dosage data (the sum of imputation probabilities $P(A1A2) + 2P(A1A1)$) including the first four principal components (PCs) as covariates as well as any PC beyond that, which were significantly associated with ASD in the sample. Combined results for iPSYCH and for iPSYCH with the PGC was achieved by meta analysing batch-wise and study-wise results using METAL[37] (July 2010 version) employing an inverse variance weighted fixed effect model[38]. Subsequently we applied a quality filter allowing only markers with an imputation info score ≥ 0.7 , MAF ≥ 0.01 and an effective sample size of at least 70% of the maximum. The effective sample size tells what the number of cases would be if a balanced design were to give equivalent statistical power. It was estimated from the number of cases, N_{ca} , and controls, N_{co} , contributing to the individual regression as $2N_{ca}N_{co}/(N_{ca} + N_{co})$.

The degree to which the deviation in the test statistics can be ascribed to cryptic relatedness and population stratification rather than to polygenicity was estimated from the intercept in LD score regression[39] (LDSC) as the ratio

$$\frac{\text{intercept} - 1}{\text{mean}(\chi^2) - 1}.$$

2.1.2 Gene-based association and gene-set analyses

MAGMA 1.06[40] was applied to the summary statistics to test for gene-based association. Using NCBI 37.3 gene definitions and restricting the analysis to SNPs located within the transcribed region (i.e. no padding was applied), mean SNP association was tested with the sum of $-\log(\text{SNP p-value})$ as test statistic. The resulting gene-based p-values were further used in gene-set enrichment analyses in MAGMA. We used the default settings where the regression is adjusted for gene size, gene density, sample size, the reciprocal of the minimal allele count, and the logarithm of all these.

One hypothesis driven analysis explored the candidate sets M13, M16 and M17 from Parikshak et al. 2013[41] as well as constrained genes[42, 43] determined from data of the Exome Aggregation Consortium (ExAC)[43]. The probability of being loss-of-function (LoF) intolerant (pLI) has been estimated based on the observed and expected protein-truncating variant (PTV) counts within each gene in the full ExAC data and are publicly available at ftp://ftp.broadinstitute.org/pub/ExAC_release/release0.3.1/functional_gene_constraint. Genes were defined as constrained if they had $pLI \geq 0.9$. Moreover, we added two small gene sets selected specifically for their association to ASD: the 65 genes identified at an $FDR < 0.1$ in [234], and the highly curated list of ASD genes(https://spark-sf.s3.amazonaws.com/SPARK_gene_list.pdf) from SPARK[235]. Another gene-set analysis was an hypothesis free analysis of the Gene Ontology[44, 45] sets ‘molecular function’ from MsigDB 6.0[46]. In general we analysed only genes outside the broad MHC region (hg19:chr6:25–35M) and included only gene sets with 10–1000 genes. However, the Sanders and the SPARK sets have no more than 1 gene in the MHC region and that was left in. All gene sets with significant enrichment were inspected to ensure that the signal was not generated by one or a few associated loci, where in reality only one of N genes in tight LD might truly be

associated.

2.1.3 MTAG analyses

MTAG[47] was run with standard settings pairing up the iPSYCH-PGC meta-analysis summary statistics with the summary statistics for major depression[48] (including the results from 23andMe[49], but excluding the Danish samples, 111 902 cases, 312 113 controls, and mean $\chi^2 = 1.477$), schizophrenia[8] (without the Danish samples, 34 129 cases, 45 512 controls, and mean $\chi^2 = 1.804$) and educational attainment[50] (328 917 samples and mean $\chi^2 = 1.648$) respectively. These samples are much better powered than the ASD sample with 18 381 cases, 27 969 controls and mean $\chi^2 = 1.201$. This poses the risk of generating signals that only have basis in the secondary phenotype. This however is countered by a more modest genetic correlation in the order of 0.2–0.4, which means that the weight given to the secondary phenotypes are equally modest ranging from 0.11 for educational attainment to 0.27 for schizophrenia with major depression slightly lower at 0.24. Hence, in general it will take a relatively strong ASD signal to generate a significant MTAG signal, and the rare cases where an exceptionally strong signal in the secondary phenotype is sufficient to drive home a significant MTAG signal will be easy to spot. This reasoning might not hold if analyzing more than two phenotypes simultaneously, where the interplay between the phenotypes gets more convoluted. Therefore, we ran the MTAG analyses pairwise rather than all together to minimize the risk of false positives.

The results were clumped and we highlighted loci of interest by selecting those that were significant at $5 \cdot 10^{-8}$ in the iPSYCH-PGC meta-analysis or the meta-analysis with the replication sample or were significant at $1.67 \cdot 10^{-8}$ in any of the three MTAG analyses. The composite GWAS consisting of the minimal p-values at each marker over these five analyses was used as a background when creating Manhattan plots for the different analyses. This way we are able to show both what is maximally achieved, where it comes from, and what the individual analysis contributes.

Moreover, we estimated max FDR — the FDR in the worst case. Briefly, the maxFDR estimation is done in the following way: For two traits, each locus will be associated with both, neither, or only one of the traits (the four states TT, FF, FT, and TF). If we knew the fraction of each type of SNP along with the distribution of effect sizes for true associations, it would be possible to estimate MTAG's FDR. These fractions are unknown, however, so we instead search through a fine grid in the space of possible fractions and calculate FDR for each point. The maximum achieved is the estimate maxFDR. To estimate FDR given the four fraction, we first assume that, conditional on a SNP being associated with one or both traits, the effect sizes are drawn from a normal or bivariate normal distribution. This implies that the unconditional distribution of effect sizes is a generalization of a spike-and-slab distribution (i.e., a mixture of normal distributions) which is a class of distributions that approximates many thick-tailed distributions. Distributions such as non-infinitesimal models where some fraction of SNPs are null for a subset of traits. For details see the supplementary note of Turley, P et al.[47]

As recommended in [47] we conducted the FDR analysis under the assumption that at least 10% of SNPs from the ASD summary statistics are tagging causal SNPs. For the secondary phenotype we allowed this to drop to 3% to accommodate a potentially much fewer causal SNPs for ASD. This resulted in estimated max FDR of 0.021 for educational attainment, 0.019 for schizophrenia and 0.012 for major depression giving a conservative estimate of max FDR for the combined analysis of 0.021. I comparison the analogous estimate for the ASD scan alone was 0.020.

2.1.4 Reviews

We compiled reviews of the loci in Table 4. Information on gene function, expression and association to disease was retrieved by targeted searches in the UniProtKB protein knowledgebase (<http://www.uniprot.org/uniprot/>), the GTEx Portal (<https://www.gtexportal.org/home/>), Pubmed (<https://www.ncbi.nlm.nih.gov/pubmed>) and SFARI Gene (<https://gene.sfari.org/>). Independent signals of association were identified for each locus using the NHGRI-EBI Catalog of published genome-wide association studies available at: www.ebi.ac.uk/gwas[51] (accessed October 10th, 2017). Each locus was defined as the $r^2 = 0.1$ clump around the index SNP (LD region from the Ricopilli output file), adding additionally 0.5 Mb to each side. The coordinates in hg19 were then lifted to hg38 using the Lift Genome Annotations function in the UCSC Genome Browser available at: <https://genome.ucsc.edu/>[52]. The hg38 coordinates were used to query the GWAS catalog, and studies filtered using the p-value threshold of $5 \cdot 10^{-8}$. All studies of mental, neurological, educational and brain related traits, having one or more SNPs within the locus reaching genome wide significance ($p=5 \times 10^{-8}$) are listed.

2.2 SNP heritability

SNP heritability was estimated using LDSC[39] for the full sample and GCTA[53–55] for sub-samples too small for LDSC.

LDSC

For LDSC we used precomputed LD scores based on the European ancestry samples of the 1000 Genomes Project[14] restricted to HapMap3[56] SNPs (downloaded from the <https://github.com/bulik/ldsc>). The summary stats with standard LDSC filtering were regressed onto these scores. For liability scale estimates, we used a population prevalence for Denmark of 1.22%[57]. Lacking proper prevalence estimates for subtypes and sex specific analyses, we scaled the full spectrum prevalence based on the composition of the case sample.

GCTA

For sub-samples too small for LDSC, the GREML approach of GCTA was used. On best guess genotypes (genotype probability > 0.8 , missing rate < 0.01 and MAF > 0.05) with INDELs removed, a genetic relatedness matrix (GRM) was fitted for the association sample (i.e. the subjects of European ancestry with $\hat{\pi} \leq 0.2$) providing a relatedness estimate for all pairwise combinations of individuals. Estimation of the phenotypic variance explained by the SNPs (REML) was performed including PC 1–4 as continuous covariates together with any other PC that was nominally significantly associated to the phenotype as well as batches as categorical indicator covariates. To test for statistical significance of differences in SNP heritabilities across subtypes, we conducted permutation test shuffling the different case labels while keeping the controls. I.e. for the subtypes childhood autism, atypical autism, Asperger’s and pervasive disorders, each permutation resulted in a different split of the cases into four parts of sizes equal to the original subtypes, which subsequently processed by GCTA together with the full control group. Thousand permutations were run for each analysis.

Partitioned heritability

Following Finucane et al.[58], we conducted an enrichment analysis of the heritability for SNPs for functional annotation and for SNPs located in cell-type-specific regulatory elements. Using first the same 24 overlapping functional annotations (stripped down from 53) as in Finucane et al., we regressed the χ^2 from the summary statistics on to the cell-type specific LD scores download from the site mentioned above with baseline scores, regression weights and allele frequencies based on European ancestry 1000 Genome Project data. The enrichment of a category was defined as the proportion of SNP heritability in the category divided by the proportion of SNPs in that category. Still following Finucane et al., we did a similar analysis using 220 cell type-specific annotations divided into 10 overlapping groups. In addition to this, we conducted an analysis based on annotation derived from data on H3K4Me1 imputed gapped peaks data from the Roadmap Epigenomics Mapping Consortium[59, 60]; more specifically information excluding the broad MHC-region (chr6:25–35MB). In line with Finucane et al. we truncate out of bounds estimates to the unit interval and does likewise for confidence intervals.

2.3 Genetic correlation and prediction

For the main samples, genetic correlations were estimated using LDSC[39] and for the analysis of ASD subtypes and subgroups where the sample size were generally small, we used GCTA[53–55]. In both cases, we followed the same procedures as explained above. The summary statistics were also uploaded to LD hub[61] for comparison to all together 219 phenotypes.

2.3.1 Polygenic risk scores

For the polygenic risk scores (PRS) we filtered and clumped the summary stats applying slight adaptations to the standard Ricioli process and parameters: We filtered at a MAF limit of 0.05, info score at 0.9 when present, and

we removed the broad MHC-region (chr6:25–35MB). When using external summary stats not processed Ricopili or imputed using different imputation references, we excluded all ambiguous markers to avoid potential strand conflicts. To improve performance of the scores and avoid including artefacts from batch effects, we restricted the summary stats to include only SNPs known to be present in the iPSYCH data at a reasonable quality (info score ≥ 0.6 and MAF ≥ 0.01) throughout all 23 sample waves. This step also checked for allele flips. Clumping was done on the filtered summary stats employing PLINK and the flags `-clump-p1 1`, `-clump-p2 1`, `-clump-r2 0.1` and `-clump-kb 500`.

PRS were generated by Ricopili (as interface to PLINK) at the default p-value thresholds ($5 \cdot 10^{-8}$, 10^{-6} , 10^{-4} , 0.001, 0.01, 0.05, 0.1, 0.2, 0.5 and 1) as a weighted sum of the allele dosages: Summing over the markers abiding by the p-value threshold in the training set and weighing by the additive scale effect measure of the marker ($\log(\text{OR})$ or β) as estimated in the training set. Scores were normalized prior to analysis.

We evaluated the predictive power using Nagelkerke's R^2 and plots of odds ratios and confidence intervals over score deciles. Both R^2 and odds ratios were estimated in regression analyses including the relevant PCs and indicator variable for genotyping waves.

Subtype analyses

The ASD subtypes are overlapping in the sense that one individual may have multiple of these sub-diagnoses. To analyse ASD subtypes in relation PRS we needed a disjoint set of individuals representing to a high degree subjects with the diagnoses in question. While lumping together 'other pervasive developmental disorder' (ICD10 F84.8) and 'pervasive developmental disorder, unspecified' (ICD10 F84.9) into one, 'pervasive disorder, mixed' (PDM), we created a set of non-overlapping hierarchical subtypes presented in Table 13 and whose counts can be seen in the last line of Table 1.

We are interested in whether the PRS for different phenotypes have different distribution or loading on ASD subtypes. In order to investigate that while taking into account both batch effect from the genotyping waves as well as PCs, we fit a multivariate regression

$$Y = XB + E,$$

where Y is a matrix of n observations on m response variables, the normalized PRS, X is a model matrix with k columns for regressors, B is the matrix of regression coefficients (one column for each response variable), and E is an error matrix. The first column of X consists of 1s for the regression constant and corresponds in B to the intercept, which is the first row. The next few columns, say k_0 , of X holds the subtype variables of interest while the subsequent ones are the wave dummy variables and PCs. This means that only the first rows of B are really of interest to us, and we take for the i th phenotype the vector $(\beta_{1i}, \dots, \beta_{k_0i})$ as a profile for how the score is loading across subtypes (β_{0i} being the intercept).

The advantage of the multivariate regression is that not only does it handle the correlation that may exist among the PRS, but most importantly it allows us to test a great number of hypotheses of interest. Eg. if the loading profile of one phenotype is different from that of another.

We test any linear null hypothesis by specifying a matrix equation:

$$H_0 : LBP = C,$$

where C is usually the zero matrix, L has k columns and selects and relates the subtypes β s within each score, and the matrix P with m rows selects and relates the scores to use. For instance to test any hypothesis on the i th score we set

$$P = (0, \dots, 0, 1, 0, \dots, 0)^T,$$

with 1 on the i th position. To select the k_0 element profile vector for testing, we specify the $(k_0 \times k)$ matrix

$$L = \begin{pmatrix} 0 & 1 & 0 & \dots & 0 & 0 & \dots & 0 \\ \vdots & \ddots & \ddots & \ddots & \vdots & \vdots & & \vdots \\ 0 & \dots & 0 & 1 & 0 & 0 & \dots & 0 \end{pmatrix}.$$

Say we want to test the 0-hypothesis that the profile for phenotype i is identical to that of phenotype j , we use the L -matrix just above and

$$P = (0, \dots, 0, 1, 0, \dots, 0, -1, 0, \dots, 0)^T,$$

where the 1 and the -1 are at the i th and j th position. This will be sensitive also an overall difference in loading, so if we want to attempt at a scale free version of the hypothesis testing just of the directions are the same (that the normalized vectors are identical), we can put

$$\mathbf{P} = (0, \dots, 0, \frac{1}{v_i}, 0, \dots, 0, -\frac{1}{v_j}, 0, \dots, 0)^T,$$

where $v_i = |(\beta_{1i}, \dots, \beta_{k_0i})|$. This will approximately be the correct test for a scale free hypothesis.

Internally trained PRS

Lacking a large ASD sample outside of iPSYCH and PGC, we trained a set of PRS for ASD internally in the following way. We divided the sample in five sub-samples of roughly equal size respecting the division into batches; see Table 2. We then ran five GWAS leaving out each group in turn from the training set and meta analysed these with the PGC results. This produced a set of PRS For each of the five sub-samples trained on their complement. Prior to analyses, each score was normalized on the group where it was defined. We evaluated the predictive power in each group and on the whole sample combined.

Supplementary Note, Table 2: Make-up of wave group for internal scoring. The table show the waves that compose the groups as well as case control counts in the groups and the complement of each group in the iPSYCH ASD GWAS plus the PGC ASD GWAS.

Group	Waves	Group		Compl+PGC	
		N cases	N ctrls	N cases	N ctrls
1	1, 3, 11 and 20	2 624	3 694	10 452	18 970
2	2, 8, 10 and 22	2 622	5 432	10 454	17 232
3	4, 9, 12, 19 and 21	2 611	4 666	10 465	17 998
4	5, 7, 13, 15 and 16	2 583	4 360	10 493	18 304
5	6, 14, 17, 18 and 23	2 636	4 512	10 440	18 152

Leveraging the power of polygenic overlap

As a simple way of exploiting the genetic overlap with other phenotype to help improve prediction of the PRS, we added PRS from a number of phenotypes in a weighted sum with the internally trained ASD score. The phenotypes we included were subjective well being[62], schizophrenia[8], depressive symptoms[62], educational attainment[50], chronotype[63], ADHD[64], MDD[65], Openness[66], Extraversion[66], Agreeableness[66] and Childhood intelligence[67]. For each phenotype we chose the one score of the ten with the highest Nagelkerke’s R^2 for ASD. The weights assigned could be any measure of ‘importance’ in context of ASD and we chose here to use the $\log(\text{OR})$ for the logistic regression of ASD status on each score as a continuous factor adjusting for relevant PCs and wave indicators. One at a time, starting with the phenotype with highest $\text{abs}(\log(\text{OR}))$ and ending with the lowest, we added a PRS weighted by its $\log(\text{OR})$ and then standardized. This way we ended up with a sequence of scores starting with $S_0 = S_{\text{ASD}}$ and continuing with:

$$S_k = \frac{S_{\text{ASD}} + \sum_{i=1}^k \log(\text{OR}_{P_i}) S_{P_i} - \mu \left(S_{\text{ASD}} + \sum_{i=1}^k \log(\text{OR}_{P_i}) S_{P_i} \right)}{\sigma \left(S_{\text{ASD}} + \sum_{i=1}^k \log(\text{OR}_{P_i}) S_{P_i} \right)},$$

where S_{ASD} is the ASD score, S_{P_i} the score for phenotype P_i , OR_{P_i} the odds ratio from the logistic regression of ASD status on S_{P_i} as a continuous factor adjusting for relevant PCs and wave indicators, and μ is the mean and σ the standard deviation. In Figure 93 we show Nagelkerke’s R^2 and in Figure 94 the decile plots over wave groups and on the full sample.

2.4 Hi-C analysis

The Hi-C data was generated from two major cortical laminae: the germinal zone (GZ), containing primarily mitotically active neural progenitors, and the cortical and subcortical plate (CP), consisting primarily of post-

mitotic neurons[68]. We first selected the top ranking loci from the ASD scan. Balancing the number of loci to work with and the depth in the p-value, we chose the top 30 loci, and of these 28 were represented in the reference set used in the Hi-C pipeline. The result was a list of 28 loci from which we derived a set of credible SNPs (putative causal SNPs) using CAVIAR[69]. To test whether credible SNPs are enriched in active marks in the fetal brain[60], we employed GREAT as previously described[68, 70]. Credible SNPs were, sub-grouped into those without known function (unannotated) and functionally annotated SNPs (SNPs in the gene promoters and SNPs that cause nonsynonymous variants) (Figure 98). Then we integrated unannotated credible SNPs with chromatin contact profiles during fetal corticogenesis[68], defining genes physically interacting with intergenic or intronic SNPs (Figure 98).

Spatiotemporal transcriptomic atlas of human brain was obtained from Kang et al[71]. We used transcriptomic profiles of multiple brain regions with developmental epochs that span prenatal (6–37 post-conception week, PCW) and postnatal (4 months–42 years) periods. Expression values were log-transformed and centered to the mean expression level for each sample using a $\text{scale}(\text{center}=T, \text{scale}=F)+1$ function in R. ASD candidate genes identified by Hi-C analyses (Figure 98) were selected for each sample and their average centered expression values were calculated and plotted.

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Part II

Supplementary Information

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3.1 Sample description

Supplementary Table 1: Descriptive statistics of key characteristics for the iPSYCH ASD GWAS sample. Top part shows the data flow from ascertainment in the registers, identification in the biobank and extraction and amplification of DNA, exclusion of related individuals to the final study sample consisting of individuals of European ancestry. Percents are relative to the register sample for the column. Everything below the line in the middle relates to the study sample of European ancestry.

[†] Median and IQR.

[§] The last of the hierarchical subtypes consists of those with a diagnosis of ‘other pervasive devopmental disorder’ or ‘pervasive developmental disorder, unspecified’ without any of the preceding subtype diagnoses. See table 13 for the definitions of the hierarchical subtypes.

Characteristics	Ctrl	ASD	CHA	ATA	Asp	OPDD	PDDU
Register sample	29659	17763	4892	2276	5910	2660	5097
Ident and WGAed	28316 (95.5%)	15744 (88.6%)	4324 (88.4%)	1995 (87.7%)	5270 (89.2%)	2394 (90.0%)	4522 (88.7%)
Gtyped and QCed	26005 (87.7%)	14970 (84.3%)	4060 (83.0%)	1889 (83.0%)	5049 (85.4%)	2286 (85.9%)	4306 (84.5%)
$\hat{\pi} \leq 0.2$	25205 (85.0%)	14337 (80.7%)	3850 (78.7%)	1800 (79.1%)	4848 (82.0%)	2194 (82.5%)	4141 (81.2%)
Eur ancestry	22664 (76.4%)	13076 (73.6%)	3310 (67.7%)	1607 (70.6%)	4622 (78.2%)	2042 (76.8%)	3753 (73.6%)
Birth years							
1981–1985	3782 (16.7%)	725 (5.5%)	97 (2.9%)	70 (4.4%)	382 (8.3%)	132 (6.5%)	158 (4.2%)
1986–1990	4775 (21.1%)	1764 (13.5%)	241 (7.3%)	194 (12.1%)	865 (18.7%)	321 (15.7%)	430 (11.5%)
1991–1995	5199 (22.9%)	3714 (28.4%)	762 (23.0%)	507 (31.5%)	1464 (31.7%)	640 (31.3%)	983 (26.2%)
1996–2000	5090 (22.5%)	4278 (32.7%)	1169 (35.3%)	544 (33.9%)	1367 (29.6%)	648 (31.7%)	1289 (34.3%)
2001–2005	3818 (16.8%)	2595 (19.8%)	1041 (31.5%)	292 (18.2%)	544 (11.8%)	301 (14.7%)	893 (23.8%)
Females	11168 (49.3%)	2820 (21.6%)	630 (19.0%)	407 (25.3%)	924 (20.0%)	463 (22.7%)	869 (23.2%)
ID (IQ<70)	109 (0.5%)	1873 (14.3%)	955 (28.9%)	460 (28.6%)	101 (2.2%)	180 (8.8%)	625 (16.7%)
Age at 1st diag [†]		10 (7 – 14)	7 (5 – 12)	11 (7 – 14)	12 (9 – 15)	11 (8 – 14)	10 (6 – 14)
ASD subtypes:							
Childhod autism		3310 (25.3%)		113 (7.0%)	147 (3.2%)	71 (3.5%)	598 (15.9%)
Atypical autism		1607 (12.3%)	113 (3.4%)		64 (1.4%)	51 (2.5%)	294 (7.8%)
Asperger		4622 (35.3%)	147 (4.4%)	64 (4.0%)		166 (8.1%)	554 (14.8%)
Other pervasive DD		2042 (15.6%)	71 (2.1%)	51 (3.2%)	166 (3.6%)		335 (8.9%)
Pervasive DD, unspec		3753 (28.7%)	598 (18.1%)	294 (18.3%)	554 (12.0%)	335 (16.4%)	
Hierar subtypes [§]			3310 (100.0%)	1494 (93.0%)	4417 (95.6%)	3855 (70.6%)	

Supplementary Table 2: Key numbers for the follow-up study. First three data rows show sample sizes, and the bottom rows show SNP counts. The numbers for the iPSYCH-PGC meta analysis and the numbers of SNPs (index SNPs, proxy SNPs and total) are displayed in the first data column. The subsequent columns provide the numbers SNPs that could be matched to index SNPs and to proxies when matching failed for index SNPs. Total SNPs for the replication samples is the total number of SNPs contributing to the analysis directly through index SNPs or via proxies when necessary.

Name	iPSYCH+PGC	BUPGEN	PAGES	Finland	deCODE Iceland	deCODE East Eur	In All Reps
Cases	18,381	164	926	159	574	296	2,119
Controls	27,969	656	3,841	526	136,968	388	142,379
Total	46,350	820	4,767	685	137,542	684	144,498
Index SNPs	88	88	61	82	71	85	45
Proxy SNPs	364	0	0	0	5	0	
Total SNPs	456	88	61	82	76	85	49

3.2 GWAS

3.2.1 The ASD GWAS and follow-up

Supplementary Table 3: Summary of the top 88 ASD associations from the iPSYCH-PGC meta analysis together with follow-up results and results of the combined analysis with the follow-up samples. The columns are a part from the basic identity and location of the loci, the p-values and ORs from the iPSYCH-PGC meta analysis, direction of effect for each follow-up sample (BUPGEN, PAGES, Finland, deCODE Iceland, deCODE East Eur; + for same direction as iPSYCH-PGC, 0 for opposite and ? for not present), p-values and ORs for follow-up together and the combined meta analysis of iPSYCH-PGC and the follow-up samples. All meta-analyses are inverse variance weighted and the p-values derived from z-scores. Loci identified by the MTAG analysis (Table 6) are marked by * (but 3 of the MTAG loci had ASD p-values below the threshold of 10^{-5}).

SNP	CHR	BP	A1A2	MAFCa	MAFCo	P	OR	Dir	P-fup	OR-fup	P-comb	OR-comb
rs910805	20	21,248,116	A/G	0.745	0.760	$2 \cdot 10^{-9}$	0.909	>+0+0<	0.312	1.039	$3 \cdot 10^{-7}$	0.927
rs10099100	8	10,576,775	C/G	0.348	0.331	$1 \cdot 10^{-8}$	1.088	>++++0<	0.310	1.036	$1 \cdot 10^{-8}$	1.080
rs71190156	20	14,836,243	GTTTT/T/G	0.460	0.481	$3 \cdot 10^{-8}$	0.925	>++++0<	0.362	0.969	$4 \cdot 10^{-8}$	0.931
rs6047270	20	21,122,212	T/C	0.335	0.319	$8 \cdot 10^{-8}$	1.084	>+0+0<	0.983	0.999	$8 \cdot 10^{-7}$	1.071
rs111931861	7	104,744,219	A/G	0.960	0.966	$1 \cdot 10^{-7}$	0.805	>+?+<	0.087	0.802	$3 \cdot 10^{-8}$	0.805
rs2391769	1	96,978,961	A/G	0.348	0.369	$1 \cdot 10^{-7}$	0.926	>+0+++<	0.687	0.986	$5 \cdot 10^{-7}$	0.935
rs138867053	19	37,439,641	A/G	0.021	0.017	$1 \cdot 10^{-7}$	1.331	>+?+0<	0.652	1.059	$4 \cdot 10^{-7}$	1.285
rs183563276m	8	48,036,474	A/G	0.987	0.989	$2 \cdot 10^{-7}$	0.658	>?000<	0.133	1.524	$4 \cdot 10^{-6}$	0.701
rs1452075*	3	62,481,063	T/C	0.738	0.721	$2 \cdot 10^{-7}$	1.084	>+0+0<	0.565	1.021	$6 \cdot 10^{-7}$	1.074
rs1222063	1	96,602,440	A/G	0.349	0.332	$3 \cdot 10^{-7}$	1.084	>+0+0<	0.315	1.035	$3 \cdot 10^{-7}$	1.075
rs142920272	17	44,301,840	T/C	0.798	0.813	$3 \cdot 10^{-7}$	0.913	>+?00<	0.982	1.001	$9 \cdot 10^{-7}$	0.919
rs55962189	20	21,242,161	A/T	0.690	0.706	$3 \cdot 10^{-7}$	0.926	>+0+0<	0.602	1.019	$6 \cdot 10^{-6}$	0.939
rs112635299	14	94,838,142	T/G	0.030	0.025	$3 \cdot 10^{-7}$	1.247	>+?+<	0.327	1.208	$2 \cdot 10^{-7}$	1.245
rs6701243	1	99,092,784	A/C	0.626	0.609	$3 \cdot 10^{-7}$	1.076	>++++0<	0.644	1.016	$1 \cdot 10^{-6}$	1.067
rs45595836	10	16,691,399	T/C	0.078	0.067	$3 \cdot 10^{-7}$	1.149	>0++++<	0.898	0.991	$3 \cdot 10^{-6}$	1.126
rs325485*	5	103,995,368	A/G	0.396	0.378	$3 \cdot 10^{-7}$	1.076	>+0+++<	0.891	1.005	$2 \cdot 10^{-6}$	1.066
rs201910565	1	96,561,801	A/AT	0.672	0.689	$3 \cdot 10^{-7}$	0.927	>++++0<	0.039	0.930	$4 \cdot 10^{-8}$	0.927
rs210894m	6	11,731,999	T/TA	0.566	0.548	$5 \cdot 10^{-7}$	1.073	>+0+0<	0.251	1.039	$4 \cdot 10^{-7}$	1.068
rs72934503*	6	98,583,488	A/G	0.523	0.537	$6 \cdot 10^{-7}$	0.932	>+?+++<	0.152	0.942	$2 \cdot 10^{-7}$	0.933
rs11185408	1	104,792,257	A/G	0.503	0.523	$7 \cdot 10^{-7}$	0.934	>+?+0<	0.756	0.987	$1 \cdot 10^{-6}$	0.939
rs141455452	17	44,019,083	T/G	0.542	0.525	$9 \cdot 10^{-7}$	1.082	>+?+0<	0.320	1.048	$6 \cdot 10^{-7}$	1.078
rs78827416	10	72,749,037	A/G	0.093	0.086	$9 \cdot 10^{-7}$	1.139	>++++0<	0.799	0.986	$2 \cdot 10^{-5}$	1.108
rs59566011	2	159,385,181	GA/G	0.586	0.569	$9 \cdot 10^{-7}$	1.073	>?0++<	0.515	1.028	$1 \cdot 10^{-6}$	1.068
rs1066089m	8	53,341,258	C/CAA	0.733	0.745	$1 \cdot 10^{-6}$	0.919	>+?00<	0.167	1.062	$5 \cdot 10^{-5}$	0.937
rs147317628	17	44,277,476	CTTTAG/C	0.212	0.195	$1 \cdot 10^{-6}$	1.087	>+++0<	0.991	0.999	$3 \cdot 10^{-6}$	1.079
rs292441	18	55,872,558	A/G	0.660	0.676	$1 \cdot 10^{-6}$	0.930	>+0+0<	0.616	0.982	$3 \cdot 10^{-6}$	0.937
chr5:168173526	5	168,173,526	A/G	0.020	0.016	$1 \cdot 10^{-6}$	1.342	>+?+0<	0.065	1.286	$2 \cdot 10^{-7}$	1.332
rs4750990	10	130,488,026	T/C	0.591	0.603	$1 \cdot 10^{-6}$	0.934	>00000<	0.168	1.048	$9 \cdot 10^{-5}$	0.950
rs117603308m	11	106,827,977	T/C	0.030	0.026	$1 \cdot 10^{-6}$	1.270	>+?0?<	0.901	0.984	$9 \cdot 10^{-6}$	1.229
rs34938366	16	72,097,255	C/CA	0.179	0.168	$2 \cdot 10^{-6}$	1.109	>+0+0<	0.717	1.014	$1 \cdot 10^{-5}$	1.086
rs35404050	12	73,196,902	T/C	0.210	0.195	$2 \cdot 10^{-6}$	1.088	>+0000<	0.902	0.995	$1 \cdot 10^{-5}$	1.073
rs11480060	20	54,230,218	C/CA	0.712	0.727	$2 \cdot 10^{-6}$	0.927	>+?000<	0.489	1.032	$2 \cdot 10^{-5}$	0.937
rs740883	6	29,575,405	A/T	0.902	0.912	$2 \cdot 10^{-6}$	0.893	>+?+<	0.386	0.937	$1 \cdot 10^{-6}$	0.896
rs16879023	6	16,753,147	A/G	0.136	0.148	$2 \cdot 10^{-6}$	0.909	>+0+0<	0.900	1.006	$2 \cdot 10^{-5}$	0.923
rs141319505	10	65,421,442	A/G	0.983	0.980	$2 \cdot 10^{-6}$	1.337	>+?+<	0.937	1.016	$4 \cdot 10^{-6}$	1.308
rs77691144	13	66,970,212	T/C	0.969	0.975	$2 \cdot 10^{-6}$	0.813	>+?+?<	0.152	0.795	$6 \cdot 10^{-7}$	0.811
rs4916723	5	87,854,395	A/C	0.560	0.572	$2 \cdot 10^{-6}$	0.935	>+0+++<	0.506	0.978	$3 \cdot 10^{-6}$	0.941
rs2635182	5	92,255,166	T/C	0.485	0.466	$2 \cdot 10^{-6}$	1.069	>+++++<	0.102	1.057	$5 \cdot 10^{-7}$	1.067
rs10110094	8	131,472,047	A/G	0.171	0.163	$2 \cdot 10^{-6}$	1.095	>+0+0<	0.154	1.068	$8 \cdot 10^{-7}$	1.091
rs12942300	17	43,859,405	A/T	0.158	0.171	$2 \cdot 10^{-6}$	0.915	>+?+<	0.478	0.958	$2 \cdot 10^{-6}$	0.918
rs78058104	15	93,953,737	A/G	0.035	0.030	$2 \cdot 10^{-6}$	1.207	>0?0+<	0.106	0.790	$3 \cdot 10^{-5}$	1.172
chr7:105064665	7	105,064,665	T/TCCCTCCCTCTCT+6	0.263	0.250	$2 \cdot 10^{-6}$	1.081	>?0+<	0.590	1.028	$3 \cdot 10^{-6}$	1.076
rs28729902	9	76,179,384	A/G	0.803	0.814	$2 \cdot 10^{-6}$	0.920	>+0000<	0.242	1.048	$1 \cdot 10^{-4}$	0.940
rs144911765	21	37,255,329	T/C	0.966	0.971	$2 \cdot 10^{-6}$	0.827	>+?+0<	0.729	1.044	$1 \cdot 10^{-5}$	0.845
rs11787216*	8	142,615,222	T/C	0.347	0.364	$3 \cdot 10^{-6}$	0.933	>00+++<	0.482	0.976	$4 \cdot 10^{-6}$	0.940
rs33966416	4	171,206,603	C/CA	0.456	0.470	$3 \cdot 10^{-6}$	0.937	>?00+<	0.197	1.058	$5 \cdot 10^{-5}$	0.947
rs200332011	3	158,001,670	CT/C	0.219	0.231	$3 \cdot 10^{-6}$	0.922	>?+00<	0.715	0.982	$6 \cdot 10^{-6}$	0.928
rs7000276	8	10,804,843	A/G	0.408	0.392	$3 \cdot 10^{-6}$	1.068	>+0+0<	0.726	0.989	$3 \cdot 10^{-5}$	1.055
rs13188074	5	113,801,423	A/G	0.408	0.391	$3 \cdot 10^{-6}$	1.068	>+++++<	0.246	1.041	$2 \cdot 10^{-6}$	1.064
rs9389208	6	135,035,609	T/C	0.374	0.359	$3 \cdot 10^{-6}$	1.070	>+0+0<	0.323	0.967	$9 \cdot 10^{-5}$	1.053
rs6692705	1	193,502,609	A/G	0.413	0.400	$3 \cdot 10^{-6}$	1.068	>+0+0<	0.118	1.053	$1 \cdot 10^{-6}$	1.066
rs76397219	8	60,390,318	A/G	0.926	0.933	$4 \cdot 10^{-6}$	0.869	>+?00<	0.859	1.017	$1 \cdot 10^{-5}$	0.882
chr11:102751102	11	102,751,102	C/<CN0>	0.955	0.963	$4 \cdot 10^{-6}$	0.851	>+?00<	0.823	1.036	$8 \cdot 10^{-6}$	0.859
chr7:71618506	7	71,618,506	T/TA	0.365	0.381	$4 \cdot 10^{-6}$	0.934	>?00+<	0.751	1.014	$2 \cdot 10^{-5}$	0.942
rs79940520	3	191,838,169	A/G	0.847	0.859	$4 \cdot 10^{-6}$	0.909	>+0+0<	0.664	0.977	$9 \cdot 10^{-6}$	0.918
rs201369005m	14	76,835,623	G/GT	0.057	0.051	$4 \cdot 10^{-6}$	1.169	>?+0+<	0.913	1.012	$9 \cdot 10^{-6}$	1.155
rs78653484	1	147,183,927	T/C	0.032	0.039	$5 \cdot 10^{-6}$	0.838	>+++0<	0.807	0.974	$1 \cdot 10^{-5}$	0.853
rs12203328	6	23,767,038	C/G	0.291	0.278	$5 \cdot 10^{-6}$	1.072	>+++++<	0.170	1.052	$2 \cdot 10^{-6}$	1.069
rs116977567	20	54,184,200	T/G	0.970	0.974	$5 \cdot 10^{-6}$	0.822	>0?00<	0.029	1.334	$3 \cdot 10^{-4}$	0.861
rs529507	11	131,773,383	A/G	0.840	0.848	$6 \cdot 10^{-6}$	0.913	>+0+++<	0.826	0.990	$2 \cdot 10^{-5}$	0.925
rs77810738	19	37,392,818	A/G	0.963	0.968	$6 \cdot 10^{-6}$	0.842	>+++<	0.289	0.898	$4 \cdot 10^{-6}$	0.849
rs113003385	12	113,568,531	A/G	0.075	0.071	$6 \cdot 10^{-6}$	1.142	>+0+0<	0.640	1.032	$1 \cdot 10^{-5}$	1.124
rs12449792	17	43,302,259	T/C	0.471	0.459	$6 \cdot 10^{-6}$	1.066	>000+0<	0.111	0.948	$4 \cdot 10^{-4}$	1.047
rs6430841	2	140,318,199	A/G	0.815	0.801	$6 \cdot 10^{-6}$	1.083	>+0+0<	0.537	1.025	$1 \cdot 10^{-5}$	1.073
rs13201465	6	134,234,093	A/G	0.984	0.987	$6 \cdot 10^{-6}$	0.755	>+?0+<	0.897	0.977	$2 \cdot 10^{-5}$	0.775
rs7578456	2	202,235,348	A/G	0.417	0.401	$7 \cdot 10^{-6}$	1.066	>+0+0<	0.726	0.988	$6 \cdot 10^{-5}$	1.053
rs34739626	6	23,870,775	T/TA	0.630	0.617	$7 \cdot 10^{-6}$	1.073	>?+00<	0.515	1.027	$8 \cdot 10^{-6}$	1.067
rs6082289	20	21,019,305	T/C	0.218	0.207	$7 \cdot 10^{-6}$	1.079	>+0000<	0.189	0.948	$3 \cdot 10^{-4}$	1.058

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Supplementary Table 3 – continued from previous page

SNP	CHR	BP	A1A2	MAFca	MAFco	P	OR	Dir	P-fup	OR-fup	P-comb	OR-comb
rs597040	20	3,663,913	C/G	0.019	0.016	7·10 ⁻⁶	1.294	>+?+0<	0.857	1.029	2·10 ⁻⁵	1.259
rs148587110	3	20,641,966	T/C	0.986	0.989	7·10 ⁻⁶	0.719	>0+?0?<	0.062	0.715	1·10 ⁻⁶	0.718
chr6:96509180	6	96,509,180	D/I	0.657	0.641	7·10 ⁻⁶	1.078	>+?+0+<	0.446	1.034	8·10 ⁻⁶	1.072
rs62256326	3	70,608,317	A/G	0.263	0.252	7·10 ⁻⁶	1.074	>+0++0<	0.528	1.024	1·10 ⁻⁵	1.066
rs116346488	4	132,217,275	A/G	0.054	0.048	8·10 ⁻⁶	1.154	>+0?00<	0.890	0.987	3·10 ⁻⁵	1.136
rs507059	6	23,909,381	A/G	0.170	0.182	8·10 ⁻⁶	0.917	>00++0<	0.993	1.000	5·10 ⁻⁵	0.931
rs2282828	6	15,392,310	T/C	0.406	0.419	8·10 ⁻⁶	0.933	>0?00+<	0.373	1.039	1·10 ⁻⁴	0.945
rs143609523	18	33,265,230	A/G	0.986	0.988	8·10 ⁻⁶	0.745	>?+?+?<	0.069	0.692	1·10 ⁻⁶	0.740
rs56054767m	4	84,621,317	C/CA	0.124	0.113	9·10 ⁻⁶	1.113	>0?++++<	0.202	1.089	4·10 ⁻⁶	1.110
rs41363353	2	63,072,403	C/G	0.835	0.848	9·10 ⁻⁶	0.919	>0+00+<	0.841	0.991	3·10 ⁻⁵	0.930
rs115833252	1	61,879,203	T/C	0.024	0.019	9·10 ⁻⁶	1.262	>+0?0?<	0.421	0.860	5·10 ⁻⁵	1.228
rs78298487	7	132,177,702	T/G	0.980	0.984	9·10 ⁻⁶	0.775	>+0?0?<	0.837	1.047	2·10 ⁻⁵	0.789
rs4609618	11	128,818,792	A/C	0.370	0.389	9·10 ⁻⁶	0.938	>+++000<	0.645	1.016	9·10 ⁻⁵	0.949
rs16933101	12	28,765,600	A/G	0.044	0.051	9·10 ⁻⁶	0.858	>+0+00<	0.873	0.989	5·10 ⁻⁵	0.883
rs113764414	10	64,649,396	A/G	0.683	0.672	9·10 ⁻⁶	1.076	>0++++<	0.016	1.090	5·10 ⁻⁷	1.078
rs17517971	13	110,828,119	A/G	0.068	0.063	1·10 ⁻⁵	1.152	>00000<	0.392	0.947	4·10 ⁻⁴	1.107
rs684502	6	89,896,927	T/G	0.554	0.566	1·10 ⁻⁵	0.939	>00+00<	0.554	1.020	1·10 ⁻⁴	0.951
rs34509057	5	153,601,262	A/G	0.238	0.226	1·10 ⁻⁵	1.076	>+++++<	0.255	1.045	6·10 ⁻⁶	1.071
rs201179706	10	30,995,488	T/C	0.494	0.508	1·10 ⁻⁵	0.934	>0?+0+<	0.945	1.003	3·10 ⁻⁵	0.941
rs71868489	2	171,338,669	C/CT	0.827	0.817	1·10 ⁻⁵	1.090	>+?0+0<	0.879	1.009	2·10 ⁻⁵	1.081

Supplementary Table 4: Review of the top loci identified in the single SNP analysis with and without follow-up sample, the three MTAG analyses, and the gene-wise MAGMA analysis (see also main Table 1, Table 3, 10 and Box 1 for further details and explanations of significance tests). Loci are $r^2 = 0.1$ clumps and the genes listed are the protein coding genes in the locus except marked with * (see below) or on chromosome 8 where the locus around rs10099100 was restricted to a $r^2 = 0.6$ clump plus 50kb flanks. See section 2.1.4 for details.

* Nearest protein coding gene within ± 1 Mb from locus.

** Results based on samples overlapping the present ASD sample, hence not independent.

† Previous GWS hits for brain disorders in locus.

Chr	Gene	Index SNP (BP)	Analysis P-value	Function	Tissue specificity	Disease	Previous GWS hits in locus [†]
1	<i>NEGR1</i>	rs1620977 (72729142)	MTAG MD: $6.7 \cdot 10^{-9}$	<i>NEGR1</i> encodes the Neuronal growth regulator 1, a GPI-anchored IgLON protein belonging to the immunoglobulin superfamily of cell adhesion molecules. <i>NEGR1</i> has been identified as a raft-associated component of the brain that is involved in neurite outgrowth[72–74] and neurodevelopmental determination of synapse number in the hippocampus[75]. <i>NEGR1</i> expression levels are affected by nutritional state in brain areas relevant to feeding[76] and <i>NEGR1</i> may also serve a role in intracellular cholesterol trafficking[77].	<i>NEGR1</i> is localized at postsynaptic sites of dendritic and somatic synapses[78] and is expressed at high levels during postnatal development in cerebral cortex, hippocampus, cerebellum, and hypothalamus[78–80].	A 1p31.1 deletion including a part of <i>NEGR1</i> (and no other genes) has been identified in two siblings with a history of neuropsychiatric and behavioral problems, learning difficulties, hypotonia, mild aortic root dilatation, hypermobility and scoliosis[81].	Meta-analysis of autism spectrum disorder and schizophrenia[24]**, Schizophrenia[82], Educational attainment[50], Depressive symptoms[62], Major depression[48], Intelligence[83]
1	<i>PTBP2</i> *	rs201910565 (96561801)	Comb: $2.5 \cdot 10^{-8}$	<i>PTBP2</i> encodes Polypyrimidine tract-binding protein 2. <i>PTBP2</i> is a <i>PTBP1</i> (Polypyrimidine tract-binding protein 1) paralog and is also known as nPTB (neuronal PTB) or brPTB (brain PTB). <i>PTBP1</i> and <i>PTBP2</i> binds to intronic polypyrimidine clusters in pre-mRNA molecules and each target large sets of exons to coordinate programs of splicing events during development[84]. During neuronal development and differentiation, several switches in the expression of <i>PTBP1</i> and <i>PTBP2</i> activates networks of new spliced isoforms[85–88].	<i>PTBP2</i> is expressed at high levels in adult brain, testis, myoblasts and lymphocytes[89, 90]. Different isoforms generated by alternative splicing of <i>PTBP2</i> are expressed in a tissue specific manner[90, 91]. In neuronal cells, <i>PTBP2</i> acts by autoregulating its own exon 10 inclusion, leading to an increased expression level of the <i>PTBP2</i> protein[92]. <i>PTBP1</i> promotes the exon 10 exclusion from <i>PTBP2</i> transcripts, leading to NMD of <i>PTBP2</i> transcripts[93].	Hemizygous deletion of varying genomic regions containing <i>PTBP2</i> , <i>DPYD</i> and <i>MIR137</i> have been reported in cases with ASD, severe speech delay and intellectual disability[94, 95]. Two brothers with ASD and intellectual disability born to consanguineous parents were found to be homozygous for a novel 5 bp indel variant located in a human accelerated region (HAR) between <i>DPYD</i> and <i>PTBP2</i> [96]. This intergenic indel variant was suggested to affect the <i>PTBP2</i> -directed enhancer activity of the region and that this effect was relatively specific to neurons[96]. Further evidence pointing to <i>PTBP2</i> as a ASD risk gene include identification of a <i>de novo</i> <i>PTBP2</i> potentially damaging missense variant in an ASD proband[97] as well as identification of a <i>de novo</i> <i>PTBP2</i> intronic insertion in a ASD proband from a simplex family[98].	Educational attainment[50]

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Supplementary Table 4 — continued from the previous page

Chr	Gene	Index SNP (BP)	Analysis P-value	Function	Tissue specificity	Disease	Previous GWS hits in locus [†]
3	<i>CADPS</i>	rs1452075 (62481063)	MTAG Edu: $3.2 \cdot 10^{-9}$	<i>CADPS</i> encodes the Calcium-dependent secretion activator 1 (CAPS-1). CAPS-1 serves an essential role in the docking and priming of dense core vesicles and synaptic vesicles in endocrine and neural cells, [99] and references herein.	In line with <i>CADPS</i> mRNA being mainly expressed in brain and pituitary (https://www.gtexportal.org), immunoreactive CAPS-1 is localized in neural and various endocrine tissues[100].	A <i>de novo</i> microdeletion including <i>CADPS</i> (and five other genes) has been described in a case with intellectual disability and autistic features with language impairment[101]. The <i>CADPS2</i> gene, encoding a CAPS-1 isoform (CAPS-2) of almost identical function is located in the 7q autism susceptibility locus, <i>AUTS1</i> . <i>CADPS2</i> knockout mice show phenotypes with translational relevance to autism and an aberrant <i>CADPS2</i> splice variant have been identified in patients with autism[102].	Cognitive decline rate in late mild cognitive impairment[103]
3	<i>FEZF2</i>		Hi-C interaction with <i>CADPS</i> locus	<i>FEZF2</i> encodes the Fez family zinc finger protein 2 (also known as known as <i>FEZL</i> and ZFP312/ZNF312). <i>FEZF2</i> is a transcriptional repressor containing 6 highly conserved C2H2 zinc-finger repeats[104]. <i>FEZF2</i> is crucial in multilineage differentiation of neurons in zebra fish[104, 105] and it regulates the differentiation of layer-5 subcortical projection neurons in mice[106]. <i>FEZF2</i> also play a role in medullary thymic epithelial cells to ensure immunologic tolerance against certain tissue antigens and suppress development of autoimmunity[107].	<i>FEZF2</i> seems to be almost exclusively expressed in amygdala, hippocampus and cortex (https://www.gtexportal.org). This is consistent with mouse <i>Fezf2</i> being selectively expressed in layer V and VI subcortical projection pyramidal neurons and their progenitor cells[108].	<i>FEZF2</i> has been associated with ASD in two cohorts of European ancestry[109] and a <i>FEZF2 de novo</i> missense variant has been identified in ASD[110]. <i>Eomesa/tbr2</i> and <i>lhx2b</i> has been identified as target genes of <i>Fezf2</i> [111]. Mutations in <i>EOMES/TBR2</i> cause microcephaly in humans[112] and <i>lhx2b</i> is a critical regulator of cell fate and axonal targeting in the developing fore-brain[113]. <i>FEZF2</i> is regulated by a non-exonic cis-regulatory element (E4) located 7.3 kb downstream of the <i>Fezf2</i> transcription start site[114]. The potential role of <i>FEZF2</i> (and related transcription factors) in autism has is discussed in detail in[115].	
4	<i>TMEM33</i>	rs16854048 (42123728)	MTAG MD: $1.3 \cdot 10^{-8}$	<i>TMEM33</i> encodes Transmembrane protein 33. <i>TMEM33</i> is a three transmembrane domain reticulon binding protein localized in the ER membrane[116]. It is likely involved in regulating the ER tubule polygonal network[116]. <i>TMEM33</i> is ER stress-inducible in cancer cells and regulates positively two main drivers of the unfolded protein response: PERK and IRE1 α [117].	<i>TMEM33</i> mRNA is ubiquitously expressed (https://www.gtexportal.org).	<i>TMEM33</i> (along with malectin and PDIA6) enhances human cytomegalovirus US2-mediated degradation of Major Histocompatibility Complex Class I molecules[118].	
4	<i>DCAF4L1</i>	rs16854048 (42123728)	MTAG MD: $1.3 \cdot 10^{-8}$	<i>DCAF4L1</i> encodes the DDB1- and CUL4-associated factor 4-like protein 1. <i>DCAF4L1</i> interactions with ubiquitin conjugation pathway proteins CUL4A, COPS5 and COPS6 have been demonstrated experimentally (IntAct database). Otherwise, <i>DCAF4L1</i> is largely uncharacterized.	<i>DCAF4L1</i> seems to be exclusively expressed in testis (https://www.gtexportal.org).	Variants in <i>DCAF4L1</i> may be involved in hemangioblastomas[119].	

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Supplementary Table 4 — continued from the previous page

Chr	Gene	Index SNP (BP)	Analysis P-value	Function	Tissue specificity	Disease	Previous GWS hits in locus [†]
4	<i>SLC30A9</i>	rs16854048 (42123728)	MTAG MD: $1.3 \cdot 10^{-8}$	<i>SLC30A9</i> encodes Zinc transporter 9. <i>SLC30A9</i> has been identified as a nuclear receptor coactivator involved in Wnt signaling[120, 121]. A later paper found that <i>SLC30A9</i> is involved in zinc transport in neuronal cells[122].	<i>SLC30A9</i> mRNA is ubiquitously expressed with high levels in fetal brain, cerebellum, skeletal muscle and kidney[122].	A single amino acid deletion (Ala350del) in <i>SLC30A9</i> has been associated with early-onset autosomal recessive cerebro-renal syndrome comprising different combinations of profound intellectual disability (very low social and verbal skills), muscle weakness, oculomotor apraxia and early onset nephropathy[122]. In neuronal cell lines, the mutation seems to affect zinc transport and thus, intracellular zinc homeostasis rather than Wnt signaling[122]. It has been shown that autism cases show developmental dysregulation in zinc uptake[123].	
4	<i>BEND4</i>	rs16854048 (42123728)	MTAG MD: $1.3 \cdot 10^{-8}$	<i>BEND4</i> encodes BEN domain-containing protein 4. <i>BEND4</i> is largely uncharacterized.	<i>BEND4</i> transcript is mainly expressed in EBV-transformed lymphocytes and testis (https://www.gtexportal.org).	Microduplications in 7q11.23 have been associated with autism in a simplex cohort with a frequency of 0.09% in 3816 ASD probands[124]. Notably, <i>BEND4</i> has been identified as a target gene of the key 7q11.23 gene, <i>GTF2I</i> [124] and is proposed as one among three genes responsible for mediating the molecular pathogenesis of both 7q-microduplication syndrome and Williams-Beuren syndrome that also comprise ASD features[124]. The evidence has mainly been obtained from patient-derived induced pluripotent stem cells and include <i>BEND4</i> sensitivity to the dosage of <i>GTF2I</i> and its LSD1-mediated repressive activity but also that <i>BEND4</i> expression is inversely correlated with the expression of <i>GTF2I</i> in human brain[124].	
5	<i>NUDT12*</i>	rs325506 (104012303)	MTAG MD: $3.3 \cdot 10^{-11}$	<i>NUDT12</i> encodes Peroxisomal NADH pyrophosphatase <i>NUDT12</i> , a peroxisomal enzymes that hydrolyses NAD(P)H to NMNH and AMP (2',5'-ADP), and diadenosine diphosphate to AMP[125]. <i>NUDT12</i> may act to regulate the concentration of peroxisomal nicotinamide nucleotide cofactors required for oxidative metabolism[125]. Besides, <i>NUDT12</i> may possess mRNA decapping activity thereby regulating RNA stability and degradation[125].	<i>NUDT12</i> is expressed in many tissues with highest levels in adrenal gland, bladder, spinal chord, transformed fibroblasts, tibial nerve and thyroid (https://www.gtexportal.org).		Educational attainment[50]

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Supplementary Table 4 — continued from the previous page

Chr	Gene	Index SNP (BP)	Analysis P-value	Function	Tissue specificity	Disease	Previous GWS hits in locus [†]
5	KCNN2		MAGMA: $1.0 \cdot 10^{-9}$	KCNN2 encodes Small conductance calcium-activated potassium channel protein 2. KCNN2 (alternatively SK or KCa2.2) is a voltage-independent Ca ²⁺ -activated K ⁺ channel that responds to changes in intracellular calcium concentration and couples calcium metabolism to potassium flux and membrane excitability. In CNS neurons, activation of KCNN2 modulates neuronal excitability by causing membrane hyperpolarization[126]. Hippocampal KCNN2 play an important role in the formation of new memory[127], in encoding and consolidation of contextual fear[128] and in drug-induced plasticity[129].	Synaptic levels of KCNN2 are regulated by the E3 ubiquitin ligase UBE3A[130], whose deficiency results in Angelman syndrome and overexpression in increased risk of ASD, respectively. At least four different isoforms of KCNN2 are expressed in human brain including the hippocampal formation, amygdala and neocortex[131].		Educational attainment[50].
6	MMS22L*	rs2388334 (98591622)	MTAG Edu: $3.3 \cdot 10^{-12}$	MMS22L encodes Protein MMS22-like. MMS22L is part of the TONSL–MMS22L complex implicated in homologous recombination, replication fork recovery, and reading of replication-dependent histone marks[132–136].	MMS22L mRNA levels are low in most tissues with highest levels in EBV-transformed lymphocytes and testis (https://www.gtexportal.org).	MMS22L might an oncogene involved in lung and esophageal carcinogenesis[137] and it is part of a 15-gene expression signature associated with the development of bone metastases in breast cancer[138].	Bipolar disorder[139, 140], Intelligence[83], Cognitive function[141], Educational attainment [50, 67, 142, 143]
6	POU3F2*	rs2388334 (98591622)	MTAG Edu: $3.3 \cdot 10^{-12}$	POU3F2 encodes POU domain, class 3, transcription factor 2 (alternatively Brn-2, Oct-7 and OTF-7). POU3F is a member of the POU-III class of neural octamer-binding transcription factors[144]. In mouse, POU3F2 is essential for the development of the PVN and SON neuronal lineages in the hypothalamus[145]. POU3F2 and POU3F3 are co-expressed in the developing neocortex where they redundantly regulate cortical neuron migration and layer production[146, 147]. POU3F2 is a downstream target of the SIM1-ARNT2 complex in the leptin-melanocortin-SIM1 pathway playing an important role in regulating expression of oxytocin in the hypothalamus[148].	POU3F2 seems to be exclusively expressed in brain and tibial nerve with lowest levels in cerebellum (https://www.gtexportal.org). POU3F2 is further plentifully expressed during all stages of neurogenesis and in distinct neural subsets in the adult central nervous system (although at lower levels), including the PVN and SON of the hypothalamus[149].	Small deletions including POU3F2 are causing susceptibility to obesity, variable developmental delay with intellectual disability and autism[148, 150]. Heterozygous POU3F2 knockout mice show deficits in adult social behavior and increased repetitive behavior in the marble burying test combined with embryonic ventricular zone and cortical plate expansion[151]. Deregulation of a β -catenin/POU3F2/Tbr2 transcriptional cascade may be responsible for embryonic expansion of basal neural progenitor cells and adult social and repetitive behavioral abnormalities[151]. Homopolymeric amino acid repeat-deleted POU3F2 mice show impairments in maternal behavior and pup recognition, cognitive deficits and impaired adult hippocampal neurogenesis[152, 153]. Besides, POU3F2 is involved in melanocytic growth, tumorigenesis and metastatic growth after dissemination of melanoma, [154] and references herein.	Bipolar disorder[139, 140], Intelligence[83], Cognitive function[141], Educational attainment [50, 67, 142, 143]

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Supplementary Table 4 — continued from the previous page

Chr	Gene	Index SNP (BP)	Analysis P-value	Function	Tissue specificity	Disease	Previous GWS hits in locus [†]
7	<i>KMT2E</i> (<i>MLL5</i>)	rs111931861 (104744219)	Comb: $3.5 \cdot 10^{-8}$	<i>KMT2E</i> (alternatively <i>MLL5</i>) encodes Histone-lysine N-methyltransferase 2E. <i>KMT2E</i> was initially categorized into the <i>KMT2</i> (<i>MLL</i>) family, however phylogenetic analysis has shown that <i>KMT2E</i> is distinct from the <i>MLL1-4</i> , <i>SETD1A</i> , and <i>SETD1B</i> group of proteins and forms a family together with another mammalian protein, the SET containing-domain 5 (<i>SETD5</i>) [155, 156]. The molecular function of <i>KMT2E</i> is uncertain since it has been suggested that <i>KMT2E</i> unlike other <i>KMT2</i> (<i>MLL</i>) proteins lack intrinsic methyltransferase activity [155]. Several lines of evidence however support that recognition of the histone H3K4me3 mark by the <i>KMT2E</i> PHD finger can facilitate the recruitment of <i>KMT2E</i> to active transcription chromatin regions [157, 158]. <i>KMT2E</i> play important roles in controlling cell cycle progression, maintaining genomic stability, as well as regulating hematopoiesis and spermatogenesis (for review see [159]).	<i>KMT2E</i> is expressed in various tissues with highest levels in cerebellum and low levels in other brain regions (https://www.gtexportal.org).	<i>KMT2E</i> has been suggested as an ASD risk gene [160]. Evidence include identification of two different <i>KMT2E de novo</i> frameshift variants in two unrelated ASD cases [161, 162] and identification of a maternally inherited frameshift variant in a Chinese ASD case [160].	Schizophrenia [8, 82], Meta-analysis of autism spectrum disorder and schizophrenia [24]**
7	<i>SRPK2</i>	rs111931861 (104744219)	Comb: $3.5 \cdot 10^{-8}$	<i>SRPK2</i> encodes the SRSF protein kinase 2 which is a cell cycle-regulated kinase that phosphorylates serine residues in the serine/arginine domain of various mRNA splicing factors and mediates pre-mRNA splicing [163, 164]. <i>SRPK2</i> also phosphorylates tau, suppresses tau-dependent microtubule polymerization, and inhibits axonal elongation in neurons [165]. Interaction of Akt-phosphorylated <i>SRPK2</i> with 14-3-3 mediates cell cycle and cell death in neurons [166]. Depletion of <i>SRPK2</i> in dentate gyrus inhibits tau phosphorylation in the APP/PS1 mouse and alleviates impaired cognitive behaviors [165].	<i>SRPK2</i> is mainly expressed in brain but also at high levels in testis (https://www.gtexportal.org)	Activity of <i>SRPK2</i> is elevated in pathological structures of human Alzheimer's disease brain [165].	
8	<i>C8orf74</i>	rs10099100 (10576775)	ASD: $1.1 \cdot 10^{-8}$ Comb: $9.6 \cdot 10^{-9}$ MTAG Edu: $1.6 \cdot 10^{-8}$	<i>C8orf74</i> encodes a protein <i>C8orf74</i> that is not yet characterized. <i>C8orf74</i> has experimentally been shown to interact with histone-lysine-N methyltransferases SUV39H1 and H2, protein arginine N-methyltransferase 6 (<i>ANM6</i>) and Lysine-specific histone demethylase 1A (<i>KDM1A</i>) (IntAct database).	<i>C8orf74</i> is abundantly expressed in testis and is furthermore detected in pineal gland tissue (https://www.gtexportal.org).		Schizophrenia [82], Extraversion [167], Neuroticism [62, 167]

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Supplementary Table 4 — continued from the previous page

Chr	Gene	Index SNP (BP)	Analysis P-value	Function	Tissue specificity	Disease	Previous GWS hits in locus [†]
8	<i>SOX7</i>	rs10099100 (10576775)	ASD: $1.1 \cdot 10^{-8}$ Comb: $9.6 \cdot 10^{-9}$ MTAG Edu: $1.6 \cdot 10^{-8}$	<i>SOX7</i> encodes Transcription factor SOX-7. <i>SOX7</i> is both a potent activator[168], and repressor[168] and it functions in several early developmental signaling pathways, including VEGF/Flk1 signaling, Wnt signaling, and Notch pathway[169]. <i>SOX7</i> is implicated in embryonic stem cell differentiation[170, 171] and in the regulation of embryonic development[172] and vascular development in particular[169]. <i>SOX7</i> acts as a tumor suppressor through the Wnt/ β -catenin signaling pathway[173].	<i>SOX7</i> mRNA is detected during embryonic development in many tissues, most abundantly in brain, heart, lung, kidney, prostate, colon and spleen[168, 169]. In adults, <i>Sox7</i> is expressed in various tissues with highest levels in vagina and ectocervix but low levels in brain tissues (https://www.gtexportal.org).		Schizophrenia[82], Extraversion[167], Neuroticism[62, 167]
8	<i>PINX1</i>	rs10099100 (10576775)	ASD: $1.1 \cdot 10^{-8}$ Comb: $9.6 \cdot 10^{-9}$ MTAG Edu: $1.6 \cdot 10^{-8}$	<i>PINX1</i> encodes PIN2/TERF1-interacting telomerase inhibitor 1, a microtubule-binding protein essential for faithful chromosome segregation[174, 175]. <i>PINX1</i> interact with telomere repeat binding factor 1 (TRF1) and telomerase[176] and can potentially inhibit telomerase activation and telomeres elongation in cancer cells[177, 178]. <i>PINX1</i> is further implicated in regulating chromosome stability[179]. <i>PINX1</i> overexpression significantly suppresses the growth of hepatocellular carcinoma cells, whereas <i>PINX1</i> inhibition potentially enhances cell growth[180]. Depletion of <i>PINX1</i> also increases tumorigenicity in nude mice[181]. <i>PINX1</i> might be a putative tumor suppressor[182].	<i>PINX1</i> is ubiquitously expressed including in the brain, where <i>PINX1</i> mRNA is more abundant in cerebellar tissue than in other brain regions (https://www.gtexportal.org).	Rare variants in the <i>PINX1</i> gene have been identified in individuals with ASD[183].	Schizophrenia[82], Extraversion[167], Neuroticism[62, 167]
8	<i>MROH5*</i>	rs11787216 (142615222)	MTAG Edu: $2.0 \cdot 10^{-9}$	<i>MROH5</i> encodes Maestro heat-like repeat family member 5. <i>MROH5</i> interactions with the <i>GPSM3</i> G-protein-signaling modulator 3 and the <i>CHCHD2</i> transcription factor have been demonstrated experimentally (IntAct database). Otherwise, <i>MROH5</i> is largely uncharacterized.	<i>MROH5</i> seems to be exclusively expressed in testis (https://www.gtexportal.org).		Educational attainment[50]

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Supplementary Table 4 — continued from the previous page

Chr	Gene	Index SNP (BP)	Analysis P-value	Function	Tissue specificity	Disease	Previous GWS hits in locus [†]
14	MARK3	rs10149470 (104017953)	MTAG MD: $8.5 \cdot 10^{-9}$	MARK3 (also known as CTAK1 or EMK2) encodes MAP/microtubule affinity-regulating kinase 3. MARK3 belongs to family of four highly conserved kinases, MARK1-4. MARKs are evolutionarily conserved and required for polarity in various species including mammals (references within[184]). MARKs have numerous different upstream regulators (e.g. Pim-1, LKB1, and CaMKI) as well as downstream substrates (e.g. Pkp2, Cdc25C, Class II HDAC, Dlg/PSD-95, and MAP2/4/TAU)[184] illuminating an extreme multifunctionality. Specifically, MARK3 negatively regulates the Hippo signaling pathway (controlling organ size in animals) and antagonizes the phosphorylation of LATS1[185]. MARK3 has also been implicated in hippocampal function[186] and CagA (Helicobacter pylori)-associated epithelial cell polarity disruption[187].	MARK3 transcripts are ubiquitously expressed with highest levels in cerebellum (https://www.gtexportal.org).	MARK3 (together with SPTBN1) has been associated with bone mineral density[188, 189]. Loss of MARK3 in mice leads to a hypermetabolic state with reduced body weight, decreased adiposity, resistance to hepatic steatosis, and hypofertility[184]. MARK1 has been associated with autism, overexpression in prefrontal cortex in post-mortem brains from patients, and accelerated evolution in the human lineage. Modulation of MARK1 expression seems to modify both dendrite length and the velocity of mitochondrial transport along microtubules in primary cultures of mouse cortical neurons[190].	Schizophrenia[8, 82], Meta-analysis of autism spectrum disorder and schizophrenia[24]**
14	CKB	rs10149470 (104017953)	MTAG MD: $8.5 \cdot 10^{-9}$	CKB encodes Creatine kinase B-type (also known as brain creatinine kinase, B-CK). CKB is one of two cytosolic creatinine kinase isoforms that control energy demand through phosphocreatine consumption for local ATP production at subcellular sites of high energy demand[191, 192]. CKB plays a crucial role in brain energy homeostasis and in GABA neurons by activating KCC2[193].	CKB is highly expressed in brain, colon, esophagus, prostate and stomach (https://www.gtexportal.org).	ASD is part of the clinical symptoms seen in creatine deficiency syndrome (CDS) along with intellectual disability, speech and language delay, and epilepsy[194]. Serum CKB has been found to be elevated in ASD and to be associated with disease severity[195]. CKB has been found to be decreased in the brains of patients with Alzheimer disease, Huntington disease, and Pick disease[196] and references herein. Mutant huntingtin (mHTT) seems to act directly on the CKB promoter to repress its activity and overexpression of CKB has been shown to rescue the ATP depletion, aggregate formation, impaired proteasome activity, and shortened neurites induced by mHTT[196]. CKB has also been implicated with schizophrenia by proteomics[197] and proteomics-based reverse genetics[198]. Upstream regions of the CKB gene has been found to be differential methylated in fibroblasts from a discordant monozygotic twin pair with Rett syndrome[199].	Schizophrenia[8, 82], Meta-analysis of autism spectrum disorder and schizophrenia[24]**
14	TRMT61A	rs10149470 (104017953)	MTAG MD: $8.5 \cdot 10^{-9}$	TRMT61A encodes tRNA (adenine(58)-N(1))-methyltransferase catalytic subunit TRMT61A. tRNA (adenine-N1)-methyltransferase catalyzes the formation of N1-methyladenine at position 58 (m1A58) in initiator methionyl-tRNA[200].	TRMT61A is ubiquitously expressed (https://www.gtexportal.org).		Schizophrenia[8, 82], Meta-analysis of autism spectrum disorder and schizophrenia[24]**

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Supplementary Table 4 — continued from the previous page

Chr	Gene	Index SNP (BP)	Analysis P-value	Function	Tissue specificity	Disease	Previous GWS hits in locus [†]
14	<i>BAG5</i>	rs10149470 (104017953)	MTAG MD: $8.5 \cdot 10^{-9}$	<i>BAG5</i> encodes BAG family molecular chaperone regulator 5 (also known as Bcl-2-associated athanogene 5). <i>BAG5</i> is special to the BAG family of proteins since it consists entirely of five Bag domains[201]. Bag domains mediates direct interaction with the ATPase domain of Hsp70/Hsc70 molecular chaperones and <i>BAG5</i> function thereby as a nucleotide exchange factor to enhance Hsp70-mediated protein refolding[202].	<i>BAG5</i> is ubiquitously expressed with highest levels in testis (https://www.gtexportal.org). <i>BAG5</i> expression has been shown to increase during endoplasmic reticulum (ER) stress in cardiomyocytes and this apparently modulates Hsps70 protein stability and reduces ER stress[203].	<i>BAG5</i> seems to be involved in neuroprotection in Parkinsons disease[204–207], Alzheimers disease[208], and spinocerebellar ataxia type-3 (SCA3)[209].	Schizophrenia[8, 82], Meta-analysis of autism spectrum disorder and schizophrenia[24]**
14	<i>APOPT1</i>	rs10149470 (104017953)	MTAG MD: $8.5 \cdot 10^{-9}$	<i>APOPT1</i> (previously <i>C14ORF153</i>) encodes Apoptogenic protein 1, mitochondrial. Mitochondrial localization of <i>APOPT1</i> has been experimentally validated[210].	<i>APOPT1</i> is ubiquitously expressed with highest levels in skeletal muscle and testis (https://www.gtexportal.org).	Mutations in <i>APOPT1</i> are causing cavitating leukoencephalopathy with Cytochrome c oxidase deficiency[210]. Alternative splicing of <i>APOPT1</i> has been suggested as the underlying mechanism of the association signal of the locus containing <i>APOPT1</i> in schizophrenia[211].	Schizophrenia[8, 82], Meta-analysis of autism spectrum disorder and schizophrenia[24]**
14	<i>KLC1</i>	rs10149470 (104017953)	MTAG MD: $8.5 \cdot 10^{-9}$	<i>KLC1</i> encodes Kinesin light chain 1. Kinesin is a microtubule-associated force-producing protein involved in organelle transport. <i>KLC1</i> may play a role in coupling of cargo to the Kinesin heavy chain or in modulation of its ATPase activity[212, 213]. Fast anterograde axonal transport of APP is mediated by direct binding between APP and <i>KLC1</i> [214].	<i>KLC1</i> is highly expressed in many tissues with very high levels in brain (https://www.gtexportal.org).	<i>KLC1</i> has been suggested to be involved in Alzheimers disease[215], age-related macular degeneration[216], and cortical cataracts[217].	Schizophrenia[8, 82], Meta-analysis of autism spectrum disorder and schizophrenia[24]**
14	<i>XRCC3</i>	rs10149470 (104017953)	MTAG MD: $8.5 \cdot 10^{-9}$	<i>XRCC3</i> encodes DNA repair protein XRCC3. <i>XRCC3</i> is part of the RAD51-related protein family[218] and forms the CX3 complex with one of them, RAD51C[219]. <i>XRCC3</i> is involved in homologous recombination repair of DNA double-strand breaks[220] and plays an important role in maintaining chromosome stability and DNA damage repair[221].	<i>XRCC3</i> is expressed in many tissues with highest levels in spleen and low levels in all brain parts except cerebellum (https://www.gtexportal.org).	<i>XRCC3</i> SNPs have been suggested as a modifier of various different cancers e.g. bladder, breast, non-melanoma skin, and colorectal cancers, [222] and references herein.	Schizophrenia[8, 82], Meta-analysis of autism spectrum disorder and schizophrenia[24]**
20	<i>MACROD2</i>	rs71190156 (14760747)	ASD $2.8 \cdot 10^{-8}$ Comb $3.0 \cdot 10^{-8}$ MTAG Edu $\cdot 10$	<i>MACROD2</i> (also known as <i>C20orf133</i>) encodes O-acetyl-ADP-ribose deacetylase <i>MACROD2</i> , an enzyme in the nucleus, that binds to mono-ADP-ribosylated (MARylated) proteins and functions as an eraser of mono-ADP-ribosylation[223]. Intracellular MARylated histones and GSK3 β are substrates of macroD2, and the removal of MAR from GSK3 β is responsible for reactivating of its kinase activity [223].	<i>MACROD2</i> is expressed in EPV-transformed lymphocytes, lung and multiple regions of the brain. Low or no expression across most other tissue (https://www.gtexportal.org).	A heterozygous <i>de novo</i> 250-kb deletion in the <i>MACROD2</i> , initially identified in a patient with Kabuki syndrome (a congenital mental retardation syndrome)[224], was later found not to be the cause of the syndrome[225].	Autism spectrum disorder[20]

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Supplementary Table 4 — continued from the previous page

Chr	Gene	Index SNP (BP)	Analysis P-value	Function	Tissue specificity	Disease	Previous GWS hits in locus [†]
20	<i>XRN2</i>	rs910805 (21248116)	ASD $2.0 \cdot 10^{-9}$ MTAG SCZ $1.5 \cdot 10^{-10}$ MTAG Edu $1.3 \cdot 10^{-8}$	<i>XRN2</i> encodes the 5'-3' exoribonuclease 2 promoting transcription termination at the 3' ends of genes[226]. In differentiated rat neurons, <i>XRN2</i> is involved in cleavage of miRNA precursors, suggesting that the gene is involved in a novel mechanism for control of miRNA expression[227].	<i>XRN2</i> is transcribed uniformly across most tissues, with lowest levels in all brain regions except cerebellum where levels are substantially higher (https://www.gtexportal.org).		
20	<i>KIZ</i>	rs910805 (21248116)	ASD $2.0 \cdot 10^{-9}$ MTAG SCZ $1.5 \cdot 10^{-10}$ MTAG Edu $1.3 \cdot 10^{-8}$	<i>KIZ</i> encodes centrosomal protein Kizuna. The gene is also known as <i>C20orf19</i> or <i>PLK1S1</i> . Protects mature centrosomes from collapse during spindle formation[228]. <i>KIZ</i> -depleted cells has been shown to grow more slowly, have nuclear abnormalities, and undergoing cell cycle arrest[228].	<i>KIZ</i> is expressed across a wide range of tissues including the brain, highest in cerebellum and cerebellar hemisphere (https://www.gtexportal.org).		
20	<i>NKX2-4</i>	rs910805 (21248116)	ASD $2.0 \cdot 10^{-9}$ MTAG SCZ $1.5 \cdot 10^{-10}$ MTAG Edu $1.3 \cdot 10^{-8}$	<i>NKX2-4</i> encodes Homeobox protein Nkx-2.4. <i>NKX2-4</i> belong to the large family of homeobox-containing transcription factors, distinguished by a 60-amino acid evolutionarily conserved DNA-binding homeodomain. Morpholino knockdown of the zebrafish orthologs of <i>NKX2-1</i> and <i>NKX2-4</i> (Nkx2.4a and Nkx2.4b) revealed a contribution of these transcription factors to neural patterning and neuronal differentiation in zebrafish brain the development of the hypothalamus, preoptic region, and pallidum [229].	<i>NKX2-4</i> is exclusively expressed in hypothalamus, pituitary and testis (https://www.gtexportal.org).	Given the expression pattern and the effects of <i>NKX2-4</i> knockdown in zebrafish, <i>NKX2-4</i> might be important in hypothalamus-pituitary function and eventually oxytocin production. Of special note, deletions affecting 20p11-20p12 have been described in patients with panhypopituitarism[230] and hypopituitarism has been associated with lower saliva oxytocin concentrations and reduced empathic ability[231].	
20	<i>NKX2-2</i>	rs910805 (21248116)	ASD $2.0 \cdot 10^{-9}$ MTAG SCZ $1.5 \cdot 10^{-10}$ MTAG Edu $1.3 \cdot 10^{-8}$	<i>NKX2-2</i> encodes Homeobox protein Nkx-2.2. This homeo domain transcription factor has an essential role in interpreting the Sonic hedgehog signals and selecting neuronal identity[232] The gene is part of the networks controlling the development of mid-brain dopaminergic neurons in the mouse brain[233].	<i>NKX2-2</i> transcripts are found in different regions of the brain and pituitary (https://www.gtexportal.org).		

3.2.2 Genetic correlation with other phenotypes

Supplementary Table 5: The complete output from LD Hub (except for ‘Years of schooling (proxy cognitive performance)’ and ‘Years of schooling 2013’ and 5 other removed out of redundancy and ASD and PGC cross-disorder due to sample overlap) with a few extra phenotypes added that had not yet been included in LD Hub. Altogether there are 234 phenotypes. The columns are trait, PMID of the publication, Category as defined by LD Hub, Ethnicity, SNP correlation, r_G , standard error of r_G , z-score of r_G , p-value for r_G , SNP heritability on the observed scale, $h^2_{G, \text{obs}}$, and its standard error, $h^2_{G, \text{Int}}$. Int is the intercept of $h^2_{G, \text{obs}}$, and it comes with its own standard error, and finally GCOV Int is the cross-trait LD Score regression intercept with standard error. The colour of the estimate of genetic correlation, r_G , signifies whether the correlation is positive, **blue**, or negative, **red**, and p-values (estimated from z-test) that are significant after Bonferroni correction on the number of tests are painted **red**. The PMID is hyperlinked to the particular pubmed page. For tables concerning genetic overlap between subtypes of ASD, see section 3.3.

* Values from in-house analyses of new summary statistics not yet included in LD Hub. The major depression is the latest PGC MD scan (excluding the iPSYCH samples).

** Values from in-house LD score pipeline as the values from LD Hub are invalid due to erroneous summary stat files at LD Hub.

Trait	PMID	Category	Ethnicity	r_G	se	Z	P	$h^2_{G, \text{obs}}$ (se)	$h^2_{G, \text{Int}}$ (se)	GCOV Int (se)
Fathers age at death	27015805	aging	European	0.138	0.090	1.540	$1.24 \cdot 10^{-1}$	0.041 (0.007)	1.017 (0.008)	-0.004 (0.007)
Mothers age at death	27015805	aging	European	0.095	0.085	1.123	$2.62 \cdot 10^{-1}$	0.041 (0.007)	1.004 (0.008)	-0.011 (0.006)
Parents age at death	27015805	aging	European	-0.023	0.100	-0.235	$8.14 \cdot 10^{-1}$	0.031 (0.007)	1.013 (0.008)	0.000 (0.007)
Birth weight	27680694	anthropometric	European	-0.008	0.045	-0.183	$8.55 \cdot 10^{-1}$	0.100 (0.007)	1.047 (0.012)	0.008 (0.006)
Body fat	26833246	anthropometric	Mixed	0.027	0.060	0.445	$6.56 \cdot 10^{-1}$	0.111 (0.009)	0.898 (0.008)	0.001 (0.006)
Body mass index	20935630	anthropometric	European	0.078	0.043	1.817	$6.93 \cdot 10^{-2}$	0.192 (0.010)	1.006 (0.013)	0.001 (0.007)
Child birth length	25281659	anthropometric	European	0.136	0.089	1.533	$1.25 \cdot 10^{-1}$	0.161 (0.025)	0.995 (0.009)	-0.015 (0.008)
Child birth weight	23202124	anthropometric	European	0.059	0.080	0.733	$4.64 \cdot 10^{-1}$	0.108 (0.020)	1.006 (0.007)	-0.008 (0.005)
Childhood obesity	22484627	anthropometric	European	0.057	0.061	0.934	$3.50 \cdot 10^{-1}$	0.439 (0.047)	0.920 (0.009)	-0.004 (0.006)
Diff in height btwn adoloes and adulth; age 14	23449627	anthropometric	European	-0.143	0.105	-1.361	$1.74 \cdot 10^{-1}$	0.448 (0.117)	0.981 (0.008)	-0.000 (0.006)
Diff in height btwn childh and adulth; age 8	23449627	anthropometric	European	-0.027	0.095	-0.290	$7.72 \cdot 10^{-1}$	0.330 (0.060)	0.973 (0.010)	-0.000 (0.007)
Extreme bmi	23563607	anthropometric	European	0.071	0.060	1.193	$2.33 \cdot 10^{-1}$	0.687 (0.059)	1.031 (0.013)	0.004 (0.009)
Extreme height	23563607	anthropometric	European	0.017	0.046	0.365	$7.15 \cdot 10^{-1}$	1.266 (0.119)	1.024 (0.023)	0.004 (0.009)
Extreme waist-to-hip ratio	23563607	anthropometric	European	0.179	0.083	2.158	$3.10 \cdot 10^{-2}$	0.361 (0.066)	0.978 (0.009)	-0.018 (0.008)
Height2010	20881960	anthropometric	European	0.036	0.034	1.067	$2.86 \cdot 10^{-1}$	0.283 (0.018)	1.022 (0.022)	-0.010 (0.008)
Height; Females at age 10 and males at age 12	23449627	anthropometric	European	0.066	0.074	0.883	$3.77 \cdot 10^{-1}$	0.416 (0.051)	0.957 (0.010)	0.000 (0.007)
Hip circumference	25673412	anthropometric	European	0.091	0.041	2.236	$2.54 \cdot 10^{-2}$	0.130 (0.006)	0.852 (0.010)	0.002 (0.006)
Infant head circumference	22504419	anthropometric	European	0.086	0.091	0.936	$3.50 \cdot 10^{-1}$	0.216 (0.048)	0.994 (0.008)	-0.013 (0.006)
Obesity class 1	23563607	anthropometric	European	0.092	0.046	2.012	$4.42 \cdot 10^{-2}$	0.216 (0.012)	1.018 (0.013)	0.004 (0.008)
Obesity class 2	23563607	anthropometric	European	0.092	0.055	1.695	$9.01 \cdot 10^{-2}$	0.181 (0.014)	1.006 (0.011)	0.006 (0.008)
Obesity class 3	23563607	anthropometric	European	0.127	0.063	2.027	$4.27 \cdot 10^{-2}$	0.119 (0.015)	0.982 (0.011)	0.003 (0.007)
Overweight	23563607	anthropometric	European	0.084	0.049	1.720	$8.55 \cdot 10^{-2}$	0.110 (0.007)	1.022 (0.011)	-0.004 (0.007)
Sitting height ratio	25865494	anthropometric	European	0.088	0.078	1.130	$2.59 \cdot 10^{-1}$	0.220 (0.029)	0.982 (0.009)	-0.010 (0.007)
Waist circumference	25673412	anthropometric	European	0.078	0.040	1.963	$4.97 \cdot 10^{-2}$	0.123 (0.005)	0.839 (0.009)	0.001 (0.006)

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Supplementary Table 5 – continued from previous page

Trait	PMID	Category	Ethnicity	r_G	se	Z	P	h_G^2 obs (se)	h_G^2 Int (se)	GCOV Int (se)
Waist-to-hip ratio	25673412	anthropometric	European	0.068	0.041	1.659	$9.71 \cdot 10^{-2}$	0.115 (0.008)	0.915 (0.012)	-0.006 (0.007)
Asthma	17611496	autoimmune	European	0.054	0.093	0.573	$5.67 \cdot 10^{-1}$	0.125 (0.030)	1.007 (0.010)	0.004 (0.007)
Celiac disease	20190752	autoimmune	European	0.062	0.087	0.715	$4.75 \cdot 10^{-1}$	0.249 (0.051)	1.085 (0.012)	0.007 (0.008)
Crohns disease	26192919	autoimmune	European	-0.027	0.056	-0.491	$6.24 \cdot 10^{-1}$	0.484 (0.064)	1.029 (0.015)	0.001 (0.007)
Eczema	26482879	autoimmune	Mixed	-0.131	0.098	-1.333	$1.82 \cdot 10^{-1}$	0.065 (0.016)	1.026 (0.009)	0.023 (0.006)
Inflammatory Bowel Disease (Euro)	26192919	autoimmune	European	-0.029	0.053	-0.550	$5.82 \cdot 10^{-1}$	0.316 (0.037)	1.064 (0.015)	0.003 (0.007)
Multiple sclerosis	21833088	autoimmune	European	-0.030	0.156	-0.191	$8.49 \cdot 10^{-1}$	0.049 (0.029)	1.064 (0.011)	-0.004 (0.008)
Primary biliary cirrhosis	26394269	autoimmune	European	-0.100	0.074	-1.350	$1.77 \cdot 10^{-1}$	0.386 (0.068)	1.002 (0.011)	0.013 (0.007)
Rheumatoid Arthritis	24390342	autoimmune	European	-0.134	0.056	-2.382	$1.72 \cdot 10^{-2}$	0.153 (0.034)	1.033 (0.026)	0.010 (0.006)
Systemic lupus erythematosus	26502338	autoimmune	European	-0.168	0.094	-1.781	$7.49 \cdot 10^{-2}$	0.378 (0.075)	1.110 (0.013)	0.011 (0.009)
Ulcerative colitis	26192919	autoimmune	European	-0.010	0.065	-0.160	$8.73 \cdot 10^{-1}$	0.238 (0.033)	1.058 (0.013)	0.005 (0.006)
Femoral neck bone mineral density	22504420	bone	Mixed	0.016	0.047	0.328	$7.43 \cdot 10^{-1}$	0.300 (0.028)	0.983 (0.012)	0.001 (0.006)
Femoral Neck bone mineral density	26367794	bone	Mixed	-0.044	0.059	-0.747	$4.55 \cdot 10^{-1}$	0.124 (0.015)	0.978 (0.009)	0.004 (0.006)
Forearm Bone mineral density	26367794	bone	Mixed	0.060	0.147	0.405	$6.86 \cdot 10^{-1}$	0.072 (0.046)	1.016 (0.008)	-0.004 (0.006)
Lumbar spine bone mineral density	22504420	bone	Mixed	0.057	0.055	1.040	$2.98 \cdot 10^{-1}$	0.262 (0.026)	1.018 (0.011)	-0.005 (0.006)
Lumbar Spine bone mineral density	26367794	bone	Mixed	-0.016	0.063	-0.258	$7.96 \cdot 10^{-1}$	0.124 (0.018)	0.983 (0.010)	0.004 (0.006)
ICV**	25607358	brainvolume	European	0.137	0.095	1.450	$1.47 \cdot 10^{-1}$	0.175 (0.048)	1.018 (0.008)	-0.004 (0.005)
Mean Accumbens	25607358	brainvolume	European	-0.081	0.145	-0.557	$5.77 \cdot 10^{-1}$	0.096 (0.037)	0.977 (0.007)	0.000 (0.006)
Mean Caudate	25607358	brainvolume	European	-0.044	0.072	-0.612	$5.40 \cdot 10^{-1}$	0.251 (0.040)	0.967 (0.008)	0.006 (0.006)
Mean Hippocampus	25607358	brainvolume	European	-0.046	0.117	-0.389	$6.97 \cdot 10^{-1}$	0.133 (0.044)	0.990 (0.009)	-0.000 (0.007)
Mean Pallidum	25607358	brainvolume	European	-0.065	0.098	-0.665	$5.06 \cdot 10^{-1}$	0.165 (0.047)	0.978 (0.009)	-0.002 (0.006)
Mean Putamen**	25607358	brainvolume	European	0.004	0.081	0.053	$9.58 \cdot 10^{-1}$	0.303 (0.052)	1.000 (0.009)	0.001 (0.006)
Mean Thalamus	25607358	brainvolume	European	0.024	0.128	0.186	$8.53 \cdot 10^{-1}$	0.128 (0.039)	0.983 (0.008)	-0.008 (0.006)
Lung adenocarcinoma	27488534	cancer	European	-0.006	0.113	-0.049	$9.61 \cdot 10^{-1}$	0.031 (0.013)	1.019 (0.008)	-0.011 (0.005)
Lung cancer	27488534	cancer	European	-0.089	0.079	-1.120	$2.63 \cdot 10^{-1}$	0.315 (0.072)	1.015 (0.009)	-0.004 (0.006)
Lung cancer (all)	24880342	cancer	European	-0.061	0.088	-0.692	$4.89 \cdot 10^{-1}$	0.126 (0.033)	1.009 (0.009)	-0.004 (0.006)
Lung cancer (squamous cell)	24880342	cancer	European	0.129	0.145	0.888	$3.75 \cdot 10^{-1}$	0.036 (0.020)	1.016 (0.007)	-0.005 (0.006)
Squamous cell lung cancer	27488534	cancer	European	0.087	0.125	0.698	$4.85 \cdot 10^{-1}$	0.031 (0.012)	1.019 (0.008)	-0.004 (0.006)
Adiponectin	22479202	cardiometabolic	Mixed	-0.125	0.079	-1.575	$1.15 \cdot 10^{-1}$	0.122 (0.027)	1.020 (0.013)	0.006 (0.007)
Coronary artery disease	26343387	cardiometabolic	Mixed	-0.050	0.041	-1.215	$2.25 \cdot 10^{-1}$	0.077 (0.006)	1.052 (0.013)	0.001 (0.006)
IQ*	28530673	cognition	European	0.199	0.048	4.122	$3.76 \cdot 10^{-5}$	0.201 (0.011)	1.007 (0.010)	0.004 (0.007)
Memory*	27046643	cognition	European	-0.021	0.066	-0.316	$7.52 \cdot 10^{-1}$	0.044 (0.006)	0.997 (0.009)	0.004 (0.006)
Reaction time*	27046643	cognition	European	-0.028	0.060	-0.469	$6.39 \cdot 10^{-1}$	0.072 (0.006)	1.014 (0.009)	-0.005 (0.006)
Verbal-Numerical reasoning*	27046643	cognition	European	0.200	0.061	3.261	$1.10 \cdot 10^{-3}$	0.217 (0.020)	1.012 (0.009)	-0.002 (0.007)
Childhood IQ	23358156	education	European	0.234	0.073	3.223	$1.30 \cdot 10^{-3}$	0.274 (0.046)	1.002 (0.011)	-0.005 (0.006)
College*	27046643	education	European	0.274	0.041	6.649	$2.94 \cdot 10^{-11}$	0.155 (0.009)	1.033 (0.011)	-0.009 (0.007)
College completion	23722424	education	European	0.167	0.055	3.032	$2.40 \cdot 10^{-3}$	0.080 (0.006)	1.020 (0.010)	0.003 (0.007)
Educational Attainment	27225129	education	European	0.199	0.033	5.958	$2.56 \cdot 10^{-9}$	0.124 (0.004)	0.942 (0.013)	-0.001 (0.008)

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Supplementary Table 5 – continued from previous page

Trait	PMID	Category	Ethnicity	r_G	se	Z	P	h_G^2 obs (se)	h_G^2 Int (se)	GCOV Int (se)
Income*	27818178	education	SES	-0.034	0.059	-0.570	$5.69 \cdot 10^{-1}$	0.062 (0.006)	1.025 (0.009)	-0.007 (0.006)
2hr glucose adjusted for BMI	20081857	glycemic	European	0.016	0.111	0.143	$8.87 \cdot 10^{-1}$	0.101 (0.036)	0.993 (0.008)	-0.005 (0.006)
Fasting glucose main effect	22581228	glycemic	European	-0.023	0.063	-0.372	$7.10 \cdot 10^{-1}$	0.097 (0.021)	1.001 (0.013)	-0.001 (0.006)
Fasting insulin main effect	22581228	glycemic	European	0.017	0.072	0.241	$8.10 \cdot 10^{-1}$	0.075 (0.011)	1.009 (0.009)	-0.002 (0.006)
Fasting proinsulin	20081858	glycemic	European	-0.052	0.087	-0.598	$5.50 \cdot 10^{-1}$	0.222 (0.099)	0.973 (0.012)	0.002 (0.006)
HbA1C	20858683	glycemic	European	0.020	0.091	0.225	$8.22 \cdot 10^{-1}$	0.059 (0.013)	1.006 (0.009)	-0.008 (0.007)
HOMA-B	20081858	glycemic	European	0.027	0.108	0.250	$8.03 \cdot 10^{-1}$	0.080 (0.015)	0.996 (0.008)	0.000 (0.007)
HOMA-IR	20081858	glycemic	European	0.029	0.107	0.271	$7.87 \cdot 10^{-1}$	0.070 (0.014)	1.002 (0.007)	-0.002 (0.007)
Type 2 Diabetes	22885922	glycemic	European	0.008	0.070	0.111	$9.11 \cdot 10^{-1}$	0.087 (0.010)	1.010 (0.009)	0.005 (0.007)
Heart rate	23583979	haematological	Mixed	-0.057	0.055	-1.037	$3.00 \cdot 10^{-1}$	0.085 (0.009)	1.011 (0.010)	0.004 (0.006)
Mean platelet volume	22139419	haematological	European	-0.072	0.071	-1.006	$3.14 \cdot 10^{-1}$	0.321 (0.054)	0.980 (0.012)	0.002 (0.007)
Platelet count	22139419	haematological	European	-0.027	0.058	-0.463	$6.43 \cdot 10^{-1}$	0.114 (0.012)	0.994 (0.011)	-0.001 (0.006)
LeptinadjBMI	26833098	hormone	European	-0.010	0.084	-0.121	$9.04 \cdot 10^{-1}$	0.096 (0.019)	1.003 (0.009)	0.000 (0.006)
LeptinnotadjBMI	26833098	hormone	European	0.070	0.082	0.849	$3.96 \cdot 10^{-1}$	0.103 (0.016)	0.992 (0.008)	-0.002 (0.006)
Chronic Kidney Disease	26831199	kidney	Mixed	-0.050	0.106	-0.476	$6.34 \cdot 10^{-1}$	0.020 (0.006)	1.013 (0.011)	0.020 (0.007)
Serum creatinine	26831199	kidney	Mixed	-0.035	0.052	-0.665	$5.06 \cdot 10^{-1}$	0.107 (0.011)	0.975 (0.018)	-0.000 (0.008)
Serum creatinine (non-diab)	26831199	kidney	Mixed	-0.056	0.051	-1.094	$2.74 \cdot 10^{-1}$	0.115 (0.013)	0.977 (0.017)	-0.000 (0.007)
Serum cystatin c	26831199	kidney	Mixed	-0.030	0.058	-0.509	$6.11 \cdot 10^{-1}$	0.178 (0.074)	0.955 (0.018)	0.003 (0.006)
Urinary alb-to-cret ratio	26631737	kidney	European	-0.037	0.091	-0.402	$6.87 \cdot 10^{-1}$	0.045 (0.009)	0.998 (0.008)	0.003 (0.006)
Urinary alb-to-cret ratio (non-diab)	26631737	kidney	European	-0.131	0.096	-1.364	$1.73 \cdot 10^{-1}$	0.050 (0.011)	0.999 (0.008)	0.010 (0.007)
HDL cholest	20686565	lipids	European	-0.019	0.050	-0.377	$7.06 \cdot 10^{-1}$	0.117 (0.029)	1.080 (0.065)	-0.002 (0.006)
LDL cholest	20686565	lipids	European	-0.005	0.054	-0.082	$9.35 \cdot 10^{-1}$	0.091 (0.035)	1.078 (0.065)	-0.004 (0.006)
Tot Cholesterol	20686565	lipids	European	0.014	0.047	0.303	$7.62 \cdot 10^{-1}$	0.123 (0.029)	1.044 (0.048)	-0.004 (0.006)
Trigly	20686565	lipids	European	0.090	0.054	1.659	$9.72 \cdot 10^{-2}$	0.167 (0.032)	0.972 (0.022)	-0.005 (0.008)
Forced expiratory volume in 1 second (FEV1)	21946350	lungfunction	European	0.202	0.080	2.516	$1.19 \cdot 10^{-2}$	0.171 (0.023)	0.882 (0.013)	0.000 (0.010)
Forced expiratory volume in 1 second (FEV1)	26635082	lungfunction	European	0.010	0.066	0.147	$8.83 \cdot 10^{-1}$	0.144 (0.018)	0.989 (0.009)	-0.004 (0.006)
Forced expiratory volume in 1 second (FEV1)	28166213	lungfunction	European	0.086	0.054	1.599	$1.10 \cdot 10^{-1}$	0.269 (0.018)	0.967 (0.009)	-0.016 (0.008)
Forced expiratory volume in 1 second (FEV1)/Forced Vital capacity(FVC)	21946350	lungfunction	European	-0.037	0.060	-0.615	$5.39 \cdot 10^{-1}$	0.125 (0.015)	0.927 (0.009)	0.007 (0.006)
Forced expiratory volume in 1 second (FEV1)/Forced Vital capacity(FVC)	26635082	lungfunction	European	-0.005	0.082	-0.059	$9.53 \cdot 10^{-1}$	0.116 (0.017)	0.980 (0.008)	-0.003 (0.006)
Forced expiratory volume in 1 second (FEV1)/Forced Vital capacity(FVC)	28166213	lungfunction	European	0.079	0.060	1.330	$1.83 \cdot 10^{-1}$	0.254 (0.022)	0.979 (0.011)	-0.013 (0.007)
Forced Vital capacity(FVC)	26635082	lungfunction	European	0.021	0.066	0.313	$7.54 \cdot 10^{-1}$	0.149 (0.016)	0.984 (0.008)	-0.004 (0.006)
Forced Vital capacity(FVC)	28166213	lungfunction	European	0.078	0.050	1.548	$1.22 \cdot 10^{-1}$	0.269 (0.017)	0.968 (0.010)	-0.015 (0.007)
18:2 linoleic acid (LA)	27005778	metabolites	European	0.085	0.117	0.728	$4.67 \cdot 10^{-1}$	0.113 (0.055)	1.008 (0.012)	-0.005 (0.006)
22:6 docosahexaenoic acid	27005778	metabolites	European	-0.050	0.095	-0.533	$5.94 \cdot 10^{-1}$	0.123 (0.039)	0.998 (0.009)	0.002 (0.006)
Acetate	27005778	metabolites	European	0.383	0.147	2.610	$9.00 \cdot 10^{-3}$	0.047 (0.019)	1.007 (0.007)	-0.012 (0.006)
Acetoacetate	27005778	metabolites	European	0.145	0.115	1.266	$2.06 \cdot 10^{-1}$	0.085 (0.030)	0.975 (0.009)	-0.005 (0.006)
Alanine	27005778	metabolites	European	-0.008	0.085	-0.089	$9.29 \cdot 10^{-1}$	0.094 (0.030)	1.007 (0.009)	0.001 (0.006)
Albumin	27005778	metabolites	European	-0.031	0.140	-0.225	$8.22 \cdot 10^{-1}$	0.061 (0.027)	0.990 (0.008)	-0.001 (0.006)

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Supplementary Table 5 – continued from previous page

Trait	PMID	Category	Ethnicity	r_G	se	Z	P	h_G^2 obs (se)	h_G^2 Int (se)	GCOV Int (se)
Apolipoprotein A-I	27005778	metabolites	European	-0.035	0.111	-0.320	$7.49 \cdot 10^{-1}$	0.091 (0.035)	0.999 (0.015)	-0.001 (0.006)
Apolipoprotein B	27005778	metabolites	European	0.077	0.115	0.673	$5.01 \cdot 10^{-1}$	0.079 (0.049)	1.010 (0.021)	-0.007 (0.006)
Av no of dbl bonds in a fatty acid chain	27005778	metabolites	European	0.135	0.085	1.593	$1.11 \cdot 10^{-1}$	0.173 (0.052)	1.002 (0.010)	-0.011 (0.006)
Av no of meth grps per a dbl bond	27005778	metabolites	European	-0.081	0.080	-1.023	$3.06 \cdot 10^{-1}$	0.213 (0.073)	0.993 (0.011)	0.009 (0.006)
Cholesterol esters in large HDL	27005778	metabolites	European	-0.118	0.103	-1.140	$2.54 \cdot 10^{-1}$	0.103 (0.037)	1.010 (0.019)	0.006 (0.006)
Cholesterol esters in large LDL	27005778	metabolites	European	0.054	0.138	0.390	$6.96 \cdot 10^{-1}$	0.062 (0.068)	1.018 (0.031)	-0.008 (0.006)
Cholesterol esters in large VLDL	27005778	metabolites	European	0.061	0.084	0.731	$4.65 \cdot 10^{-1}$	0.166 (0.037)	0.978 (0.009)	0.003 (0.006)
Cholesterol esters in medium HDL	27005778	metabolites	European	-0.195	0.136	-1.432	$1.52 \cdot 10^{-1}$	0.051 (0.033)	1.003 (0.011)	0.002 (0.006)
Cholesterol esters in medium LDL	27005778	metabolites	European	0.081	0.129	0.622	$5.34 \cdot 10^{-1}$	0.069 (0.065)	1.015 (0.029)	-0.009 (0.006)
Cholesterol esters in medium VLDL	27005778	metabolites	European	0.105	0.083	1.271	$2.04 \cdot 10^{-1}$	0.144 (0.040)	0.989 (0.010)	-0.005 (0.006)
Citrate	27005778	metabolites	European	0.072	0.113	0.641	$5.22 \cdot 10^{-1}$	0.068 (0.022)	1.013 (0.008)	-0.013 (0.006)
Conc of chylom and lrgst VLDL prt	27005778	metabolites	European	-0.143	0.103	-1.381	$1.67 \cdot 10^{-1}$	0.121 (0.029)	0.987 (0.008)	0.006 (0.006)
Conc of IDL prt	27005778	metabolites	European	0.001	0.135	0.009	$9.93 \cdot 10^{-1}$	0.059 (0.054)	1.025 (0.025)	-0.006 (0.006)
Conc of large HDL prt	27005778	metabolites	European	-0.144	0.101	-1.430	$1.53 \cdot 10^{-1}$	0.109 (0.037)	1.010 (0.020)	0.006 (0.006)
Conc of large LDL prt	27005778	metabolites	European	0.032	0.135	0.235	$8.14 \cdot 10^{-1}$	0.061 (0.066)	1.022 (0.030)	-0.007 (0.006)
Conc of large VLDL prt	27005778	metabolites	European	0.093	0.086	1.091	$2.75 \cdot 10^{-1}$	0.137 (0.037)	0.972 (0.009)	-0.000 (0.006)
Conc of medium HDL prt	27005778	metabolites	European	-0.182	0.115	-1.585	$1.13 \cdot 10^{-1}$	0.067 (0.031)	0.995 (0.009)	0.002 (0.006)
Conc of medium LDL prt	27005778	metabolites	European	0.069	0.125	0.548	$5.84 \cdot 10^{-1}$	0.070 (0.063)	1.016 (0.028)	-0.009 (0.006)
Conc of medium VLDL prt	27005778	metabolites	European	0.084	0.087	0.960	$3.37 \cdot 10^{-1}$	0.144 (0.038)	0.984 (0.010)	0.001 (0.006)
Conc of small LDL prt	27005778	metabolites	European	0.067	0.105	0.640	$5.22 \cdot 10^{-1}$	0.094 (0.053)	1.007 (0.022)	-0.008 (0.006)
Conc of small VLDL prt	27005778	metabolites	European	0.116	0.080	1.446	$1.48 \cdot 10^{-1}$	0.151 (0.039)	0.990 (0.010)	-0.005 (0.006)
Conc of very large HDL prt	27005778	metabolites	European	-0.156	0.148	-1.057	$2.91 \cdot 10^{-1}$	0.049 (0.033)	1.009 (0.019)	0.008 (0.006)
Conc of very large VLDL prt	27005778	metabolites	European	0.044	0.104	0.420	$6.74 \cdot 10^{-1}$	0.135 (0.033)	0.980 (0.009)	-0.001 (0.006)
Conc of very small VLDL prt	27005778	metabolites	European	0.039	0.094	0.412	$6.80 \cdot 10^{-1}$	0.104 (0.041)	1.010 (0.016)	-0.005 (0.006)
Creatinine	27005778	metabolites	European	0.164	0.089	1.843	$6.54 \cdot 10^{-2}$	0.112 (0.028)	1.013 (0.009)	-0.011 (0.006)
Desc of av fatty acid chain length;	27005778	metabolites	European	0.024	0.101	0.235	$8.15 \cdot 10^{-1}$	0.120 (0.039)	0.977 (0.009)	-0.000 (0.006)
Free cholest	27005778	metabolites	European	-0.061	0.159	-0.387	$6.99 \cdot 10^{-1}$	0.071 (0.049)	1.022 (0.013)	-0.006 (0.006)
Free cholest in IDL	27005778	metabolites	European	0.016	0.137	0.119	$9.06 \cdot 10^{-1}$	0.053 (0.054)	1.029 (0.028)	-0.006 (0.006)
Free cholest in large HDL	27005778	metabolites	European	-0.125	0.104	-1.211	$2.26 \cdot 10^{-1}$	0.088 (0.033)	1.017 (0.019)	0.005 (0.006)
Free cholest in large LDL	27005778	metabolites	European	-0.010	0.143	-0.070	$9.44 \cdot 10^{-1}$	0.049 (0.065)	1.024 (0.034)	-0.006 (0.006)
Free cholest in large VLDL	27005778	metabolites	European	0.077	0.085	0.900	$3.68 \cdot 10^{-1}$	0.129 (0.032)	0.987 (0.009)	0.000 (0.006)
Free cholest in medium HDL	27005778	metabolites	European	-0.208	0.109	-1.916	$5.54 \cdot 10^{-2}$	0.067 (0.027)	0.999 (0.011)	0.002 (0.006)
Free cholest in medium VLDL	27005778	metabolites	European	0.089	0.092	0.971	$3.32 \cdot 10^{-1}$	0.108 (0.035)	0.998 (0.009)	-0.001 (0.006)
Free cholest in small VLDL	27005778	metabolites	European	0.165	0.097	1.709	$8.75 \cdot 10^{-2}$	0.102 (0.036)	1.004 (0.011)	-0.008 (0.006)
Free cholest in very large HDL	27005778	metabolites	European	-0.162	0.166	-0.974	$3.30 \cdot 10^{-1}$	0.035 (0.029)	1.022 (0.019)	0.007 (0.006)
Free cholest to esterified cholest ratio	27005778	metabolites	European	0.004	0.185	0.024	$9.81 \cdot 10^{-1}$	0.051 (0.060)	1.016 (0.016)	-0.007 (0.006)
Glucose	27005778	metabolites	European	0.043	0.100	0.432	$6.66 \cdot 10^{-1}$	0.086 (0.024)	0.995 (0.008)	-0.008 (0.006)
Glutamine	27005778	metabolites	European	0.108	0.124	0.870	$3.84 \cdot 10^{-1}$	0.057 (0.023)	1.020 (0.010)	-0.009 (0.006)

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Supplementary Table 5 – continued from previous page

Trait	PMID	Category	Ethnicity	r_G	se	Z	P	h_G^2 obs (se)	h_G^2 Int (se)	GCOV Int (se)
Glycop acetyls; mainly a1-acid glycop	27005778	metabolites	European	0.029	0.099	0.296	$7.67 \cdot 10^{-1}$	0.107 (0.030)	0.983 (0.008)	-0.001 (0.006)
Isoleucine	27005778	metabolites	European	0.089	0.108	0.822	$4.11 \cdot 10^{-1}$	0.065 (0.025)	0.998 (0.008)	-0.003 (0.006)
Leucine	27005778	metabolites	European	0.127	0.133	0.959	$3.38 \cdot 10^{-1}$	0.047 (0.023)	1.008 (0.008)	-0.003 (0.006)
Mean diameter for HDL prt	27005778	metabolites	European	-0.116	0.117	-0.994	$3.20 \cdot 10^{-1}$	0.088 (0.041)	1.022 (0.026)	0.007 (0.006)
Mean diameter for LDL prt	27005778	metabolites	European	-0.020	0.132	-0.154	$8.78 \cdot 10^{-1}$	0.048 (0.031)	1.001 (0.015)	-0.001 (0.005)
Mean diameter for VLDL prt	27005778	metabolites	European	0.119	0.092	1.286	$1.99 \cdot 10^{-1}$	0.121 (0.039)	0.997 (0.011)	0.002 (0.006)
Mono-unsaturated fatty acids	27005778	metabolites	European	-0.116	0.111	-1.044	$2.97 \cdot 10^{-1}$	0.105 (0.045)	0.988 (0.008)	0.003 (0.006)
Omega-3 fatty acids	27005778	metabolites	European	-0.037	0.094	-0.390	$6.97 \cdot 10^{-1}$	0.136 (0.043)	0.996 (0.009)	0.003 (0.006)
Omega-9 and saturated fatty acids	27005778	metabolites	European	-0.130	0.131	-0.997	$3.19 \cdot 10^{-1}$	0.082 (0.045)	0.998 (0.009)	0.003 (0.006)
Phenylalanine	27005778	metabolites	European	0.007	0.119	0.063	$9.50 \cdot 10^{-1}$	0.057 (0.024)	1.004 (0.008)	-0.002 (0.006)
PhosLip in chylom and lrgst VLDL prt	27005778	metabolites	European	-0.024	0.094	-0.256	$7.98 \cdot 10^{-1}$	0.119 (0.028)	0.975 (0.008)	0.003 (0.006)
PhosLip in IDL	27005778	metabolites	European	0.021	0.149	0.140	$8.88 \cdot 10^{-1}$	0.043 (0.052)	1.029 (0.027)	-0.006 (0.006)
PhosLip in large HDL	27005778	metabolites	European	-0.151	0.102	-1.490	$1.36 \cdot 10^{-1}$	0.106 (0.036)	1.008 (0.019)	0.005 (0.006)
PhosLip in large LDL	27005778	metabolites	European	-0.006	0.141	-0.039	$9.69 \cdot 10^{-1}$	0.048 (0.059)	1.024 (0.030)	-0.007 (0.006)
PhosLip in large VLDL	27005778	metabolites	European	0.024	0.094	0.258	$7.96 \cdot 10^{-1}$	0.116 (0.033)	0.990 (0.009)	0.004 (0.006)
PhosLip in medium HDL	27005778	metabolites	European	-0.202	0.109	-1.851	$6.42 \cdot 10^{-2}$	0.067 (0.028)	0.996 (0.010)	0.002 (0.006)
PhosLip in medium LDL	27005778	metabolites	European	0.079	0.120	0.662	$5.08 \cdot 10^{-1}$	0.067 (0.055)	1.012 (0.027)	-0.010 (0.006)
PhosLip in medium VLDL	27005778	metabolites	European	0.079	0.092	0.862	$3.89 \cdot 10^{-1}$	0.106 (0.035)	0.999 (0.009)	-0.001 (0.006)
PhosLip in small VLDL	27005778	metabolites	European	0.126	0.092	1.377	$1.69 \cdot 10^{-1}$	0.106 (0.037)	1.003 (0.011)	-0.006 (0.006)
PhosLip in very large HDL	27005778	metabolites	European	-0.045	0.121	-0.370	$7.12 \cdot 10^{-1}$	0.068 (0.038)	1.018 (0.023)	0.004 (0.006)
PhosLip in very large VLDL	27005778	metabolites	European	0.018	0.094	0.197	$8.44 \cdot 10^{-1}$	0.117 (0.030)	0.982 (0.008)	0.003 (0.006)
PhosLip in very small VLDL	27005778	metabolites	European	0.017	0.119	0.143	$8.86 \cdot 10^{-1}$	0.067 (0.044)	1.022 (0.020)	-0.006 (0.006)
Ratio of bisal grps to dbl bonds	27005778	metabolites	European	-0.013	0.064	-0.211	$8.33 \cdot 10^{-1}$	0.306 (0.102)	0.975 (0.012)	0.000 (0.006)
Ratio of bisal grps to tot fatty acids	27005778	metabolites	European	0.039	0.070	0.560	$5.75 \cdot 10^{-1}$	0.287 (0.091)	0.983 (0.011)	-0.006 (0.006)
Serum tot cholest	27005778	metabolites	European	0.065	0.131	0.497	$6.19 \cdot 10^{-1}$	0.059 (0.047)	1.024 (0.021)	-0.008 (0.006)
Serum tot triglycerides	27005778	metabolites	European	0.063	0.079	0.798	$4.25 \cdot 10^{-1}$	0.133 (0.037)	0.989 (0.010)	-0.001 (0.006)
Tot cholest in HDL	27005778	metabolites	European	-0.158	0.105	-1.507	$1.32 \cdot 10^{-1}$	0.085 (0.033)	1.012 (0.017)	0.004 (0.006)
Tot cholest in IDL	27005778	metabolites	European	0.003	0.148	0.017	$9.87 \cdot 10^{-1}$	0.052 (0.060)	1.027 (0.028)	-0.006 (0.006)
Tot cholest in large HDL	27005778	metabolites	European	-0.158	0.106	-1.488	$1.37 \cdot 10^{-1}$	0.085 (0.034)	1.016 (0.020)	0.007 (0.006)
Tot cholest in large LDL	27005778	metabolites	European	0.025	0.139	0.178	$8.59 \cdot 10^{-1}$	0.051 (0.063)	1.022 (0.032)	-0.007 (0.006)
Tot cholest in large VLDL	27005778	metabolites	European	0.073	0.086	0.846	$3.98 \cdot 10^{-1}$	0.121 (0.032)	0.990 (0.008)	0.001 (0.006)
Tot cholest in LDL	27005778	metabolites	European	0.085	0.133	0.640	$5.22 \cdot 10^{-1}$	0.059 (0.068)	1.019 (0.034)	-0.009 (0.006)
Tot cholest in medium HDL	27005778	metabolites	European	-0.222	0.125	-1.785	$7.42 \cdot 10^{-2}$	0.051 (0.029)	1.001 (0.011)	0.003 (0.006)
Tot cholest in medium LDL	27005778	metabolites	European	0.084	0.131	0.638	$5.23 \cdot 10^{-1}$	0.059 (0.063)	1.018 (0.032)	-0.010 (0.006)
Tot cholest in medium VLDL	27005778	metabolites	European	0.103	0.088	1.178	$2.39 \cdot 10^{-1}$	0.117 (0.037)	0.996 (0.010)	-0.004 (0.006)
Tot cholest in small LDL	27005778	metabolites	European	0.135	0.136	0.990	$3.22 \cdot 10^{-1}$	0.066 (0.056)	1.018 (0.027)	-0.009 (0.006)
Tot cholest in small VLDL	27005778	metabolites	European	0.186	0.118	1.584	$1.13 \cdot 10^{-1}$	0.072 (0.036)	1.014 (0.013)	-0.010 (0.006)
Tot cholest in very large HDL	27005778	metabolites	European	-0.195	0.200	-0.977	$3.29 \cdot 10^{-1}$	0.028 (0.028)	1.017 (0.016)	0.007 (0.006)

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Supplementary Table 5 – continued from previous page

Trait	PMID	Category	Ethnicity	r_G	se	Z	P	h_G^2 obs (se)	h_G^2 Int (se)	GCOV Int (se)
Tot lipids in chylom and lrgst VLDL prt	27005778	metabolites	European	-0.027	0.093	-0.292	$7.71 \cdot 10^{-1}$	0.141 (0.029)	0.982 (0.008)	0.003 (0.006)
Tot lipids in IDL	27005778	metabolites	European	0.002	0.140	0.014	$9.89 \cdot 10^{-1}$	0.056 (0.057)	1.027 (0.026)	-0.006 (0.006)
Tot lipids in large HDL	27005778	metabolites	European	-0.137	0.101	-1.353	$1.76 \cdot 10^{-1}$	0.108 (0.037)	1.011 (0.020)	0.006 (0.006)
Tot lipids in large LDL	27005778	metabolites	European	0.038	0.138	0.279	$7.81 \cdot 10^{-1}$	0.061 (0.068)	1.021 (0.031)	-0.008 (0.006)
Tot lipids in large VLDL	27005778	metabolites	European	0.042	0.094	0.449	$6.54 \cdot 10^{-1}$	0.141 (0.033)	0.985 (0.009)	0.003 (0.007)
Tot lipids in medium HDL	27005778	metabolites	European	-0.197	0.121	-1.633	$1.03 \cdot 10^{-1}$	0.062 (0.031)	0.997 (0.010)	0.002 (0.006)
Tot lipids in medium LDL	27005778	metabolites	European	0.066	0.126	0.520	$6.03 \cdot 10^{-1}$	0.071 (0.065)	1.014 (0.029)	-0.008 (0.006)
Tot lipids in medium VLDL	27005778	metabolites	European	0.083	0.086	0.973	$3.30 \cdot 10^{-1}$	0.141 (0.037)	0.986 (0.009)	-0.001 (0.006)
Tot lipids in small HDL	27005778	metabolites	European	-0.020	0.133	-0.149	$8.82 \cdot 10^{-1}$	0.051 (0.030)	1.004 (0.009)	-0.006 (0.006)
Tot lipids in small LDL	27005778	metabolites	European	0.078	0.116	0.671	$5.02 \cdot 10^{-1}$	0.081 (0.057)	1.010 (0.025)	-0.008 (0.006)
Tot lipids in small VLDL	27005778	metabolites	European	0.121	0.084	1.441	$1.50 \cdot 10^{-1}$	0.142 (0.039)	0.993 (0.010)	-0.006 (0.006)
Tot lipids in very large HDL	27005778	metabolites	European	-0.066	0.149	-0.443	$6.58 \cdot 10^{-1}$	0.044 (0.035)	1.024 (0.021)	0.004 (0.006)
Tot lipids in very large VLDL	27005778	metabolites	European	0.037	0.090	0.408	$6.83 \cdot 10^{-1}$	0.152 (0.032)	0.973 (0.008)	0.000 (0.006)
Tot lipids in very small VLDL	27005778	metabolites	European	0.031	0.101	0.309	$7.57 \cdot 10^{-1}$	0.092 (0.041)	1.016 (0.016)	-0.005 (0.006)
Trigly in chylom and lrgst VLDL prt	27005778	metabolites	European	-0.039	0.091	-0.425	$6.71 \cdot 10^{-1}$	0.107 (0.029)	0.984 (0.009)	0.006 (0.006)
Trigly in IDL	27005778	metabolites	European	0.027	0.096	0.280	$7.80 \cdot 10^{-1}$	0.098 (0.041)	1.013 (0.019)	-0.005 (0.006)
Trigly in large VLDL	27005778	metabolites	European	0.044	0.093	0.476	$6.34 \cdot 10^{-1}$	0.116 (0.031)	0.987 (0.009)	0.004 (0.006)
Trigly in medium VLDL	27005778	metabolites	European	0.079	0.100	0.789	$4.30 \cdot 10^{-1}$	0.094 (0.032)	1.000 (0.009)	0.002 (0.006)
Trigly in small HDL	27005778	metabolites	European	0.157	0.114	1.372	$1.70 \cdot 10^{-1}$	0.059 (0.029)	1.002 (0.010)	-0.007 (0.005)
Trigly in small VLDL	27005778	metabolites	European	0.125	0.086	1.458	$1.45 \cdot 10^{-1}$	0.124 (0.037)	0.993 (0.010)	-0.004 (0.006)
Trigly in very large HDL	27005778	metabolites	European	-0.003	0.113	-0.027	$9.78 \cdot 10^{-1}$	0.066 (0.040)	1.023 (0.037)	0.001 (0.006)
Trigly in very large VLDL	27005778	metabolites	European	0.040	0.085	0.472	$6.37 \cdot 10^{-1}$	0.136 (0.030)	0.974 (0.008)	0.001 (0.006)
Trigly in very small VLDL	27005778	metabolites	European	0.085	0.079	1.069	$2.85 \cdot 10^{-1}$	0.143 (0.039)	0.996 (0.012)	-0.005 (0.006)
Tyrosine	27005778	metabolites	European	-0.072	0.107	-0.678	$4.98 \cdot 10^{-1}$	0.067 (0.029)	1.004 (0.008)	0.004 (0.006)
Valine	27005778	metabolites	European	0.081	0.116	0.699	$4.84 \cdot 10^{-1}$	0.062 (0.022)	1.014 (0.008)	-0.001 (0.006)
Ferritin	25352340	metal	European	0.008	0.101	0.074	$9.41 \cdot 10^{-1}$	0.086 (0.030)	1.030 (0.011)	-0.003 (0.007)
Transferrin	25352340	metal	European	0.039	0.085	0.451	$6.52 \cdot 10^{-1}$	0.162 (0.079)	1.062 (0.027)	-0.001 (0.007)
Alzheimers disease	24162737	neurological	European	0.055	0.104	0.525	$6.00 \cdot 10^{-1}$	0.039 (0.029)	1.074 (0.042)	-0.002 (0.006)
Amyotrophic lateral sclerosis	27455348	neurological	European	-0.005	0.125	-0.041	$9.67 \cdot 10^{-1}$	0.051 (0.014)	0.993 (0.008)	-0.003 (0.006)
Parkinsons disease	19915575	neurological	European	0.103	0.071	1.463	$1.44 \cdot 10^{-1}$	0.341 (0.127)	1.130 (0.011)	-0.008 (0.005)
Male pat baldness*	28196072	other	European	-0.133	0.043	-3.106	$1.90 \cdot 10^{-3}$	0.298 (0.049)	1.038 (0.028)	0.017 (0.007)
SCDC*	28044064	other	European	0.375	0.126	2.986	$2.80 \cdot 10^{-3}$	0.304 (0.096)	0.986 (0.008)	-0.010 (0.006)
Self-rated health*	27864402	other	European	0.081	0.059	1.386	$1.66 \cdot 10^{-1}$	0.095 (0.006)	1.018 (0.009)	0.010 (0.007)
Self-reported tiredness*	28194004	other	European	0.331	0.055	5.992	$2.07 \cdot 10^{-9}$	0.068 (0.006)	0.994 (0.008)	-0.007 (0.006)
Urate	23263486	other	European	0.012	0.042	0.284	$7.77 \cdot 10^{-1}$	0.186 (0.068)	0.934 (0.070)	0.002 (0.007)
Neo-conscientiousness	21173776	personality	European	-0.173	0.152	-1.142	$2.54 \cdot 10^{-1}$	0.072 (0.032)	1.002 (0.009)	-0.009 (0.007)
Neo-openness to experience	21173776	personality	European	0.339	0.105	3.246	$1.20 \cdot 10^{-3}$	0.123 (0.029)	0.985 (0.008)	0.002 (0.006)
Neuroticism	24828478	personality	European	0.217	0.116	1.879	$6.03 \cdot 10^{-2}$	0.012 (0.004)	1.018 (0.008)	-0.006 (0.007)

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Supplementary Table 5 – continued from previous page

Trait	PMID	Category	Ethnicity	r_G	se	Z	P	h_G^2 obs (se)	h_G^2 Int (se)	GCOV Int (se)
Neuroticism	27089181	personality	European	0.270	0.067	4.065	$4.81 \cdot 10^{-5}$	0.090 (0.009)	0.987 (0.017)	-0.012 (0.011)
ADHD*	30478444	psychiatric	Mixed	0.360	0.051	7.101	$1.24 \cdot 10^{-12}$	0.229 (0.015)	1.034 (0.010)	0.351 (0.008)
Anorexia Nervosa	24514567	psychiatric	European	0.059	0.064	0.923	$3.56 \cdot 10^{-1}$	0.388 (0.033)	0.960 (0.009)	0.008 (0.006)
Bipolar disorder	21926972	psychiatric	European	0.110	0.066	1.677	$9.36 \cdot 10^{-2}$	0.396 (0.041)	1.038 (0.009)	0.004 (0.007)
Depressive symptoms	27089181	psychiatric	European	0.320	0.063	5.106	$3.28 \cdot 10^{-7}$	0.046 (0.004)	1.007 (0.009)	-0.001 (0.007)
Major depression*	29700475	psychiatric	European	0.412	0.039	10.454	$1.40 \cdot 10^{-25}$	0.056 (0.003)	1.006 (0.012)	-0.000 (0.007)
Schizophrenia	25056061	psychiatric	Mixed	0.211	0.048	4.410	$1.03 \cdot 10^{-5}$	0.424 (0.020)	1.110 (0.016)	0.018 (0.009)
Subjective well being	27089181	psychiatric	European	-0.491	0.055	-8.876	$6.93 \cdot 10^{-19}$	0.025 (0.002)	1.002 (0.009)	0.009 (0.007)
Age at Menarche	25231870	reproductive	European	-0.108	0.036	-3.029	$2.50 \cdot 10^{-3}$	0.211 (0.011)	0.931 (0.015)	-0.002 (0.007)
Age at Menopause	26414677	reproductive	European	-0.014	0.054	-0.263	$7.93 \cdot 10^{-1}$	0.135 (0.017)	0.992 (0.017)	0.005 (0.007)
Age of first birth	27798627	reproductive	European	0.005	0.051	0.104	$9.17 \cdot 10^{-1}$	0.062 (0.004)	0.957 (0.011)	-0.003 (0.007)
Number of children ever born	27798627	reproductive	European	-0.024	0.051	-0.464	$6.42 \cdot 10^{-1}$	0.026 (0.002)	0.968 (0.008)	-0.009 (0.006)
Social deprivation*	27864402	SES	European	0.224	0.075	2.985	$2.80 \cdot 10^{-3}$	0.038 (0.005)	1.012 (0.009)	0.004 (0.006)
Chronotype	27494321	sleeping	European	-0.209	0.047	-4.420	$9.88 \cdot 10^{-6}$	0.103 (0.006)	1.011 (0.010)	0.009 (0.006)
Sleep duration	27494321	sleeping	European	-0.080	0.077	-1.043	$2.97 \cdot 10^{-1}$	0.053 (0.006)	1.027 (0.010)	0.006 (0.007)
Age of smoking initiation	20418890	smokingbehaviour	European	-0.127	0.135	-0.943	$3.46 \cdot 10^{-1}$	0.060 (0.020)	1.000 (0.008)	0.007 (0.007)
Cigarettes smoked per day	20418890	smokingbehaviour	European	0.080	0.104	0.762	$4.46 \cdot 10^{-1}$	0.055 (0.017)	1.009 (0.008)	-0.003 (0.006)
Ever vs never smoked	20418890	smokingbehaviour	European	0.063	0.062	1.012	$3.12 \cdot 10^{-1}$	0.068 (0.007)	1.008 (0.007)	-0.009 (0.005)
Former vs Current smoker	20418890	smokingbehaviour	European	0.046	0.112	0.409	$6.83 \cdot 10^{-1}$	0.054 (0.012)	1.009 (0.008)	0.006 (0.006)
Serumurate overweight	25811787	uricacid	European	-0.082	0.068	-1.194	$2.33 \cdot 10^{-1}$	0.535 (0.348)	0.969 (0.037)	0.005 (0.006)

3.2.3 MTAG analysis and tophits in related phenotypes

Supplementary Table 6: Top hits of MTAG analyses: An extended version of Table 1b of the main manuscript including also the effect size and p-value for the contributing secondary phenotypes as well as the original GWAS signal: Loci from the three MTAG analyses with schizophrenia (SCZ)[8], educational attainment (Edu)[50] and major depression (MD)[48] — loci not already represented in the ASD scan. Independent loci ($r^2 < 0.1$, distance $> 400\text{kb}$) with index variant (Index var), chromosome (CHR), chromosomal position (BP), MTAG estimate of effect (β) with respect to A1, standard error of β (SE), the association p-value of the index variant (P), and max FDR (mFDR), alleles (A1/A2), allele frequency of A1 (FRQ). The “Analysis” column shows the analysis that generated the particular MTAG result. The corresponding P-value (P) and effect size (β) from the ASD scan (logistic regression and inverse variance weighted meta-analysis) and the secondary phenotype (logistic regression and inverse variance weighted meta-analysis except for educational attainment which linear regression) is provided together with the weight assigned to the secondary phenotype by MTAG (Weight). All p-values are derived from z-scores. The column “Nearest genes” lists nearest genes from within 50kb of the $r^2 \geq 0.6$ LD friends of the index variant. The loci also found in the 88-loci follow-up sample (Table 3) are marked with *. They all show same direction of effect. Figures 49–55 show regional plot for the 7 loci.

Index var	CHR	BP	MTAG				A1/A2	FRQ	Analysis	ASD scan		Secondary phenotype			Nearest genes
			P	β	SE	mFDR				P	β	P	β	Weight	
rs2388334*	6	98,591,622	$3.34 \cdot 10^{-12}$	-0.065	0.009	0.021	A/G	0.517	ASD-Edu	$1.00 \cdot 10^{-6}$	-0.068	$6.56 \cdot 10^{-21}$	-0.023	0.108	MMS22L, POU3F2
rs325506*	5	104,012,303	$3.26 \cdot 10^{-11}$	0.057	0.009	0.012	C/G	0.423	ASD-MD	$3.50 \cdot 10^{-7}$	0.071	$2.17 \cdot 10^{-7}$	0.026	0.244	NUD12
rs11787216*	8	142,615,222	$2.00 \cdot 10^{-9}$	-0.058	0.010	0.021	T/C	0.364	ASD-Edu	$2.59 \cdot 10^{-6}$	-0.030	$6.12 \cdot 10^{-10}$	-0.016	0.108	MROH5
rs1452075*	3	62,481,063	$3.17 \cdot 10^{-9}$	0.061	0.010	0.021	T/C	0.721	ASD-Edu	$2.07 \cdot 10^{-7}$	0.035	$7.44 \cdot 10^{-5}$	0.011	0.108	CADPS
rs1620977	1	72,729,142	$6.66 \cdot 10^{-9}$	0.056	0.010	0.012	A/G	0.260	ASD-MD	$1.19 \cdot 10^{-4}$	0.062	$7.64 \cdot 10^{-9}$	0.032	0.244	NEGR1
rs10149470	14	104,017,953	$8.52 \cdot 10^{-9}$	-0.049	0.008	0.012	A/G	0.487	ASD-MD	$8.49 \cdot 10^{-5}$	-0.056	$3.57 \cdot 10^{-8}$	-0.028	0.244	MARK3, CKB, TRMT61A, BAG5, APOPT1, KLC1, XRCC3
rs16854048	4	42,123,728	$1.29 \cdot 10^{-8}$	0.069	0.012	0.012	A/C	0.858	ASD-MD	$5.87 \cdot 10^{-5}$	0.082	$2.44 \cdot 10^{-7}$	0.038	0.244	SLC30A9, BEND4, TMEM33, DCAF4L1

Supplementary Table 7: Look-up in the ASD scan of the top major depression loci[48] shown together with the results from the corresponding MTAG analysis. The 40 loci represented in the ASD summary statistics are shown. The columns are chromosome (CHR), marker name in the major depression scan (SNP), chromosomal position (BP), logistic regression effect size (β), standard error of effect (SE), p-value (P) in major depression, ASD and the ASD-major-depression MTAG analysis, allele names (A1A2), minor allele frequency in ASD cases (FRQ A) and controls (FRQ U). The results are presented in order of the p-values in the major depression scan, and statistically significant MTAG results ($P < 1.667 \cdot 10^{-8}$) are presented in **red**. The effect measures of ASD and major depression were estimated using logistic regression and inverse variance weighted meta-analysis, and all p-values were estimated from the z-scores.

CHR	SNP	BP	Major Depression			ASD			ASD-MD MTAG			A1/A2	FRQ A	FRQ U
			β	SE	P	β	SE	P	β	SE	P			
13	rs12552	53,625,781	0.039	0.005	$6.10 \cdot 10^{-19}$	0.032	0.014	$2.18 \cdot 10^{-2}$	0.047	0.009	$3.72 \cdot 10^{-8}$	A/G	0.434	0.424
1	rs1432639	72,813,218	0.039	0.005	$4.60 \cdot 10^{-15}$	0.036	0.014	$1.04 \cdot 10^{-2}$	0.045	0.009	$1.32 \cdot 10^{-7}$	A/C	0.611	0.598
15	rs8025231	37,648,402	-0.030	0.005	$2.40 \cdot 10^{-12}$	0.014	0.014	$3.15 \cdot 10^{-1}$	-0.017	0.009	$4.78 \cdot 10^{-2}$	A/C	0.560	0.556
1	rs12129573	73,768,366	0.039	0.005	$4.00 \cdot 10^{-12}$	0.026	0.014	$7.23 \cdot 10^{-2}$	0.037	0.009	$2.41 \cdot 10^{-5}$	A/C	0.371	0.364
18	rs12958048	53,101,598	0.030	0.005	$3.60 \cdot 10^{-11}$	-0.004	0.015	$7.78 \cdot 10^{-1}$	0.021	0.009	$1.82 \cdot 10^{-2}$	A/G	0.327	0.328
5	chr5:87992715.1	87,992,715	-0.030	0.005	$7.90 \cdot 10^{-11}$	-0.003	0.014	$8.31 \cdot 10^{-1}$				T/TA	0.407	0.405
10	rs61867293	106,563,924	-0.041	0.006	$7.00 \cdot 10^{-10}$	-0.051	0.017	$2.86 \cdot 10^{-3}$	-0.050	0.010	$1.24 \cdot 10^{-6}$	T/C	0.207	0.216
14	rs915057	64,686,207	-0.030	0.005	$7.60 \cdot 10^{-10}$	-0.026	0.014	$6.74 \cdot 10^{-2}$	-0.033	0.009	$9.39 \cdot 10^{-5}$	A/G	0.430	0.439
5	rs11135349	164,523,472	-0.030	0.005	$1.10 \cdot 10^{-9}$	0.013	0.014	$3.69 \cdot 10^{-1}$	-0.014	0.009	$9.49 \cdot 10^{-2}$	A/C	0.441	0.432
11	rs1806153	31,850,105	0.039	0.006	$1.20 \cdot 10^{-9}$	0.058	0.017	$6.59 \cdot 10^{-4}$	0.057	0.010	$4.14 \cdot 10^{-8}$	T/G	0.222	0.213
14	rs4904738	42,179,732	-0.030	0.005	$2.60 \cdot 10^{-9}$	-0.013	0.014	$3.46 \cdot 10^{-1}$	-0.026	0.009	$2.13 \cdot 10^{-3}$	T/C	0.561	0.566
3	rs7430565	158,107,180	-0.030	0.005	$2.90 \cdot 10^{-9}$	-0.025	0.014	$7.67 \cdot 10^{-2}$	-0.034	0.009	$7.51 \cdot 10^{-5}$	A/G	0.566	0.570
14	rs10149470	104,017,953	-0.030	0.005	$3.10 \cdot 10^{-9}$	-0.056	0.014	$8.49 \cdot 10^{-5}$	-0.049	0.008	$8.52 \cdot 10^{-9}$	A/G	0.476	0.487
4	rs34215985	42,047,778	-0.041	0.006	$3.10 \cdot 10^{-9}$	-0.057	0.017	$9.48 \cdot 10^{-4}$	-0.055	0.010	$1.59 \cdot 10^{-7}$	C/G	0.202	0.211
2	rs11682175	57,987,593	-0.030	0.005	$4.70 \cdot 10^{-9}$	-0.024	0.014	$8.48 \cdot 10^{-2}$	-0.033	0.008	$1.01 \cdot 10^{-4}$	T/C	0.529	0.536
9	rs10959913	11,544,964	0.030	0.006	$5.10 \cdot 10^{-9}$	-0.031	0.016	$5.80 \cdot 10^{-2}$	0.007	0.010	$4.69 \cdot 10^{-1}$	T/G	0.746	0.751
5	rs4869056	166,992,078	-0.030	0.005	$6.80 \cdot 10^{-9}$	-0.025	0.014	$7.84 \cdot 10^{-2}$	-0.032	0.009	$2.51 \cdot 10^{-4}$	A/G	0.629	0.635
16	rs8063603	6,310,645	-0.030	0.005	$6.90 \cdot 10^{-9}$	-0.042	0.015	$5.62 \cdot 10^{-3}$	-0.041	0.009	$6.97 \cdot 10^{-6}$	A/G	0.662	0.673
5	rs116755193	124,251,883	-0.030	0.005	$7.00 \cdot 10^{-9}$	-0.009	0.015	$5.37 \cdot 10^{-1}$	-0.025	0.009	$3.64 \cdot 10^{-3}$	T/C	0.386	0.391
22	rs5758265	41,617,897	0.030	0.005	$7.60 \cdot 10^{-9}$	0.024	0.016	$1.20 \cdot 10^{-1}$	0.036	0.010	$1.70 \cdot 10^{-4}$	A/G	0.278	0.271
17	rs17727765	27,576,962	-0.051	0.009	$8.50 \cdot 10^{-9}$	0.014	0.027	$5.92 \cdot 10^{-1}$	-0.029	0.016	$7.11 \cdot 10^{-2}$	T/C	0.923	0.923
9	rs7856424	119,733,595	-0.030	0.005	$8.50 \cdot 10^{-9}$	0.000	0.015	$9.94 \cdot 10^{-1}$	-0.021	0.009	$2.80 \cdot 10^{-2}$	T/C	0.287	0.286
16	rs7198928	7,666,402	0.030	0.005	$1.00 \cdot 10^{-8}$	-0.005	0.014	$7.22 \cdot 10^{-1}$	0.019	0.009	$2.54 \cdot 10^{-2}$	T/C	0.610	0.610
1	rs2389016	80,799,329	0.030	0.005	$1.00 \cdot 10^{-8}$	0.011	0.015	$4.86 \cdot 10^{-1}$	0.029	0.009	$2.35 \cdot 10^{-3}$	T/C	0.284	0.283
1	rs4261101	90,796,053	-0.030	0.005	$1.00 \cdot 10^{-8}$	-0.011	0.014	$4.67 \cdot 10^{-1}$	-0.026	0.009	$2.74 \cdot 10^{-3}$	A/G	0.357	0.357
18	rs62099069	36,883,737	-0.030	0.005	$1.30 \cdot 10^{-8}$	-0.008	0.014	$5.53 \cdot 10^{-1}$	-0.024	0.009	$5.74 \cdot 10^{-3}$	A/T	0.414	0.417
7	rs12666117	109,105,611	0.030	0.005	$1.40 \cdot 10^{-8}$	0.007	0.014	$6.22 \cdot 10^{-1}$	0.024	0.008	$3.86 \cdot 10^{-3}$	A/G	0.474	0.474
18	rs11663393	50,614,732	0.030	0.005	$1.60 \cdot 10^{-8}$	0.005	0.014	$7.09 \cdot 10^{-1}$	0.022	0.008	$9.85 \cdot 10^{-3}$	A/G	0.490	0.493
16	rs7200826	13,066,833	0.030	0.006	$2.40 \cdot 10^{-8}$	0.003	0.016	$8.61 \cdot 10^{-1}$	0.022	0.010	$2.40 \cdot 10^{-2}$	T/C	0.257	0.258
2	rs1226412	157,111,313	0.030	0.006	$2.40 \cdot 10^{-8}$	0.012	0.017	$4.84 \cdot 10^{-1}$	0.032	0.011	$2.60 \cdot 10^{-3}$	T/C	0.802	0.801
9	rs1354115	2,983,774	0.030	0.005	$2.40 \cdot 10^{-8}$	-0.009	0.014	$5.22 \cdot 10^{-1}$	0.015	0.009	$9.35 \cdot 10^{-2}$	A/C	0.626	0.631
13	rs4143229	44,327,799	-0.051	0.009	$2.50 \cdot 10^{-8}$	-0.002	0.027	$9.35 \cdot 10^{-1}$	-0.037	0.017	$2.57 \cdot 10^{-2}$	A/C	0.929	0.930
7	rs10950398	12,264,871	0.030	0.005	$2.60 \cdot 10^{-8}$	-0.017	0.024	$2.16 \cdot 10^{-1}$	0.010	0.009	$2.34 \cdot 10^{-1}$	A/G	0.418	0.424
9	rs7029033	126,682,068	0.049	0.009	$2.70 \cdot 10^{-8}$	-0.014	0.025	$5.87 \cdot 10^{-1}$	0.027	0.015	$7.65 \cdot 10^{-2}$	T/C	0.080	0.083
6	rs9402472	99,566,521	0.030	0.006	$2.80 \cdot 10^{-8}$	0.004	0.016	$8.31 \cdot 10^{-1}$	0.023	0.010	$2.14 \cdot 10^{-2}$	A/G	0.235	0.234
12	rs4074723	23,947,737	-0.030	0.005	$3.10 \cdot 10^{-8}$	-0.010	0.014	$4.78 \cdot 10^{-1}$	-0.023	0.009	$7.03 \cdot 10^{-3}$	A/C	0.403	0.404
1	rs9427672	197,754,741	-0.030	0.006	$3.10 \cdot 10^{-8}$	0.041	0.017	$1.49 \cdot 10^{-2}$	-0.005	0.010	$6.26 \cdot 10^{-1}$	A/G	0.227	0.218
1	rs159963	8,504,421	-0.030	0.005	$3.20 \cdot 10^{-8}$	-0.021	0.014	$1.36 \cdot 10^{-1}$	-0.031	0.009	$3.32 \cdot 10^{-4}$	A/C	0.569	0.577
16	rs11643192	72,214,276	0.030	0.005	$3.40 \cdot 10^{-8}$	0.035	0.014	$1.31 \cdot 10^{-2}$	0.038	0.009	$1.14 \cdot 10^{-5}$	A/C	0.388	0.375
3	chr3:44287760.1	44,287,760	0.030	0.005	$4.60 \cdot 10^{-8}$	0.010	0.015	$4.99 \cdot 10^{-1}$				A/AT	0.670	0.670

Supplementary Table 8: Look-up in the ASD scan of the top educational attainment loci[50] shown together with the results from the corresponding MTAG analysis. The 73 loci represented in the ASD summary statistics are shown. The columns are chromosome (CHR), marker name in educational attainment scan (SNP), chromosomal position (BP), linear regression effect size (β), standard error of effect (SE), p-value (P) in educational attainment, ASD and the ASD-educational-attainment MTAG analysis, allele names (A1A2), minor allele frequency in ASD cases (FRQ A) and controls (FRQ U). The results are presented in order of the p-values in the major depression scan, and statistically significant MTAG results ($P < 1.667 \cdot 10^{-8}$) are presented in **red**. The effect measures of ASD were estimated using logistic regression and inverse variance weighted meta-analysis and those for educational attainment by linear regression. P-values were estimated from the z-scores.

CHR	SNP	BP	Educational Attainment			ASD			ASD-Edu MTAG			A1/A2	FRQ A	FRQ U
			β	SE	P	β	SE	P	β	SE	P			
3	rs11712056	49,914,397	0.025	0.002	$7.53 \cdot 10^{-24}$	0.026	0.014	$6.01 \cdot 10^{-2}$	0.040	0.009	$2.04 \cdot 10^{-5}$	T/C	0.563	0.558
3	rs148734725	49,406,708	0.027	0.003	$1.25 \cdot 10^{-23}$	0.028	0.015	$6.21 \cdot 10^{-2}$	0.043	0.010	$2.29 \cdot 10^{-5}$	A/G	0.304	0.298
9	rs13294439	23,358,875	-0.025	0.003	$6.63 \cdot 10^{-23}$	-0.040	0.014	$4.71 \cdot 10^{-3}$	-0.048	0.009	$3.22 \cdot 10^{-7}$	A/C	0.589	0.596
6	rs9320913	98,584,733	0.024	0.002	$2.05 \cdot 10^{-21}$	0.067	0.014	$1.51 \cdot 10^{-6}$	0.064	0.009	$4.66 \cdot 10^{-12}$	A/C	0.498	0.483

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Supplementary Table 8 – continued from previous page

CHR	SNP	BP	Educational Attainment			ASD			ASD-Edu MTAG			A1/A2	FRQ A	FRQ U
			β	SE	P	β	SE	P	β	SE	P			
2	rs12987662	100,821,548	0.022	0.003	$3.25 \cdot 10^{-18}$	0.005	0.014	$7.14 \cdot 10^{-1}$	0.023	0.009	$1.31 \cdot 10^{-2}$	A/C	0.399	0.399
13	rs9537821	58,402,771	0.023	0.003	$8.61 \cdot 10^{-17}$	-0.023	0.015	$1.26 \cdot 10^{-1}$	0.006	0.010	$5.63 \cdot 10^{-1}$	A/G	0.708	0.709
1	rs34305371	72,733,610	0.036	0.004	$2.34 \cdot 10^{-16}$	0.024	0.025	$3.35 \cdot 10^{-1}$	0.047	0.016	$3.40 \cdot 10^{-3}$	A/G	0.095	0.092
12	rs7306755	123,767,929	0.025	0.003	$4.23 \cdot 10^{-16}$	0.002	0.017	$9.03 \cdot 10^{-1}$	0.024	0.012	$3.50 \cdot 10^{-2}$	A/G	0.202	0.202
2	rs17824247	144,152,539	-0.018	0.003	$5.29 \cdot 10^{-13}$	-0.026	0.014	$6.79 \cdot 10^{-2}$	-0.033	0.009	$4.48 \cdot 10^{-4}$	T/C	0.574	0.578
5	rs61160187	60,111,579	-0.018	0.003	$5.93 \cdot 10^{-13}$	-0.026	0.014	$7.07 \cdot 10^{-2}$	-0.033	0.009	$4.86 \cdot 10^{-4}$	A/G	0.593	0.598
10	rs11191193	103,802,408	0.019	0.003	$6.97 \cdot 10^{-13}$	-0.005	0.014	$7.35 \cdot 10^{-1}$	0.014	0.010	$1.51 \cdot 10^{-1}$	A/G	0.634	0.630
1	rs11588857	204,587,047	0.022	0.003	$1.31 \cdot 10^{-12}$	0.001	0.017	$9.35 \cdot 10^{-1}$	0.021	0.011	$6.94 \cdot 10^{-2}$	A/G	0.213	0.214
12	rs2456973	56,416,928	-0.018	0.003	$1.58 \cdot 10^{-12}$	-0.015	0.015	$2.95 \cdot 10^{-1}$	-0.027	0.010	$6.33 \cdot 10^{-3}$	A/C	0.673	0.676
4	rs4863692	140,764,124	0.018	0.003	$3.80 \cdot 10^{-12}$	0.001	0.015	$9.67 \cdot 10^{-1}$	0.017	0.010	$8.16 \cdot 10^{-2}$	T/G	0.312	0.312
18	rs12969294	35,186,122	-0.018	0.003	$1.11 \cdot 10^{-11}$	-0.001	0.015	$9.22 \cdot 10^{-1}$	-0.017	0.010	$7.87 \cdot 10^{-2}$	A/G	0.336	0.336
3	rs112634398	50,075,494	0.041	0.006	$2.45 \cdot 10^{-11}$	-0.002	0.035	$9.62 \cdot 10^{-1}$	0.034	0.021	$1.12 \cdot 10^{-1}$	A/G	0.952	0.951
1	rs1008078	91,189,731	-0.016	0.003	$7.88 \cdot 10^{-11}$	-0.034	0.014	$1.70 \cdot 10^{-2}$	-0.036	0.009	$1.09 \cdot 10^{-4}$	T/C	0.399	0.409
14	rs17119973	84,913,111	-0.018	0.003	$8.39 \cdot 10^{-11}$	-0.021	0.016	$1.77 \cdot 10^{-1}$	-0.030	0.010	$3.99 \cdot 10^{-3}$	A/G	0.260	0.266
1	rs11210860	43,982,527	0.016	0.003	$9.73 \cdot 10^{-11}$	0.016	0.014	$2.54 \cdot 10^{-1}$	0.025	0.010	$7.50 \cdot 10^{-3}$	A/G	0.385	0.384
10	rs12772375	104,082,688	-0.016	0.003	$1.36 \cdot 10^{-10}$	-0.020	0.014	$1.61 \cdot 10^{-1}$	-0.027	0.009	$3.63 \cdot 10^{-3}$	T/G	0.424	0.429
14	rs1043209	23,373,986	0.016	0.003	$1.37 \cdot 10^{-10}$	-0.005	0.014	$7.01 \cdot 10^{-1}$	0.011	0.009	$2.27 \cdot 10^{-1}$	A/G	0.607	0.609
2	rs16845580	161,920,884	0.016	0.003	$2.07 \cdot 10^{-10}$	0.011	0.014	$4.52 \cdot 10^{-1}$	0.022	0.010	$2.29 \cdot 10^{-2}$	T/C	0.626	0.625
12	rs7955289	14,653,667	0.016	0.003	$3.49 \cdot 10^{-10}$	-0.004	0.014	$8.00 \cdot 10^{-1}$	0.012	0.010	$1.95 \cdot 10^{-1}$	A/T	0.618	0.618
5	rs10061788	87,934,707	0.021	0.003	$5.92 \cdot 10^{-10}$	-0.025	0.019	$1.88 \cdot 10^{-1}$	0.003	0.012	$7.93 \cdot 10^{-1}$	A/G	0.172	0.171
2	rs2457660	60,757,419	-0.016	0.003	$7.33 \cdot 10^{-10}$	-0.008	0.015	$5.82 \cdot 10^{-1}$	-0.019	0.010	$4.19 \cdot 10^{-2}$	T/C	0.623	0.626
1	rs2992632	243,503,764	0.017	0.003	$7.64 \cdot 10^{-10}$	-0.008	0.015	$5.85 \cdot 10^{-1}$	0.010	0.010	$3.24 \cdot 10^{-1}$	A/T	0.708	0.710
9	rs1871109	1,746,016	-0.015	0.002	$1.18 \cdot 10^{-9}$	-0.008	0.014	$5.86 \cdot 10^{-1}$				T/G	0.555	0.559
5	rs1402025	113,987,898	0.018	0.003	$1.51 \cdot 10^{-9}$	0.062	0.017	$1.57 \cdot 10^{-4}$	0.055	0.011	$3.67 \cdot 10^{-7}$	T/C	0.774	0.763
6	rs6231335	98,187,291	-0.016	0.003	$1.62 \cdot 10^{-9}$	-0.002	0.016	$9.17 \cdot 10^{-1}$	-0.016	0.010	$1.15 \cdot 10^{-1}$	T/C	0.692	0.694
3	rs62263923	85,674,790	-0.016	0.003	$1.63 \cdot 10^{-9}$	0.001	0.015	$9.53 \cdot 10^{-1}$	-0.014	0.010	$1.55 \cdot 10^{-1}$	A/G	0.645	0.646
3	rs35761247	48,623,124	0.034	0.006	$2.59 \cdot 10^{-9}$	-0.004	0.033	$9.11 \cdot 10^{-1}$	0.029	0.021	$1.76 \cdot 10^{-1}$	A/G	0.049	0.049
2	rs13402908	100,333,377	-0.015	0.002	$2.91 \cdot 10^{-9}$	0.012	0.014	$4.02 \cdot 10^{-1}$	-0.006	0.009	$5.12 \cdot 10^{-1}$	T/C	0.461	0.454
2	rs4500960	162,818,621	-0.014	0.002	$6.55 \cdot 10^{-9}$	-0.025	0.014	$7.43 \cdot 10^{-2}$	-0.029	0.009	$1.78 \cdot 10^{-3}$	T/C	0.459	0.464
17	rs192818565	43,991,515	0.019	0.003	$7.69 \cdot 10^{-9}$	-0.077	0.017	$9.01 \cdot 10^{-6}$				T/G	0.795	0.810
7	rs12671937	92,654,365	0.014	0.003	$1.02 \cdot 10^{-8}$	-0.041	0.014	$4.22 \cdot 10^{-3}$	-0.012	0.009	$1.85 \cdot 10^{-1}$	A/G	0.525	0.534
7	rs17167170	133,302,345	0.017	0.003	$1.62 \cdot 10^{-8}$	0.041	0.017	$1.72 \cdot 10^{-2}$	0.042	0.012	$2.55 \cdot 10^{-4}$	A/G	0.805	0.800
5	rs2964197	57,535,206	0.014	0.002	$2.01 \cdot 10^{-8}$	0.022	0.014	$1.17 \cdot 10^{-1}$	0.027	0.009	$4.12 \cdot 10^{-3}$	T/C	0.485	0.475
5	rs4493682	45,188,024	0.019	0.003	$2.27 \cdot 10^{-8}$	-0.005	0.019	$7.85 \cdot 10^{-1}$	0.014	0.012	$2.67 \cdot 10^{-1}$	C/G	0.168	0.165
8	rs12682297	145,712,860	-0.014	0.002	$2.29 \cdot 10^{-8}$	0.036	0.015	$1.34 \cdot 10^{-2}$	0.009	0.009	$3.23 \cdot 10^{-1}$	A/T	0.483	0.474
2	rs4851251	100,753,490	-0.015	0.003	$2.83 \cdot 10^{-8}$	0.001	0.015	$9.51 \cdot 10^{-1}$	-0.013	0.010	$1.93 \cdot 10^{-1}$	T/C	0.280	0.280
11	rs7945718	12,748,819	0.014	0.003	$2.85 \cdot 10^{-8}$	0.005	0.014	$7.27 \cdot 10^{-1}$	0.016	0.009	$9.03 \cdot 10^{-2}$	A/G	0.608	0.605
2	rs55830725	237,056,854	-0.018	0.003	$3.05 \cdot 10^{-8}$	0.008	0.019	$6.85 \cdot 10^{-1}$	-0.012	0.012	$3.32 \cdot 10^{-1}$	A/T	0.168	0.166
2	rs1606974	51,873,599	0.021	0.004	$4.03 \cdot 10^{-8}$	-0.012	0.021	$5.44 \cdot 10^{-1}$				A/G	0.132	0.135
1	rs1777827	211,613,114	0.014	0.003	$4.19 \cdot 10^{-8}$	0.008	0.014	$5.76 \cdot 10^{-1}$	0.018	0.010	$6.04 \cdot 10^{-2}$	A/G	0.618	0.619
9	rs895606	88,003,668	0.013	0.002	$6.90 \cdot 10^{-8}$	-0.006	0.014	$6.65 \cdot 10^{-1}$	0.008	0.009	$3.63 \cdot 10^{-1}$	A/G	0.436	0.434
12	rs7131944	92,159,557	0.013	0.003	$1.74 \cdot 10^{-7}$	0.005	0.014	$7.08 \cdot 10^{-1}$	0.016	0.009	$1.01 \cdot 10^{-1}$	A/T	0.613	0.607
1	rs2568955	72,762,169	-0.015	0.003	$1.82 \cdot 10^{-7}$	-0.036	0.016	$2.54 \cdot 10^{-2}$	-0.035	0.010	$6.49 \cdot 10^{-4}$	T/C	0.281	0.293
7	rs11768238	135,227,513	-0.014	0.003	$2.12 \cdot 10^{-7}$	-0.004	0.015	$7.87 \cdot 10^{-1}$	-0.015	0.010	$1.26 \cdot 10^{-1}$	A/G	0.328	0.327
1	rs301800	8,490,603	0.017	0.003	$2.43 \cdot 10^{-7}$	0.016	0.018	$3.74 \cdot 10^{-1}$	0.026	0.012	$3.46 \cdot 10^{-2}$	T/C	0.175	0.169
21	rs2837992	42,620,520	0.013	0.003	$3.08 \cdot 10^{-7}$	0.004	0.015	$7.94 \cdot 10^{-1}$	0.014	0.010	$1.33 \cdot 10^{-1}$	T/C	0.375	0.373
2	rs76076331	10,977,585	0.018	0.004	$3.72 \cdot 10^{-7}$	-0.065	0.020	$1.30 \cdot 10^{-3}$	-0.024	0.013	$6.87 \cdot 10^{-2}$	T/C	0.136	0.146
9	rs7854982	124,644,562	-0.013	0.002	$4.05 \cdot 10^{-7}$	0.030	0.014	$3.12 \cdot 10^{-2}$	0.008	0.009	$4.16 \cdot 10^{-1}$	T/C	0.469	0.461
4	rs12646808	3,249,828	0.013	0.003	$4.40 \cdot 10^{-7}$	-0.007	0.015	$6.29 \cdot 10^{-1}$	0.008	0.010	$4.37 \cdot 10^{-1}$	T/C	0.654	0.654
2	rs6739979	193,731,929	-0.013	0.003	$6.53 \cdot 10^{-7}$	0.015	0.014	$2.99 \cdot 10^{-1}$	-0.002	0.010	$8.19 \cdot 10^{-1}$	T/C	0.628	0.627
5	rs324886	87,896,602	-0.012	0.003	$9.67 \cdot 10^{-7}$	0.056	0.014	$7.94 \cdot 10^{-5}$	0.024	0.009	$1.04 \cdot 10^{-2}$	T/C	0.403	0.395
2	rs11690172	57,387,094	0.012	0.003	$1.44 \cdot 10^{-6}$	-0.005	0.014	$7.47 \cdot 10^{-1}$	0.008	0.009	$3.82 \cdot 10^{-1}$	A/G	0.586	0.584
7	rs12531458	39,090,698	0.011	0.002	$3.47 \cdot 10^{-6}$	0.015	0.014	$2.90 \cdot 10^{-1}$	0.020	0.009	$3.18 \cdot 10^{-2}$	A/C	0.524	0.520
5	rs62379838	120,102,028	0.012	0.003	$4.00 \cdot 10^{-6}$	-0.010	0.015	$5.09 \cdot 10^{-1}$	0.005	0.010	$6.17 \cdot 10^{-1}$	T/C	0.696	0.696
5	rs2431108	103,947,968	0.012	0.003	$4.56 \cdot 10^{-6}$	-0.048	0.015	$1.08 \cdot 10^{-3}$	-0.020	0.010	$4.63 \cdot 10^{-2}$	T/C	0.666	0.677
2	rs10496091	61,482,261	-0.012	0.003	$5.61 \cdot 10^{-6}$	0.026	0.015	$9.64 \cdot 10^{-2}$	0.005	0.010	$6.36 \cdot 10^{-1}$	A/G	0.278	0.272
6	rs7767938	153,367,613	0.013	0.003	$5.75 \cdot 10^{-6}$	-0.016	0.016	$3.14 \cdot 10^{-1}$	0.002	0.011	$8.79 \cdot 10^{-1}$	T/C	0.746	0.750
2	rs2245901	194,296,294	-0.011	0.003	$5.98 \cdot 10^{-6}$	0.010	0.014	$4.98 \cdot 10^{-1}$	-0.004	0.009	$6.44 \cdot 10^{-1}$	A/G	0.408	0.407
7	rs113520408	128,402,782	0.013	0.003	$6.59 \cdot 10^{-6}$	-0.010	0.016	$5.27 \cdot 10^{-1}$	0.005	0.010	$6.17 \cdot 10^{-1}$	A/G	0.265	0.266
3	rs6799130	160,847,801	-0.011	0.002	$8.01 \cdot 10^{-6}$	-0.005	0.014	$7.04 \cdot 10^{-1}$	-0.014	0.009	$1.45 \cdot 10^{-1}$	C/G	0.536	0.540
2	rs11689269	15,621,917	0.012	0.003	$8.35 \cdot 10^{-6}$	-0.029	0.015	$4.68 \cdot 10^{-2}$	-0.008	0.010	$4.21 \cdot 10^{-1}$	C/G	0.326	0.334
12	rs572016	121,279,083	0.011	0.002	$1.08 \cdot 10^{-5}$	0.009	0.014	$5.40 \cdot 10^{-1}$	0.015	0.009	$9.64 \cdot 10^{-2}$	A/G	0.509	0.510
22	rs165633	29,880,773	-0.012	0.003	$1.47 \cdot 10^{-5}$	-0.018	0.017	$2.72 \cdot 10^{-1}$	-0.023	0.011	$3.49 \cdot 10^{-2}$	A/G	0.761	0.764
3	rs62259535	48,939,052	0.032	0.008	$2.35 \cdot 10^{-5}$	0.065	0.037	$7.62 \cdot 10^{-2}$	0.064	0.023	$6.39 \cdot 10^{-3}$	A/G	0.963	0.959
2	rs114598875	60,976,384	-0.014	0.003	$2.71 \cdot 10^{-5}$	-0.006	0.017	$7.05 \cdot 10^{-1}$	-0.016	0.011				

Supplementary Table 9: Look-up in the ASD scan of the top schizophrenia loci[8] shown together with the results from the corresponding MTAG analysis. The 119 loci represented in the ASD summary statistics directly or by proxy are shown. The columns are chromosome (CHR), marker name in schizophrenia scan (SNP), chromosomal position (BP), logistic regression effect size (β), standard error of effect (SE), p-value (P) in schizophrenia, ASD and the ASD-schizophrenia MTAG analysis, allele names (A1A2), minor allele frequency in ASD cases (FRQ A) and controls (FRQ U), and proxies where appropriate, - elsewhere (Proxy). The results are presented in order of the p-values in the major depression scan. There were no statistically significant MTAG results ($P < 1.667 \cdot 10^{-8}$) in this range of loci. The effect measures of ASD and schizophrenia were estimated using logistic regression and inverse variance weighted meta-analysis, and all p-values were estimated from the z-scores.

CHR	SNP	BP	Schizophrenia			ASD			ASD-SCZ MTAG			A1/A2	FRQ A	FRQ U	Proxy
			β	SE	P	β	SE	P	β	SE	P				
6	rs115329265	28,712,247	0.186	0.016	$3.48 \cdot 10^{-31}$	0.029	0.020	$1.49 \cdot 10^{-1}$				A/G	0.863	0.860	-
1	rs1702294	98,501,984	-0.119	0.013	$3.36 \cdot 10^{-19}$	-0.037	0.018	$3.65 \cdot 10^{-2}$	-0.046	0.012	$6.01 \cdot 10^{-5}$	T/C	0.190	0.199	-
10	rs11191419	104,612,335	-0.099	0.011	$6.20 \cdot 10^{-19}$	-0.008	0.015	$5.71 \cdot 10^{-1}$	-0.025	0.010	$8.15 \cdot 10^{-3}$	A/T	0.350	0.351	-
12	rs2007044	2,344,960	-0.092	0.011	$3.22 \cdot 10^{-18}$	0.030	0.014	$3.59 \cdot 10^{-2}$	-0.002	0.009	$8.48 \cdot 10^{-1}$	A/G	0.606	0.596	-
8	rs4129585	143,312,933	0.083	0.010	$1.74 \cdot 10^{-15}$	0.008	0.014	$5.90 \cdot 10^{-1}$	0.021	0.009	$2.15 \cdot 10^{-2}$	A/C	0.440	0.438	-
4	rs35518360	103,146,890	-0.155	0.020	$7.98 \cdot 10^{-15}$	-0.047	0.029	$1.05 \cdot 10^{-1}$	-0.063	0.019	$1.16 \cdot 10^{-3}$	A/T	0.933	0.940	-
7	chr7:2025096:1	2,025,096	-0.081	0.011	$8.20 \cdot 10^{-15}$	-0.003	0.014	$8.61 \cdot 10^{-1}$				A/ACT	0.416	0.417	-
5	rs4391122	60,598,543	-0.081	0.010	$1.10 \cdot 10^{-14}$	0.030	0.014	$3.03 \cdot 10^{-2}$	0.002	0.009	$8.39 \cdot 10^{-1}$	A/G	0.514	0.506	-
12	rs2851447	123,665,113	-0.089	0.012	$1.86 \cdot 10^{-14}$	0.010	0.016	$5.58 \cdot 10^{-1}$	-0.015	0.011	$1.64 \cdot 10^{-1}$	C/G	0.762	0.762	-
15	rs4702	91,426,560	-0.081	0.011	$8.30 \cdot 10^{-14}$	-0.023	0.014	$1.05 \cdot 10^{-1}$	-0.031	0.009	$9.12 \cdot 10^{-4}$	A/G	0.560	0.567	-
3	rs75968099	36,858,583	0.082	0.011	$1.05 \cdot 10^{-13}$	-0.007	0.014	$6.13 \cdot 10^{-1}$	0.012	0.010	$2.09 \cdot 10^{-1}$	T/C	0.362	0.365	-
14	rs12887734	104,046,834	0.084	0.011	$1.36 \cdot 10^{-13}$	0.033	0.015	$2.78 \cdot 10^{-2}$	0.039	0.010	$1.04 \cdot 10^{-4}$	T/G	0.301	0.296	-
15	rs8042374	78,908,032	0.089	0.012	$2.44 \cdot 10^{-13}$	0.016	0.016	$3.17 \cdot 10^{-1}$	0.028	0.011	$9.32 \cdot 10^{-3}$	A/G	0.772	0.770	-
7	rs13240464	110,898,915	0.080	0.011	$3.03 \cdot 10^{-13}$	0.004	0.015	$7.69 \cdot 10^{-1}$	0.021	0.010	$3.69 \cdot 10^{-2}$	T/C	0.674	0.679	-
11	rs10791097	130,718,630	0.074	0.010	$1.09 \cdot 10^{-12}$	0.000	0.014	$9.76 \cdot 10^{-1}$	0.017	0.009	$7.43 \cdot 10^{-2}$	T/G	0.452	0.449	-
2	rs11693094	185,601,420	-0.074	0.010	$1.53 \cdot 10^{-12}$	0.029	0.014	$3.52 \cdot 10^{-2}$	0.003	0.009	$7.14 \cdot 10^{-1}$	T/C	0.477	0.475	-
10	rs7893279	18,745,105	0.117	0.017	$1.97 \cdot 10^{-12}$	0.051	0.022	$2.17 \cdot 10^{-2}$	0.054	0.014	$1.99 \cdot 10^{-4}$	T/G	0.891	0.885	-
12	rs12826178	57,622,371	-0.167	0.024	$2.02 \cdot 10^{-12}$	-0.045	0.028	$1.04 \cdot 10^{-1}$	-0.056	0.018	$1.69 \cdot 10^{-3}$	T/G	0.069	0.072	-
1	rs12129573	73,768,366	0.075	0.011	$2.03 \cdot 10^{-12}$	0.026	0.014	$7.23 \cdot 10^{-2}$	0.031	0.010	$1.27 \cdot 10^{-3}$	A/C	0.371	0.364	-
2	rs6704768	233,592,501	-0.073	0.010	$2.32 \cdot 10^{-12}$	0.030	0.014	$3.28 \cdot 10^{-2}$	0.003	0.009	$7.57 \cdot 10^{-1}$	A/G	0.564	0.557	-
11	rs55661361	124,613,957	-0.077	0.011	$2.80 \cdot 10^{-12}$	-0.048	0.015	$1.19 \cdot 10^{-3}$	-0.047	0.010	$9.97 \cdot 10^{-7}$	A/G	0.336	0.348	-
18	rs9636107	53,200,117	-0.073	0.010	$3.34 \cdot 10^{-12}$	-0.019	0.014	$1.74 \cdot 10^{-1}$	-0.028	0.009	$2.86 \cdot 10^{-3}$	A/G	0.541	0.549	-
11	chr11:46350213:D	46,350,213	-0.098	0.015	$1.26 \cdot 10^{-11}$	0.038	0.020	$5.48 \cdot 10^{-2}$				A/AG	0.157	0.152	rs61126341:1
3	chr3:180594593:1	180,594,593	-0.090	0.013	$1.30 \cdot 10^{-11}$	-0.009	0.017	$5.88 \cdot 10^{-1}$				T/TA	0.783	0.788	-
20	rs6065094	37,453,194	-0.074	0.011	$1.46 \cdot 10^{-11}$	-0.001	0.015	$9.31 \cdot 10^{-1}$	-0.017	0.010	$9.35 \cdot 10^{-2}$	A/G	0.306	0.305	-
2	rs11682175	57,987,593	-0.070	0.010	$1.47 \cdot 10^{-11}$	-0.024	0.014	$8.48 \cdot 10^{-2}$	-0.031	0.009	$7.62 \cdot 10^{-4}$	T/C	0.529	0.536	-
15	rs950169	84,706,461	-0.080	0.012	$1.62 \cdot 10^{-11}$	-0.036	0.016	$1.81 \cdot 10^{-2}$	-0.040	0.010	$8.67 \cdot 10^{-5}$	T/C	0.269	0.277	-
18	rs72934570	53,533,189	-0.135	0.020	$1.97 \cdot 10^{-11}$	-0.038	0.025	$1.22 \cdot 10^{-1}$	-0.050	0.016	$2.05 \cdot 10^{-3}$	T/C	0.085	0.089	-
2	rs6434928	198,304,577	-0.074	0.011	$2.06 \cdot 10^{-11}$	-0.021	0.015	$1.48 \cdot 10^{-1}$	-0.029	0.010	$3.13 \cdot 10^{-3}$	A/G	0.678	0.681	-
22	rs9607782	41,587,556	0.083	0.012	$2.07 \cdot 10^{-11}$	0.025	0.016	$1.16 \cdot 10^{-1}$	0.034	0.011	$1.64 \cdot 10^{-3}$	A/T	0.248	0.243	-
8	rs36068923	111,485,761	-0.085	0.013	$2.61 \cdot 10^{-11}$	-0.038	0.017	$2.60 \cdot 10^{-2}$	-0.042	0.011	$1.85 \cdot 10^{-4}$	A/G	0.787	0.791	-
3	rs17194490	2,547,786	0.096	0.014	$2.69 \cdot 10^{-11}$	0.031	0.019	$1.05 \cdot 10^{-1}$	0.041	0.013	$1.11 \cdot 10^{-3}$	T/G	0.165	0.160	-
11	rs2514218	113,392,994	-0.075	0.011	$2.75 \cdot 10^{-11}$	0.006	0.015	$6.54 \cdot 10^{-1}$	-0.012	0.010	$2.06 \cdot 10^{-1}$	T/C	0.347	0.343	-
11	rs75059851	133,822,569	0.087	0.013	$3.87 \cdot 10^{-11}$	0.004	0.017	$7.90 \cdot 10^{-1}$	0.021	0.011	$5.19 \cdot 10^{-2}$	A/G	0.776	0.771	-
3	rs2535627	52,845,105	0.068	0.010	$4.26 \cdot 10^{-11}$	0.052	0.014	$1.91 \cdot 10^{-4}$	0.047	0.009	$2.51 \cdot 10^{-7}$	T/C	0.530	0.516	-
16	rs12691307	29,939,877	0.071	0.011	$4.55 \cdot 10^{-11}$	0.023	0.014	$1.05 \cdot 10^{-1}$	0.029	0.009	$1.55 \cdot 10^{-3}$	A/G	0.456	0.449	-
3	rs7432375	136,288,405	-0.069	0.011	$7.26 \cdot 10^{-11}$	0.017	0.014	$2.20 \cdot 10^{-1}$	-0.004	0.009	$6.32 \cdot 10^{-1}$	A/G	0.414	0.412	-
18	chr18:52749216:D	52,749,216	0.069	0.011	$8.03 \cdot 10^{-11}$	0.005	0.014	$7.12 \cdot 10^{-1}$				GA/G	0.588	0.585	-
5	rs111294930	152,177,121	0.090	0.014	$1.06 \cdot 10^{-10}$	-0.004	0.015	$7.83 \cdot 10^{-1}$	0.012	0.010	$2.17 \cdot 10^{-1}$	A/G	0.693	0.691	-
5	rs2973155	152,608,619	-0.069	0.011	$1.11 \cdot 10^{-10}$	-0.015	0.014	$3.03 \cdot 10^{-1}$	-0.023	0.010	$1.50 \cdot 10^{-2}$	T/C	0.354	0.358	-
17	rs4523957	2,208,899	0.069	0.011	$2.86 \cdot 10^{-10}$	0.011	0.014	$4.60 \cdot 10^{-1}$	0.021	0.009	$2.51 \cdot 10^{-2}$	T/G	0.624	0.619	-
7	rs12704290	86,427,626	-0.101	0.016	$3.33 \cdot 10^{-10}$	-0.030	0.021	$1.57 \cdot 10^{-1}$	-0.042	0.014	$3.11 \cdot 10^{-3}$	A/G	0.120	0.122	-
15	rs12903146	61,854,663	0.065	0.010	$3.38 \cdot 10^{-10}$	-0.011	0.014	$4.11 \cdot 10^{-1}$	0.007	0.009	$4.44 \cdot 10^{-1}$	A/G	0.541	0.543	-
1	rs11210892	44,100,084	-0.069	0.011	$3.39 \cdot 10^{-10}$	-0.011	0.015	$4.60 \cdot 10^{-1}$	-0.022	0.010	$2.60 \cdot 10^{-2}$	A/G	0.672	0.675	-
19	rs2905426	19,478,022	-0.069	0.011	$3.63 \cdot 10^{-10}$	-0.025	0.014	$7.88 \cdot 10^{-2}$	-0.029	0.010	$1.99 \cdot 10^{-3}$	T/G	0.621	0.626	-
1	rs140505938	150,031,490	-0.090	0.014	$4.49 \cdot 10^{-10}$	0.028	0.018	$1.24 \cdot 10^{-1}$	-0.001	0.012	$9.33 \cdot 10^{-1}$	T/C	0.170	0.167	-
1	rs4648845	2,387,101	0.070	0.011	$8.70 \cdot 10^{-10}$	-0.025	0.016	$1.18 \cdot 10^{-1}$	0.001	0.009	$9.43 \cdot 10^{-1}$	T/C	0.505	0.513	-
16	rs7405404	13,749,859	0.075	0.012	$1.00 \cdot 10^{-9}$	0.057	0.017	$5.67 \cdot 10^{-4}$	0.054	0.011	$1.47 \cdot 10^{-6}$	T/C	0.226	0.215	-
7	rs6466055	104,929,064	0.066	0.011	$1.13 \cdot 10^{-9}$	0.050	0.015	$5.75 \cdot 10^{-4}$	0.046	0.010	$1.82 \cdot 10^{-6}$	A/C	0.357	0.345	-
1	chr1:8424984:D	8,424,984	0.068	0.011	$1.17 \cdot 10^{-9}$	0.004	0.015	$7.76 \cdot 10^{-1}$				GA/G	0.342	0.343	-
12	rs4766428	110,723,245	0.065	0.011	$1.40 \cdot 10^{-9}$	0.002	0.014	$8.59 \cdot 10^{-1}$	0.016	0.009	$8.39 \cdot 10^{-2}$	T/C	0.445	0.442	-
4	rs10520163	170,626,552	0.063	0.010	$1.47 \cdot 10^{-9}$	0.030	0.014	$2.90 \cdot 10^{-2}$	0.032	0.009	$4.54 \cdot 10^{-4}$	T/C	0.502	0.497	-
6	rs117074560	96,459,651	-0.163	0.027	$1.64 \cdot 10^{-9}$	-0.029	0.033	$3.81 \cdot 10^{-1}$	-0.051	0.022	$2.06 \cdot 10^{-2}$	T/C	0.046	0.047	-
22	rs6002655	42,603,814	0.064	0.011	$1.71 \cdot 10^{-9}$	0.031	0.014	$3.05 \cdot 10^{-2}$	0.033	0.009	$4.15 \cdot 10^{-4}$	T/C	0.431	0.424	-
2	chr2:146436222:1	146,436,222	0.083	0.014	$1.81 \cdot 10^{-9}$	-0.016	0.018	$3.83 \cdot 10^{-1}$				TC/T	0.172	0.175	rs56807175:1
11	rs9420	57,510,294	0.066	0.011	$2.24 \cdot 10^{-9}$	0.032	0.015	$2.74 \cdot 10^{-2}$	0.033	0.010	$7.00 \cdot 10^{-4}$	A/G	0.331	0.324	-
11	rs11027857	24,403,620	0.062	0.010	$2.55 \cdot 10^{-9}$	0.002	0.014	$8.77 \cdot 10^{-1}$	0.015	0.009	$9.56 \cdot 10^{-2}$	A/G	0.511	0.509	-
1	rs1498232	30,433,951	0.067	0.011	$2.86 \cdot 10^{-9}$	0.019	0.015	$1.91 \cdot 10^{-1}$	0.027	0.010	$6.59 \cdot 10^{-3}$	T/C	0.321	0.319	-
7	rs3735025	137,074,844	0.064	0.011	$3.28 \cdot 10^{-9}$	0.030	0.015	$4.19 \cdot 10^{-2}$	0.033	0.010	$7.12 \cdot 10^{-4}$	T/C	0.654	0.648	-
9	rs11139497	84,739,941	0.066	0.011	$3.61 \cdot 10^{-9}$	0.018	0.015	$2.26 \cdot 10^{-1}$	0.026	0.010	$9.44 \cdot 10^{-3}$	A/T	0.319	0.314	-

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Supplementary Table 9 – continued from previous page

CHR	SNP	BP	Schizophrenia			ASD			ASD-SCZ MTAG			A1/A2	FRQ A	FRQ U	Proxy
			β	SE	P	β	SE	P	β	SE	P				
1	rs77149735	243,555,105	0.276	0.047	$3.73 \cdot 10^{-9}$	0.107	0.041	$8.11 \cdot 10^{-3}$				A/G	0.032	0.030	-
15	rs56205728	40,567,237	0.071	0.012	$4.18 \cdot 10^{-9}$	0.006	0.016	$7.09 \cdot 10^{-1}$	0.017	0.010	$9.38 \cdot 10^{-2}$	A/G	0.281	0.278	-
19	rs2053079	30,987,423	-0.071	0.012	$4.49 \cdot 10^{-9}$	0.023	0.016	$1.59 \cdot 10^{-1}$	-0.001	0.011	$9.18 \cdot 10^{-1}$	A/G	0.761	0.755	-
5	rs16867576	88,746,331	0.097	0.017	$4.61 \cdot 10^{-9}$	0.033	0.020	$9.65 \cdot 10^{-2}$	0.039	0.013	$2.93 \cdot 10^{-3}$	A/G	0.860	0.854	-
3	rs4330281	17,859,366	-0.062	0.011	$4.64 \cdot 10^{-9}$	0.014	0.014	$3.05 \cdot 10^{-1}$	-0.003	0.009	$7.24 \cdot 10^{-1}$	T/C	0.512	0.506	-
5	rs3849046	137,851,192	0.061	0.010	$4.67 \cdot 10^{-9}$	0.018	0.014	$1.94 \cdot 10^{-1}$	0.024	0.009	$8.27 \cdot 10^{-3}$	T/C	0.555	0.553	-
14	rs2693698	99,719,219	-0.063	0.011	$4.80 \cdot 10^{-9}$	-0.018	0.014	$2.09 \cdot 10^{-1}$	-0.024	0.009	$1.05 \cdot 10^{-2}$	A/G	0.463	0.469	-
14	rs2332700	72,417,326	0.070	0.012	$4.86 \cdot 10^{-9}$	0.034	0.016	$3.37 \cdot 10^{-2}$	0.037	0.011	$4.84 \cdot 10^{-4}$	C/G	0.260	0.255	-
5	rs1501357	45,364,875	-0.077	0.013	$5.00 \cdot 10^{-9}$	0.000	0.018	$9.76 \cdot 10^{-1}$	-0.017	0.012	$1.71 \cdot 10^{-1}$	T/C	0.822	0.823	-
8	rs6984242	60,700,469	-0.061	0.011	$6.00 \cdot 10^{-9}$	-0.004	0.014	$7.74 \cdot 10^{-1}$	-0.016	0.009	$9.08 \cdot 10^{-2}$	A/G	0.608	0.612	-
1	chr1:243881945:1	243,881,945	0.066	0.011	$6.53 \cdot 10^{-9}$	-0.008	0.015	$5.90 \cdot 10^{-1}$				A/AT	0.317	0.317	-
5	rs79212538	151,993,104	0.144	0.025	$7.00 \cdot 10^{-9}$	0.039	0.033	$2.37 \cdot 10^{-1}$	0.055	0.022	$1.28 \cdot 10^{-2}$	T/G	0.047	0.046	-
2	rs3768644	72,361,505	-0.101	0.017	$7.39 \cdot 10^{-9}$	-0.012	0.023	$6.09 \cdot 10^{-1}$	-0.026	0.015	$7.96 \cdot 10^{-2}$	A/G	0.106	0.107	-
11	rs77502336	123,394,636	0.064	0.011	$7.54 \cdot 10^{-9}$	0.016	0.015	$3.04 \cdot 10^{-1}$	0.025	0.010	$1.27 \cdot 10^{-2}$	C/G	0.309	0.303	-
2	rs6704641	200,164,252	0.078	0.014	$8.33 \cdot 10^{-9}$	-0.016	0.018	$3.83 \cdot 10^{-1}$	0.007	0.012	$5.77 \cdot 10^{-1}$	A/G	0.817	0.818	-
2	rs59979824	193,848,340	-0.065	0.011	$8.41 \cdot 10^{-9}$	-0.021	0.015	$1.48 \cdot 10^{-1}$	-0.027	0.010	$4.63 \cdot 10^{-3}$	A/C	0.344	0.351	-
4	rs1106568	176,861,301	-0.068	0.012	$9.47 \cdot 10^{-9}$	-0.024	0.016	$1.23 \cdot 10^{-1}$	-0.030	0.011	$4.89 \cdot 10^{-3}$	A/G	0.742	0.744	-
8	rs10503253	4,180,844	0.071	0.012	$1.06 \cdot 10^{-8}$	-0.001	0.017	$9.43 \cdot 10^{-1}$	0.015	0.011	$1.87 \cdot 10^{-1}$	A/C	0.207	0.208	-
5	rs10043984	137,712,121	0.067	0.012	$1.09 \cdot 10^{-8}$	0.033	0.016	$4.21 \cdot 10^{-2}$	0.035	0.011	$1.05 \cdot 10^{-3}$	T/C	0.249	0.239	-
2	rs11685299	225,391,296	-0.063	0.011	$1.12 \cdot 10^{-8}$	0.037	0.015	$1.22 \cdot 10^{-2}$	0.009	0.010	$3.79 \cdot 10^{-1}$	A/C	0.333	0.322	-
8	rs7819570	89,588,626	0.076	0.013	$1.22 \cdot 10^{-8}$	0.006	0.018	$7.34 \cdot 10^{-1}$	0.021	0.012	$7.97 \cdot 10^{-2}$	T/G	0.179	0.178	-
18	rs715170	53,795,514	-0.067	0.012	$1.27 \cdot 10^{-8}$	0.048	0.016	$2.05 \cdot 10^{-3}$	0.016	0.010	$1.14 \cdot 10^{-1}$	T/C	0.275	0.265	-
16	rs9922678	9,946,319	0.065	0.011	$1.28 \cdot 10^{-8}$	0.026	0.015	$8.46 \cdot 10^{-2}$	0.031	0.010	$2.90 \cdot 10^{-3}$	A/G	0.284	0.276	-
18	rs78322266	53,063,676	0.173	0.030	$1.32 \cdot 10^{-8}$	0.060	0.042	$1.55 \cdot 10^{-1}$	0.081	0.029	$5.13 \cdot 10^{-3}$	T/G	0.029	0.026	-
14	rs2068012	30,190,316	-0.070	0.012	$1.41 \cdot 10^{-8}$	-0.010	0.016	$5.46 \cdot 10^{-1}$	-0.020	0.011	$6.11 \cdot 10^{-2}$	T/C	0.759	0.759	-
3	rs832187	63,833,050	-0.060	0.011	$1.43 \cdot 10^{-8}$	-0.025	0.015	$8.42 \cdot 10^{-2}$	-0.029	0.010	$2.48 \cdot 10^{-3}$	T/C	0.637	0.644	-
16	rs8044995	68,189,340	0.078	0.014	$1.51 \cdot 10^{-8}$	-0.017	0.019	$3.52 \cdot 10^{-1}$	0.007	0.012	$5.68 \cdot 10^{-1}$	A/G	0.167	0.170	-
2	chr2:149429178:D	149,429,178	-0.154	0.027	$1.59 \cdot 10^{-8}$	0.055	0.030	$6.71 \cdot 10^{-2}$				A/AT	0.059	0.057	rs200327371:1
17	rs8082590	17,958,402	-0.063	0.011	$1.77 \cdot 10^{-8}$	0.037	0.015	$1.51 \cdot 10^{-2}$	0.008	0.010	$4.44 \cdot 10^{-1}$	A/G	0.698	0.694	-
15	rs12148337	70,589,272	0.058	0.010	$1.79 \cdot 10^{-8}$	0.006	0.014	$6.67 \cdot 10^{-1}$	0.016	0.009	$7.73 \cdot 10^{-2}$	T/C	0.456	0.453	-
16	rs12325245	58,681,393	-0.084	0.015	$1.87 \cdot 10^{-8}$	-0.013	0.020	$1.81 \cdot 10^{-1}$	-0.028	0.014	$3.99 \cdot 10^{-2}$	A/T	0.863	0.867	-
12	rs2239063	2,511,831	0.065	0.012	$1.93 \cdot 10^{-8}$	-0.004	0.016	$8.00 \cdot 10^{-1}$	0.013	0.010	$2.05 \cdot 10^{-1}$	A/C	0.725	0.727	-
5	rs12522290	152,797,656	0.081	0.014	$1.99 \cdot 10^{-8}$	0.012	0.018	$5.08 \cdot 10^{-1}$	0.024	0.012	$4.97 \cdot 10^{-2}$	C/G	0.826	0.823	-
1	rs10803138	243,555,219	-0.069	0.012	$2.03 \cdot 10^{-8}$	0.010	0.016	$5.28 \cdot 10^{-1}$	-0.008	0.011	$4.33 \cdot 10^{-1}$	A/G	0.259	0.258	-
8	rs73229090	27,442,127	-0.097	0.017	$2.10 \cdot 10^{-8}$	-0.030	0.022	$1.81 \cdot 10^{-1}$	-0.039	0.015	$7.31 \cdot 10^{-3}$	A/C	0.110	0.113	-
12	rs324017	57,487,814	-0.064	0.011	$2.13 \cdot 10^{-8}$	0.002	0.015	$8.94 \cdot 10^{-1}$	-0.012	0.010	$2.40 \cdot 10^{-1}$	A/C	0.292	0.294	-
10	rs55833108	104,741,583	0.073	0.013	$2.23 \cdot 10^{-8}$	0.007	0.017	$6.96 \cdot 10^{-1}$	0.021	0.011	$6.23 \cdot 10^{-2}$	T/G	0.208	0.209	-
3	rs9841616	181,167,585	-0.078	0.014	$2.35 \cdot 10^{-8}$	-0.020	0.019	$2.86 \cdot 10^{-1}$	-0.031	0.013	$1.49 \cdot 10^{-2}$	A/T	0.158	0.157	-
1	rs76869799	97,834,525	-0.167	0.030	$2.64 \cdot 10^{-8}$	0.048	0.042	$2.49 \cdot 10^{-1}$	-0.004	0.026	$8.65 \cdot 10^{-1}$	C/G	0.968	0.967	-
6	rs1339227	73,155,701	-0.060	0.011	$2.69 \cdot 10^{-8}$	-0.026	0.015	$7.58 \cdot 10^{-2}$	-0.028	0.010	$2.81 \cdot 10^{-3}$	T/C	0.358	0.369	-
7	chr7:24747494:D	24,747,494	0.096	0.017	$2.85 \cdot 10^{-8}$	-0.065	0.023	$5.06 \cdot 10^{-3}$				CTA/C	0.896	0.901	-
5	rs4388249	109,036,066	0.073	0.013	$3.05 \cdot 10^{-8}$	0.012	0.019	$5.20 \cdot 10^{-1}$	0.023	0.012	$6.00 \cdot 10^{-2}$	T/C	0.174	0.172	-
4	rs215411	23,423,603	0.062	0.011	$3.06 \cdot 10^{-8}$	0.026	0.015	$6.98 \cdot 10^{-2}$	0.029	0.010	$2.28 \cdot 10^{-3}$	A/T	0.348	0.346	-
5	rs11740474	153,680,747	-0.060	0.011	$3.15 \cdot 10^{-8}$	-0.020	0.014	$1.57 \cdot 10^{-1}$	-0.025	0.009	$6.17 \cdot 10^{-3}$	A/T	0.572	0.575	-
22	rs1023500	42,340,844	0.073	0.013	$3.43 \cdot 10^{-8}$	0.023	0.018	$2.14 \cdot 10^{-1}$	0.031	0.012	$9.73 \cdot 10^{-3}$	T/C	0.821	0.821	-
11	rs12421382	109,378,071	-0.061	0.011	$3.70 \cdot 10^{-8}$	-0.020	0.015	$1.76 \cdot 10^{-1}$	-0.026	0.010	$7.52 \cdot 10^{-3}$	T/C	0.333	0.341	-
7	rs211829	110,048,893	0.059	0.011	$3.71 \cdot 10^{-8}$	0.045	0.014	$1.60 \cdot 10^{-3}$	0.039	0.009	$3.77 \cdot 10^{-5}$	T/C	0.631	0.619	-
12	rs679087	29,917,265	-0.061	0.011	$3.91 \cdot 10^{-8}$	0.001	0.014	$9.38 \cdot 10^{-1}$	-0.012	0.010	$2.23 \cdot 10^{-1}$	A/C	0.361	0.363	-
2	rs75575209	58,138,192	-0.103	0.019	$3.95 \cdot 10^{-8}$	-0.016	0.025	$5.20 \cdot 10^{-1}$	-0.033	0.017	$4.94 \cdot 10^{-2}$	A/T	0.914	0.917	-
7	rs7801375	131,567,263	-0.079	0.014	$4.42 \cdot 10^{-8}$	-0.026	0.019	$1.60 \cdot 10^{-1}$	-0.034	0.012	$6.59 \cdot 10^{-3}$	A/G	0.158	0.162	-
1	rs14403	243,663,893	-0.068	0.013	$4.42 \cdot 10^{-8}$	0.000	0.017	$9.88 \cdot 10^{-1}$	-0.015	0.011	$1.63 \cdot 10^{-1}$	T/C	0.221	0.223	-
1	rs6670165	177,280,121	0.072	0.013	$4.45 \cdot 10^{-8}$	0.033	0.017	$5.74 \cdot 10^{-2}$	0.037	0.012	$1.50 \cdot 10^{-3}$	T/C	0.202	0.198	-
1	rs7523273	207,977,083	0.061	0.011	$4.47 \cdot 10^{-8}$	-0.010	0.015	$5.00 \cdot 10^{-1}$	0.006	0.010	$5.36 \cdot 10^{-1}$	A/G	0.645	0.642	-
20	rs7267348	48,131,036	-0.065	0.012	$4.56 \cdot 10^{-8}$	-0.027	0.016	$8.79 \cdot 10^{-2}$	-0.032	0.011	$3.14 \cdot 10^{-3}$	T/C	0.754	0.761	-
12	rs4240748	92,246,786	-0.059	0.011	$4.59 \cdot 10^{-8}$	0.006	0.014	$6.66 \cdot 10^{-1}$	-0.009	0.010	$3.64 \cdot 10^{-1}$	C/G	0.376	0.375	-
2	rs2909457	162,845,855	-0.058	0.011	$4.62 \cdot 10^{-8}$	0.020	0.014	$1.46 \cdot 10^{-1}$	0.000	0.009	$9.99 \cdot 10^{-1}$	A/G	0.558	0.553	-
19	rs56873913	50,091,199	0.069	0.013	$4.69 \cdot 10^{-8}$	0.004	0.016	$8.06 \cdot 10^{-1}$	0.017	0.011	$1.13 \cdot 10^{-1}$	T/G	0.753	0.750	-
12	rs10860964	103,596,455	0.059	0.011	$4.84 \cdot 10^{-8}$	0.010	0.014	$4.79 \cdot 10^{-1}$	0.018	0.009	$6.07 \cdot 10^{-2}$	T/C	0.622	0.620	-
5	chr5:140143664:1	140,143,664	0.056	0.010	$4.85 \cdot 10^{-8}$	0.015	0.014	$2.79 \cdot 10^{-1}$				CATTGAAGAAA/C	0.474	0.470	rs111896713:1

3.2.4 Gene-based association and gene-set enrichment

Supplementary Table 10: Top 25 results from Magma gene-based association from the iPSYCH-PGC meta analysis of ASD. **Red** means the gene is significant after Bonferroni correction, *slanted* gene name that no single SNP in the gene was genome wide significant, **bold face** gene name that no single SNP in the gene was significant at the Bonferroni level (significance from z-scores).

Gene	Chr	BP start	BP stop	N SNPs	N param	N	Z-stat	P	Bonf P
XRN2	20	21,283,942	21,370,463	174	17	21,104	6.003	$9.69 \cdot 10^{-10}$	$1.73 \cdot 10^{-5}$
KCNN2	5	113,698,016	113,832,197	306	39	21,009	5.995	$1.02 \cdot 10^{-9}$	$1.82 \cdot 10^{-5}$
KIZ	20	21,106,624	21,227,260	200	12	21,080	5.725	$5.17 \cdot 10^{-9}$	$9.23 \cdot 10^{-5}$
KANSL1	17	44,107,282	44,302,740	89	14	21,038	5.169	$1.18 \cdot 10^{-7}$	$2.10 \cdot 10^{-3}$
MACROD2	20	13,976,146	16,033,842	6,037	256	21,104	5.137	$1.40 \cdot 10^{-7}$	$2.49 \cdot 10^{-3}$
WNT3	17	44,841,686	44,896,082	74	11	20,952	4.934	$4.03 \cdot 10^{-7}$	$7.19 \cdot 10^{-3}$
MAPT	17	43,971,748	44,105,700	72	8	21,020	4.891	$5.01 \cdot 10^{-7}$	$8.94 \cdot 10^{-3}$
MFHAS1	8	8,641,999	8,751,131	497	28	21,081	4.870	$5.58 \cdot 10^{-7}$	$9.96 \cdot 10^{-3}$
XKR6	8	10,753,654	11,058,875	1,013	28	21,102	4.798	$8.01 \cdot 10^{-7}$	$1.43 \cdot 10^{-2}$
MSRA	8	9,911,830	10,286,401	1,547	62	21,112	4.771	$9.15 \cdot 10^{-7}$	$1.63 \cdot 10^{-2}$
CRHR1	17	43,697,710	43,913,194	276	21	21,133	4.740	$1.07 \cdot 10^{-6}$	$1.91 \cdot 10^{-2}$
SOX7	8	10,581,278	10,588,022	24	5	21,199	4.709	$1.24 \cdot 10^{-6}$	$2.22 \cdot 10^{-2}$
NTM	11	131,240,371	132,206,716	2,938	213	21,113	4.698	$1.32 \cdot 10^{-6}$	$2.35 \cdot 10^{-2}$
MMP12	11	102,733,464	102,745,764	22	8	21,104	4.584	$2.28 \cdot 10^{-6}$	$4.07 \cdot 10^{-2}$
BLK	8	11,351,521	11,422,108	269	21	21,161	4.569	$2.45 \cdot 10^{-6}$	$4.37 \cdot 10^{-2}$
MANBA	4	103,552,643	103,682,151	210	13	21,114	4.468	$3.96 \cdot 10^{-6}$	$7.06 \cdot 10^{-2}$
ADTRP	6	11,713,888	11,779,280	247	25	21,083	4.463	$4.04 \cdot 10^{-6}$	$7.21 \cdot 10^{-2}$
WDPCP	2	63,348,518	63,815,867	955	25	21,121	4.413	$5.11 \cdot 10^{-6}$	$9.11 \cdot 10^{-2}$
PINX1	8	10,622,884	10,697,299	416	17	21,155	4.326	$7.60 \cdot 10^{-6}$	$1.36 \cdot 10^{-1}$
PKP4	2	159,313,476	159,537,941	605	19	21,070	4.312	$8.11 \cdot 10^{-6}$	$1.45 \cdot 10^{-1}$
PLEKHM1	17	43,513,266	43,568,146	64	9	20,916	4.289	$8.99 \cdot 10^{-6}$	$1.60 \cdot 10^{-1}$
C8orf74	8	10,530,147	10,558,103	31	9	21,199	4.225	$1.20 \cdot 10^{-5}$	$2.14 \cdot 10^{-1}$
MDH1	2	63,815,743	63,834,331	38	6	21,008	4.203	$1.32 \cdot 10^{-5}$	$2.35 \cdot 10^{-1}$
HDAC4	2	239,969,864	240,322,643	1,109	80	21,125	4.165	$1.56 \cdot 10^{-5}$	$2.77 \cdot 10^{-1}$
WNT5B	12	1,726,222	1,756,378	115	29	21,191	4.054	$2.52 \cdot 10^{-5}$	$4.50 \cdot 10^{-1}$

Supplementary Table 11: Gene set analysis of candidate gene sets, M13, M16 and M17 from [41], constrained genes of [42, 43] with a high probability of being loss-of-function intolerant ($pLI > 0.9$), the 65 genes at $FDR < 0.1$ in [234], and the highly curated list of ASD genes (https://spark-sf.s3.amazonaws.com/SPARK_gene_list.pdf) from SPARK[235]. The effect sizes estimated by MAGMA indicate the extent to which the z-scores for the genes in the set are higher than those in the complement, adjusted among other features the gene length (see section 2.1.2 for details). The β_{std} are semi-standardized regression coefficients, and the p-values are from a z-test.

Gene set	N Genes	β	β_{std}	$SD(\beta)$	P
M13	603	-0.015	-0.003	0.035	0.662
M16	401	0.069	0.010	0.042	0.050
M17	694	-0.022	-0.004	0.033	0.749
Constrained	2,939	0.038	0.014	0.017	0.014
Sanders	65	0.164	0.010	0.107	0.063
SPARK	77	0.272	0.017	0.100	0.003

Supplementary Table 12: Top 25 genesets from gene set analysis of 863 gene sets from the Gene Oncology[44, 45] ‘molecular functions’ of MsigDB[46] including ‘BioCarta’. MAGMA estimates the effect sizes in a regression adjusted among other features the gene length (see section 2.1.2 for details). The β s indicate the extent to which the z-scores for the genes in the set are higher than those in the complement, the β_{std} are semi-standardized regression coefficients, and p-values are from a z-test.

Gene set	N Genes	β	β_{std}	SD(β)	P
GO ION GATED CHANNEL ACTIVITY	41	0.435	0.021	0.144	0.001
GO PHOSPHATIDYLINOSITOL MONOPHOSPHATE PHOSPHATASE ACTIVITY	11	0.724	0.018	0.244	0.002
GO CALCIUM ACTIVATED POTASSIUM CHANNEL ACTIVITY	17	0.686	0.021	0.234	0.002
GO UBIQUITIN LIKE PROTEIN SPECIFIC PROTEASE ACTIVITY	85	0.236	0.016	0.082	0.002
GO PHOSPHATIDYLINOSITOL PHOSPHATE BINDING	106	0.241	0.019	0.085	0.002
GO PHOSPHATIDYLINOSITOL BINDING	184	0.174	0.018	0.062	0.003
GO CALCIUM ACTIVATED CATION CHANNEL ACTIVITY	28	0.475	0.019	0.170	0.003
GO RNA POLYMERASE II TRANSCRIPTION COREPRESSOR ACTIVITY	22	0.475	0.017	0.172	0.003
GO HISTONE DEMETHYLASE ACTIVITY	19	0.523	0.017	0.192	0.003
GO PROTEOGLYCAN BINDING	26	0.450	0.017	0.173	0.005
GO CYSTEINE TYPE PEPTIDASE ACTIVITY	144	0.170	0.015	0.066	0.005
GO OXIDOREDUCTASE ACTIVITY OXIDIZING METAL IONS	16	0.550	0.017	0.216	0.005
GO PROTEIN SERINE THREONINE PHOSPHATASE ACTIVITY	61	0.263	0.015	0.104	0.006
GO PHOSPHATIDYLINOSITOL 3 4 5 TRISPHOSPHATE BINDING	30	0.405	0.017	0.160	0.006
GO PHOSPHOLIPID BINDING	326	0.117	0.016	0.048	0.007
GO INSULIN LIKE GROWTH FACTOR BINDING	25	0.434	0.016	0.181	0.008
GO OXIDOREDUCTASE ACTIVITY ACTING ON PEROXIDE AS ACCEPTOR	35	0.343	0.015	0.143	0.008
GO KINASE BINDING	577	0.083	0.015	0.035	0.008
GO PHOSPHORIC ESTER HYDROLASE ACTIVITY	339	0.106	0.015	0.045	0.009
GO MHC PROTEIN BINDING	22	0.400	0.014	0.172	0.010
GO DIPEPTIDASE ACTIVITY	14	0.456	0.013	0.204	0.013
GO COA HYDROLASE ACTIVITY	19	0.445	0.015	0.201	0.013
GO NOTCH BINDING	18	0.469	0.015	0.211	0.013
GO PROTEIN TYROSINE PHOSPHATASE ACTIVITY	92	0.196	0.014	0.089	0.014
GO DIPEPTIDYL PEPTIDASE ACTIVITY	11	0.489	0.012	0.225	0.015

3.3 Polygenic qualities of subtypes

This section presents results on heritability of and genetic overlap between subtypes of ASD. For results on genetic correlation with other phenotypes, see Table 5.

3.3.1 Heritability and genetic correlation across subtypes

Supplementary Table 13: Definition of the hierarchical ASD subtypes from the ICD10 diagnosis categories. See Table 1 for counts.

Name	Diagnosis
(h)CHA	Childhood autism (ICD10 F84.0)
hATA	those with ATA and no CHA (ICD10 F84.1)
hAsp	Asperger’s syndrome and no CHA and no ATA (ICD10 F84.5)
hPDM	those with PDM (pervasive disorders, unspecified and other, ICD10 F84.8+9) and none of the above.

Supplementary Table 14: Heritability estimates from GCTA for subtypes and substrata on the observed as well as the liability scale. The number of samples are: ASD 13 076, ASD wID 1 873, ASD woID 11 203, CHA and hCHA 3 310, ATA 1 607, Asp 4 622, OPDD 2 042, PDDU 3 753, PDM 5 460, hATA 1 494, hAsp 4 417, hPDM 3 855. For a graphical representation, see Figure 82.

Summary Stats	Pop Prev	h^2_{G} Obs	$SE(h^2_{\text{G}} \text{ Obs})$	h^2_{G} Liab	$SE(h^2_{\text{G}} \text{ Liab})$
ASD	0.012	0.129	0.008	0.080	0.005
ASD wID	0.002	0.020	0.009	0.029	0.013
ASD woID	0.010	0.137	0.008	0.086	0.005
CHA	0.003	0.049	0.009	0.048	0.009
ATA	0.002	0.008	0.009	0.012	0.013
Asp	0.004	0.120	0.010	0.097	0.008
OPDD	0.002	0.024	0.009	0.031	0.012
PDDU	0.004	0.047	0.009	0.042	0.008
PDM	0.005	0.062	0.009	0.047	0.007
hCHA	0.003	0.049	0.009	0.048	0.009
hATA	0.001	0.010	0.009	0.016	0.014
hAsp	0.004	0.116	0.010	0.096	0.008
hPDM	0.004	0.050	0.009	0.045	0.008

Supplementary Table 15: GCTA based genetic correlation, r_{G} , between ASD subtypes. The number of samples are: ASD 13 076, ASD wID 1 873, ASD woID 11 203, CHA and hCHA 3 310, ATA 1 607, Asp 4 622, OPDD 2 042, PDDU 3 753, PDM 5 460, hATA 1 494, hAsp 4 417, hPDM 3 855. For a graphical presentation, see Figure 83.

Trait 1	Trait 2	$h^2_{\text{G},1}$	$se(h^2_{\text{G},1})$	$h^2_{\text{G},2}$	$se(h^2_{\text{G},2})$	r_{G}	$se(r_{\text{G}})$
ASD woID	ASD wID	0.140	0.009	0.065	0.040	1.000	0.312
CHA	hATA	0.060	0.012	0.070	0.026	0.891	0.250
CHA	hAsp	0.083	0.018	0.136	0.015	1.000	0.141
CHA	hPDM	0.093	0.017	0.083	0.015	0.965	0.151
hATA	hPDM	0.038	0.028	0.056	0.012	1.000	0.459
hATA	hAsp	0.065	0.030	0.141	0.012	0.718	0.206
hAsp	hPDM	0.170	0.016	0.077	0.016	0.955	0.121
CHA woID	hPDM woID	0.070	0.020	0.066	0.014	1.000	0.229
hATA woID	hAsp woID	0.080	0.037	0.131	0.012	0.908	0.241
CHA woID	hATA woID	0.062	0.013	0.034	0.025	1.000	0.424
hATA woID	hPDM woID	0.038	0.032	0.048	0.011	1.000	0.522
hAsp woID	hPDM woID	0.160	0.015	0.066	0.017	1.000	0.156
CHA woID	hAsp woID	0.093	0.022	0.159	0.014	0.893	0.132

3.4 Hi-C analysis

Supplementary Table 16: Altogether 380 credible SNPs were identified from 28 loci that could be assigned to 95 genes (of which 40 are protein-coding genes), including 39 promoter SNPs assigned to 9 genes, and 16 functional SNPs within 8 genes. Hi-C identified 86 genes interacting with credible SNPs in either GZ or CP. Among these genes, 34 genes are interacting with credible SNPs in both CP and GZ, which represent a high-confidence gene list (these are coloured [blue](#)).

CHR	BP	CredSNP	IndexSNP	Index P	ENSGID_CP	ENSGID_GZ	ENSGID Promoter	ENSGID functional	Chrom HMM	Histone	HGNC_CP	HGNC_GZ	HGNC Promoter	HGNC functional
chr10	72,748,374	chr10:72748374	rs78827416	9.0e-07	NA	NA	NA	NA	15_Quies		NA	NA	NA	NA
chr17	44,104,576	chr17:44104576	rs142920272, rs147317628	2.9e-07, 1.1e-06	ENSG00000131484(3.118e-13), ENSG00000176681(5.735e-113), ENSG00000214425(3.118e-13), ENSG00000228696(5.735e-113), ENSG00000260075(2.751e-15), ENSG00000266918(3.118e-13)	ENSG00000073969(1.064e-07), ENSG00000120071(2.801e-05), ENSG00000136448(5.713e-08), ENSG00000172992(5.713e-08), ENSG00000228696(6.468e-09), ENSG00000238723(2.941e-05), ENSG00000262500(5.235e-05)	NA	NA	5_TxWk	LRR37A, LRR37A4P, ARL17B, NSFP1	NSF, KANSL1, NMT1, DCAKD, ARL17B	NA	NA	
chr17	44,122,345	chr17:44122345	rs142920272, rs147317628	2.9e-07, 1.1e-06	ENSG00000108433(7.417e-05), ENSG00000120071(2.093e-15), ENSG00000176681(4.187e-13), ENSG00000214401(7.753e-08), ENSG00000228696(4.187e-13), ENSG00000238723(2.093e-15), ENSG00000262500(1.238e-14), ENSG00000262633(7.417e-05), ENSG00000262879(7.417e-05), ENSG00000263142(7.417e-05)	ENSG00000108433(4.841e-06), ENSG00000120071(1.708e-15), ENSG00000176681(1.036e-29), ENSG00000214401(8.406e-08), ENSG00000228696(1.036e-29), ENSG00000238723(1.708e-15), ENSG00000262500(5.602e-15), ENSG00000262633(4.841e-06), ENSG00000262879(4.841e-06), ENSG00000263142(4.841e-06), ENSG00000266497(2.301e-06)	NA	NA	5_TxWk	GOSR2, KANSL1, LRR37A, KANSL1-AS1, ARL17B, LRR37A17P	GOSR2, KANSL1, LRR37A, KANSL1-AS1, ARL17B, LRR37A17P	NA	NA	
chr17	44,122,348	chr17:44122348	rs142920272, rs147317628	2.9e-07, 1.1e-06	ENSG00000108433(7.417e-05), ENSG00000120071(2.093e-15), ENSG00000176681(4.187e-13), ENSG00000214401(7.753e-08), ENSG00000228696(4.187e-13), ENSG00000238723(2.093e-15), ENSG00000262500(1.238e-14), ENSG00000262633(7.417e-05), ENSG00000262879(7.417e-05), ENSG00000263142(7.417e-05)	ENSG00000108433(4.841e-06), ENSG00000120071(1.708e-15), ENSG00000176681(1.036e-29), ENSG00000214401(8.406e-08), ENSG00000228696(1.036e-29), ENSG00000238723(1.708e-15), ENSG00000262500(5.602e-15), ENSG00000262633(4.841e-06), ENSG00000262879(4.841e-06), ENSG00000263142(4.841e-06), ENSG00000266497(2.301e-06)	NA	NA	5_TxWk	GOSR2, KANSL1, LRR37A, KANSL1-AS1, ARL17B, LRR37A17P	GOSR2, KANSL1, LRR37A, KANSL1-AS1, ARL17B, LRR37A17P	NA	NA	
chr17	44,122,354	chr17:44122354	rs147317628	1.1e-06	ENSG00000108433(7.417e-05), ENSG00000120071(2.093e-15), ENSG00000176681(4.187e-13), ENSG00000214401(7.753e-08), ENSG00000228696(4.187e-13), ENSG00000238723(2.093e-15), ENSG00000262500(1.238e-14), ENSG00000262633(7.417e-05), ENSG00000262879(7.417e-05), ENSG00000263142(7.417e-05)	ENSG00000108433(4.841e-06), ENSG00000120071(1.708e-15), ENSG00000176681(1.036e-29), ENSG00000214401(8.406e-08), ENSG00000228696(1.036e-29), ENSG00000238723(1.708e-15), ENSG00000262500(5.602e-15), ENSG00000262633(4.841e-06), ENSG00000262879(4.841e-06), ENSG00000263142(4.841e-06), ENSG00000266497(2.301e-06)	NA	NA	5_TxWk	GOSR2, KANSL1, LRR37A, KANSL1-AS1, ARL17B, LRR37A17P	GOSR2, KANSL1, LRR37A, KANSL1-AS1, ARL17B, LRR37A17P	NA	NA	
chr17	44,125,277	chr17:44125277	rs142920272, rs147317628	2.9e-07, 1.1e-06			ENSG00000120071		5_TxWk				KANSL1	
chr17	44,126,473	chr17:44126473	rs142920272, rs147317628	2.9e-07, 1.1e-06	ENSG00000108433(7.417e-05), ENSG00000120071(2.093e-15), ENSG00000176681(4.187e-13), ENSG00000214401(7.753e-08), ENSG00000228696(4.187e-13), ENSG00000238723(2.093e-15), ENSG00000262500(1.238e-14), ENSG00000262633(7.417e-05), ENSG00000262879(7.417e-05), ENSG00000263142(7.417e-05)	ENSG00000108433(4.841e-06), ENSG00000120071(1.708e-15), ENSG00000176681(1.036e-29), ENSG00000214401(8.406e-08), ENSG00000228696(1.036e-29), ENSG00000238723(1.708e-15), ENSG00000262500(5.602e-15), ENSG00000262633(4.841e-06), ENSG00000262879(4.841e-06), ENSG00000263142(4.841e-06), ENSG00000266497(2.301e-06)	NA	NA	5_TxWk	GOSR2, KANSL1, LRR37A, KANSL1-AS1, ARL17B, LRR37A17P	GOSR2, KANSL1, LRR37A, KANSL1-AS1, ARL17B, LRR37A17P	NA	NA	

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Supplementary Table 16 – continued from previous page

CHR	BP	CredSNP	IndexSNP	Index P	ENSGID_CP	ENSGID_GZ	ENSGID Promoter	ENSGID functional	Chrom HMM	Histone	HGNC_CP	HGNC_GZ	HGNC Promoter	HGNC functional
chr17	44,126,477	chr17:44126477	rs142920272	2.9e-07	ENSG00000108433(7.417e-05), ENSG00000120071(2.093e-15), ENSG00000176681(4.187e-13), ENSG00000214401(7.753e-08), ENSG00000228696(4.187e-13), ENSG00000238723(2.093e-15), ENSG00000262500(1.238e-14), ENSG00000262633(7.417e-05), ENSG00000262879(7.417e-05), ENSG00000263142(7.417e-05)	ENSG00000108433(4.841e-06), ENSG00000120071(1.708e-15), ENSG00000176681(1.036e-29), ENSG00000214401(8.406e-08), ENSG00000228696(1.036e-29), ENSG00000238723(1.708e-15), ENSG00000262500(5.602e-15), ENSG00000262633(4.841e-06), ENSG00000262879(4.841e-06), ENSG00000263142(4.841e-06), ENSG00000266497(2.301e-06)	NA	NA	5_TxWk	GOSR2, KANSL1, LRR37A, KANSL1-AS1, ARL17B, LRR37A17P	GOSR2, KANSL1, LRR37A, KANSL1-AS1, ARL17B, LRR37A17P	NA	NA	
chr17	44,126,478	chr17:44126478	rs142920272, rs147317628	2.9e-07, 1.1e-06	ENSG00000108433(7.417e-05), ENSG00000120071(2.093e-15), ENSG00000176681(4.187e-13), ENSG00000214401(7.753e-08), ENSG00000228696(4.187e-13), ENSG00000238723(2.093e-15), ENSG00000262500(1.238e-14), ENSG00000262633(7.417e-05), ENSG00000262879(7.417e-05), ENSG00000263142(7.417e-05)	ENSG00000108433(4.841e-06), ENSG00000120071(1.708e-15), ENSG00000176681(1.036e-29), ENSG00000214401(8.406e-08), ENSG00000228696(1.036e-29), ENSG00000238723(1.708e-15), ENSG00000262500(5.602e-15), ENSG00000262633(4.841e-06), ENSG00000262879(4.841e-06), ENSG00000263142(4.841e-06), ENSG00000266497(2.301e-06)	NA	NA	5_TxWk	GOSR2, KANSL1, LRR37A, KANSL1-AS1, ARL17B, LRR37A17P	GOSR2, KANSL1, LRR37A, KANSL1-AS1, ARL17B, LRR37A17P	NA	NA	
chr17	44,227,623	chr17:44227623	rs142920272, rs147317628	2.9e-07, 1.1e-06	ENSG00000267198(1.976e-05)	ENSG00000131484(1.08e-05), ENSG00000176681(1.083e-66), ENSG00000214425(1.08e-05), ENSG00000228696(1.083e-66), ENSG00000267198(5.005e-08)	NA	NA	5_TxWk			LRR37A, LRR37A4P, ARL17B	NA	NA
chr17	44,227,623	chr17:44227623m	rs142920272, rs147317628	2.9e-07, 1.1e-06	ENSG00000267198(1.976e-05)	ENSG00000131484(1.08e-05), ENSG00000176681(1.083e-66), ENSG00000214425(1.08e-05), ENSG00000228696(1.083e-66), ENSG00000267198(5.005e-08)	NA	NA	5_TxWk			LRR37A, LRR37A4P, ARL17B	NA	NA
chr17	44,228,169	chr17:44228169	rs142920272, rs147317628	2.9e-07, 1.1e-06	ENSG00000267198(1.976e-05)	ENSG00000131484(1.08e-05), ENSG00000176681(1.083e-66), ENSG00000214425(1.08e-05), ENSG00000228696(1.083e-66), ENSG00000267198(5.005e-08)	NA	NA	5_TxWk			LRR37A, LRR37A4P, ARL17B	NA	NA
chr17	44,228,529	chr17:44228529	rs142920272, rs147317628	2.9e-07, 1.1e-06	ENSG00000267198(1.976e-05)	ENSG00000131484(1.08e-05), ENSG00000176681(1.083e-66), ENSG00000214425(1.08e-05), ENSG00000228696(1.083e-66), ENSG00000267198(5.005e-08)	NA	NA	5_TxWk			LRR37A, LRR37A4P, ARL17B	NA	NA
chr17	44,228,609	chr17:44228609	rs147317628	1.1e-06	ENSG00000267198(1.976e-05)	ENSG00000131484(1.08e-05), ENSG00000176681(1.083e-66), ENSG00000214425(1.08e-05), ENSG00000228696(1.083e-66), ENSG00000267198(5.005e-08)	NA	NA	5_TxWk			LRR37A, LRR37A4P, ARL17B	NA	NA
chr17	44,228,770	chr17:44228770	rs147317628	1.1e-06	ENSG00000267198(1.976e-05)	ENSG00000131484(1.08e-05), ENSG00000176681(1.083e-66), ENSG00000214425(1.08e-05), ENSG00000228696(1.083e-66), ENSG00000267198(5.005e-08)	NA	NA	5_TxWk			LRR37A, LRR37A4P, ARL17B	NA	NA
chr17	44,230,647	chr17:44230647	rs142920272, rs147317628	2.9e-07, 1.1e-06	ENSG00000073969(6.506e-16), ENSG00000176681(6.285e-64), ENSG00000228696(6.285e-64), ENSG00000262879(1.715e-07), ENSG00000263142(1.622e-06)	ENSG00000073969(3.809e-94), ENSG00000131484(1.089e-13), ENSG00000176681(8.213e-05), ENSG00000214425(1.089e-13), ENSG00000225190(2.05e-06), ENSG00000228696(8.213e-05), ENSG00000264225(2.05e-06), ENSG00000266918(1.089e-13)	NA	NA	5_TxWk	NSF, LRR37A, ARL17B, LRR37A17P	NSF, LRR37A, LRR37A4P, PLEKHM1, ARL17B, RN7SL730P	NA	NA	
chr17	44,231,117	chr17:44231117	rs147317628	1.1e-06	ENSG00000073969(6.506e-16), ENSG00000176681(6.285e-64), ENSG00000228696(6.285e-64), ENSG00000262879(1.715e-07), ENSG00000263142(1.622e-06)	ENSG00000073969(3.809e-94), ENSG00000131484(1.089e-13), ENSG00000176681(8.213e-05), ENSG00000214425(1.089e-13), ENSG00000225190(2.05e-06), ENSG00000228696(8.213e-05), ENSG00000264225(2.05e-06), ENSG00000266918(1.089e-13)	NA	NA	5_TxWk	H3K4me1 NSF, LRR37A, ARL17B, LRR37A17P	NSF, LRR37A, LRR37A4P, PLEKHM1, ARL17B, RN7SL730P	NA	NA	

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Supplementary Table 16 – continued from previous page

CHR	BP	CredSNP	IndexSNP	Index P	ENSGID_CP	ENSGID_GZ	ENSGID Promoter	ENSGID functional	Chrom HMM	Histone	HGNC_CP	HGNC_GZ	HGNC Promoter	HGNC functional
chr17	44,231,295	chr17:44231295	rs142920272	2.9e-07	ENSG00000073969(6.506e-16), ENSG00000176681(6.285e-64), ENSG00000228696(6.285e-64), ENSG00000262879(1.715e-07), ENSG00000263142(1.622e-06)	ENSG00000073969(3.809e-94), ENSG00000131484(1.089e-13), ENSG00000176681(8.213e-05), ENSG00000214425(1.089e-13), ENSG00000225190(2.05e-06), ENSG00000228696(8.213e-05), ENSG00000264225(2.05e-06), ENSG00000266918(1.089e-13)	NA	NA	7_Enh	H3K4me1	NSF, LRRC37A, ARL17B, LRRC37A17P	NSF, LRRC37A, LRRC37A4P, PLEKHM1, ARL17B, RN7SL730P	NA	NA
chr17	44,231,326	chr17:44231326	rs147317628	1.1e-06	ENSG00000073969(6.506e-16), ENSG00000176681(6.285e-64), ENSG00000228696(6.285e-64), ENSG00000262879(1.715e-07), ENSG00000263142(1.622e-06)	ENSG00000073969(3.809e-94), ENSG00000131484(1.089e-13), ENSG00000176681(8.213e-05), ENSG00000214425(1.089e-13), ENSG00000225190(2.05e-06), ENSG00000228696(8.213e-05), ENSG00000264225(2.05e-06), ENSG00000266918(1.089e-13)	NA	NA	7_Enh		NSF, LRRC37A, ARL17B, LRRC37A17P	NSF, LRRC37A, LRRC37A4P, PLEKHM1, ARL17B, RN7SL730P	NA	NA
chr17	44,232,959	chr17:44232959	rs142920272, rs147317628	2.9e-07, 1.1e-06	ENSG00000073969(6.506e-16), ENSG00000176681(6.285e-64), ENSG00000228696(6.285e-64), ENSG00000262879(1.715e-07), ENSG00000263142(1.622e-06)	ENSG00000073969(3.809e-94), ENSG00000131484(1.089e-13), ENSG00000176681(8.213e-05), ENSG00000214425(1.089e-13), ENSG00000225190(2.05e-06), ENSG00000228696(8.213e-05), ENSG00000264225(2.05e-06), ENSG00000266918(1.089e-13)	NA	NA	5_TxWk		NSF, LRRC37A, ARL17B, LRRC37A17P	NSF, LRRC37A, LRRC37A4P, PLEKHM1, ARL17B, RN7SL730P	NA	NA
chr17	44,233,433	chr17:44233433	rs142920272	2.9e-07	ENSG00000073969(6.506e-16), ENSG00000176681(6.285e-64), ENSG00000228696(6.285e-64), ENSG00000262879(1.715e-07), ENSG00000263142(1.622e-06)	ENSG00000073969(3.809e-94), ENSG00000131484(1.089e-13), ENSG00000176681(8.213e-05), ENSG00000214425(1.089e-13), ENSG00000225190(2.05e-06), ENSG00000228696(8.213e-05), ENSG00000264225(2.05e-06), ENSG00000266918(1.089e-13)	NA	NA	5_TxWk		NSF, LRRC37A, ARL17B, LRRC37A17P	NSF, LRRC37A, LRRC37A4P, PLEKHM1, ARL17B, RN7SL730P	NA	NA
chr17	44,233,589	chr17:44233589	rs147317628	1.1e-06	ENSG00000073969(6.506e-16), ENSG00000176681(6.285e-64), ENSG00000228696(6.285e-64), ENSG00000262879(1.715e-07), ENSG00000263142(1.622e-06)	ENSG00000073969(3.809e-94), ENSG00000131484(1.089e-13), ENSG00000176681(8.213e-05), ENSG00000214425(1.089e-13), ENSG00000225190(2.05e-06), ENSG00000228696(8.213e-05), ENSG00000264225(2.05e-06), ENSG00000266918(1.089e-13)	NA	NA	5_TxWk		NSF, LRRC37A, ARL17B, LRRC37A17P	NSF, LRRC37A, LRRC37A4P, PLEKHM1, ARL17B, RN7SL730P	NA	NA
chr17	44,234,060	chr17:44234060	rs147317628	1.1e-06	ENSG00000073969(6.506e-16), ENSG00000176681(6.285e-64), ENSG00000228696(6.285e-64), ENSG00000262879(1.715e-07), ENSG00000263142(1.622e-06)	ENSG00000073969(3.809e-94), ENSG00000131484(1.089e-13), ENSG00000176681(8.213e-05), ENSG00000214425(1.089e-13), ENSG00000225190(2.05e-06), ENSG00000228696(8.213e-05), ENSG00000264225(2.05e-06), ENSG00000266918(1.089e-13)	NA	NA	5_TxWk		NSF, LRRC37A, ARL17B, LRRC37A17P	NSF, LRRC37A, LRRC37A4P, PLEKHM1, ARL17B, RN7SL730P	NA	NA
chr17	44,234,265	chr17:44234265	rs147317628	1.1e-06	ENSG00000073969(6.506e-16), ENSG00000176681(6.285e-64), ENSG00000228696(6.285e-64), ENSG00000262879(1.715e-07), ENSG00000263142(1.622e-06)	ENSG00000073969(3.809e-94), ENSG00000131484(1.089e-13), ENSG00000176681(8.213e-05), ENSG00000214425(1.089e-13), ENSG00000225190(2.05e-06), ENSG00000228696(8.213e-05), ENSG00000264225(2.05e-06), ENSG00000266918(1.089e-13)	NA	NA	5_TxWk		NSF, LRRC37A, ARL17B, LRRC37A17P	NSF, LRRC37A, LRRC37A4P, PLEKHM1, ARL17B, RN7SL730P	NA	NA
chr17	44,236,725	chr17:44236725	rs142920272, rs147317628	2.9e-07, 1.1e-06	ENSG00000073969(6.506e-16), ENSG00000176681(6.285e-64), ENSG00000228696(6.285e-64), ENSG00000262879(1.715e-07), ENSG00000263142(1.622e-06)	ENSG00000073969(3.809e-94), ENSG00000131484(1.089e-13), ENSG00000176681(8.213e-05), ENSG00000214425(1.089e-13), ENSG00000225190(2.05e-06), ENSG00000228696(8.213e-05), ENSG00000264225(2.05e-06), ENSG00000266918(1.089e-13)	NA	NA	5_TxWk		NSF, LRRC37A, ARL17B, LRRC37A17P	NSF, LRRC37A, LRRC37A4P, PLEKHM1, ARL17B, RN7SL730P	NA	NA
chr17	44,237,068	chr17:44237068	rs142920272	2.9e-07	ENSG00000073969(6.506e-16), ENSG00000176681(6.285e-64), ENSG00000228696(6.285e-64), ENSG00000262879(1.715e-07), ENSG00000263142(1.622e-06)	ENSG00000073969(3.809e-94), ENSG00000131484(1.089e-13), ENSG00000176681(8.213e-05), ENSG00000214425(1.089e-13), ENSG00000225190(2.05e-06), ENSG00000228696(8.213e-05), ENSG00000264225(2.05e-06), ENSG00000266918(1.089e-13)	NA	NA	5_TxWk		NSF, LRRC37A, ARL17B, LRRC37A17P	NSF, LRRC37A, LRRC37A4P, PLEKHM1, ARL17B, RN7SL730P	NA	NA

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Supplementary Table 16 – continued from previous page

CHR	BP	CredSNP	IndexSNP	Index P	ENSGID_CP	ENSGID_GZ	ENSGID Promoter	ENSGID functional	Chrom HMM	Histone	HGNC_CP	HGNC_GZ	HGNC Promoter	HGNC functional
chr17	44,237,372	chr17:44237372	rs142920272	2.9e-07	ENSG00000073969(6.506e-16), ENSG00000176681(6.285e-64), ENSG00000228696(6.285e-64), ENSG00000262879(1.715e-07), ENSG00000263142(1.622e-06)	ENSG00000073969(3.809e-94), ENSG00000131484(1.089e-13), ENSG00000176681(8.213e-05), ENSG00000214425(1.089e-13), ENSG00000225190(2.05e-06), ENSG00000228696(8.213e-05), ENSG00000264225(2.05e-06), ENSG00000266918(1.089e-13)	NA	NA	5_TxWk		NSF, LRRC37A, ARL17B, LRRC37A17P	NSF, LRRC37A, LRRC37A4P, PLEKHM1, ARL17B, RN7SL730P	NA	NA
chr17	44,237,790	chr17:44237790	rs142920272, rs147317628	2.9e-07, 1.1e-06	ENSG00000073969(6.506e-16), ENSG00000176681(6.285e-64), ENSG00000228696(6.285e-64), ENSG00000262879(1.715e-07), ENSG00000263142(1.622e-06)	ENSG00000073969(3.809e-94), ENSG00000131484(1.089e-13), ENSG00000176681(8.213e-05), ENSG00000214425(1.089e-13), ENSG00000225190(2.05e-06), ENSG00000228696(8.213e-05), ENSG00000264225(2.05e-06), ENSG00000266918(1.089e-13)	NA	NA	5_TxWk		NSF, LRRC37A, ARL17B, LRRC37A17P	NSF, LRRC37A, LRRC37A4P, ARL17B, RN7SL730P	NA	NA
chr17	44,238,130	chr17:44238130	rs142920272, rs147317628	2.9e-07, 1.1e-06	ENSG00000073969(6.506e-16), ENSG00000176681(6.285e-64), ENSG00000228696(6.285e-64), ENSG00000262879(1.715e-07), ENSG00000263142(1.622e-06)	ENSG00000073969(3.809e-94), ENSG00000131484(1.089e-13), ENSG00000176681(8.213e-05), ENSG00000214425(1.089e-13), ENSG00000225190(2.05e-06), ENSG00000228696(8.213e-05), ENSG00000264225(2.05e-06), ENSG00000266918(1.089e-13)	NA	NA	5_TxWk		NSF, LRRC37A, ARL17B, LRRC37A17P	NSF, LRRC37A, LRRC37A4P, PLEKHM1, ARL17B, RN7SL730P	NA	NA
chr17	44,238,424	chr17:44238424	rs142920272, rs147317628	2.9e-07, 1.1e-06	ENSG00000073969(6.506e-16), ENSG00000176681(6.285e-64), ENSG00000228696(6.285e-64), ENSG00000262879(1.715e-07), ENSG00000263142(1.622e-06)	ENSG00000073969(3.809e-94), ENSG00000131484(1.089e-13), ENSG00000176681(8.213e-05), ENSG00000214425(1.089e-13), ENSG00000225190(2.05e-06), ENSG00000228696(8.213e-05), ENSG00000264225(2.05e-06), ENSG00000266918(1.089e-13)	NA	NA	5_TxWk		NSF, LRRC37A, ARL17B, LRRC37A17P	NSF, LRRC37A, LRRC37A4P, PLEKHM1, ARL17B, RN7SL730P	NA	NA
chr17	44,238,571	chr17:44238571	rs142920272, rs147317628	2.9e-07, 1.1e-06	ENSG00000073969(6.506e-16), ENSG00000176681(6.285e-64), ENSG00000228696(6.285e-64), ENSG00000262879(1.715e-07), ENSG00000263142(1.622e-06)	ENSG00000073969(3.809e-94), ENSG00000131484(1.089e-13), ENSG00000176681(8.213e-05), ENSG00000214425(1.089e-13), ENSG00000225190(2.05e-06), ENSG00000228696(8.213e-05), ENSG00000264225(2.05e-06), ENSG00000266918(1.089e-13)	NA	NA	5_TxWk		NSF, LRRC37A, ARL17B, LRRC37A17P	NSF, LRRC37A, LRRC37A4P, PLEKHM1, ARL17B, RN7SL730P	NA	NA
chr17	44,239,958	chr17:44239958	rs142920272	2.9e-07	ENSG00000073969(6.506e-16), ENSG00000176681(6.285e-64), ENSG00000228696(6.285e-64), ENSG00000262879(1.715e-07), ENSG00000263142(1.622e-06)	ENSG00000073969(3.809e-94), ENSG00000131484(1.089e-13), ENSG00000176681(8.213e-05), ENSG00000214425(1.089e-13), ENSG00000225190(2.05e-06), ENSG00000228696(8.213e-05), ENSG00000264225(2.05e-06), ENSG00000266918(1.089e-13)	NA	NA	5_TxWk		NSF, LRRC37A, ARL17B, LRRC37A17P	NSF, LRRC37A, LRRC37A4P, ARL17B, RN7SL730P	NA	NA
chr17	44,241,304	chr17:44241304	rs147317628	1.1e-06	ENSG00000176681(0), ENSG00000228696(0), ENSG00000260075(8.89e-10)	ENSG00000176681(2.828e-09), ENSG00000228696(2.597e-31), ENSG00000238083(1.646e-06), ENSG00000260075(3.47e-99), ENSG00000266497(1.646e-06)	NA	NA	5_TxWk		LRRC37A, ARL17B, NSFP1	LRRC37A, ARL17B, LRRC37A2, NSFP1	NA	NA
chr17	44,242,536	chr17:44242536	rs142920272, rs147317628	2.9e-07, 1.1e-06	ENSG00000176681(0), ENSG00000228696(0), ENSG00000260075(8.89e-10)	ENSG00000176681(2.828e-09), ENSG00000228696(2.597e-31), ENSG00000238083(1.646e-06), ENSG00000260075(3.47e-99), ENSG00000266497(1.646e-06)	NA	NA	5_TxWk		LRRC37A, ARL17B, NSFP1	LRRC37A, ARL17B, LRRC37A2, NSFP1	NA	NA
chr17	44,242,574	chr17:44242574	rs142920272, rs147317628	2.9e-07, 1.1e-06	ENSG00000176681(0), ENSG00000228696(0), ENSG00000260075(8.89e-10)	ENSG00000176681(2.828e-09), ENSG00000228696(2.597e-31), ENSG00000238083(1.646e-06), ENSG00000260075(3.47e-99), ENSG00000266497(1.646e-06)	NA	NA	5_TxWk		LRRC37A, ARL17B, NSFP1	LRRC37A, ARL17B, LRRC37A2, NSFP1	NA	NA
chr17	44,242,788	chr17:44242788	rs142920272, rs147317628	2.9e-07, 1.1e-06	ENSG00000176681(0), ENSG00000228696(0), ENSG00000260075(8.89e-10)	ENSG00000176681(2.828e-09), ENSG00000228696(2.597e-31), ENSG00000238083(1.646e-06), ENSG00000260075(3.47e-99), ENSG00000266497(1.646e-06)	NA	NA	5_TxWk		LRRC37A, ARL17B, NSFP1	LRRC37A, ARL17B, LRRC37A2, NSFP1	NA	NA

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Supplementary Table 16 – continued from previous page

CHR	BP	CredSNP	IndexSNP	Index P	ENSGID_CP	ENSGID_GZ	ENSGID Promoter	ENSGID functional	Chrom HMM	Histone	HGNC_CP	HGNC_GZ	HGNC Promoter	HGNC functional
chr17	44,243,543	chr17:44243543	rs142920272, rs147317628	2.9e-07, 1.1e-06	ENSG00000176681(0), ENSG00000228696(0), ENSG00000260075(8.89e-10)	ENSG00000176681(2.828e-09), ENSG00000228696(2.597e-31), ENSG00000238083(1.646e-06), ENSG00000260075(3.47e-99), ENSG00000266497(1.646e-06)	NA	NA	5_TxWk		LRRC37A, ARL17B, NSFP1	LRRC37A, ARL17B, LRRC37A2, NSFP1	NA	NA
chr17	44,243,979	chr17:44243979	rs142920272	2.9e-07	ENSG00000176681(0), ENSG00000228696(0), ENSG00000260075(8.89e-10)	ENSG00000176681(2.828e-09), ENSG00000228696(2.597e-31), ENSG00000238083(1.646e-06), ENSG00000260075(3.47e-99), ENSG00000266497(1.646e-06)	NA	NA	7_Enh	H3K4me1	LRRC37A, ARL17B, NSFP1	LRRC37A, ARL17B, LRRC37A2, NSFP1	NA	NA
chr17	44,244,397	chr17:44244397	rs142920272	2.9e-07	ENSG00000176681(0), ENSG00000228696(0), ENSG00000260075(8.89e-10)	ENSG00000176681(2.828e-09), ENSG00000228696(2.597e-31), ENSG00000238083(1.646e-06), ENSG00000260075(3.47e-99), ENSG00000266497(1.646e-06)	NA	NA	7_Enh		LRRC37A, ARL17B, NSFP1	LRRC37A, ARL17B, LRRC37A2, NSFP1	NA	NA
chr17	44,244,581	chr17:44244581	rs147317628	1.1e-06	ENSG00000176681(0), ENSG00000228696(0), ENSG00000260075(8.89e-10)	ENSG00000176681(2.828e-09), ENSG00000228696(2.597e-31), ENSG00000238083(1.646e-06), ENSG00000260075(3.47e-99), ENSG00000266497(1.646e-06)	NA	NA	5_TxWk		LRRC37A, ARL17B, NSFP1	LRRC37A, ARL17B, LRRC37A2, NSFP1	NA	NA
chr17	44,245,766	chr17:44245766	rs142920272, rs147317628	2.9e-07, 1.1e-06	ENSG00000176681(0), ENSG00000228696(0), ENSG00000260075(8.89e-10)	ENSG00000176681(2.828e-09), ENSG00000228696(2.597e-31), ENSG00000238083(1.646e-06), ENSG00000260075(3.47e-99), ENSG00000266497(1.646e-06)	NA	NA	5_TxWk		LRRC37A, ARL17B, NSFP1	LRRC37A, ARL17B, LRRC37A2, NSFP1	NA	NA
chr17	44,245,876	chr17:44245876	rs147317628	1.1e-06	ENSG00000176681(0), ENSG00000228696(0), ENSG00000260075(8.89e-10)	ENSG00000176681(2.828e-09), ENSG00000228696(2.597e-31), ENSG00000238083(1.646e-06), ENSG00000260075(3.47e-99), ENSG00000266497(1.646e-06)	NA	NA	5_TxWk		LRRC37A, ARL17B, NSFP1	LRRC37A, ARL17B, LRRC37A2, NSFP1	NA	NA
chr17	44,246,405	chr17:44246405	rs142920272, rs147317628	2.9e-07, 1.1e-06	ENSG00000176681(0), ENSG00000228696(0), ENSG00000260075(8.89e-10)	ENSG00000176681(2.828e-09), ENSG00000228696(2.597e-31), ENSG00000238083(1.646e-06), ENSG00000260075(3.47e-99), ENSG00000266497(1.646e-06)	NA	NA	5_TxWk		LRRC37A, ARL17B, NSFP1	LRRC37A, ARL17B, LRRC37A2, NSFP1	NA	NA
chr17	44,246,997	chr17:44246997	rs142920272, rs147317628	2.9e-07, 1.1e-06	ENSG00000176681(0), ENSG00000228696(0), ENSG00000260075(8.89e-10)	ENSG00000176681(2.828e-09), ENSG00000228696(2.597e-31), ENSG00000238083(1.646e-06), ENSG00000260075(3.47e-99), ENSG00000266497(1.646e-06)	NA	NA	7_Enh		LRRC37A, ARL17B, NSFP1	LRRC37A, ARL17B, LRRC37A2, NSFP1	NA	NA
chr17	44,247,314	chr17:44247314	rs142920272, rs147317628	2.9e-07, 1.1e-06	ENSG00000176681(0), ENSG00000228696(0), ENSG00000260075(8.89e-10)	ENSG00000176681(2.828e-09), ENSG00000228696(2.597e-31), ENSG00000238083(1.646e-06), ENSG00000260075(3.47e-99), ENSG00000266497(1.646e-06)	NA	NA	7_Enh	H3K4me1	LRRC37A, ARL17B, NSFP1	LRRC37A, ARL17B, LRRC37A2, NSFP1	NA	NA
chr17	44,248,042	chr17:44248042	rs142920272, rs147317628	2.9e-07, 1.1e-06	ENSG00000176681(0), ENSG00000228696(0), ENSG00000260075(8.89e-10)	ENSG00000176681(2.828e-09), ENSG00000228696(2.597e-31), ENSG00000238083(1.646e-06), ENSG00000260075(3.47e-99), ENSG00000266497(1.646e-06)	NA	NA	6_EnhG	H3K36me3	LRRC37A, ARL17B, NSFP1	LRRC37A, ARL17B, LRRC37A2, NSFP1	NA	NA
chr17	44,248,814	chr17:44248814	rs142920272, rs147317628	2.9e-07, 1.1e-06			ENSG00000120071		5_TxWk				KANSL1	
chr17	44,249,800	chr17:44249800	rs142920272, rs147317628	2.9e-07, 1.1e-06			ENSG00000120071		5_TxWk	DHS:H3K4me1			KANSL1	
chr17	44,250,616	chr17:44250616	rs142920272	2.9e-07			ENSG00000120071		5_TxWk				KANSL1	
chr17	44,250,669	chr17:44250669	rs142920272, rs147317628	2.9e-07, 1.1e-06			ENSG00000120071		5_TxWk				KANSL1	
chr17	44,251,548	chr17:44251548	rs147317628	1.1e-06			ENSG00000120071		5_TxWk	H3K4me1			KANSL1	
chr17	44,251,582	chr17:44251582	rs142920272, rs147317628	2.9e-07, 1.1e-06			ENSG00000120071		5_TxWk	H3K4me1			KANSL1	
chr17	44,251,698	chr17:44251698	rs142920272	2.9e-07	ENSG00000004897(3.479e-05), ENSG00000214425(0), ENSG00000267198(0), ENSG00000267246(0)	ENSG00000073969(9.158e-12), ENSG00000120071(2.834e-05), ENSG00000131484(1.201e-19), ENSG00000186868(2.834e-05), ENSG00000214425(1.201e-19), ENSG00000228696(1.46e-32), ENSG00000260075(1.46e-32), ENSG00000266918(1.201e-19)	NA	NA	5_TxWk	H3K4me1	CDC27, LRRC37A4P	NSF, KANSL1, MAPT, LRRC37A4P, ARL17B, NSFP1	NA	NA

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Supplementary Table 16 – continued from previous page

CHR	BP	CredSNP	IndexSNP	Index P	ENSGID_CP	ENSGID_GZ	ENSGID Promoter	ENSGID functional	Chrom HMM	Histone	HGNC_CP	HGNC_GZ	HGNC Promoter	HGNC functional
chr17	44,251,907	chr17:44251907	rs142920272, rs147317628	2.9e-07, 1.1e-06	ENSG00000004897(3.479e-05), ENSG00000214425(0), ENSG00000267198(0), ENSG00000267246(0)	ENSG00000073969(9.158e-12), ENSG00000120071(2.834e-05), ENSG00000131484(1.201e-19), ENSG00000186868(2.834e-05), ENSG00000214425(1.201e-19), ENSG00000228696(1.46e-32), ENSG00000260075(1.46e-32), ENSG00000266918(1.201e-19)	NA	NA	5_TxWk		CDC27, LRRC37A4P	NSF, KANSL1, MAPT, LRRC37A4P, ARL17B, NSFP1	NA	NA
chr17	44,251,972	chr17:44251972	rs142920272, rs147317628	2.9e-07, 1.1e-06	ENSG00000004897(3.479e-05), ENSG00000214425(0), ENSG00000267198(0), ENSG00000267246(0)	ENSG00000073969(9.158e-12), ENSG00000120071(2.834e-05), ENSG00000131484(1.201e-19), ENSG00000186868(2.834e-05), ENSG00000214425(1.201e-19), ENSG00000228696(1.46e-32), ENSG00000260075(1.46e-32), ENSG00000266918(1.201e-19)	NA	NA	5_TxWk		CDC27, LRRC37A4P	NSF, KANSL1, MAPT, LRRC37A4P, ARL17B, NSFP1	NA	NA
chr17	44,252,098	chr17:44252098	rs147317628	1.1e-06	ENSG00000004897(3.479e-05), ENSG00000214425(0), ENSG00000267198(0), ENSG00000267246(0)	ENSG00000073969(9.158e-12), ENSG00000120071(2.834e-05), ENSG00000131484(1.201e-19), ENSG00000186868(2.834e-05), ENSG00000214425(1.201e-19), ENSG00000228696(1.46e-32), ENSG00000260075(1.46e-32), ENSG00000266918(1.201e-19)	NA	NA	5_TxWk		CDC27, LRRC37A4P	NSF, KANSL1, MAPT, LRRC37A4P, ARL17B, NSFP1	NA	NA
chr17	44,252,197	chr17:44252197	rs147317628	1.1e-06	ENSG00000004897(3.479e-05), ENSG00000214425(0), ENSG00000267198(0), ENSG00000267246(0)	ENSG00000073969(9.158e-12), ENSG00000120071(2.834e-05), ENSG00000131484(1.201e-19), ENSG00000186868(2.834e-05), ENSG00000214425(1.201e-19), ENSG00000228696(1.46e-32), ENSG00000260075(1.46e-32), ENSG00000266918(1.201e-19)	NA	NA	5_TxWk		CDC27, LRRC37A4P	NSF, KANSL1, MAPT, LRRC37A4P, ARL17B, NSFP1	NA	NA
chr17	44,252,416	chr17:44252416	rs142920272, rs147317628	2.9e-07, 1.1e-06	ENSG00000004897(3.479e-05), ENSG00000214425(0), ENSG00000267198(0), ENSG00000267246(0)	ENSG00000073969(9.158e-12), ENSG00000120071(2.834e-05), ENSG00000131484(1.201e-19), ENSG00000186868(2.834e-05), ENSG00000214425(1.201e-19), ENSG00000228696(1.46e-32), ENSG00000260075(1.46e-32), ENSG00000266918(1.201e-19)	NA	NA	5_TxWk		CDC27, LRRC37A4P	NSF, KANSL1, MAPT, LRRC37A4P, ARL17B, NSFP1	NA	NA
chr17	44,253,203	chr17:44253203	rs142920272, rs147317628	2.9e-07, 1.1e-06	ENSG00000004897(3.479e-05), ENSG00000214425(0), ENSG00000267198(0), ENSG00000267246(0)	ENSG00000073969(9.158e-12), ENSG00000120071(2.834e-05), ENSG00000131484(1.201e-19), ENSG00000186868(2.834e-05), ENSG00000214425(1.201e-19), ENSG00000228696(1.46e-32), ENSG00000260075(1.46e-32), ENSG00000266918(1.201e-19)	NA	NA	5_TxWk		CDC27, LRRC37A4P	NSF, KANSL1, MAPT, LRRC37A4P, ARL17B, NSFP1	NA	NA
chr17	44,254,379	chr17:44254379	rs142920272	2.9e-07	ENSG00000004897(3.479e-05), ENSG00000214425(0), ENSG00000267198(0), ENSG00000267246(0)	ENSG00000073969(9.158e-12), ENSG00000120071(2.834e-05), ENSG00000131484(1.201e-19), ENSG00000186868(2.834e-05), ENSG00000214425(1.201e-19), ENSG00000228696(1.46e-32), ENSG00000260075(1.46e-32), ENSG00000266918(1.201e-19)	NA	NA	5_TxWk		CDC27, LRRC37A4P	NSF, KANSL1, MAPT, LRRC37A4P, ARL17B, NSFP1	NA	NA
chr17	44,254,494	chr17:44254494	rs147317628	1.1e-06	ENSG00000004897(3.479e-05), ENSG00000214425(0), ENSG00000267198(0), ENSG00000267246(0)	ENSG00000073969(9.158e-12), ENSG00000120071(2.834e-05), ENSG00000131484(1.201e-19), ENSG00000186868(2.834e-05), ENSG00000214425(1.201e-19), ENSG00000228696(1.46e-32), ENSG00000260075(1.46e-32), ENSG00000266918(1.201e-19)	NA	NA	5_TxWk		CDC27, LRRC37A4P	NSF, KANSL1, MAPT, LRRC37A4P, ARL17B, NSFP1	NA	NA
chr17	44,254,686	chr17:44254686	rs142920272, rs147317628	2.9e-07, 1.1e-06	ENSG00000004897(3.479e-05), ENSG00000214425(0), ENSG00000267198(0), ENSG00000267246(0)	ENSG00000073969(9.158e-12), ENSG00000120071(2.834e-05), ENSG00000131484(1.201e-19), ENSG00000186868(2.834e-05), ENSG00000214425(1.201e-19), ENSG00000228696(1.46e-32), ENSG00000260075(1.46e-32), ENSG00000266918(1.201e-19)	NA	NA	5_TxWk		CDC27, LRRC37A4P	NSF, KANSL1, MAPT, LRRC37A4P, ARL17B, NSFP1	NA	NA

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Supplementary Table 16 – continued from previous page

CHR	BP	CredSNP	IndexSNP	Index P	ENSGID_CP	ENSGID_GZ	ENSGID Promoter	ENSGID functional	Chrom HMM	Histone	HGNC_CP	HGNC_GZ	HGNC Promoter	HGNC functional
chr17	44,254,993	chr17:44254993	rs142920272	2.9e-07	ENSG00000004897(3.479e-05), ENSG00000214425(0), ENSG00000267198(0), ENSG00000267246(0)	ENSG00000073969(9.158e-12), ENSG00000120071(2.834e-05), ENSG00000131484(1.201e-19), ENSG00000186868(2.834e-05), ENSG00000214425(1.201e-19), ENSG00000228696(1.46e-32), ENSG00000260075(1.46e-32), ENSG00000266918(1.201e-19)	NA	NA	5_TxWk		CDC27, LRR37A4P	NSF, KANSL1, MAPT, LRR37A4P, ARL17B, NSFP1	NA	NA
chr17	44,255,532	chr17:44255532	rs147317628	1.1e-06	ENSG00000004897(3.479e-05), ENSG00000214425(0), ENSG00000267198(0), ENSG00000267246(0)	ENSG00000073969(9.158e-12), ENSG00000120071(2.834e-05), ENSG00000131484(1.201e-19), ENSG00000186868(2.834e-05), ENSG00000214425(1.201e-19), ENSG00000228696(1.46e-32), ENSG00000260075(1.46e-32), ENSG00000266918(1.201e-19)	NA	NA	5_TxWk		CDC27, LRR37A4P	NSF, KANSL1, MAPT, LRR37A4P, ARL17B, NSFP1	NA	NA
chr17	44,255,777	chr17:44255777	rs142920272, rs147317628	2.9e-07, 1.1e-06	ENSG00000004897(3.479e-05), ENSG00000214425(0), ENSG00000267198(0), ENSG00000267246(0)	ENSG00000073969(9.158e-12), ENSG00000120071(2.834e-05), ENSG00000131484(1.201e-19), ENSG00000186868(2.834e-05), ENSG00000214425(1.201e-19), ENSG00000228696(1.46e-32), ENSG00000260075(1.46e-32), ENSG00000266918(1.201e-19)	NA	NA	5_TxWk		CDC27, LRR37A4P	NSF, KANSL1, MAPT, LRR37A4P, ARL17B, NSFP1	NA	NA
chr17	44,257,783	chr17:44257783	rs142920272	2.9e-07	ENSG00000004897(3.479e-05), ENSG00000214425(0), ENSG00000267198(0), ENSG00000267246(0)	ENSG00000073969(9.158e-12), ENSG00000120071(2.834e-05), ENSG00000131484(1.201e-19), ENSG00000186868(2.834e-05), ENSG00000214425(1.201e-19), ENSG00000228696(1.46e-32), ENSG00000260075(1.46e-32), ENSG00000266918(1.201e-19)	NA	NA	5_TxWk		CDC27, LRR37A4P	NSF, KANSL1, MAPT, LRR37A4P, ARL17B, NSFP1	NA	NA
chr17	44,257,788	chr17:44257788	rs147317628	1.1e-06	ENSG00000004897(3.479e-05), ENSG00000214425(0), ENSG00000267198(0), ENSG00000267246(0)	ENSG00000073969(9.158e-12), ENSG00000120071(2.834e-05), ENSG00000131484(1.201e-19), ENSG00000186868(2.834e-05), ENSG00000214425(1.201e-19), ENSG00000228696(1.46e-32), ENSG00000260075(1.46e-32), ENSG00000266918(1.201e-19)	NA	NA	5_TxWk		CDC27, LRR37A4P	NSF, KANSL1, MAPT, LRR37A4P, ARL17B, NSFP1	NA	NA
chr17	44,258,354	chr17:44258354	rs142920272, rs147317628	2.9e-07, 1.1e-06	ENSG00000004897(3.479e-05), ENSG00000214425(0), ENSG00000267198(0), ENSG00000267246(0)	ENSG00000073969(9.158e-12), ENSG00000120071(2.834e-05), ENSG00000131484(1.201e-19), ENSG00000186868(2.834e-05), ENSG00000214425(1.201e-19), ENSG00000228696(1.46e-32), ENSG00000260075(1.46e-32), ENSG00000266918(1.201e-19)	NA	NA	5_TxWk		CDC27, LRR37A4P	NSF, KANSL1, MAPT, LRR37A4P, ARL17B, NSFP1	NA	NA
chr17	44,258,422	chr17:44258422	rs142920272, rs147317628	2.9e-07, 1.1e-06	ENSG00000004897(3.479e-05), ENSG00000214425(0), ENSG00000267198(0), ENSG00000267246(0)	ENSG00000073969(9.158e-12), ENSG00000120071(2.834e-05), ENSG00000131484(1.201e-19), ENSG00000186868(2.834e-05), ENSG00000214425(1.201e-19), ENSG00000228696(1.46e-32), ENSG00000260075(1.46e-32), ENSG00000266918(1.201e-19)	NA	NA	5_TxWk		CDC27, LRR37A4P	NSF, KANSL1, MAPT, LRR37A4P, ARL17B, NSFP1	NA	NA
chr17	44,258,954	chr17:44258954	rs142920272, rs147317628	2.9e-07, 1.1e-06	ENSG00000004897(3.479e-05), ENSG00000214425(0), ENSG00000267198(0), ENSG00000267246(0)	ENSG00000073969(9.158e-12), ENSG00000120071(2.834e-05), ENSG00000131484(1.201e-19), ENSG00000186868(2.834e-05), ENSG00000214425(1.201e-19), ENSG00000228696(1.46e-32), ENSG00000260075(1.46e-32), ENSG00000266918(1.201e-19)	NA	NA	5_TxWk		CDC27, LRR37A4P	NSF, KANSL1, MAPT, LRR37A4P, ARL17B, NSFP1	NA	NA
chr17	44,259,040	chr17:44259040	rs142920272	2.9e-07	ENSG00000004897(3.479e-05), ENSG00000214425(0), ENSG00000267198(0), ENSG00000267246(0)	ENSG00000073969(9.158e-12), ENSG00000120071(2.834e-05), ENSG00000131484(1.201e-19), ENSG00000186868(2.834e-05), ENSG00000214425(1.201e-19), ENSG00000228696(1.46e-32), ENSG00000260075(1.46e-32), ENSG00000266918(1.201e-19)	NA	NA	5_TxWk		CDC27, LRR37A4P	NSF, KANSL1, MAPT, LRR37A4P, ARL17B, NSFP1	NA	NA

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Supplementary Table 16 – continued from previous page

CHR	BP	CredSNP	IndexSNP	Index P	ENSGID_CP	ENSGID_GZ	ENSGID Promoter	ENSGID functional	Chrom HMM	Histone	HGNC_CP	HGNC_GZ	HGNC Promoter	HGNC functional
chr17	44,259,792	chr17:44259792	rs142920272, rs147317628	2.9e-07, 1.1e-06	ENSG0000004897(3.479e-05), ENSG00000214425(0), ENSG00000267198(0), ENSG00000267246(0)	ENSG00000073969(9.158e-12), ENSG00000120071(2.834e-05), ENSG00000131484(1.201e-19), ENSG00000186868(2.834e-05), ENSG00000214425(1.201e-19), ENSG00000228696(1.46e-32), ENSG00000260075(1.46e-32), ENSG00000266918(1.201e-19)	NA	NA	5_TxWk		CDC27, LRR37A4P	NSF, KANSL1, MAPT, LRR37A4P, ARL17B, NSFP1	NA	NA
chr17	44,260,416	chr17:44260416	rs142920272	2.9e-07	ENSG00000120071(9.613e-15), ENSG00000186868(1.943e-06), ENSG00000228696(1.763e-05), ENSG00000260075(1.763e-05), ENSG00000262372(2.229e-05)	ENSG00000131484(6.359e-16), ENSG00000176681(0), ENSG00000185829(4.033e-101), ENSG00000214425(6.359e-16), ENSG00000228696(0), ENSG00000238083(4.033e-101), ENSG00000262879(5.357e-06)	NA	NA	5_TxWk	H3K4me1	KANSL1, MAPT, ARL17B, NSFP1	LRR37A, ARL17A, LRR37A4P, ARL17B, LRR37A2	NA	NA
chr17	44,260,939	chr17:44260939	rs147317628	1.1e-06	ENSG00000120071(9.613e-15), ENSG00000186868(1.943e-06), ENSG00000228696(1.763e-05), ENSG00000260075(1.763e-05), ENSG00000262372(2.229e-05)	ENSG00000131484(6.359e-16), ENSG00000176681(0), ENSG00000185829(4.033e-101), ENSG00000214425(6.359e-16), ENSG00000228696(0), ENSG00000238083(4.033e-101), ENSG00000262879(5.357e-06)	NA	NA	7_Enh	H3K4me1	KANSL1, MAPT, ARL17B, NSFP1	LRR37A, ARL17A, LRR37A4P, ARL17B, LRR37A2	NA	NA
chr17	44,260,967	chr17:44260967	rs142920272, rs147317628	2.9e-07, 1.1e-06	ENSG00000120071(9.613e-15), ENSG00000186868(1.943e-06), ENSG00000228696(1.763e-05), ENSG00000260075(1.763e-05), ENSG00000262372(2.229e-05)	ENSG00000131484(6.359e-16), ENSG00000176681(0), ENSG00000185829(4.033e-101), ENSG00000214425(6.359e-16), ENSG00000228696(0), ENSG00000238083(4.033e-101), ENSG00000262879(5.357e-06)	NA	NA	7_Enh	H3K4me1	KANSL1, MAPT, ARL17B, NSFP1	LRR37A, ARL17A, LRR37A4P, ARL17B, LRR37A2	NA	NA
chr17	44,261,613	chr17:44261613	rs142920272, rs147317628	2.9e-07, 1.1e-06	ENSG00000120071(9.613e-15), ENSG00000186868(1.943e-06), ENSG00000228696(1.763e-05), ENSG00000260075(1.763e-05), ENSG00000262372(2.229e-05)	ENSG00000131484(6.359e-16), ENSG00000176681(0), ENSG00000185829(4.033e-101), ENSG00000214425(6.359e-16), ENSG00000228696(0), ENSG00000238083(4.033e-101), ENSG00000262879(5.357e-06)	NA	NA	7_Enh	DHS	KANSL1, MAPT, ARL17B, NSFP1	LRR37A, ARL17A, LRR37A4P, ARL17B, LRR37A2	NA	NA
chr17	44,261,753	chr17:44261753	rs147317628	1.1e-06	ENSG00000120071(9.613e-15), ENSG00000186868(1.943e-06), ENSG00000228696(1.763e-05), ENSG00000260075(1.763e-05), ENSG00000262372(2.229e-05)	ENSG00000131484(6.359e-16), ENSG00000176681(0), ENSG00000185829(4.033e-101), ENSG00000214425(6.359e-16), ENSG00000228696(0), ENSG00000238083(4.033e-101), ENSG00000262879(5.357e-06)	NA	NA	7_Enh		KANSL1, MAPT, ARL17B, NSFP1	LRR37A, ARL17A, LRR37A4P, ARL17B, LRR37A2	NA	NA
chr17	44,262,203	chr17:44262203	rs142920272, rs147317628	2.9e-07, 1.1e-06	ENSG00000120071(9.613e-15), ENSG00000186868(1.943e-06), ENSG00000228696(1.763e-05), ENSG00000260075(1.763e-05), ENSG00000262372(2.229e-05)	ENSG00000131484(6.359e-16), ENSG00000176681(0), ENSG00000185829(4.033e-101), ENSG00000214425(6.359e-16), ENSG00000228696(0), ENSG00000238083(4.033e-101), ENSG00000262879(5.357e-06)	NA	NA	7_Enh	H3K4me1	KANSL1, MAPT, ARL17B, NSFP1	LRR37A, ARL17A, LRR37A4P, ARL17B, LRR37A2	NA	NA
chr17	44,262,403	chr17:44262403	rs142920272, rs147317628	2.9e-07, 1.1e-06	ENSG00000120071(9.613e-15), ENSG00000186868(1.943e-06), ENSG00000228696(1.763e-05), ENSG00000260075(1.763e-05), ENSG00000262372(2.229e-05)	ENSG00000131484(6.359e-16), ENSG00000176681(0), ENSG00000185829(4.033e-101), ENSG00000214425(6.359e-16), ENSG00000228696(0), ENSG00000238083(4.033e-101), ENSG00000262879(5.357e-06)	NA	NA	7_Enh	H3K4me1	KANSL1, MAPT, ARL17B, NSFP1	LRR37A, ARL17A, LRR37A4P, ARL17B, LRR37A2	NA	NA
chr17	44,262,418	chr17:44262418	rs142920272	2.9e-07	ENSG00000120071(9.613e-15), ENSG00000186868(1.943e-06), ENSG00000228696(1.763e-05), ENSG00000260075(1.763e-05), ENSG00000262372(2.229e-05)	ENSG00000131484(6.359e-16), ENSG00000176681(0), ENSG00000185829(4.033e-101), ENSG00000214425(6.359e-16), ENSG00000228696(0), ENSG00000238083(4.033e-101), ENSG00000262879(5.357e-06)	NA	NA	7_Enh	H3K4me1	KANSL1, MAPT, ARL17B, NSFP1	LRR37A, ARL17A, LRR37A4P, ARL17B, LRR37A2	NA	NA
chr17	44,263,022	chr17:44263022	rs142920272	2.9e-07	ENSG00000120071(9.613e-15), ENSG00000186868(1.943e-06), ENSG00000228696(1.763e-05), ENSG00000260075(1.763e-05), ENSG00000262372(2.229e-05)	ENSG00000131484(6.359e-16), ENSG00000176681(0), ENSG00000185829(4.033e-101), ENSG00000214425(6.359e-16), ENSG00000228696(0), ENSG00000238083(4.033e-101), ENSG00000262879(5.357e-06)	NA	NA	7_Enh	H3K4me1	KANSL1, MAPT, ARL17B, NSFP1	LRR37A, ARL17A, LRR37A4P, ARL17B, LRR37A2	NA	NA

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Supplementary Table 16 – continued from previous page

CHR	BP	CredSNP	IndexSNP	Index P	ENSGID_CP	ENSGID_GZ	ENSGID Promoter	ENSGID functional	Chrom HMM	Histone	HGNC_CP	HGNC_GZ	HGNC Promoter	HGNC functional
chr17	44,263,341	chr17:44263341	rs142920272, rs147317628	2.9e-07, 1.1e-06	ENSG00000120071(9.613e-15), ENSG00000186868(1.943e-06), ENSG00000228696(1.763e-05), ENSG00000260075(1.763e-05), ENSG00000262372(2.229e-05)	ENSG00000131484(6.359e-16), ENSG00000176681(0), ENSG00000185829(4.033e-101), ENSG00000214425(6.359e-16), ENSG00000228696(0), ENSG00000238083(4.033e-101), ENSG00000262879(5.357e-06)	NA	NA	7_Enh	H3K4me1	KANSL1, MAPT, ARL17B, NSFP1	LRR37A, ARL17A, LRR37A4P, ARL17B, LRR37A2	NA	NA
chr17	44,263,729	chr17:44263729	rs147317628	1.1e-06	ENSG00000120071(9.613e-15), ENSG00000186868(1.943e-06), ENSG00000228696(1.763e-05), ENSG00000260075(1.763e-05), ENSG00000262372(2.229e-05)	ENSG00000131484(6.359e-16), ENSG00000176681(0), ENSG00000185829(4.033e-101), ENSG00000214425(6.359e-16), ENSG00000228696(0), ENSG00000238083(4.033e-101), ENSG00000262879(5.357e-06)	NA	NA	7_Enh		KANSL1, MAPT, ARL17B, NSFP1	LRR37A, ARL17A, LRR37A4P, ARL17B, LRR37A2	NA	NA
chr17	44,263,965	chr17:44263965	rs147317628	1.1e-06	ENSG00000120071(9.613e-15), ENSG00000186868(1.943e-06), ENSG00000228696(1.763e-05), ENSG00000260075(1.763e-05), ENSG00000262372(2.229e-05)	ENSG00000131484(6.359e-16), ENSG00000176681(0), ENSG00000185829(4.033e-101), ENSG00000214425(6.359e-16), ENSG00000228696(0), ENSG00000238083(4.033e-101), ENSG00000262879(5.357e-06)	NA	NA	7_Enh		KANSL1, MAPT, ARL17B, NSFP1	LRR37A, ARL17A, LRR37A4P, ARL17B, LRR37A2	NA	NA
chr17	44,264,045	chr17:44264045	rs142920272, rs147317628	2.9e-07, 1.1e-06	ENSG00000120071(9.613e-15), ENSG00000186868(1.943e-06), ENSG00000228696(1.763e-05), ENSG00000260075(1.763e-05), ENSG00000262372(2.229e-05)	ENSG00000131484(6.359e-16), ENSG00000176681(0), ENSG00000185829(4.033e-101), ENSG00000214425(6.359e-16), ENSG00000228696(0), ENSG00000238083(4.033e-101), ENSG00000262879(5.357e-06)	NA	NA	7_Enh		KANSL1, MAPT, ARL17B, NSFP1	LRR37A, ARL17A, LRR37A4P, ARL17B, LRR37A2	NA	NA
chr17	44,264,269	chr17:44264269	rs142920272, rs147317628	2.9e-07, 1.1e-06	ENSG00000120071(9.613e-15), ENSG00000186868(1.943e-06), ENSG00000228696(1.763e-05), ENSG00000260075(1.763e-05), ENSG00000262372(2.229e-05)	ENSG00000131484(6.359e-16), ENSG00000176681(0), ENSG00000185829(4.033e-101), ENSG00000214425(6.359e-16), ENSG00000228696(0), ENSG00000238083(4.033e-101), ENSG00000262879(5.357e-06)	NA	NA	7_Enh	H3K4me1	KANSL1, MAPT, ARL17B, NSFP1	LRR37A, ARL17A, LRR37A4P, ARL17B, LRR37A2	NA	NA
chr17	44,264,717	chr17:44264717	rs142920272, rs147317628	2.9e-07, 1.1e-06	ENSG00000120071(9.613e-15), ENSG00000186868(1.943e-06), ENSG00000228696(1.763e-05), ENSG00000260075(1.763e-05), ENSG00000262372(2.229e-05)	ENSG00000131484(6.359e-16), ENSG00000176681(0), ENSG00000185829(4.033e-101), ENSG00000214425(6.359e-16), ENSG00000228696(0), ENSG00000238083(4.033e-101), ENSG00000262879(5.357e-06)	NA	NA	7_Enh	H3K4me1	KANSL1, MAPT, ARL17B, NSFP1	LRR37A, ARL17A, LRR37A4P, ARL17B, LRR37A2	NA	NA
chr17	44,264,943	chr17:44264943	rs142920272, rs147317628	2.9e-07, 1.1e-06	ENSG00000120071(9.613e-15), ENSG00000186868(1.943e-06), ENSG00000228696(1.763e-05), ENSG00000260075(1.763e-05), ENSG00000262372(2.229e-05)	ENSG00000131484(6.359e-16), ENSG00000176681(0), ENSG00000185829(4.033e-101), ENSG00000214425(6.359e-16), ENSG00000228696(0), ENSG00000238083(4.033e-101), ENSG00000262879(5.357e-06)	NA	NA	7_Enh		KANSL1, MAPT, ARL17B, NSFP1	LRR37A, ARL17A, LRR37A4P, ARL17B, LRR37A2	NA	NA
chr17	44,265,192	chr17:44265192	rs147317628	1.1e-06	ENSG00000120071(9.613e-15), ENSG00000186868(1.943e-06), ENSG00000228696(1.763e-05), ENSG00000260075(1.763e-05), ENSG00000262372(2.229e-05)	ENSG00000131484(6.359e-16), ENSG00000176681(0), ENSG00000185829(4.033e-101), ENSG00000214425(6.359e-16), ENSG00000228696(0), ENSG00000238083(4.033e-101), ENSG00000262879(5.357e-06)	NA	NA	7_Enh		KANSL1, MAPT, ARL17B, NSFP1	LRR37A, ARL17A, LRR37A4P, ARL17B, LRR37A2	NA	NA
chr17	44,265,328	chr17:44265328	rs142920272, rs147317628	2.9e-07, 1.1e-06	ENSG00000120071(9.613e-15), ENSG00000186868(1.943e-06), ENSG00000228696(1.763e-05), ENSG00000260075(1.763e-05), ENSG00000262372(2.229e-05)	ENSG00000131484(6.359e-16), ENSG00000176681(0), ENSG00000185829(4.033e-101), ENSG00000214425(6.359e-16), ENSG00000228696(0), ENSG00000238083(4.033e-101), ENSG00000262879(5.357e-06)	NA	NA	7_Enh		KANSL1, MAPT, ARL17B, NSFP1	LRR37A, ARL17A, LRR37A4P, ARL17B, LRR37A2	NA	NA
chr17	44,265,702	chr17:44265702	rs147317628	1.1e-06	ENSG00000120071(9.613e-15), ENSG00000186868(1.943e-06), ENSG00000228696(1.763e-05), ENSG00000260075(1.763e-05), ENSG00000262372(2.229e-05)	ENSG00000131484(6.359e-16), ENSG00000176681(0), ENSG00000185829(4.033e-101), ENSG00000214425(6.359e-16), ENSG00000228696(0), ENSG00000238083(4.033e-101), ENSG00000262879(5.357e-06)	NA	NA	2_TssAFlnk	H3K4me1	KANSL1, MAPT, ARL17B, NSFP1	LRR37A, ARL17A, LRR37A4P, ARL17B, LRR37A2	NA	NA

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Supplementary Table 16 – continued from previous page

CHR	BP	CredSNP	IndexSNP	Index P	ENSGID_CP	ENSGID_GZ	ENSGID Promoter	ENSGID functional	Chrom HMM	Histone	HGNC_CP	HGNC_GZ	HGNC Promoter	HGNC functional
chr17	44,266,022	chr17:44266022	rs142920272, rs147317628	2.9e-07, 1.1e-06	ENSG00000120071(9.613e-15), ENSG00000186868(1.943e-06), ENSG00000228696(1.763e-05), ENSG00000260075(1.763e-05), ENSG00000262372(2.229e-05)	ENSG00000131484(6.359e-16), ENSG00000176681(0), ENSG00000185829(4.033e-101), ENSG00000214425(6.359e-16), ENSG00000228696(0), ENSG00000238083(4.033e-101), ENSG00000262879(5.357e-06)	NA	NA	7_Enh		KANSL1, MAPT, ARL17B, NSFP1	LRRC37A, ARL17A, LRRC37A4P, ARL17B, LRRC37A2	NA	NA
chr17	44,266,972	chr17:44266972	rs147317628	1.1e-06	ENSG00000120071(9.613e-15), ENSG00000186868(1.943e-06), ENSG00000228696(1.763e-05), ENSG00000260075(1.763e-05), ENSG00000262372(2.229e-05)	ENSG00000131484(6.359e-16), ENSG00000176681(0), ENSG00000185829(4.033e-101), ENSG00000214425(6.359e-16), ENSG00000228696(0), ENSG00000238083(4.033e-101), ENSG00000262879(5.357e-06)	NA	NA	7_Enh	H3K4me1	KANSL1, MAPT, ARL17B, NSFP1	LRRC37A, ARL17A, LRRC37A4P, ARL17B, LRRC37A2	NA	NA
chr17	44,267,617	chr17:44267617	rs147317628	1.1e-06	ENSG00000120071(9.613e-15), ENSG00000186868(1.943e-06), ENSG00000228696(1.763e-05), ENSG00000260075(1.763e-05), ENSG00000262372(2.229e-05)	ENSG00000131484(6.359e-16), ENSG00000176681(0), ENSG00000185829(4.033e-101), ENSG00000214425(6.359e-16), ENSG00000228696(0), ENSG00000238083(4.033e-101), ENSG00000262879(5.357e-06)	NA	NA	2_TssAFlnk		KANSL1, MAPT, ARL17B, NSFP1	LRRC37A, ARL17A, LRRC37A4P, ARL17B, LRRC37A2	NA	NA
chr17	44,269,546	chr17:44269546	rs142920272, rs147317628	2.9e-07, 1.1e-06			ENSG00000214401		8_ZNF/Rpts	DHS:H3K4me3			KANSL1- AS1	
chr17	44,269,676	chr17:44269676	rs142920272, rs147317628	2.9e-07, 1.1e-06			ENSG00000214401		8_ZNF/Rpts	H3K4me3			KANSL1- AS1	
chr17	44,271,152	chr17:44271152	rs142920272, rs147317628	2.9e-07, 1.1e-06			ENSG00000120071		1_TssA	DHS:H3K4me3			KANSL1	
chr17	44,271,430	chr17:44271430	rs142920272, rs147317628	2.9e-07, 1.1e-06			ENSG00000120071, ENSG00000214401		1_TssA	DHS			KANSL1, KANSL1- AS1	
chr17	44,272,266	chr17:44272266	rs142920272	2.9e-07			ENSG00000120071, ENSG00000214401		5_TxWk				KANSL1, KANSL1- AS1	
chr17	44,272,552	chr17:44272552	rs142920272	2.9e-07			ENSG00000120071, ENSG00000214401		5_TxWk	DHS			KANSL1, KANSL1- AS1	
chr17	44,272,679	chr17:44272679	rs142920272, rs147317628	2.9e-07, 1.1e-06			ENSG00000120071, ENSG00000214401		5_TxWk				KANSL1, KANSL1- AS1	
chr17	44,272,928	chr17:44272928	rs142920272	2.9e-07			ENSG00000120071, ENSG00000214401		5_TxWk				KANSL1, KANSL1- AS1	
chr17	44,273,264	chr17:44273264	rs142920272	2.9e-07			ENSG00000214401		5_TxWk				KANSL1- AS1	
chr17	44,273,448	chr17:44273448	rs142920272, rs147317628	2.9e-07, 1.1e-06	ENSG00000120071(5.738e-08), ENSG00000228696(6.206e-181), ENSG00000260075(6.206e-181)	ENSG00000120071(8.6e-08), ENSG00000185829(0), ENSG00000238083(0), ENSG00000262500(1.699e-08)	NA	NA	5_TxWk		KANSL1, ARL17B, NSFP1	KANSL1, ARL17A, LRRC37A2	NA	NA
chr17	44,273,448	chr17:44273448m	rs142920272	2.9e-07	ENSG00000120071(5.738e-08), ENSG00000228696(6.206e-181), ENSG00000260075(6.206e-181)	ENSG00000120071(8.6e-08), ENSG00000185829(0), ENSG00000238083(0), ENSG00000262500(1.699e-08)	NA	NA	5_TxWk		KANSL1, ARL17B, NSFP1	KANSL1, ARL17A, LRRC37A2	NA	NA
chr17	44,275,172	chr17:44275172	rs142920272, rs147317628	2.9e-07, 1.1e-06	ENSG00000120071(5.738e-08), ENSG00000228696(6.206e-181), ENSG00000260075(6.206e-181)	ENSG00000120071(8.6e-08), ENSG00000185829(0), ENSG00000238083(0), ENSG00000262500(1.699e-08)	NA	NA	5_TxWk		KANSL1, ARL17B, NSFP1	KANSL1, ARL17A, LRRC37A2	NA	NA
chr17	44,275,619	chr17:44275619	rs147317628	1.1e-06	ENSG00000120071(5.738e-08), ENSG00000228696(6.206e-181), ENSG00000260075(6.206e-181)	ENSG00000120071(8.6e-08), ENSG00000185829(0), ENSG00000238083(0), ENSG00000262500(1.699e-08)	NA	NA	5_TxWk		KANSL1, ARL17B, NSFP1	KANSL1, ARL17A, LRRC37A2	NA	NA
chr17	44,276,578	chr17:44276578	rs147317628	1.1e-06	ENSG00000120071(5.738e-08), ENSG00000228696(6.206e-181), ENSG00000260075(6.206e-181)	ENSG00000120071(8.6e-08), ENSG00000185829(0), ENSG00000238083(0), ENSG00000262500(1.699e-08)	NA	NA	5_TxWk		KANSL1, ARL17B, NSFP1	KANSL1, ARL17A, LRRC37A2	NA	NA

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Supplementary Table 16 – continued from previous page

CHR	BP	CredSNP	IndexSNP	Index P	ENSGID_CP	ENSGID_GZ	ENSGID Promoter	ENSGID functional	Chrom HMM	Histone	HGNC_CP	HGNC_GZ	HGNC Promoter	HGNC functional
chr17	44,276,618	chr17:44276618	rs142920272	2.9e-07	ENSG00000120071(5.738e-08), ENSG00000228696(6.206e-181), ENSG00000260075(6.206e-181)	ENSG00000120071(8.6e-08), ENSG00000185829(0), ENSG00000238083(0), ENSG00000262500(1.699e-08)	NA	NA	5_TxWk		KANSL1, ARL17B, NSFP1	KANSL1, ARL17A, LRR37A2	NA	NA
chr17	44,276,821	chr17:44276821	rs142920272, rs147317628	2.9e-07, 1.1e-06	ENSG00000120071(5.738e-08), ENSG00000228696(6.206e-181), ENSG00000260075(6.206e-181)	ENSG00000120071(8.6e-08), ENSG00000185829(0), ENSG00000238083(0), ENSG00000262500(1.699e-08)	NA	NA	5_TxWk		KANSL1, ARL17B, NSFP1	KANSL1, ARL17A, LRR37A2	NA	NA
chr17	44,277,691	chr17:44277691	rs142920272, rs147317628	2.9e-07, 1.1e-06	ENSG00000120071(5.738e-08), ENSG00000228696(6.206e-181), ENSG00000260075(6.206e-181)	ENSG00000120071(8.6e-08), ENSG00000185829(0), ENSG00000238083(0), ENSG00000262500(1.699e-08)	NA	NA	5_TxWk		KANSL1, ARL17B, NSFP1	KANSL1, ARL17A, LRR37A2	NA	NA
chr17	44,277,818	chr17:44277818	rs147317628	1.1e-06	ENSG00000120071(5.738e-08), ENSG00000228696(6.206e-181), ENSG00000260075(6.206e-181)	ENSG00000120071(8.6e-08), ENSG00000185829(0), ENSG00000238083(0), ENSG00000262500(1.699e-08)	NA	NA	5_TxWk		KANSL1, ARL17B, NSFP1	KANSL1, ARL17A, LRR37A2	NA	NA
chr17	44,277,923	chr17:44277923	rs142920272, rs147317628	2.9e-07, 1.1e-06	ENSG00000120071(5.738e-08), ENSG00000228696(6.206e-181), ENSG00000260075(6.206e-181)	ENSG00000120071(8.6e-08), ENSG00000185829(0), ENSG00000238083(0), ENSG00000262500(1.699e-08)	NA	NA	5_TxWk		KANSL1, ARL17B, NSFP1	KANSL1, ARL17A, LRR37A2	NA	NA
chr17	44,281,452	chr17:44281452	rs147317628	1.1e-06	ENSG00000006062(7.889e-05), ENSG00000120071(6.771e-18), ENSG00000186868(4.302e-05), ENSG00000228696(8.171e-82), ENSG00000260075(8.171e-82), ENSG00000262500(4.334e-75)	ENSG00000120071(8.907e-09), ENSG00000131484(1.778e-10), ENSG00000176681(0), ENSG00000185829(2.706e-16), ENSG00000214425(1.778e-10), ENSG00000225190(1.778e-10), ENSG00000228696(0), ENSG00000238083(2.706e-16), ENSG00000262500(1.013e-28), ENSG00000262879(1.557e-07)	NA	NA	5_TxWk	MAP3K14, KANSL1, MAPT, ARL17B, NSFP1	KANSL1, LRR37A, ARL17A, LRR37A4P, PLEKHM1, ARL17B, LRR37A2	NA	NA	
chr17	44,283,139	chr17:44283139	rs147317628	1.1e-06	ENSG00000006062(7.889e-05), ENSG00000120071(6.771e-18), ENSG00000186868(4.302e-05), ENSG00000228696(8.171e-82), ENSG00000260075(8.171e-82), ENSG00000262500(4.334e-75)	ENSG00000120071(8.907e-09), ENSG00000131484(1.778e-10), ENSG00000176681(0), ENSG00000185829(2.706e-16), ENSG00000214425(1.778e-10), ENSG00000225190(1.778e-10), ENSG00000228696(0), ENSG00000238083(2.706e-16), ENSG00000262500(1.013e-28), ENSG00000262879(1.557e-07)	NA	NA	5_TxWk	MAP3K14, KANSL1, MAPT, ARL17B, NSFP1	KANSL1, LRR37A, ARL17A, LRR37A4P, PLEKHM1, ARL17B, LRR37A2	NA	NA	
chr17	44,283,479	chr17:44283479	rs142920272	2.9e-07	ENSG00000006062(7.889e-05), ENSG00000120071(6.771e-18), ENSG00000186868(4.302e-05), ENSG00000228696(8.171e-82), ENSG00000260075(8.171e-82), ENSG00000262500(4.334e-75)	ENSG00000120071(8.907e-09), ENSG00000131484(1.778e-10), ENSG00000176681(0), ENSG00000185829(2.706e-16), ENSG00000214425(1.778e-10), ENSG00000225190(1.778e-10), ENSG00000228696(0), ENSG00000238083(2.706e-16), ENSG00000262500(1.013e-28), ENSG00000262879(1.557e-07)	NA	NA	8_ZNF/Rpts	MAP3K14, KANSL1, MAPT, ARL17B, NSFP1	KANSL1, LRR37A, ARL17A, LRR37A4P, PLEKHM1, ARL17B, LRR37A2	NA	NA	
chr17	44,283,761	chr17:44283761	rs142920272, rs147317628	2.9e-07, 1.1e-06	ENSG00000006062(7.889e-05), ENSG00000120071(6.771e-18), ENSG00000186868(4.302e-05), ENSG00000228696(8.171e-82), ENSG00000260075(8.171e-82), ENSG00000262500(4.334e-75)	ENSG00000120071(8.907e-09), ENSG00000131484(1.778e-10), ENSG00000176681(0), ENSG00000185829(2.706e-16), ENSG00000214425(1.778e-10), ENSG00000225190(1.778e-10), ENSG00000228696(0), ENSG00000238083(2.706e-16), ENSG00000262500(1.013e-28), ENSG00000262879(1.557e-07)	NA	NA	8_ZNF/Rpts	MAP3K14, KANSL1, MAPT, ARL17B, NSFP1	KANSL1, LRR37A, ARL17A, LRR37A4P, PLEKHM1, ARL17B, LRR37A2	NA	NA	
chr17	44,284,542	chr17:44284542	rs142920272, rs147317628	2.9e-07, 1.1e-06	ENSG00000006062(7.889e-05), ENSG00000120071(6.771e-18), ENSG00000186868(4.302e-05), ENSG00000228696(8.171e-82), ENSG00000260075(8.171e-82), ENSG00000262500(4.334e-75)	ENSG00000120071(8.907e-09), ENSG00000131484(1.778e-10), ENSG00000176681(0), ENSG00000185829(2.706e-16), ENSG00000214425(1.778e-10), ENSG00000225190(1.778e-10), ENSG00000228696(0), ENSG00000238083(2.706e-16), ENSG00000262500(1.013e-28), ENSG00000262879(1.557e-07)	NA	NA	5_TxWk	MAP3K14, KANSL1, MAPT, ARL17B, NSFP1	KANSL1, LRR37A, ARL17A, LRR37A4P, PLEKHM1, ARL17B, LRR37A2	NA	NA	

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Supplementary Table 16 – continued from previous page

CHR	BP	CredSNP	IndexSNP	Index P	ENSGID_CP	ENSGID_GZ	ENSGID Promoter	ENSGID functional	Chrom HMM	Histone	HGNC_CP	HGNC_GZ	HGNC Promoter	HGNC functional
chr17	44,285,142	chr17:44285142	rs142920272,rs147317628	2.9e-07,1.1e-06	ENSG00000006062(7.889e-05), ENSG00000120071(6.771e-18), ENSG00000186868(4.302e-05), ENSG00000228696(8.171e-82), ENSG00000260075(8.171e-82), ENSG00000262500(4.334e-75)	ENSG00000120071(8.907e-09), ENSG00000131484(1.778e-10), ENSG00000176681(0), ENSG00000185829(2.706e-16), ENSG00000214425(1.778e-10), ENSG00000225190(1.778e-10), ENSG00000228696(0), ENSG00000238083(2.706e-16), ENSG00000262500(1.013e-28), ENSG00000262879(1.557e-07)	NA	NA	5_TxWk	MAP3K14, KANSL1, MAPT, ARL17B, NSFP1	KANSL1, LRRC37A, ARL17A, LRRC37A4P, PLEKHM1, ARL17B, LRRC37A2	NA	NA	
chr17	44,285,531	chr17:44285531	rs142920272,rs147317628	2.9e-07,1.1e-06	ENSG00000006062(7.889e-05), ENSG00000120071(6.771e-18), ENSG00000186868(4.302e-05), ENSG00000228696(8.171e-82), ENSG00000260075(8.171e-82), ENSG00000262500(4.334e-75)	ENSG00000120071(8.907e-09), ENSG00000131484(1.778e-10), ENSG00000176681(0), ENSG00000185829(2.706e-16), ENSG00000214425(1.778e-10), ENSG00000225190(1.778e-10), ENSG00000228696(0), ENSG00000238083(2.706e-16), ENSG00000262500(1.013e-28), ENSG00000262879(1.557e-07)	NA	NA	5_TxWk	MAP3K14, KANSL1, MAPT, ARL17B, NSFP1	KANSL1, LRRC37A, ARL17A, LRRC37A4P, PLEKHM1, ARL17B, LRRC37A2	NA	NA	
chr17	44,286,089	chr17:44286089	rs142920272,rs147317628	2.9e-07,1.1e-06	ENSG00000006062(7.889e-05), ENSG00000120071(6.771e-18), ENSG00000186868(4.302e-05), ENSG00000228696(8.171e-82), ENSG00000260075(8.171e-82), ENSG00000262500(4.334e-75)	ENSG00000120071(8.907e-09), ENSG00000131484(1.778e-10), ENSG00000176681(0), ENSG00000185829(2.706e-16), ENSG00000214425(1.778e-10), ENSG00000225190(1.778e-10), ENSG00000228696(0), ENSG00000238083(2.706e-16), ENSG00000262500(1.013e-28), ENSG00000262879(1.557e-07)	NA	NA	5_TxWk	MAP3K14, KANSL1, MAPT, ARL17B, NSFP1	KANSL1, LRRC37A, ARL17A, LRRC37A4P, PLEKHM1, ARL17B, LRRC37A2	NA	NA	
chr17	44,286,128	chr17:44286128	rs142920272,rs147317628	2.9e-07,1.1e-06	ENSG00000006062(7.889e-05), ENSG00000120071(6.771e-18), ENSG00000186868(4.302e-05), ENSG00000228696(8.171e-82), ENSG00000260075(8.171e-82), ENSG00000262500(4.334e-75)	ENSG00000120071(8.907e-09), ENSG00000131484(1.778e-10), ENSG00000176681(0), ENSG00000185829(2.706e-16), ENSG00000214425(1.778e-10), ENSG00000225190(1.778e-10), ENSG00000228696(0), ENSG00000238083(2.706e-16), ENSG00000262500(1.013e-28), ENSG00000262879(1.557e-07)	NA	NA	5_TxWk	MAP3K14, KANSL1, MAPT, ARL17B, NSFP1	KANSL1, LRRC37A, ARL17A, LRRC37A4P, PLEKHM1, ARL17B, LRRC37A2	NA	NA	
chr17	44,286,198	chr17:44286198	rs142920272	2.9e-07	ENSG00000006062(7.889e-05), ENSG00000120071(6.771e-18), ENSG00000186868(4.302e-05), ENSG00000228696(8.171e-82), ENSG00000260075(8.171e-82), ENSG00000262500(4.334e-75)	ENSG00000120071(8.907e-09), ENSG00000131484(1.778e-10), ENSG00000176681(0), ENSG00000185829(2.706e-16), ENSG00000214425(1.778e-10), ENSG00000225190(1.778e-10), ENSG00000228696(0), ENSG00000238083(2.706e-16), ENSG00000262500(1.013e-28), ENSG00000262879(1.557e-07)	NA	NA	5_TxWk	MAP3K14, KANSL1, MAPT, ARL17B, NSFP1	KANSL1, LRRC37A, ARL17A, LRRC37A4P, PLEKHM1, ARL17B, LRRC37A2	NA	NA	
chr17	44,287,310	chr17:44287310	rs147317628	1.1e-06	ENSG00000006062(7.889e-05), ENSG00000120071(6.771e-18), ENSG00000186868(4.302e-05), ENSG00000228696(8.171e-82), ENSG00000260075(8.171e-82), ENSG00000262500(4.334e-75)	ENSG00000120071(8.907e-09), ENSG00000131484(1.778e-10), ENSG00000176681(0), ENSG00000185829(2.706e-16), ENSG00000214425(1.778e-10), ENSG00000225190(1.778e-10), ENSG00000228696(0), ENSG00000238083(2.706e-16), ENSG00000262500(1.013e-28), ENSG00000262879(1.557e-07)	NA	NA	5_TxWk	H3K36me3 MAP3K14, KANSL1, MAPT, ARL17B, NSFP1	KANSL1, LRRC37A, ARL17A, LRRC37A4P, PLEKHM1, ARL17B, LRRC37A2	NA	NA	
chr17	44,288,114	chr17:44288114	rs142920272,rs147317628	2.9e-07,1.1e-06	ENSG00000006062(7.889e-05), ENSG00000120071(6.771e-18), ENSG00000186868(4.302e-05), ENSG00000228696(8.171e-82), ENSG00000260075(8.171e-82), ENSG00000262500(4.334e-75)	ENSG00000120071(8.907e-09), ENSG00000131484(1.778e-10), ENSG00000176681(0), ENSG00000185829(2.706e-16), ENSG00000214425(1.778e-10), ENSG00000225190(1.778e-10), ENSG00000228696(0), ENSG00000238083(2.706e-16), ENSG00000262500(1.013e-28), ENSG00000262879(1.557e-07)	NA	NA	5_TxWk	MAP3K14, KANSL1, MAPT, ARL17B, NSFP1	KANSL1, LRRC37A, ARL17A, LRRC37A4P, PLEKHM1, ARL17B, LRRC37A2	NA	NA	

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Supplementary Table 16 – continued from previous page

CHR	BP	CredSNP	IndexSNP	Index P	ENSGID_CP	ENSGID_GZ	ENSGID Promoter	ENSGID functional	Chrom HMM	Histone	HGNC_CP	HGNC_GZ	HGNC Promoter	HGNC functional
chr17	44,295,157	chr17:44295157	rs147317628	1.1e-06	ENSG00000108379(6.796e-05), ENSG00000120071(1.371e-15), ENSG00000158955(6.796e-05), ENSG00000186868(1.113e-08), ENSG00000225190(1.173e-17), ENSG00000262372(2.024e-10), ENSG00000262539(4.07e-09), ENSG00000264589(1.113e-08), ENSG00000266497(5.756e-25)	ENSG00000004897(3.849e-37), ENSG00000108379(1.454e-06), ENSG00000120071(1.453e-15), ENSG00000159314(1.815e-07), ENSG00000186868(7.968e-07), ENSG00000225190(2.236e-12), ENSG00000236234(2.236e-12), ENSG00000239291(8.887e-15), ENSG00000256762(7.075e-06), ENSG00000260075(1.26e-41), ENSG00000262265(8.887e-15), ENSG00000262372(1.283e-13), ENSG00000262879(1.357e-09), ENSG00000264038(3.203e-07), ENSG00000264225(2.236e-12), ENSG00000266497(9.576e-13)	NA	NA	5_TxWk	WNT3, KANSL1, WNT9B, MAPT, PLEKHM1, MAPT-AS1	CDC27, WNT3, KANSL1, ARHGAP27, MAPT, PLEKHM1, STH, NSFP1, RN7SL730P	NA	NA	
chr17	44,298,102	chr17:44298102	rs142920272, rs147317628	2.9e-07, 1.1e-06	ENSG00000108379(6.796e-05), ENSG00000120071(1.371e-15), ENSG00000158955(6.796e-05), ENSG00000186868(1.113e-08), ENSG00000225190(1.173e-17), ENSG00000262372(2.024e-10), ENSG00000262539(4.07e-09), ENSG00000264589(1.113e-08), ENSG00000266497(5.756e-25)	ENSG00000004897(3.849e-37), ENSG00000108379(1.454e-06), ENSG00000120071(1.453e-15), ENSG00000159314(1.815e-07), ENSG00000186868(7.968e-07), ENSG00000225190(2.236e-12), ENSG00000236234(2.236e-12), ENSG00000239291(8.887e-15), ENSG00000256762(7.075e-06), ENSG00000260075(1.26e-41), ENSG00000262265(8.887e-15), ENSG00000262372(1.283e-13), ENSG00000262879(1.357e-09), ENSG00000264038(3.203e-07), ENSG00000264225(2.236e-12), ENSG00000266497(9.576e-13)	NA	NA	5_TxWk	WNT3, KANSL1, WNT9B, MAPT, PLEKHM1, MAPT-AS1	CDC27, WNT3, KANSL1, ARHGAP27, MAPT, PLEKHM1, STH, NSFP1, RN7SL730P	NA	NA	
chr17	44,298,209	chr17:44298209	rs147317628	1.1e-06	ENSG00000108379(6.796e-05), ENSG00000120071(1.371e-15), ENSG00000158955(6.796e-05), ENSG00000186868(1.113e-08), ENSG00000225190(1.173e-17), ENSG00000262372(2.024e-10), ENSG00000262539(4.07e-09), ENSG00000264589(1.113e-08), ENSG00000266497(5.756e-25)	ENSG00000004897(3.849e-37), ENSG00000108379(1.454e-06), ENSG00000120071(1.453e-15), ENSG00000159314(1.815e-07), ENSG00000186868(7.968e-07), ENSG00000225190(2.236e-12), ENSG00000236234(2.236e-12), ENSG00000239291(8.887e-15), ENSG00000256762(7.075e-06), ENSG00000260075(1.26e-41), ENSG00000262265(8.887e-15), ENSG00000262372(1.283e-13), ENSG00000262879(1.357e-09), ENSG00000264038(3.203e-07), ENSG00000264225(2.236e-12), ENSG00000266497(9.576e-13)	NA	NA	7_Enh	WNT3, KANSL1, WNT9B, MAPT, PLEKHM1, MAPT-AS1	CDC27, WNT3, KANSL1, ARHGAP27, MAPT, PLEKHM1, STH, NSFP1, RN7SL730P	NA	NA	
chr17	44,302,881	chr17:44302881	rs147317628	1.1e-06			ENSG00000120071		9_Het	DHS:H3K4me3		KANSL1		
chr17	44,305,689	chr17:44305689	rs147317628	1.1e-06	ENSG00000120071(6.42e-18), ENSG00000186868(5.937e-08), ENSG00000225190(1.552e-14), ENSG00000262372(3.52e-09), ENSG00000264589(5.937e-08), ENSG00000266497(1.564e-21)	ENSG00000004897(1.111e-36), ENSG00000108379(2.723e-06), ENSG00000120071(5.97e-15), ENSG00000159314(1.602e-07), ENSG00000186868(1.111e-06), ENSG00000225190(2.259e-11), ENSG00000236234(2.259e-11), ENSG00000239291(4.569e-13), ENSG00000256762(5.986e-06), ENSG00000260075(1.762e-39), ENSG00000262265(4.569e-13), ENSG00000262372(1.069e-12), ENSG00000262879(4.408e-09), ENSG00000264038(8.752e-07), ENSG00000264225(2.259e-11), ENSG00000266497(7.218e-12)	NA	NA	15_Quies	KANSL1, MAPT, PLEKHM1, MAPT-AS1	CDC27, WNT3, KANSL1, ARHGAP27, MAPT, PLEKHM1, STH, NSFP1, RN7SL730P	NA	NA	

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Supplementary Table 16 – continued from previous page

CHR	BP	CredSNP	IndexSNP	Index P	ENSGID_CP	ENSGID_GZ	ENSGID Promoter	ENSGID functional	Chrom HMM	Histone	HGNC_CP	HGNC_GZ	HGNC Promoter	HGNC functional
chr17	44,311,099	chr17:44311099	rs147317628	1.1e-06	ENSG00000120071(1.001e-75), ENSG00000186868(4.221e-08), ENSG00000225190(1.513e-15), ENSG00000262372(1.712e-09), ENSG00000264589(4.221e-08), ENSG00000266497(2.367e-21)	ENSG00000004897(5.898e-35), ENSG00000108379(3.433e-06), ENSG00000120071(6.373e-29), ENSG00000159314(2.318e-07), ENSG00000186868(5.531e-07), ENSG00000214401(1.613e-08), ENSG00000225190(2.693e-11), ENSG00000236234(2.693e-11), ENSG00000239291(2.76e-13), ENSG00000256762(5.458e-06), ENSG00000260075(2.959e-37), ENSG00000262265(2.76e-13), ENSG00000262372(5.801e-12), ENSG00000262879(1.682e-08), ENSG00000264038(1.084e-06), ENSG00000264225(2.693e-11), ENSG00000266497(1.002e-11)	NA	NA	15_Quies	KANSL1, MAPT, PLEKHM1, MAPT-AS1	CDC27, WNT3, KANSL1, ARHGAP27, MAPT, KANSL1-AS1, PLEKHM1, STH, NSFP1, RN7SL730P	NA	NA	
chr20	21,117,240	chr20:21117240	rs6047270	7.7e-08	NA	NA	NA	NA	15_Quies		NA	NA	NA	NA
chr2	159,384,735	chr2:159384735	rs59566011	9.3e-07	ENSG00000153237(3.184e-05), ENSG00000227480(3.184e-05)	NA	NA	NA	15_Quies		CCDC148, CCDC148-AS1	NA	NA	NA
chr5	168,173,526	chr5:168173526	chr5:168173526	1.3e-06	NA	NA	NA	NA	14_ReprPCWk		NA	NA	NA	NA
chr6	98,584,711	chr6:98584711	rs72934503	5.9e-07	NA	NA	NA	NA	15_Quies		NA	NA	NA	NA
chr8	10,575,786	chr8:10575786	rs10099100	1.1e-08	NA	ENSG00000171056(9.841e-05), ENSG00000171060(1.154e-09), ENSG00000183638(1.154e-09), ENSG00000212433(1.154e-09), ENSG00000252565(9.841e-05), ENSG00000254093(9.841e-05), ENSG00000258724(9.841e-05)	NA	NA	14_ReprPCWk		NA	SOX7, C8orf74, RP1L1, RNA5SP252, PINX1	NA	NA
chr17	44,212,310	rs10221243	rs142920272, rs147317628	2.9e-07, 1.1e-06	ENSG00000120071(1.173e-15), ENSG00000159314(3.167e-06), ENSG00000225190(3.167e-06), ENSG00000238723(1.173e-15), ENSG00000260075(3.877e-18), ENSG00000262500(4.273e-15), ENSG00000262879(3.174e-29), ENSG00000263142(3.174e-29)	ENSG00000131484(6.934e-14), ENSG00000159314(2.906e-07), ENSG00000176681(0), ENSG00000214425(6.934e-14), ENSG00000225190(1.09e-11), ENSG00000228696(0), ENSG00000236234(1.09e-11)	NA	NA	5_TxWk	KANSL1, ARHGAP27, PLEKHM1, NSFP1, LRRC37A17P	ARHGAP27, LRRC37A, LRRC37A4P, PLEKHM1, ARL17B	NA	NA	
chr8	53,374,007	rs1036715	rs10666089m	1.0e-06			ENSG00000147488		15_Quies				ST18	
chr6	98,572,120	rs10457441	rs72934503	5.9e-07	NA	NA	NA	NA	15_Quies		NA	NA	NA	NA
chr1	104,791,770	rs10625106	rs11185408	7.0e-07	NA	NA	NA	NA	15_Quies		NA	NA	NA	NA
chr20	21,258,707	rs10627346	rs910805	2.0e-09	NA	NA	NA	NA	14_ReprPCWk		NA	NA	NA	NA
chr8	53,341,258	rs10666089	rs10666089m	1.0e-06	NA	NA	NA	NA	15_Quies		NA	NA	NA	NA
chr8	53,341,258	rs10666089m	rs10666089m	1.0e-06	NA	NA	NA	NA	15_Quies		NA	NA	NA	NA
chr20	21,151,377	rs1072271	rs6047270	7.7e-08	NA	NA	NA	NA	15_Quies		NA	NA	NA	NA
chr1	96,961,107	rs11165656	rs2391769	1.1e-07	NA	NA	NA	NA	15_Quies		NA	NA	NA	NA
chr1	104,792,257	rs11185408	rs11185408	7.0e-07	NA	NA	NA	NA	15_Quies		NA	NA	NA	NA
chr18	55,882,032	rs111876941	rs292441	1.1e-06	NA	ENSG00000049759(3.605e-07)	NA	NA	7_Enh	H3K4me1	NA	NEDD4L	NA	NA
chr7	104,744,219	rs111931861	rs111931861	1.1e-07				ENSG000000005483	5_TxWk					KMT2E
chr14	94,838,142	rs112635299	rs112635299	3.0e-07	NA	NA	NA	NA	14_ReprPCWk		NA	NA	NA	NA
chr20	21,272,136	rs11277342	rs910805	2.0e-09	NA	NA	NA	NA	14_ReprPCWk		NA	NA	NA	NA
chr17	44,256,811	rs113089855	rs147317628	1.1e-06	ENSG00000004897(3.479e-05), ENSG00000214425(0), ENSG00000267198(0), ENSG00000267246(0)	ENSG000000073969(9.158e-12), ENSG00000120071(2.834e-05), ENSG00000131484(1.201e-19), ENSG00000186868(2.834e-05), ENSG00000214425(1.201e-19), ENSG00000228696(1.46e-32), ENSG00000260075(1.46e-32), ENSG00000266918(1.201e-19)	NA	NA	5_TxWk	CDC27, LRRC37A4P	NSF, KANSL1, MAPT, LRRC37A4P, ARL17B, NSFP1	NA	NA	
chr17	44,270,809	rs113417378	rs142920272, rs147317628	2.9e-07, 1.1e-06			ENSG00000120071, ENSG00000214401		5_TxWk	DHS			KANSL1, KANSL1-AS1	
chr8	53,372,983	rs11452991	rs10666089m	1.0e-06	NA	NA	NA	NA	15_Quies		NA	NA	NA	NA
chr20	21,145,353	rs11475262	rs6047270	7.7e-08			ENSG00000228604		5_TxWk					

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Supplementary Table 16 – continued from previous page

CHR	BP	CredSNP	IndexSNP	Index P	ENSGID_CP	ENSGID_GZ	ENSGID Promoter	ENSGID functional	Chrom HMM	Histone	HGNC_CP	HGNC_GZ	HGNC Promoter	HGNC functional
chr20	14,822,173	rs11481126	rs11481126	1.3e-07	NA	NA	NA	NA	15_Quies		NA	NA	NA	NA
chr20	21,136,056	rs11483719	rs6047270	7.7e-08	NA	NA	NA	NA	5_TxWk		NA	NA	NA	NA
chr2	159,377,277	rs11675009	rs59566011	9.3e-07				ENSG00000144283	15_Quies					PKP4
chr2	159,376,514	rs11695939	rs59566011	9.3e-07				ENSG00000144283	15_Quies					PKP4
chr11	106,827,977	rs117603308	rs117603308n	1.4e-06	ENSG00000254580(9.283e-05)	NA	NA	NA	5_TxWk			NA	NA	NA
chr11	106,827,977	rs117603308m	rs117603308m	1.4e-06	ENSG00000254580(9.283e-05)	NA	NA	NA	5_TxWk			NA	NA	NA
chr17	44,298,631	rs117662214	rs142920272, rs147317628	2.9e-07, 1.1e-06	ENSG00000108379(6.796e-05), ENSG00000120071(1.371e-15), ENSG00000158955(6.796e-05), ENSG00000186868(1.113e-08), ENSG00000225190(1.173e-17), ENSG00000262372(2.024e-10), ENSG00000262539(4.07e-09), ENSG00000264589(1.113e-08), ENSG00000266497(5.756e-25)	ENSG00000004897(3.849e-37), ENSG00000108379(1.454e-06), ENSG00000120071(1.453e-15), ENSG00000159314(1.815e-07), ENSG00000186868(7.968e-07), ENSG00000225190(2.236e-12), ENSG00000236234(2.236e-12), ENSG00000239291(8.887e-15), ENSG00000256762(7.075e-06), ENSG00000260075(1.26e-41), ENSG00000262265(8.887e-15), ENSG00000262372(1.283e-13), ENSG00000262879(1.357e-09), ENSG00000264038(3.203e-07), ENSG00000264225(2.236e-12), ENSG00000266497(9.576e-13)	NA	NA	15_Quies	WNT3, KANSL1, WNT9B, MAPT, PLEKHM1, MAPT-AS1	CDC27, WNT3, KANSL1, ARHGAP27, MAPT, PLEKHM1, STH, NSFP1, RN7SL730P	NA	NA	
chr1	104,790,871	rs11806291	rs11185408	7.0e-07	NA	NA	NA	NA	15_Quies		NA	NA	NA	NA
chr1	96,586,426	rs12089599	rs201910565	3.4e-07	NA	NA	NA	NA	15_Quies		NA	NA	NA	NA
chr6	98,576,223	rs12202969	rs72934503	5.9e-07	NA	NA	NA	NA	5_TxWk	DHS	NA	NA	NA	NA
chr6	98,579,481	rs12204181	rs72934503	5.9e-07	NA	NA	NA	NA	7_Enh	H3K4me1	NA	NA	NA	NA
chr6	98,582,900	rs12206087	rs72934503	5.9e-07	NA	NA	NA	NA	15_Quies		NA	NA	NA	NA
chr1	96,602,440	rs1222063	rs1222063	2.6e-07	NA	NA	NA	NA	15_Quies		NA	NA	NA	NA
chr20	21,130,707	rs13041255	rs6047270	7.7e-08	NA	NA	NA	NA	5_TxWk		NA	NA	NA	NA
chr8	53,333,088	rs1365880	rs10666089m	1.0e-06	ENSG00000147485(8.966e-05), ENSG00000147488(5.221e-11)	ENSG00000147488(7.94e-08), ENSG00000253844(9.555e-06)	NA	NA	5_TxWk		PXDNL, ST18	ST18	NA	NA
chr6	98,583,487	rs138027849	rs72934503	5.9e-07	NA	NA	NA	NA	15_Quies		NA	NA	NA	NA
chr19	37,439,641	rs138867053	rs138867053	1.2e-07	NA	NA	NA	NA	4_Tx		NA	NA	NA	NA
chr20	21,263,168	rs139520783	rs910805	2.0e-09	NA	NA	NA	NA	14_ReprPCWk		NA	NA	NA	NA
chr10	72,749,057	rs140093403	rs78827416	9.0e-07	NA	NA	NA	NA	15_Quies		NA	NA	NA	NA
chr17	44,305,817	rs140256385	rs147317628	1.1e-06	ENSG00000120071(6.42e-18), ENSG00000186868(5.937e-08), ENSG00000225190(1.552e-14), ENSG00000262372(3.52e-09), ENSG00000264589(5.937e-08), ENSG00000266497(1.564e-21)	ENSG00000004897(1.111e-36), ENSG00000108379(2.723e-06), ENSG00000120071(5.97e-15), ENSG00000159314(1.602e-07), ENSG00000186868(1.111e-06), ENSG00000225190(2.259e-11), ENSG00000236234(2.259e-11), ENSG00000239291(4.569e-13), ENSG00000256762(5.986e-06), ENSG00000260075(1.762e-39), ENSG00000262265(4.569e-13), ENSG00000262372(1.069e-12), ENSG00000262879(4.408e-09), ENSG00000264038(8.752e-07), ENSG00000264225(2.259e-11), ENSG00000266497(7.218e-12)	NA	NA	15_Quies	KANSL1, MAPT, PLEKHM1, MAPT-AS1	CDC27, WNT3, KANSL1, ARHGAP27, MAPT, PLEKHM1, STH, NSFP1, RN7SL730P	NA	NA	
chr17	44,019,083	rs141455452	rs141455452	8.9e-07				ENSG00000186868	7_Enh	H3K4me1				MAPT

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Supplementary Table 16 – continued from previous page

CHR	BP	CredSNP	IndexSNP	Index P	ENSGID_CP	ENSGID_GZ	ENSGID Promoter	ENSGID functional	Chrom HMM	Histone	HGNC_CP	HGNC_GZ	HGNC Promoter	HGNC functional
chr17	44,325,593	rs142380704	rs147317628	1.1e-06	ENSG00000120071(4.406e-45), ENSG00000225190(1.681e-08), ENSG00000262372(6.759e-07), ENSG00000266497(1.582e-08)	ENSG0000004897(2.896e-17), ENSG00000073969(2.982e-90), ENSG00000120071(4.184e-10), ENSG00000159314(0.0001098), ENSG00000186868(5.377e-07), ENSG00000225190(5.302e-06), ENSG00000236234(5.302e-06), ENSG00000239291(1.338e-06), ENSG00000256762(0.0001159), ENSG00000260075(3.06e-13), ENSG00000262265(1.338e-06), ENSG00000262372(7.016e-07), ENSG00000264038(0.0001098), ENSG00000264070(2.942e-32), ENSG00000264225(5.302e-06)	NA	NA	15_Quies		KANSL1, PLEKHM1	CDC27, NSF, KANSL1, ARHGAP27, MAPT, PLEKHM1, STH, NSFP1, DND1P1, RN7SL730P	NA	NA
chr17	44,301,840	rs142920272	rs142920272, rs147317628	2.9e-07, 1.1e-06			ENSG00000120071		15_Quies				KANSL1	
chr17	44,296,523	rs144416125	rs142920272	2.9e-07	ENSG00000108379(6.796e-05), ENSG00000120071(1.371e-15), ENSG00000158955(6.796e-05), ENSG00000186868(1.113e-08), ENSG00000225190(1.173e-17), ENSG00000262372(2.024e-10), ENSG00000262539(4.07e-09), ENSG00000264589(1.113e-08), ENSG00000266497(5.756e-25)	ENSG0000004897(3.849e-37), ENSG00000108379(1.454e-06), ENSG00000120071(1.453e-15), ENSG00000159314(1.815e-07), ENSG00000186868(7.968e-07), ENSG00000225190(2.236e-12), ENSG00000236234(2.236e-12), ENSG00000239291(8.887e-15), ENSG00000256762(7.075e-06), ENSG00000260075(1.26e-41), ENSG00000262265(8.887e-15), ENSG00000262372(1.283e-13), ENSG00000262879(1.357e-09), ENSG00000264038(3.203e-07), ENSG00000264225(2.236e-12), ENSG00000266497(9.576e-13)	NA	NA	5_TxWk		WNT3, KANSL1, WNT9B, MAPT, PLEKHM1, MAPT-AS1	CDC27, WNT3, KANSL1, ARHGAP27, MAPT, PLEKHM1, STH, NSFP1, RN7SL730P	NA	NA
chr3	62,481,063	rs1452075	rs1452075	2.1e-07	NA	ENSG00000153266(1.642e-05), ENSG00000241472(1.642e-05)	NA	NA	15_Quies		NA	FEZF2, PTPRG-AS1	NA	NA
chr17	44,277,476	rs147317628	rs142920272, rs147317628	2.9e-07, 1.1e-06	ENSG00000120071(5.738e-08), ENSG00000228696(6.206e-181), ENSG00000260075(6.206e-181)	ENSG00000120071(8.6e-08), ENSG00000185829(0), ENSG00000238083(0), ENSG00000262500(1.699e-08)	NA	NA	5_TxWk		KANSL1, ARL17B, NSFP1	KANSL1, ARL17A, LRR37A2	NA	NA
chr6	98,553,894	rs1487441	rs72934503	5.9e-07	ENSG00000187472(2.472e-05)	ENSG00000236920(6.072e-05), ENSG00000271860(6.072e-05)	NA	NA	15_Quies				NA	NA
chr6	98,565,211	rs1487445	rs72934503	5.9e-07	NA	NA	NA	NA	7_Enh		NA	NA	NA	NA
chr17	44,285,982	rs149187563	rs147317628	1.1e-06	ENSG00000006062(7.889e-05), ENSG00000120071(6.771e-18), ENSG00000186868(4.302e-05), ENSG00000228696(8.171e-82), ENSG00000260075(8.171e-82), ENSG00000262500(4.334e-75)	ENSG00000120071(8.907e-09), ENSG00000131484(1.778e-10), ENSG00000176681(0), ENSG00000185829(2.706e-16), ENSG00000214425(1.778e-10), ENSG00000225190(1.778e-10), ENSG00000228696(0), ENSG00000238083(2.706e-16), ENSG00000262500(1.013e-28), ENSG00000262879(1.557e-07)	NA	NA	5_TxWk		MAP3K14, KANSL1, MAPT, ARL17B, NSFP1	KANSL1, LRR37A, ARL17A, LRR37A4P, PLEKHM1, ARL17B, LRR37A2	NA	NA
chr19	37,980,494	rs149923766	rs138867053	1.2e-07	NA	NA	NA	NA	9_Het		NA	NA	NA	NA
chr8	48,402,014	rs150271817	rs183563276n	1.9e-07				ENSG00000164808	15_Quies					SPIDR
chr1	96,543,995	rs150859	rs201910565	3.4e-07	NA	NA	NA	NA	15_Quies		NA	NA	NA	NA
chr8	53,367,501	rs1582846	rs10666089m	1.0e-06	NA	NA	NA	NA	15_Quies		NA	NA	NA	NA
chr8	53,371,160	rs1627458	rs10666089m	1.0e-06	NA	NA	NA	NA	15_Quies		NA	NA	NA	NA
chr8	53,367,337	rs1660550	rs10666089m	1.0e-06	NA	NA	NA	NA	15_Quies		NA	NA	NA	NA
chr8	53,343,969	rs1660554	rs10666089m	1.0e-06	NA	NA	NA	NA	15_Quies	H3K4me1	NA	NA	NA	NA
chr17	44,186,063	rs16940904	rs147317628	1.1e-06	ENSG00000131484(6.16e-22), ENSG00000176681(4.632e-15), ENSG00000214425(6.16e-22), ENSG00000228696(4.28e-59), ENSG00000238083(1.129e-08), ENSG00000260075(4.28e-59), ENSG00000266497(1.129e-08), ENSG00000266918(6.16e-22)	ENSG00000120071(3.253e-05), ENSG00000238723(3.253e-05), ENSG00000262500(3.755e-05)	NA	NA	15_Quies		LRR37A, LRR37A4P, ARL17B, LRR37A2, NSFP1	KANSL1	NA	NA

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Supplementary Table 16 – continued from previous page

CHR	BP	CredSNP	IndexSNP	Index P	ENSGID_CP	ENSGID_GZ	ENSGID Promoter	ENSGID functional	Chrom HMM	Histone	HGNC_CP	HGNC_GZ	HGNC Promoter	HGNC functional
chr8	53,365,864	rs176184	rs10666089m	1.0e-06	NA	NA	NA	NA	15_Quies		NA	NA	NA	NA
chr17	44,196,015	rs17661141	rs147317628	1.1e-06	ENSG00000108433(1.024e-12), ENSG00000238083(1.869e-07), ENSG00000262633(1.024e-12), ENSG00000262879(1.024e-12), ENSG00000263142(1.024e-12), ENSG00000264070(2.525e-10), ENSG00000266497(1.869e-07)	ENSG00000262633(7.751e-14), ENSG00000262879(7.751e-14), ENSG00000263142(7.751e-14)	NA	NA	15_Quies	H3K4me1	GOSR2, LRR37A2, LRR37A17P, DND1P1	LRR37A17P	NA	NA
chr6	98,585,502	rs17814604	rs72934503	5.9e-07	NA	NA	NA	NA	15_Quies	DHS	NA	NA	NA	NA
chr8	53,346,704	rs182262	rs10666089m	1.0e-06	NA	NA	NA	NA	15_Quies		NA	NA	NA	NA
chr8	48,036,474	rs183563276	rs183563276m	1.9e-07	ENSG00000188873(7.505e-06), ENSG00000248347(7.505e-06), ENSG00000248498(2.381e-05), ENSG00000251470(7.116e-08)	ENSG00000164808(4.108e-06), ENSG00000251470(6.413e-14)	NA	NA	15_Quies		RPL10AP2, ASNSP1, ASNSP4	SPIDR, ASNSP4	NA	NA
chr8	48,036,474	rs183563276m	rs183563276m	1.9e-07	ENSG00000188873(7.505e-06), ENSG00000248347(7.505e-06), ENSG00000248498(2.381e-05), ENSG00000251470(7.116e-08)	ENSG00000164808(4.108e-06), ENSG00000251470(6.413e-14)	NA	NA	15_Quies		RPL10AP2, ASNSP1, ASNSP4	SPIDR, ASNSP4	NA	NA
chr6	98,576,688	rs1872841	rs72934503	5.9e-07	NA	NA	NA	NA	5_TxWk		NA	NA	NA	NA
chr8	53,365,524	rs188209	rs10666089m	1.0e-06	NA	NA	NA	NA	15_Quies		NA	NA	NA	NA
chr20	21,128,127	rs1884763	rs6047270	7.7e-08	NA	NA	NA	NA	5_TxWk		NA	NA	NA	NA
chr17	44,299,864	rs201443147	rs142920272, rs147317628	2.9e-07, 1.1e-06	ENSG00000108379(6.796e-05), ENSG00000120071(1.371e-15), ENSG00000158955(6.796e-05), ENSG00000186868(1.113e-08), ENSG00000225190(1.173e-17), ENSG00000262372(2.024e-10), ENSG00000262539(4.07e-09), ENSG00000264589(1.113e-08), ENSG00000266497(5.756e-25)	ENSG00000004897(3.849e-37), ENSG00000108379(1.454e-06), ENSG00000120071(1.453e-15), ENSG00000159314(1.815e-07), ENSG00000186868(7.968e-07), ENSG00000225190(2.236e-12), ENSG00000236234(2.236e-12), ENSG00000239291(8.887e-15), ENSG00000256762(7.075e-06), ENSG00000260075(1.26e-41), ENSG0000026265(8.887e-15), ENSG00000262372(1.283e-13), ENSG00000262879(1.357e-09), ENSG00000264038(3.203e-07), ENSG00000264225(2.236e-12), ENSG00000266497(9.576e-13)	NA	NA	15_Quies		WNT3, KANSL1, WNT9B, MAPT, PLEKHM1, MAPT-AS1	CDC27, WNT3, KANSL1, ARHGAP27, MAPT, PLEKHM1, STH, NSFP1, RN7SL730P	NA	NA
chr20	21,298,446	rs202829	rs910805	2.0e-09	ENSG00000188559(5.21e-05)	ENSG00000088930(8.115e-07)	NA	NA	15_Quies		RALGAPA2	XRN2	NA	NA
chr20	21,312,288	rs202837	rs910805	2.0e-09	NA	NA	NA	NA	4_Tx		NA	NA	NA	NA
chr17	44,155,732	rs2066899	rs142920272, rs147317628	2.9e-07, 1.1e-06	ENSG00000108433(1.738e-17), ENSG00000120071(9.114e-05), ENSG00000176681(2.996e-19), ENSG00000214401(9.114e-05), ENSG00000228696(2.996e-19), ENSG00000260075(7.901e-09), ENSG00000262633(1.738e-17), ENSG00000262879(1.738e-17), ENSG00000263142(1.738e-17)	ENSG00000073969(0), ENSG00000225190(2.979e-06)	NA	NA	15_Quies		GOSR2, KANSL1, LRR37A, KANSL1-AS1, ARL17B, NSFP1, LRR37A17P	NSF, PLEKHM1	NA	NA
chr20	21,140,342	rs2093069	rs6047270	7.7e-08			ENSG00000228604		5_TxWk					
chr17	44,168,677	rs2097760	rs142920272	2.9e-07	ENSG00000120071(1.319e-14), ENSG00000214401(1.319e-14), ENSG00000260075(9.54e-06)	ENSG00000073969(1.964e-37), ENSG00000120071(1.862e-09), ENSG00000176681(5.414e-10), ENSG00000228696(5.414e-10), ENSG00000238723(1.862e-09), ENSG00000262500(1.094e-09)	NA	NA	15_Quies		KANSL1, KANSL1-AS1, NSFP1	NSF, KANSL1, LRR37A, ARL17B	NA	NA
chr20	21,156,960	rs2103976	rs6047270	7.7e-08	NA	NA	NA	NA	15_Quies		NA	NA	NA	NA
chr6	11,731,999	rs210894	rs210894m	4.9e-07				ENSG00000111863	7_Enh					ADTRP
chr6	11,731,999	rs210894m	rs210894m	4.9e-07				ENSG00000111863	7_Enh					ADTRP
chr5	104,013,782	rs21126	rs325485	3.3e-07	NA	ENSG00000251574(2.86e-08), ENSG00000253584(2.86e-08)	NA	NA	15_Quies		NA		NA	NA

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Supplementary Table 16 – continued from previous page

CHR	BP	CredSNP	IndexSNP	Index P	ENSGID_CP	ENSGID_GZ	ENSGID Promoter	ENSGID functional	Chrom HMM	Histone	HGNC_CP	HGNC_GZ	HGNC Promoter	HGNC functional
chr17	44,268,488	rs21421298	rs142920272, rs147317628	2.9e-07, 1.1e-06	ENSG00000120071(9.613e-15), ENSG00000186868(1.943e-06), ENSG00000228696(1.763e-05), ENSG00000260075(1.763e-05), ENSG00000262372(2.229e-05)	ENSG00000131484(6.359e-16), ENSG00000176681(0), ENSG00000185829(4.033e-101), ENSG00000214425(6.359e-16), ENSG00000228696(0), ENSG00000238083(4.033e-101), ENSG00000262879(5.357e-06)	NA	NA	1_TssA		KANSL1, MAPT, ARL17B, NSFP1	LRR37A, ARL17A, LRR37A4P, ARL17B, LRR37A2	NA	NA
chr20	21,151,852	rs2180581	rs6047270	7.7e-08	NA	NA	NA	NA	15_Quies		NA	NA	NA	NA
chr20	14,766,216	rs2224275	rs11481126	1.3e-07	NA	NA	NA	NA	15_Quies		NA	NA	NA	NA
chr1	96,526,728	rs222887	rs201910565	3.4e-07	NA	ENSG00000117569(4.103e-05)	NA	NA	15_Quies		NA	PTBP2	NA	NA
chr1	96,525,762	rs222888	rs201910565	3.4e-07	NA	ENSG00000117569(4.103e-05)	NA	NA	15_Quies		NA	PTBP2	NA	NA
chr1	96,508,040	rs222901	rs201910565	3.4e-07	NA	NA	NA	NA	15_Quies		NA	NA	NA	NA
chr1	96,552,025	rs223245	rs201910565	3.4e-07	NA	NA	NA	NA	15_Quies		NA	NA	NA	NA
chr1	96,522,970	rs223250	rs201910565	3.4e-07	NA	ENSG00000117569(4.103e-05)	NA	NA	15_Quies		NA	PTBP2	NA	NA
chr20	21,142,813	rs2236178	rs6047270	7.7e-08	NA	NA	NA	NA	5_TxWk		NA	NA	NA	NA
chr17	44,294,786	rs2266497	rs147317628	1.1e-06	ENSG00000108379(6.796e-05), ENSG00000120071(1.371e-15), ENSG00000158955(6.796e-05), ENSG00000186868(1.113e-08), ENSG00000225190(1.173e-17), ENSG00000262372(2.024e-10), ENSG00000262539(4.07e-09), ENSG00000264589(1.113e-08), ENSG00000266497(5.756e-25)	ENSG00000004897(3.849e-37), ENSG00000108379(1.454e-06), ENSG00000120071(1.453e-15), ENSG00000159314(1.815e-07), ENSG00000186868(7.968e-07), ENSG00000225190(2.236e-12), ENSG00000236234(2.236e-12), ENSG00000239291(8.887e-15), ENSG00000256762(7.075e-06), ENSG00000260075(1.26e-41), ENSG00000262655(8.887e-15), ENSG00000262372(1.283e-13), ENSG00000262879(1.357e-09), ENSG00000264038(3.203e-07), ENSG00000264225(2.236e-12), ENSG00000266497(9.576e-13)	NA	NA	5_TxWk		WNT3, KANSL1, WNT9B, MAPT, PLEKHM1, MAPT-AS1	CDC27, WNT3, KANSL1, ARHGAP27, MAPT, PLEKHM1, STH, NSFP1, RN7SL730P	NA	NA
chr1	104,789,431	rs2317081	rs11185408	7.0e-07	NA	NA	NA	NA	15_Quies		NA	NA	NA	NA
chr20	21,153,138	rs2328608	rs6047270	7.7e-08	NA	NA	NA	NA	15_Quies		NA	NA	NA	NA
chr20	21,156,578	rs2328609	rs6047270	7.7e-08	NA	NA	NA	NA	15_Quies		NA	NA	NA	NA
chr1	96,978,961	rs2391769	rs2391769	1.1e-07	NA	NA	NA	NA	7_Enh	DHS	NA	NA	NA	NA
chr17	44,293,963	rs2458204	rs142920272, rs147317628	2.9e-07, 1.1e-06			ENSG00000238723		5_TxWk					
chr17	44,273,218	rs2532233	rs142920272, rs147317628	2.9e-07, 1.1e-06			ENSG00000120071, ENSG00000214401		5_TxWk	H3K4me1			KANSL1, KANSL1- AS1	
chr17	44,266,227	rs2532239	rs142920272, rs147317628	2.9e-07, 1.1e-06	ENSG00000120071(9.613e-15), ENSG00000186868(1.943e-06), ENSG00000228696(1.763e-05), ENSG00000260075(1.763e-05), ENSG00000262372(2.229e-05)	ENSG00000131484(6.359e-16), ENSG00000176681(0), ENSG00000185829(4.033e-101), ENSG00000214425(6.359e-16), ENSG00000228696(0), ENSG00000238083(4.033e-101), ENSG00000262879(5.357e-06)	NA	NA	7_Enh	H3K4me1	KANSL1, MAPT, ARL17B, NSFP1	LRR37A, ARL17A, LRR37A4P, ARL17B, LRR37A2	NA	NA
chr17	44,246,624	rs2532276	rs142920272, rs147317628	2.9e-07, 1.1e-06	ENSG00000176681(0), ENSG00000228696(0), ENSG00000260075(8.89e-10)	ENSG00000176681(2.828e-09), ENSG00000228696(2.597e-31), ENSG00000238083(1.646e-06), ENSG00000260075(3.47e-99), ENSG00000266497(1.646e-06)	NA	NA	5_TxWk		LRR37A, ARL17B, NSFP1	LRR37A, ARL17B, LRR37A2, NSFP1	NA	NA

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Supplementary Table 16 – continued from previous page

CHR	BP	CredSNP	IndexSNP	Index P	ENSGID_CP	ENSGID_GZ	ENSGID Promoter	ENSGID functional	Chrom HMM	Histone	HGNC_CP	HGNC_GZ	HGNC Promoter	HGNC functional
chr17	44,307,193	rs2532395	rs147317628	1.1e-06	ENSG00000120071(6.42e-18), ENSG00000186868(5.937e-08), ENSG00000225190(1.552e-14), ENSG00000262372(3.52e-09), ENSG00000264589(5.937e-08), ENSG00000266497(1.564e-21)	ENSG00000004897(1.111e-36), ENSG00000108379(2.723e-06), ENSG00000120071(5.97e-15), ENSG00000159314(1.602e-07), ENSG00000186868(1.111e-06), ENSG00000225190(2.259e-11), ENSG00000236234(2.259e-11), ENSG00000239291(4.569e-13), ENSG00000256762(5.986e-06), ENSG00000260075(1.762e-39), ENSG00000262265(4.569e-13), ENSG00000262372(1.069e-12), ENSG00000262879(4.408e-09), ENSG00000264038(8.752e-07), ENSG00000264225(2.259e-11), ENSG00000266497(7.218e-12)	NA	NA	15_Quies	KANSL1, MAPT, PLEKHM1, MAPT-AS1	CDC27, WNT3, KANSL1, ARHGAP27, MAPT, PLEKHM1, STH, NSFP1, RN7SL730P	NA	NA	
chr17	44,294,983	rs2532413	rs142920272, rs147317628	2.9e-07, 1.1e-06	ENSG00000108379(6.796e-05), ENSG00000120071(1.371e-15), ENSG00000158955(6.796e-05), ENSG00000186868(1.113e-08), ENSG00000225190(1.173e-17), ENSG00000262372(2.024e-10), ENSG00000262539(4.07e-09), ENSG00000264589(1.113e-08), ENSG00000266497(5.756e-25)	ENSG00000004897(3.849e-37), ENSG00000108379(1.454e-06), ENSG00000120071(1.453e-15), ENSG00000159314(1.815e-07), ENSG00000186868(7.968e-07), ENSG00000225190(2.236e-12), ENSG00000236234(2.236e-12), ENSG00000239291(8.887e-15), ENSG00000256762(7.075e-06), ENSG00000260075(1.26e-41), ENSG00000262265(8.887e-15), ENSG00000262372(1.283e-13), ENSG00000262879(1.357e-09), ENSG00000264038(3.203e-07), ENSG00000264225(2.236e-12), ENSG00000266497(9.576e-13)	NA	NA	5_TxWk	WNT3, KANSL1, WNT9B, MAPT, PLEKHM1, MAPT-AS1	CDC27, WNT3, KANSL1, ARHGAP27, MAPT, PLEKHM1, STH, NSFP1, RN7SL730P	NA	NA	
chr17	44,289,220	rs2532417	rs142920272, rs147317628	2.9e-07, 1.1e-06	ENSG00000006062(7.889e-05), ENSG00000120071(6.771e-18), ENSG00000186868(4.302e-05), ENSG00000228696(8.171e-82), ENSG00000260075(8.171e-82), ENSG00000262500(4.334e-75)	ENSG00000120071(8.907e-09), ENSG00000131484(1.778e-10), ENSG00000176681(0), ENSG00000185829(2.706e-16), ENSG00000214425(1.778e-10), ENSG00000225190(1.778e-10), ENSG00000228696(0), ENSG00000238083(2.706e-16), ENSG00000262500(1.013e-28), ENSG00000262879(1.557e-07)	NA	NA	5_TxWk	MAP3K14, KANSL1, MAPT, ARL17B, NSFP1	KANSL1, LRRC37A, ARL17A, LRRC37A4P, PLEKHM1, ARL17B, LRRC37A2	NA	NA	
chr17	44,288,579	rs2532418	rs142920272, rs147317628	2.9e-07, 1.1e-06	ENSG00000006062(7.889e-05), ENSG00000120071(6.771e-18), ENSG00000186868(4.302e-05), ENSG00000228696(8.171e-82), ENSG00000260075(8.171e-82), ENSG00000262500(4.334e-75)	ENSG00000120071(8.907e-09), ENSG00000131484(1.778e-10), ENSG00000176681(0), ENSG00000185829(2.706e-16), ENSG00000214425(1.778e-10), ENSG00000225190(1.778e-10), ENSG00000228696(0), ENSG00000238083(2.706e-16), ENSG00000262500(1.013e-28), ENSG00000262879(1.557e-07)	NA	NA	5_TxWk	MAP3K14, KANSL1, MAPT, ARL17B, NSFP1	KANSL1, LRRC37A, ARL17A, LRRC37A4P, PLEKHM1, ARL17B, LRRC37A2	NA	NA	
chr17	44,285,952	rs2532423	rs147317628	1.1e-06	ENSG00000006062(7.889e-05), ENSG00000120071(6.771e-18), ENSG00000186868(4.302e-05), ENSG00000228696(8.171e-82), ENSG00000260075(8.171e-82), ENSG00000262500(4.334e-75)	ENSG00000120071(8.907e-09), ENSG00000131484(1.778e-10), ENSG00000176681(0), ENSG00000185829(2.706e-16), ENSG00000214425(1.778e-10), ENSG00000225190(1.778e-10), ENSG00000228696(0), ENSG00000238083(2.706e-16), ENSG00000262500(1.013e-28), ENSG00000262879(1.557e-07)	NA	NA	5_TxWk	MAP3K14, KANSL1, MAPT, ARL17B, NSFP1	KANSL1, LRRC37A, ARL17A, LRRC37A4P, PLEKHM1, ARL17B, LRRC37A2	NA	NA	
chr8	53,373,448	rs2582639	rs10666089m	1.0e-06	NA	NA	NA	NA	15_Quies		NA	NA	NA	NA

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Supplementary Table 16 – continued from previous page

CHR	BP	CredSNP	IndexSNP	Index P	ENSGID_CP	ENSGID_GZ	ENSGID Promoter	ENSGID functional	Chrom HMM	Histone	HGNC_CP	HGNC_GZ	HGNC Promoter	HGNC functional
chr17	44,324,539	rs2668639	rs147317628	1.1e-06	ENSG00000120071(4.406e-45), ENSG00000225190(1.681e-08), ENSG00000262372(6.759e-07), ENSG00000266497(1.582e-08)	ENSG00000004897(2.896e-17), ENSG00000073969(2.982e-90), ENSG00000120071(4.184e-10), ENSG00000159314(0.0001098), ENSG00000186868(5.377e-07), ENSG00000225190(5.302e-06), ENSG00000236234(5.302e-06), ENSG00000239291(1.338e-06), ENSG00000256762(0.0001159), ENSG00000260075(3.06e-13), ENSG00000262265(1.338e-06), ENSG00000262372(7.016e-07), ENSG00000264038(0.0001098), ENSG00000264070(2.942e-32), ENSG00000264225(5.302e-06)	NA	NA	15_Quies	KANSL1, PLEKHM1	CDC27, NSF, KANSL1, ARHGAP27, MAPT, PLEKHM1, STH, NSFP1, DND1P1, RN7SL730P	NA	NA	
chr17	44,288,640	rs2668645	rs142920272	2.9e-07	ENSG00000006062(7.889e-05), ENSG00000120071(6.771e-18), ENSG00000186868(4.302e-05), ENSG00000228696(8.171e-82), ENSG00000260075(8.171e-82), ENSG00000262500(4.334e-75)	ENSG00000120071(8.907e-09), ENSG00000131484(1.778e-10), ENSG00000176681(0), ENSG00000185829(2.706e-16), ENSG00000214425(1.778e-10), ENSG00000225190(1.778e-10), ENSG00000228696(0), ENSG00000238083(2.706e-16), ENSG00000262500(1.013e-28), ENSG00000262879(1.557e-07)	NA	NA	5_TxWk	MAP3K14, KANSL1, MAPT, ARL17B, NSFP1	KANSL1, LRR37A, ARL17A, LRR37A4P, PLEKHM1, ARL17B, LRR37A2	NA	NA	
chr17	44,288,156	rs2668653	rs142920272, rs147317628	2.9e-07, 1.1e-06	ENSG00000006062(7.889e-05), ENSG00000120071(6.771e-18), ENSG00000186868(4.302e-05), ENSG00000228696(8.171e-82), ENSG00000260075(8.171e-82), ENSG00000262500(4.334e-75)	ENSG00000120071(8.907e-09), ENSG00000131484(1.778e-10), ENSG00000176681(0), ENSG00000185829(2.706e-16), ENSG00000214425(1.778e-10), ENSG00000225190(1.778e-10), ENSG00000228696(0), ENSG00000238083(2.706e-16), ENSG00000262500(1.013e-28), ENSG00000262879(1.557e-07)	NA	NA	5_TxWk	MAP3K14, KANSL1, MAPT, ARL17B, NSFP1	KANSL1, LRR37A, ARL17A, LRR37A4P, PLEKHM1, ARL17B, LRR37A2	NA	NA	
chr17	44,290,759	rs2668662	rs142920272, rs147317628	2.9e-07, 1.1e-06	ENSG00000108379(6.796e-05), ENSG00000120071(1.371e-15), ENSG00000158955(6.796e-05), ENSG00000186868(1.113e-08), ENSG00000225190(1.173e-17), ENSG00000262372(2.024e-10), ENSG00000262539(4.07e-09), ENSG00000264589(1.113e-08), ENSG00000266497(5.756e-25)	ENSG00000004897(3.849e-37), ENSG00000108379(1.454e-06), ENSG00000120071(1.453e-15), ENSG00000159314(1.815e-07), ENSG00000186868(7.968e-07), ENSG00000225190(2.236e-12), ENSG00000236234(2.236e-12), ENSG00000239291(8.887e-15), ENSG00000256762(7.075e-06), ENSG00000260075(1.26e-41), ENSG00000262265(8.887e-15), ENSG00000262372(1.283e-13), ENSG00000262879(1.357e-09), ENSG00000264038(3.203e-07), ENSG00000264225(2.236e-12), ENSG00000266497(9.576e-13)	NA	NA	5_TxWk	WNT3, KANSL1, WNT9B, MAPT, PLEKHM1, MAPT-AS1	CDC27, WNT3, KANSL1, ARHGAP27, MAPT, PLEKHM1, STH, NSFP1, RN7SL730P	NA	NA	
chr17	44,290,047	rs2668665	rs142920272, rs147317628	2.9e-07, 1.1e-06	ENSG00000108379(6.796e-05), ENSG00000120071(1.371e-15), ENSG00000158955(6.796e-05), ENSG00000186868(1.113e-08), ENSG00000225190(1.173e-17), ENSG00000262372(2.024e-10), ENSG00000262539(4.07e-09), ENSG00000264589(1.113e-08), ENSG00000266497(5.756e-25)	ENSG00000004897(3.849e-37), ENSG00000108379(1.454e-06), ENSG00000120071(1.453e-15), ENSG00000159314(1.815e-07), ENSG00000186868(7.968e-07), ENSG00000225190(2.236e-12), ENSG00000236234(2.236e-12), ENSG00000239291(8.887e-15), ENSG00000256762(7.075e-06), ENSG00000260075(1.26e-41), ENSG00000262265(8.887e-15), ENSG00000262372(1.283e-13), ENSG00000262879(1.357e-09), ENSG00000264038(3.203e-07), ENSG00000264225(2.236e-12), ENSG00000266497(9.576e-13)	NA	NA	5_TxWk	WNT3, KANSL1, WNT9B, MAPT, PLEKHM1, MAPT-AS1	CDC27, WNT3, KANSL1, ARHGAP27, MAPT, PLEKHM1, STH, NSFP1, RN7SL730P	NA	NA	

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Supplementary Table 16 – continued from previous page

CHR	BP	CredSNP	IndexSNP	Index P	ENSGID_CP	ENSGID_GZ	ENSGID Promoter	ENSGID functional	Chrom HMM	Histone	HGNC_CP	HGNC_GZ	HGNC Promoter	HGNC functional
chr17	44,289,628	rs2668670	rs147317628	1.1e-06	ENSG0000006062(7.889e-05), ENSG00000120071(6.771e-18), ENSG00000186868(4.302e-05), ENSG00000228696(8.171e-82), ENSG00000260075(8.171e-82), ENSG00000262500(4.334e-75)	ENSG00000120071(8.907e-09), ENSG00000131484(1.778e-10), ENSG00000176681(0), ENSG00000185829(2.706e-16), ENSG00000214425(1.778e-10), ENSG00000225190(1.778e-10), ENSG00000228696(0), ENSG00000238083(2.706e-16), ENSG00000262500(1.013e-28), ENSG00000262879(1.557e-07)	NA	NA	5_TxWk		MAP3K14, KANSL1, MAPT, ARL17B, NSFP1	KANSL1, LRR37A, ARL17A, LRR37A4P, PLEKHM1, ARL17B, LRR37A2	NA	NA
chr17	44,292,319	rs2668694	rs147317628	1.1e-06	ENSG00000108379(6.796e-05), ENSG00000120071(1.371e-15), ENSG00000158955(6.796e-05), ENSG00000186868(1.113e-08), ENSG00000225190(1.173e-17), ENSG00000262372(2.024e-10), ENSG00000262539(4.07e-09), ENSG00000264589(1.113e-08), ENSG00000266497(5.756e-25)	ENSG00000004897(3.849e-37), ENSG00000108379(1.454e-06), ENSG00000120071(1.453e-15), ENSG00000159314(1.815e-07), ENSG00000186868(7.968e-07), ENSG00000225190(2.236e-12), ENSG00000236234(2.236e-12), ENSG00000239291(8.887e-15), ENSG00000256762(7.075e-06), ENSG00000260075(1.26e-41), ENSG00000262265(8.887e-15), ENSG00000262372(1.283e-13), ENSG00000262879(1.357e-09), ENSG00000264038(3.203e-07), ENSG00000264225(2.236e-12), ENSG00000266497(9.576e-13)	NA	NA	5_TxWk	H3K36me3	WNT3, KANSL1, WNT9B, MAPT, PLEKHM1, MAPT-AS1	CDC27, WNT3, KANSL1, ARHGAP27, MAPT, PLEKHM1, STH, NSFP1, RN7SL730P	NA	NA
chr17	44,266,531	rs2696566	rs142920272, rs147317628	2.9e-07, 1.1e-06	ENSG00000120071(9.613e-15), ENSG00000186868(1.943e-06), ENSG00000228696(1.763e-05), ENSG00000260075(1.763e-05), ENSG00000262372(2.229e-05)	ENSG00000131484(6.359e-16), ENSG00000176681(0), ENSG00000185829(4.033e-101), ENSG00000214425(6.359e-16), ENSG00000228696(0), ENSG00000238083(4.033e-101), ENSG00000262879(5.357e-06)	NA	NA	7_Enh	H3K4me1	KANSL1, MAPT, ARL17B, NSFP1	LRR37A, ARL17A, LRR37A4P, ARL17B, LRR37A2	NA	NA
chr17	44,266,342	rs2696568	rs142920272, rs147317628	2.9e-07, 1.1e-06	ENSG00000120071(9.613e-15), ENSG00000186868(1.943e-06), ENSG00000228696(1.763e-05), ENSG00000260075(1.763e-05), ENSG00000262372(2.229e-05)	ENSG00000131484(6.359e-16), ENSG00000176681(0), ENSG00000185829(4.033e-101), ENSG00000214425(6.359e-16), ENSG00000228696(0), ENSG00000238083(4.033e-101), ENSG00000262879(5.357e-06)	NA	NA	7_Enh	H3K4me1	KANSL1, MAPT, ARL17B, NSFP1	LRR37A, ARL17A, LRR37A4P, ARL17B, LRR37A2	NA	NA
chr17	44,293,205	rs2696609	rs142920272	2.9e-07			ENSG00000238723		5_TxWk					
chr17	44,270,059	rs2696633	rs147317628	1.1e-06			ENSG00000214401		8_ZNF/Rpts	DHS:H3K4me3			KANSL1- AS1	
chr17	44,291,479	rs2732601	rs142920272	2.9e-07	ENSG00000108379(6.796e-05), ENSG00000120071(1.371e-15), ENSG00000158955(6.796e-05), ENSG00000186868(1.113e-08), ENSG00000225190(1.173e-17), ENSG00000262372(2.024e-10), ENSG00000262539(4.07e-09), ENSG00000264589(1.113e-08), ENSG00000266497(5.756e-25)	ENSG00000004897(3.849e-37), ENSG00000108379(1.454e-06), ENSG00000120071(1.453e-15), ENSG00000159314(1.815e-07), ENSG00000186868(7.968e-07), ENSG00000225190(2.236e-12), ENSG00000236234(2.236e-12), ENSG00000239291(8.887e-15), ENSG00000256762(7.075e-06), ENSG00000260075(1.26e-41), ENSG00000262265(8.887e-15), ENSG00000262372(1.283e-13), ENSG00000262879(1.357e-09), ENSG00000264038(3.203e-07), ENSG00000264225(2.236e-12), ENSG00000266497(9.576e-13)	NA	NA	5_TxWk		WNT3, KANSL1, WNT9B, MAPT, PLEKHM1, MAPT-AS1	CDC27, WNT3, KANSL1, ARHGAP27, MAPT, PLEKHM1, STH, NSFP1, RN7SL730P	NA	NA

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Supplementary Table 16 – continued from previous page

CHR	BP	CredSNP	IndexSNP	Index P	ENSGID_CP	ENSGID_GZ	ENSGID Promoter	ENSGID functional	Chrom HMM	Histone	HGNC_CP	HGNC_GZ	HGNC Promoter	HGNC functional
chr17	44,291,365	rs2732606	rs142920272	2.9e-07	ENSG00000108379(6.796e-05), ENSG00000120071(1.371e-15), ENSG00000158955(6.796e-05), ENSG00000186868(1.113e-08), ENSG00000225190(1.173e-17), ENSG00000262372(2.024e-10), ENSG00000262539(4.07e-09), ENSG00000264589(1.113e-08), ENSG00000266497(5.756e-25)	ENSG00000004897(3.849e-37), ENSG00000108379(1.454e-06), ENSG00000120071(1.453e-15), ENSG00000159314(1.815e-07), ENSG00000186868(7.968e-07), ENSG00000225190(2.236e-12), ENSG00000236234(2.236e-12), ENSG00000239291(8.887e-15), ENSG00000256762(7.075e-06), ENSG00000260075(1.26e-41), ENSG00000262265(8.887e-15), ENSG00000262372(1.283e-13), ENSG00000262879(1.357e-09), ENSG00000264038(3.203e-07), ENSG00000264225(2.236e-12), ENSG00000266497(9.576e-13)	NA	NA	5_TxWk	WNT3, KANSL1, WNT9B, MAPT, PLEKHM1, MAPT-AS1	CDC27, WNT3, KANSL1, ARHGAP27, MAPT, PLEKHM1, STH, NSFP1, RN7SL730P	NA	NA	
chr17	44,289,101	rs2732629	rs147317628	1.1e-06	ENSG00000006062(7.889e-05), ENSG00000120071(6.771e-18), ENSG00000186868(4.302e-05), ENSG00000228696(8.171e-82), ENSG00000260075(8.171e-82), ENSG00000262500(4.334e-75)	ENSG00000120071(8.907e-09), ENSG00000131484(1.778e-10), ENSG00000176681(0), ENSG00000185829(2.706e-16), ENSG00000214425(1.778e-10), ENSG00000225190(1.778e-10), ENSG00000228696(0), ENSG00000238083(2.706e-16), ENSG00000262500(1.013e-28), ENSG00000262879(1.557e-07)	NA	NA	5_TxWk	MAP3K14, KANSL1, MAPT, ARL17B, NSFP1	KANSL1, LRRC37A, ARL17A, LRRC37A4P, PLEKHM1, ARL17B, LRRC37A2	NA	NA	
chr17	44,289,232	rs2732631	rs142920272, rs147317628	2.9e-07, 1.1e-06	ENSG00000006062(7.889e-05), ENSG00000120071(6.771e-18), ENSG00000186868(4.302e-05), ENSG00000228696(8.171e-82), ENSG00000260075(8.171e-82), ENSG00000262500(4.334e-75)	ENSG00000120071(8.907e-09), ENSG00000131484(1.778e-10), ENSG00000176681(0), ENSG00000185829(2.706e-16), ENSG00000214425(1.778e-10), ENSG00000225190(1.778e-10), ENSG00000228696(0), ENSG00000238083(2.706e-16), ENSG00000262500(1.013e-28), ENSG00000262879(1.557e-07)	NA	NA	5_TxWk	MAP3K14, KANSL1, MAPT, ARL17B, NSFP1	KANSL1, LRRC37A, ARL17A, LRRC37A4P, PLEKHM1, ARL17B, LRRC37A2	NA	NA	
chr8	53,351,137	rs284800	rs10666089m	1.0e-06	ENSG00000147488(7.9e-07)	NA	NA	NA	15_Quies		ST18	NA	NA	NA
chr8	53,348,475	rs284801	rs10666089m	1.0e-06	NA	NA	NA	NA	15_Quies		NA	NA	NA	NA
chr8	53,368,585	rs284803	rs10666089m	1.0e-06	NA	NA	NA	NA	15_Quies		NA	NA	NA	NA
chr8	53,343,063	rs284810	rs10666089m	1.0e-06	NA	NA	NA	NA	15_Quies		NA	NA	NA	NA
chr8	53,338,714	rs284813	rs10666089m	1.0e-06	ENSG00000147485(8.966e-05), ENSG00000147488(5.221e-11)	ENSG00000147488(7.94e-08), ENSG00000253844(9.555e-06)	NA	NA	15_Quies		PXDNL, ST18	ST18	NA	NA
chr8	53,360,144	rs284814	rs10666089m	1.0e-06	NA	NA	NA	NA	15_Quies		NA	NA	NA	NA
chr8	53,361,934	rs284815	rs10666089m	1.0e-06	NA	NA	NA	NA	15_Quies		NA	NA	NA	NA
chr8	53,364,500	rs284819	rs10666089m	1.0e-06	NA	NA	NA	NA	15_Quies		NA	NA	NA	NA
chr20	21,158,371	rs2876596	rs6047270	7.7e-08	NA	NA	NA	NA	15_Quies		NA	NA	NA	NA
chr14	94,844,947	rs28929474	rs112635299	3.0e-07				ENSG00000197249	14_ReprPCWk					SERPINA1
chr18	55,888,468	rs292453	rs292441	1.1e-06			ENSG00000049759		5_TxWk				NEDD4L	
chr5	103,995,368	rs325485	rs325485	3.3e-07	NA	NA	NA	NA	15_Quies		NA	NA	NA	NA
chr5	104,007,433	rs325501	rs325485	3.3e-07	NA	NA	NA	NA	15_Quies		NA	NA	NA	NA
chr5	104,012,303	rs325506	rs325485	3.3e-07	NA	ENSG00000251574(2.86e-08), ENSG00000253584(2.86e-08)	NA	NA	15_Quies		NA	NA	NA	NA
chr8	53,336,933	rs34006528	rs10666089m	1.0e-06	ENSG00000147485(8.966e-05), ENSG00000147488(5.221e-11)	ENSG00000147488(7.94e-08), ENSG00000253844(9.555e-06)	NA	NA	15_Quies	DHS	PXDNL, ST18	ST18	NA	NA
chr6	98,591,074	rs34645063	rs72934503	5.9e-07	NA	NA	NA	NA	9_Het		NA	NA	NA	NA
chr17	44,288,672	rs34898647	rs142920272, rs147317628	2.9e-07, 1.1e-06	ENSG00000006062(7.889e-05), ENSG00000120071(6.771e-18), ENSG00000186868(4.302e-05), ENSG00000228696(8.171e-82), ENSG00000260075(8.171e-82), ENSG00000262500(4.334e-75)	ENSG00000120071(8.907e-09), ENSG00000131484(1.778e-10), ENSG00000176681(0), ENSG00000185829(2.706e-16), ENSG00000214425(1.778e-10), ENSG00000225190(1.778e-10), ENSG00000228696(0), ENSG00000238083(2.706e-16), ENSG00000262500(1.013e-28), ENSG00000262879(1.557e-07)	NA	NA	5_TxWk	MAP3K14, KANSL1, MAPT, ARL17B, NSFP1	KANSL1, LRRC37A, ARL17A, LRRC37A4P, PLEKHM1, ARL17B, LRRC37A2	NA	NA	

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Supplementary Table 16 – continued from previous page

CHR	BP	CredSNP	IndexSNP	Index P	ENSGID_CP	ENSGID_GZ	ENSGID Promoter	ENSGID functional	Chrom HMM	Histone	HGNC_CP	HGNC_GZ	HGNC Promoter	HGNC functional
chr8	53,353,660	rs366249	rs10666089m	1.0e-06	ENSG00000147488(7.9e-07)	NA	NA	NA	15_Quies		ST18	NA	NA	NA
chr6	98,584,720	rs368928614	rs72934503	5.9e-07	NA	NA	NA	NA	15_Quies		NA	NA	NA	NA
chr17	44,311,200	rs373392968	rs147317628	1.1e-06	ENSG00000120071(1.001e-75), ENSG00000186868(4.221e-08), ENSG00000225190(1.513e-15), ENSG00000262372(1.712e-09), ENSG00000264589(4.221e-08), ENSG00000266497(2.367e-21)	ENSG00000004897(5.898e-35), ENSG00000108379(3.433e-06), ENSG00000120071(6.373e-29), ENSG00000159314(2.318e-07), ENSG00000186868(5.531e-07), ENSG00000214401(1.613e-08), ENSG00000225190(2.693e-11), ENSG00000236234(2.693e-11), ENSG00000239291(2.76e-13), ENSG00000256762(5.458e-06), ENSG00000260075(2.959e-37), ENSG00000262265(2.76e-13), ENSG00000262372(5.801e-12), ENSG00000262879(1.682e-08), ENSG00000264038(1.084e-06), ENSG00000264225(2.693e-11), ENSG00000266497(1.002e-11)	NA	NA	15_Quies	KANSL1, MAPT, PLEKHM1, MAPT-AS1	CDC27, WNT3, KANSL1, ARHGAP27, MAPT, KANSL1-AS1, PLEKHM1, STH, NSFP1, RN7SL730P	NA	NA	
chr20	21,343,944	rs3848794	rs910805	2.0e-09	ENSG00000225803(2.479e-05)	ENSG00000088930(1.351e-06)	NA	NA	4_Tx		MRPS11P1	XRN2	NA	NA
chr1	96,984,894	rs3851274	rs2391769	1.1e-07	NA	NA	NA	NA	5_TxWk		NA	NA	NA	NA
chr20	14,832,798	rs386257	rs11481126	1.3e-07	NA	NA	NA	NA	15_Quies		NA	NA	NA	NA
chr17	44,165,169	rs3865315	rs142920272, rs147317628	2.9e-07, 1.1e-06	ENSG00000120071(1.319e-14), ENSG00000214401(1.319e-14), ENSG00000260075(9.54e-06)	ENSG00000073969(1.964e-37), ENSG00000120071(1.862e-09), ENSG00000176681(5.414e-10), ENSG00000228696(5.414e-10), ENSG00000238723(1.862e-09), ENSG00000262500(1.094e-09)	NA	NA	15_Quies		KANSL1, KANSL1-AS1, NSFP1	NSF, KANSL1, LRRC37A, ARL17B	NA	NA
chr20	14,817,453	rs389171	rs11481126	1.3e-07	NA	NA	NA	NA	15_Quies		NA	NA	NA	NA
chr20	14,825,067	rs447564	rs11481126	1.3e-07	NA	NA	NA	NA	15_Quies		NA	NA	NA	NA
chr10	16,691,399	rs45595836	rs45595836	3.1e-07	NA	ENSG00000148484(8.329e-05)	NA	NA	15_Quies		NA	RSU1	NA	NA
chr17	44,110,670	rs4597358	rs142920272, rs147317628	2.9e-07, 1.1e-06			ENSG00000120071		4_Tx	H3K36me3			KANSL1	
chr20	21,147,968	rs4625995	rs6047270	7.7e-08			ENSG00000214535		5_TxWk				RPS15AP1	
chr6	11,729,460	rs4713834	rs210894m	4.9e-07				ENSG00000111863	15_Quies					ADTRP
chr10	130,488,026	rs4750990	rs4750990	1.4e-06	NA	NA	NA	NA	14_ReprPCWk		NA	NA	NA	NA
chr18	55,880,518	rs477257	rs292441	1.1e-06	NA	ENSG00000049759(3.605e-07)	NA	NA	7_Enh		NA	NEDD4L	NA	NA
chr18	55,881,771	rs479865	rs292441	1.1e-06	NA	ENSG00000049759(3.605e-07)	NA	NA	7_Enh		NA	NEDD4L	NA	NA
chr20	21,133,212	rs4813414	rs6047270	7.7e-08	NA	NA	NA	NA	5_TxWk		NA	NA	NA	NA
chr20	21,124,872	rs4815022	rs6047270	7.7e-08			ENSG00000088970		15_Quies				PLK1S1	
chr20	21,138,367	rs4815024	rs6047270	7.7e-08	NA	NA	NA	NA	5_TxWk		NA	NA	NA	NA
chr20	21,149,971	rs4815027	rs6047270	7.7e-08	NA	NA	NA	NA	15_Quies		NA	NA	NA	NA
chr20	21,344,454	rs4815040	rs910805	2.0e-09	ENSG00000225803(2.479e-05)	ENSG00000088930(1.351e-06)	NA	NA	5_TxWk		MRPS11P1	XRN2	NA	NA
chr18	55,881,103	rs482805	rs292441	1.1e-06	NA	ENSG00000049759(3.605e-07)	NA	NA	7_Enh		NA	NEDD4L	NA	NA
chr18	55,877,856	rs526730	rs292441	1.1e-06	ENSG00000198796(4.493e-05), ENSG00000267579(4.493e-05)	NA	NA	NA	5_TxWk		ALPK2	NA	NA	NA
chr18	55,880,322	rs535188	rs292441	1.1e-06	NA	ENSG00000049759(3.605e-07)	NA	NA	5_TxWk		NA	NEDD4L	NA	NA
chr17	44,316,076	rs55653937	rs147317628	1.1e-06	ENSG00000120071(1.001e-75), ENSG00000186868(4.221e-08), ENSG00000225190(1.513e-15), ENSG00000262372(1.712e-09), ENSG00000264589(4.221e-08), ENSG00000266497(2.367e-21)	ENSG00000004897(5.898e-35), ENSG00000108379(3.433e-06), ENSG00000120071(6.373e-29), ENSG00000159314(2.318e-07), ENSG00000186868(5.531e-07), ENSG00000214401(1.613e-08), ENSG00000225190(2.693e-11), ENSG00000236234(2.693e-11), ENSG00000239291(2.76e-13), ENSG00000256762(5.458e-06), ENSG00000260075(2.959e-37), ENSG00000262265(2.76e-13), ENSG00000262372(5.801e-12), ENSG00000262879(1.682e-08), ENSG00000264038(1.084e-06), ENSG00000264225(2.693e-11), ENSG00000266497(1.002e-11)	NA	NA	15_Quies	KANSL1, MAPT, PLEKHM1, MAPT-AS1	CDC27, WNT3, KANSL1, ARHGAP27, MAPT, KANSL1-AS1, PLEKHM1, STH, NSFP1, RN7SL730P	NA	NA	

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Supplementary Table 16 – continued from previous page

CHR	BP	CredSNP	IndexSNP	Index P	ENSGID_CP	ENSGID_GZ	ENSGID Promoter	ENSGID functional	Chrom HMM	Histone	HGNC_CP	HGNC_GZ	HGNC Promoter	HGNC functional
chr20	21,242,161	rs55962189	rs55962189	3.0e-07	NA	ENSG00000226405(8.388e-08)	NA	NA	15_Quies		NA		NA	NA
chr18	55,879,203	rs566343	rs292441	1.1e-06	ENSG00000198796(4.493e-05), ENSG00000267579(4.493e-05)	NA	NA	NA	5_TxWk		ALPK2	NA	NA	NA
chr18	55,872,566	rs56724831	rs292441	1.1e-06	ENSG00000198796(4.493e-05), ENSG00000267579(4.493e-05)	NA	NA	NA	7_Enh	H3K4me1	ALPK2	NA	NA	NA
chr18	55,882,192	rs574626	rs292441	1.1e-06	NA	ENSG00000049759(3.605e-07)	NA	NA	7_Enh	H3K4me1	NA	NEDD4L	NA	NA
chr18	55,877,842	rs576451	rs292441	1.1e-06	ENSG00000198796(4.493e-05), ENSG00000267579(4.493e-05)	NA	NA	NA	5_TxWk		ALPK2	NA	NA	NA
chr20	21,401,057	rs57976745	rs910805	2.0e-09	NA	NA	NA	NA	13_ReprPC	H3K27me3:H3K9	NA	NA	NA	NA
chr1	96,588,432	rs58752701	rs201910565	3.4e-07	NA	NA	NA	NA	15_Quies		NA	NA	NA	NA
chr1	96,588,438	rs60135683	rs201910565	3.4e-07	NA	NA	NA	NA	15_Quies		NA	NA	NA	NA
chr19	37,284,873	rs60288454	rs138867053	1.2e-07	ENSG00000120784(1.563e-05), ENSG00000171817(1.563e-05)	ENSG00000120784(2.492e-05), ENSG00000171817(2.492e-05), ENSG00000226686(4.354e-05), ENSG00000232677(1.903e-05), ENSG00000267053(1.903e-05), ENSG00000267605(4.354e-05)	NA	NA	9_Het		ZFP30, ZNF540	ZFP30, ZNF540, LINC00665	NA	NA
chr20	21,121,739	rs6035788	rs6047270	7.7e-08	NA	NA	NA	NA	15_Quies		NA	NA	NA	NA
chr20	21,135,718	rs6035791	rs6047270	7.7e-08	NA	NA	NA	NA	5_TxWk		NA	NA	NA	NA
chr20	21,138,989	rs6035792	rs6047270	7.7e-08	NA	NA	NA	NA	5_TxWk		NA	NA	NA	NA
chr20	21,139,252	rs6035793	rs6047270	7.7e-08	NA	NA	NA	NA	5_TxWk		NA	NA	NA	NA
chr20	21,145,885	rs6035795	rs6047270	7.7e-08	NA	NA	NA	NA	5_TxWk		NA	NA	NA	NA
chr20	21,151,348	rs6035800	rs6047270	7.7e-08	NA	NA	NA	NA	15_Quies		NA	NA	NA	NA
chr20	21,153,782	rs6035803	rs6047270	7.7e-08	NA	NA	NA	NA	15_Quies		NA	NA	NA	NA
chr20	21,243,293	rs6035821	rs910805	2.0e-09	NA	ENSG00000226405(8.388e-08)	NA	NA	15_Quies		NA		NA	NA
chr20	21,332,449	rs6035852	rs910805	2.0e-09	NA	ENSG00000235065(1.844e-05)	NA	NA	5_TxWk		NA	RPL24P2	NA	NA
chr8	10,572,617	rs60410697	rs10099100	1.1e-08	NA	ENSG00000171056(9.841e-05), ENSG00000171060(1.154e-09), ENSG00000183638(1.154e-09), ENSG00000212433(1.154e-09), ENSG00000252565(9.841e-05), ENSG00000254093(9.841e-05), ENSG00000258724(9.841e-05)	NA	NA	14_ReprPCWk		NA	SOX7, C8orf74, RP1L1, RNA5SP252, PINX1	NA	NA
chr20	21,126,194	rs6047271	rs6047270	7.7e-08			ENSG00000088970		15_Quies				PLK1S1	
chr20	21,134,799	rs6047273	rs6047270	7.7e-08	NA	NA	NA	NA	5_TxWk		NA	NA	NA	NA
chr20	21,139,382	rs6047278	rs6047270	7.7e-08			ENSG00000228604		5_TxWk					
chr20	21,143,935	rs6047280	rs6047270	7.7e-08			ENSG00000228604		5_TxWk					
chr20	21,146,468	rs6047282	rs6047270	7.7e-08	NA	NA	NA	NA	5_TxWk		NA	NA	NA	NA
chr20	21,147,009	rs6047283	rs6047270	7.7e-08	NA	NA	NA	NA	5_TxWk		NA	NA	NA	NA
chr20	21,151,324	rs6047288	rs6047270	7.7e-08	NA	NA	NA	NA	15_Quies		NA	NA	NA	NA
chr20	21,158,239	rs6047296	rs6047270	7.7e-08	NA	NA	NA	NA	15_Quies		NA	NA	NA	NA
chr20	21,331,541	rs6047396	rs910805	2.0e-09	NA	ENSG00000235065(1.844e-05)	NA	NA	5_TxWk	H3K36me3	NA	RPL24P2	NA	NA
chr20	21,152,231	rs6082338	rs6047270	7.7e-08	NA	NA	NA	NA	15_Quies		NA	NA	NA	NA
chr20	21,348,430	rs6082397	rs910805	2.0e-09	ENSG00000225803(2.479e-05)	ENSG00000088930(1.351e-06)	NA	NA	5_TxWk		MRPS11P1	XRN2	NA	NA
chr20	14,761,232	rs6110430	rs11481126	1.3e-07	NA	NA	NA	NA	15_Quies		NA	NA	NA	NA
chr20	21,150,267	rs6132402	rs6047270	7.7e-08	NA	NA	NA	NA	15_Quies		NA	NA	NA	NA
chr20	21,146,816	rs6137276	rs6047270	7.7e-08	NA	NA	NA	NA	5_TxWk		NA	NA	NA	NA
chr17	44,185,431	rs62061820	rs142920272, rs147317628	2.9e-07, 1.1e-06	ENSG00000131484(6.16e-22), ENSG00000176681(4.632e-15), ENSG00000214425(6.16e-22), ENSG00000228696(4.28e-59), ENSG00000238083(1.129e-08), ENSG00000260075(4.28e-59), ENSG00000266497(1.129e-08), ENSG00000266918(6.16e-22)	ENSG00000120071(3.253e-05), ENSG00000238723(3.253e-05), ENSG00000262500(3.755e-05)	NA	NA	15_Quies		LRR37A, LRR37A4P, ARL17B, LRR37A2, NSFP1	KANSL1	NA	NA

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Supplementary Table 16 – continued from previous page

CHR	BP	CredSNP	IndexSNP	Index P	ENSGID_CP	ENSGID_GZ	ENSGID Promoter	ENSGID functional	Chrom HMM	Histone	HGNC_CP	HGNC_GZ	HGNC Promoter	HGNC functional
chr17	44,096,541	rs62062287	rs147317628	1.1e-06	ENSG00000131484(4.629e-14), ENSG00000176681(2.617e-38), ENSG00000214425(4.629e-14), ENSG00000228696(2.617e-38), ENSG00000266918(4.629e-14)	ENSG00000120071(7.247e-07), ENSG00000228696(1.262e-09), ENSG00000238723(7.247e-07), ENSG00000262500(4.782e-07), ENSG00000262539(1.124e-06)	NA	NA	5_TxWk		LRR37A, LRR37A4P, ARL17B	KANSL1, ARL17B	NA	NA
chr17	44,096,541	rs62062287m	rs147317628	1.1e-06	ENSG00000131484(4.629e-14), ENSG00000176681(2.617e-38), ENSG00000214425(4.629e-14), ENSG00000228696(2.617e-38), ENSG00000266918(4.629e-14)	ENSG00000120071(7.247e-07), ENSG00000228696(1.262e-09), ENSG00000238723(7.247e-07), ENSG00000262500(4.782e-07), ENSG00000262539(1.124e-06)	NA	NA	5_TxWk		LRR37A, LRR37A4P, ARL17B	KANSL1, ARL17B	NA	NA
chr17	44,096,553	rs62062288	rs142920272, rs147317628	2.9e-07, 1.1e-06	ENSG00000131484(4.629e-14), ENSG00000176681(2.617e-38), ENSG00000214425(4.629e-14), ENSG00000228696(2.617e-38), ENSG00000266918(4.629e-14)	ENSG00000120071(7.247e-07), ENSG00000228696(1.262e-09), ENSG00000238723(7.247e-07), ENSG00000262500(4.782e-07), ENSG00000262539(1.124e-06)	NA	NA	5_TxWk		LRR37A, LRR37A4P, ARL17B	KANSL1, ARL17B	NA	NA
chr1	96,549,641	rs638729	rs201910565	3.4e-07	NA	NA	NA	NA	15_Quies		NA	NA	NA	NA
chr17	44,208,312	rs6503457	rs142920272, rs147317628	2.9e-07, 1.1e-06	ENSG00000185829(6.167e-37), ENSG00000238083(6.167e-37), ENSG00000262633(3.592e-08), ENSG00000262879(3.592e-08), ENSG00000263142(3.592e-08), ENSG00000263650(3.592e-08), ENSG00000265315(7.022e-29), ENSG00000266497(2.372e-25)	ENSG00000073969(2.084e-07), ENSG00000228696(1.23e-44), ENSG00000260075(1.23e-44), ENSG00000266497(2.657e-06)	NA	NA	7_Enh	H3K4me1	ARL17A, LRR37A2, LRR37A17P, RN7SL270P, RN7SL199P	NSF, ARL17B, NSFP1	NA	NA
chr17	44,278,110	rs66498281	rs147317628	1.1e-06	ENSG00000120071(5.738e-08), ENSG00000228696(6.206e-181), ENSG00000260075(6.206e-181)	ENSG00000120071(8.6e-08), ENSG00000185829(0), ENSG00000238083(0), ENSG00000262500(1.699e-08)	NA	NA	5_TxWk		KANSL1, ARL17B, NSFP1	KANSL1, ARL17A, LRR37A2	NA	NA
chr1	96,511,272	rs66512	rs201910565	3.4e-07	NA	NA	NA	NA	15_Quies		NA	NA	NA	NA
chr1	96,522,935	rs6682371	rs201910565	3.4e-07	NA	ENSG00000117569(4.103e-05)	NA	NA	15_Quies		NA	PTBP2	NA	NA
chr1	99,092,784	rs6701243	rs6701243	3.1e-07	NA	NA	NA	NA	15_Quies		NA	NA	NA	NA
chr2	159,379,244	rs6734020	rs59566011	9.3e-07				ENSG00000144283	15_Quies					PKP4
chr2	159,378,458	rs6743102	rs59566011	9.3e-07				ENSG00000144283	15_Quies					PKP4
chr20	21,139,624	rs68145616	rs6047270	7.7e-08			ENSG00000228604		5_TxWk					
chr20	21,139,623	rs68145616mm	rs6047270	7.7e-08			ENSG00000228604		5_TxWk					
chr20	14,836,243	rs71190156	rs71190156	2.8e-08	NA	NA	NA	NA	15_Quies		NA	NA	NA	NA
chr6	98,561,749	rs71806471	rs72934503	5.9e-07	NA	NA	NA	NA	5_TxWk		NA	NA	NA	NA
chr17	44,210,933	rs7207582	rs142920272	2.9e-07	ENSG00000120071(1.173e-15), ENSG00000159314(3.167e-06), ENSG00000225190(3.167e-06), ENSG00000238723(1.173e-15), ENSG00000260075(3.877e-18), ENSG00000262500(4.273e-15), ENSG00000262879(3.174e-29), ENSG00000263142(3.174e-29)	ENSG00000131484(6.934e-14), ENSG00000159314(2.906e-07), ENSG00000176681(0), ENSG00000214425(6.934e-14), ENSG00000225190(1.09e-11), ENSG00000228696(0), ENSG00000236234(1.09e-11)	NA	NA	5_TxWk		KANSL1, ARHGAP27, LRR37A, PLEKHM1, NSFP1, LRR37A17P	ARHGAP27, LRR37A, LRR37A4P, PLEKHM1, ARL17B	NA	NA
chr20	21,140,171	rs721785	rs6047270	7.7e-08			ENSG00000228604		5_TxWk					
chr17	44,126,365	rs7218319	rs142920272, rs147317628	2.9e-07, 1.1e-06	ENSG00000108433(7.417e-05), ENSG00000120071(2.093e-15), ENSG00000176681(4.187e-13), ENSG00000214401(7.753e-08), ENSG00000228696(4.187e-13), ENSG00000238723(2.093e-15), ENSG00000262500(1.238e-14), ENSG00000262633(7.417e-05), ENSG00000262879(7.417e-05), ENSG00000263142(7.417e-05)	ENSG00000108433(4.841e-06), ENSG00000120071(1.708e-15), ENSG00000176681(1.036e-29), ENSG00000214401(8.406e-08), ENSG00000228696(1.036e-29), ENSG00000238723(1.708e-15), ENSG00000262500(5.602e-15), ENSG00000262633(4.841e-06), ENSG00000262879(4.841e-06), ENSG00000263142(4.841e-06), ENSG00000266497(2.301e-06)	NA	NA	5_TxWk		GOSR2, KANSL1, LRR37A, KANSL1-AS1, ARL17B, LRR37A17P	GOSR2, KANSL1, LRR37A, KANSL1-AS1, ARL17B, LRR37A17P	NA	NA
chr17	44,138,201	rs7220752	rs147317628	1.1e-06	ENSG00000073969(8.129e-26), ENSG00000108433(8.236e-05), ENSG00000176681(6.545e-66), ENSG00000228696(6.545e-66), ENSG00000262633(8.236e-05)	ENSG00000176681(2.694e-19), ENSG00000185829(3.87e-268), ENSG00000228696(2.694e-19), ENSG00000238083(3.87e-268), ENSG00000265315(6.759e-53), ENSG00000266497(7.335e-05)	NA	NA	5_TxWk		NSF, GOSR2, LRR37A, ARL17B	LRR37A, ARL17A, ARL17B, LRR37A2, RN7SL199P	NA	NA
chr17	44,116,950	rs7221390	rs142920272	2.9e-07			ENSG00000120071		5_TxWk				KANSL1	
chr20	21,267,360	rs726025	rs910805	2.0e-09	NA	NA	NA	NA	14_ReprPCWk		NA	NA	NA	NA

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Supplementary Table 16 – continued from previous page

CHR	BP	CredSNP	IndexSNP	Index P	ENSGID_CP	ENSGID_GZ	ENSGID Promoter	ENSGID functional	Chrom HMM	Histone	HGNC_CP	HGNC_GZ	HGNC Promoter	HGNC functional
chr6	98,583,488	rs72934503	rs72934503	5.9e-07	NA	NA	NA	NA	15_Quies		NA	NA	NA	NA
chr20	21,277,361	rs73128960	rs910805	2.0e-09	NA	NA	NA	NA	13_ReprPC	H3K27me3	NA	NA	NA	NA
chr20	21,279,983	rs73128966	rs910805	2.0e-09	NA	NA	NA	NA	14_ReprPCWk		NA	NA	NA	NA
chr17	44,110,271	rs7350980	rs142920272, rs147317628	2.9e-07, 1.1e-06			ENSG00000120071		5_TxWk				KANSL1	
chr2	159,379,571	rs7421094	rs59566011	9.3e-07				ENSG00000144283	15_Quies					PKP4
chr17	44,290,849	rs76475191	rs142920272	2.9e-07	ENSG00000108379(6.796e-05), ENSG00000120071(1.371e-15), ENSG00000158955(6.796e-05), ENSG00000186868(1.113e-08), ENSG00000225190(1.173e-17), ENSG00000262372(2.024e-10), ENSG00000262539(4.07e-09), ENSG00000264589(1.113e-08), ENSG00000266497(5.756e-25)	ENSG0000004897(3.849e-37), ENSG00000108379(1.454e-06), ENSG00000120071(1.453e-15), ENSG00000159314(1.815e-07), ENSG00000186868(7.968e-07), ENSG00000225190(2.236e-12), ENSG00000236234(2.236e-12), ENSG00000239291(8.887e-15), ENSG00000256762(7.075e-06), ENSG00000260075(1.26e-41), ENSG00000262265(8.887e-15), ENSG00000262372(1.283e-13), ENSG00000262879(1.357e-09), ENSG00000264038(3.203e-07), ENSG00000264225(2.236e-12), ENSG00000266497(9.576e-13)	NA	NA	5_TxWk	WNT3, KANSL1, WNT9B, MAPT, PLEKHM1, MAPT-AS1	CDC27, WNT3, KANSL1, ARHGAP27, MAPT, PLEKHM1, STH, NSFP1, RN7SL730P	NA	NA	
chr1	96,968,232	rs76504869	rs2391769	1.1e-07	NA	NA	NA	NA	15_Quies		NA	NA	NA	NA
chr18	55,881,169	rs76747718	rs292441	1.1e-06	NA	ENSG00000049759(3.605e-07)	NA	NA	7_Enh		NA	NEDD4L	NA	NA
chr6	98,566,506	rs77910749	rs72934503	5.9e-07	NA	NA	NA	NA	14_ReprPCWk	DHS	NA	NA	NA	NA
chr8	10,573,702	rs7820334	rs10099100	1.1e-08	NA	ENSG00000171056(9.841e-05), ENSG00000171060(1.154e-09), ENSG00000183638(1.154e-09), ENSG00000212433(1.154e-09), ENSG00000252565(9.841e-05), ENSG00000254093(9.841e-05), ENSG00000258724(9.841e-05)	NA	NA	14_ReprPCWk		NA	SOX7, C8orf74, RP1L1, RNA5SP252, PINX1	NA	NA
chr17	44,296,230	rs78358711	rs142920272, rs147317628	2.9e-07, 1.1e-06	ENSG00000108379(6.796e-05), ENSG00000120071(1.371e-15), ENSG00000158955(6.796e-05), ENSG00000186868(1.113e-08), ENSG00000225190(1.173e-17), ENSG00000262372(2.024e-10), ENSG00000262539(4.07e-09), ENSG00000264589(1.113e-08), ENSG00000266497(5.756e-25)	ENSG0000004897(3.849e-37), ENSG00000108379(1.454e-06), ENSG00000120071(1.453e-15), ENSG00000159314(1.815e-07), ENSG00000186868(7.968e-07), ENSG00000225190(2.236e-12), ENSG00000236234(2.236e-12), ENSG00000239291(8.887e-15), ENSG00000256762(7.075e-06), ENSG00000260075(1.26e-41), ENSG00000262265(8.887e-15), ENSG00000262372(1.283e-13), ENSG00000262879(1.357e-09), ENSG00000264038(3.203e-07), ENSG00000264225(2.236e-12), ENSG00000266497(9.576e-13)	NA	NA	5_TxWk	WNT3, KANSL1, WNT9B, MAPT, PLEKHM1, MAPT-AS1	CDC27, WNT3, KANSL1, ARHGAP27, MAPT, PLEKHM1, STH, NSFP1, RN7SL730P	NA	NA	
chr19	37,621,419	rs78410150	rs138867053	1.2e-07				ENSG00000196967	8_ZNF/Rpts					ZNF585A
chr8	48,504,452	rs78611701	rs183563276n	1.9e-07				ENSG00000272972	15_Quies					NA
chr10	72,749,037	rs78827416	rs78827416	9.0e-07	NA	NA	NA	NA	15_Quies		NA	NA	NA	NA
chr2	159,375,792	rs78932389	rs59566011	9.3e-07				ENSG00000144283	15_Quies					PKP4
chr1	104,791,770	rs79783584	rs11185408	7.0e-07	NA	NA	NA	NA	15_Quies		NA	NA	NA	NA

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Supplementary Table 16 – continued from previous page

CHR	BP	CredSNP	IndexSNP	Index P	ENSGID_CP	ENSGID_GZ	ENSGID Promoter	ENSGID functional	Chrom HMM	Histone	HGNC_CP	HGNC_GZ	HGNC Promoter	HGNC functional
chr17	44,290,910	rs79861768	rs147317628	1.1e-06	ENSG00000108379(6.796e-05), ENSG00000120071(1.371e-15), ENSG00000158955(6.796e-05), ENSG00000186868(1.113e-08), ENSG00000225190(1.173e-17), ENSG00000262372(2.024e-10), ENSG00000262539(4.07e-09), ENSG00000264589(1.113e-08), ENSG00000266497(5.756e-25)	ENSG00000004897(3.849e-37), ENSG00000108379(1.454e-06), ENSG00000120071(1.453e-15), ENSG00000159314(1.815e-07), ENSG00000186868(7.968e-07), ENSG00000225190(2.236e-12), ENSG00000236234(2.236e-12), ENSG00000239291(8.887e-15), ENSG00000256762(7.075e-06), ENSG00000260075(1.26e-41), ENSG00000262265(8.887e-15), ENSG00000262372(1.283e-13), ENSG00000262879(1.357e-09), ENSG00000264038(3.203e-07), ENSG00000264225(2.236e-12), ENSG00000266497(9.576e-13)	NA	NA	5_TxWk	WNT3, KANSL1, WNT9B, MAPT, PLEKHM1, MAPT-AS1	CDC27, WNT3, KANSL1, ARHGAP27, MAPT, PLEKHM1, STH, NSFP1, RN7SL730P	NA	NA	
chr17	44,208,674	rs8070942	rs147317628	1.1e-06	ENSG00000185829(6.167e-37), ENSG00000238083(6.167e-37), ENSG00000262633(3.592e-08), ENSG00000262879(3.592e-08), ENSG00000263142(3.592e-08), ENSG00000263650(3.592e-08), ENSG00000265315(7.022e-29), ENSG00000266497(2.372e-25)	ENSG00000073969(2.084e-07), ENSG00000228696(1.23e-44), ENSG00000260075(1.23e-44), ENSG00000266497(2.657e-06)	NA	NA	7_Enh	H3K4me1	ARL17A, LRRC37A2, LRRC37A17P, RN7SL270P, RN7SL199P	NSF, ARL17B, NSFP1	NA	NA
chr17	44,115,440	rs8077487	rs142920272, rs147317628	2.9e-07, 1.1e-06	ENSG00000120071(2.025e-10), ENSG00000131484(3.196e-08), ENSG00000176681(5.016e-43), ENSG00000214401(2.347e-05), ENSG00000214425(3.196e-08), ENSG00000228696(5.016e-43), ENSG00000238723(2.025e-10), ENSG00000260075(8.608e-13), ENSG00000262500(1.634e-09), ENSG00000266918(3.196e-08)	ENSG00000120071(1.235e-13), ENSG00000176681(8.657e-28), ENSG00000214425(1.962e-99), ENSG00000228696(8.657e-28), ENSG00000238723(1.235e-13), ENSG00000260075(2.132e-197), ENSG00000262500(4.59e-12), ENSG00000267198(1.962e-99), ENSG00000267246(1.962e-99)	NA	NA	5_TxWk	KANSL1, LRRC37A, KANSL1-AS1, LRRC37A4P, ARL17B, NSFP1	KANSL1, LRRC37A, LRRC37A4P, ARL17B, NSFP1	NA	NA	
chr17	44,162,597	rs8080583	rs147317628	1.1e-06	ENSG00000120071(1.319e-14), ENSG00000214401(1.319e-14), ENSG00000260075(9.54e-06)	ENSG00000073969(1.964e-37), ENSG00000120071(1.862e-09), ENSG00000176681(5.414e-10), ENSG00000228696(5.414e-10), ENSG00000238723(1.862e-09), ENSG00000262500(1.094e-09)	NA	NA	15_Quies		KANSL1, KANSL1-AS1, NSFP1	NSF, KANSL1, LRRC37A, ARL17B	NA	NA
chr20	21,168,395	rs8120293	rs6047270	7.7e-08	NA	NA	NA	NA	14_ReprPCWk		NA	NA	NA	NA
chr20	21,248,116	rs910805	rs910805	2.0e-09	NA	ENSG00000226405(8.388e-08)	NA	NA	14_ReprPCWk		NA	NA	NA	NA
chr17	44,187,257	rs9303525	rs142920272, rs147317628	2.9e-07, 1.1e-06	ENSG00000131484(6.16e-22), ENSG00000176681(4.632e-15), ENSG00000214425(6.16e-22), ENSG00000228696(4.28e-59), ENSG00000238083(1.129e-08), ENSG00000260075(4.28e-59), ENSG00000266497(1.129e-08), ENSG00000266918(6.16e-22)	ENSG00000120071(3.253e-05), ENSG00000238723(3.253e-05), ENSG00000262500(3.755e-05)	NA	NA	15_Quies	LRRC37A, LRRC37A4P, ARL17B, LRRC37A2, NSFP1	KANSL1	NA	NA	
chr3	62,478,786	rs9311841	rs1452075	2.1e-07			ENSG00000163618		15_Quies				CADPS	
chr6	11,730,878	rs9366877	rs210894m	4.9e-07				ENSG00000111863	15_Quies					ADTRP
chr6	98,577,689	rs9372734	rs72934503	5.9e-07	NA	NA	NA	NA	5_TxWk		NA	NA	NA	NA
chr6	98,575,726	rs9388171	rs72934503	5.9e-07	NA	NA	NA	NA	5_TxWk		NA	NA	NA	NA
chr6	98,549,801	rs9401593	rs72934503	5.9e-07	NA	NA	NA	NA	15_Quies		NA	NA	NA	NA
chr6	98,574,560	rs968050	rs72934503	5.9e-07	NA	NA	NA	NA	5_TxWk		NA	NA	NA	NA
chr1	96,968,371	rs9804071	rs2391769	1.1e-07	NA	NA	NA	NA	15_Quies		NA	NA	NA	NA
chr17	44,091,886	rs9891103	rs147317628	1.1e-06	ENSG00000131484(4.629e-14), ENSG00000176681(2.617e-38), ENSG00000214425(4.629e-14), ENSG00000228696(2.617e-38), ENSG00000266918(4.629e-14)	ENSG00000120071(7.247e-07), ENSG00000228696(1.262e-09), ENSG00000238723(7.247e-07), ENSG00000262500(4.782e-07), ENSG00000262539(1.124e-06)	NA	NA	5_TxWk	LRRC37A, LRRC37A4P, ARL17B	KANSL1, ARL17B	NA	NA	

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Supplementary Table 16 – continued from previous page

CHR	BP	CredSNP	IndexSNP	Index P	ENSGID_CP	ENSGID_GZ	ENSGID Promoter	ENSGID functional	Chrom HMM	Histone	HGNC_CP	HGNC_GZ	HGNC Promoter	HGNC functional
chr17	44,151,546	rs9907738	rs142920272, rs147317628	2.9e-07, 1.1e-06	ENSG00000108433(1.738e-17), ENSG00000120071(9.114e-05), ENSG00000176681(2.996e-19), ENSG00000214401(9.114e-05), ENSG00000228696(2.996e-19), ENSG00000260075(7.901e-08), ENSG00000262633(1.738e-17), ENSG00000262879(1.738e-17), ENSG00000263142(1.738e-17)	ENSG00000073969(0), ENSG00000225190(2.979e-06)	NA	NA	15_Quies		GOSR2, KANSI1, LRR37A, KANSI1-AS1, ARL17B, NSFP1, LRR37A17P	NSF, PLEKHM1	NA	NA
chr17	44,212,782	rs9915547	rs142920272, rs147317628	2.9e-07, 1.1e-06	ENSG00000120071(1.173e-15), ENSG00000159314(3.167e-06), ENSG00000225190(3.167e-06), ENSG00000238723(1.173e-15), ENSG00000260075(3.877e-18), ENSG00000262500(4.273e-15), ENSG00000262879(3.174e-29), ENSG00000263142(3.174e-29)	ENSG00000131484(6.934e-14), ENSG00000159314(2.906e-07), ENSG00000176681(0), ENSG00000214425(6.934e-14), ENSG00000225190(1.09e-11), ENSG00000228696(0), ENSG00000236234(1.09e-11)	NA	NA	5_TxWk		KANSI1, ARHGAP27, PLEKHM1, NSFP1, LRR37A17P	ARHGAP27, LRR37A, LRR37A4P, PLEKHM1, ARL17B	NA	NA
chr3	62,471,282	rs9968060	rs1452075	2.1e-07	ENSG00000163618(1.026e-06)	ENSG00000153266(9.164e-05), ENSG00000163618(6.154e-05), ENSG00000241472(9.164e-05)	NA	NA	5_TxWk		CADPS	FEZF2, CADPS, PTPRG-AS1	NA	NA

Supplementary figures

List of Supplementary Figures

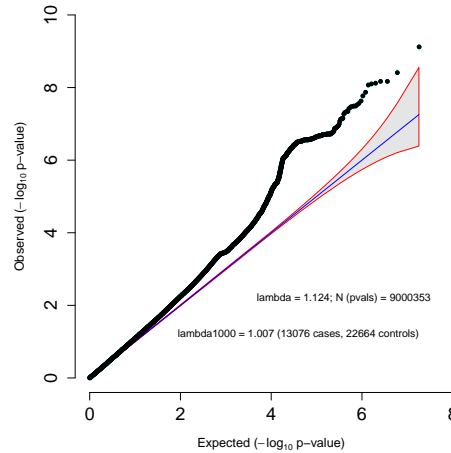
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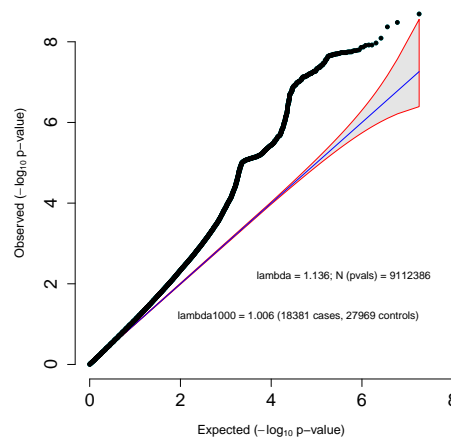
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4.1 GWAS

4.1.1 Qq plots



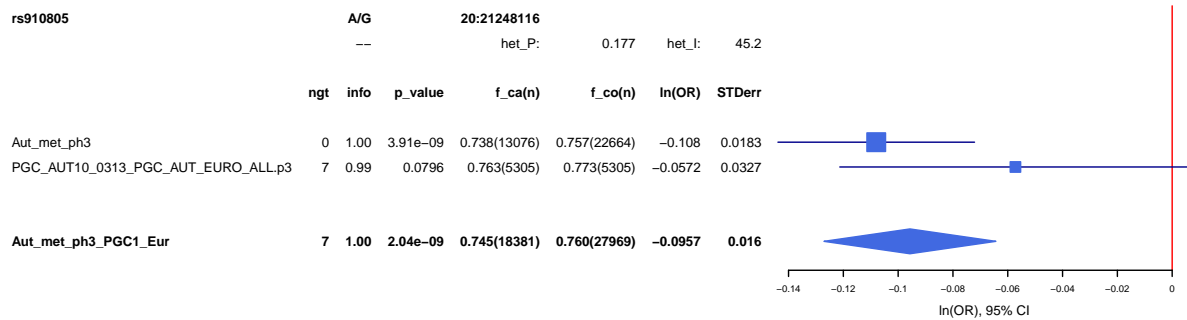
Supplementary Figure 1: Qq (quantile-quantile) plot for the 13076 cases and 22664 controls in the iPSYCH ASD GWAS. Association quantiles of the $-\log_{10}$ p-values (achieved from z-score from logistic regression in genotyping waves and subsequent inverse variant weighted meta-analysis across waves) are plotted against the quantiles expected under the null. The shading indicates 95%-confidence region under the null. The genomic inflation factor, lambda, is the observed median χ^2 test statistic divided by the median expected χ^2 test statistic under the null hypothesis, lambda1000 estimates what lambda would be equivalent to in a sample of 500 cases and 500 controls.



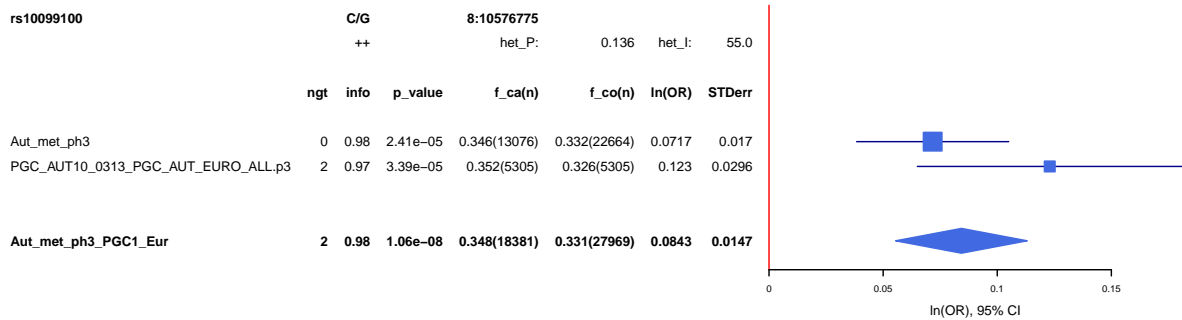
Supplementary Figure 2: Qq (quantile-quantile) plot for the iPSYCH-PGC meta analysis composed of 18381 cases and 27969 controls. Association quantiles of the p-values (achieved from z-score from logistic regression in genotyping waves and subsequent inverse variant weighted meta-analysis across waves) are plotted against the quantiles expected under the null. The shading indicates 95%-confidence region under the null. The genomic inflation factor, lambda, is the observed median χ^2 test statistic divided by the median expected χ^2 test statistic under the null hypothesis, lambda1000 estimates what lambda would be equivalent to in a sample of 500 cases and 500 controls.

4.1.2 Forest plots

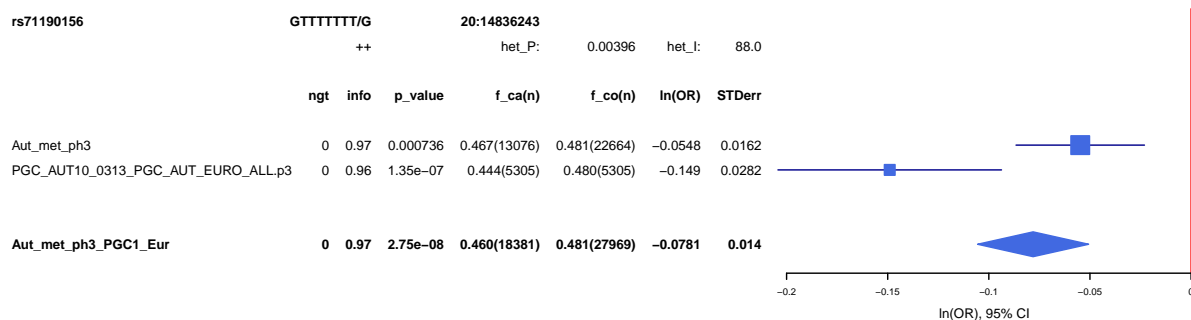
In figures 3–25 we provide the forest plots for the top signals of the iPSYCH-PGC meta analysis.



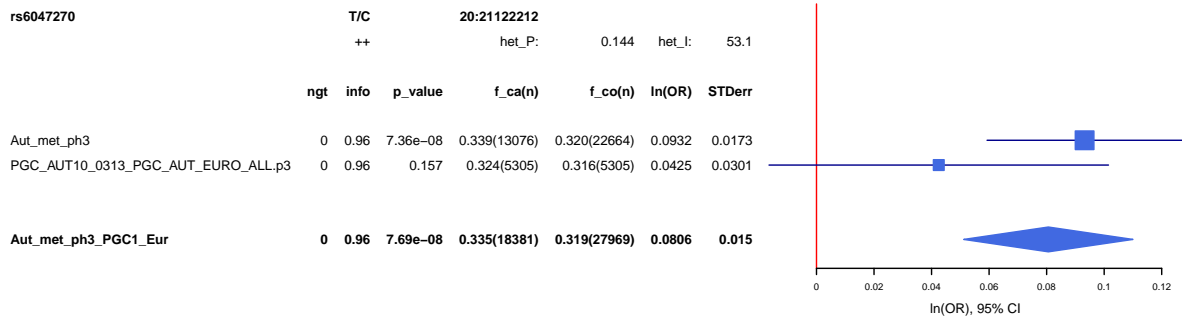
Supplementary Figure 3: Forest plot for rs910805 for meta analysis of iPSYCH ASD GWAS (13 076 cases and 22 664 controls) and the PGC ASD GWAS (5305 cases and 5305 pseudo-controls). The plot shows the effect estimate with 95%-confidence interval for rs910805 in the iPSYCH sample (Aut_met_ph3), PGC (PGC_AUT10_0313_PGC_AUT_EURO_ALL.p3) and the the inverse variance weighted meta analysis (Aut_met_ph3_PGC1_Eur).



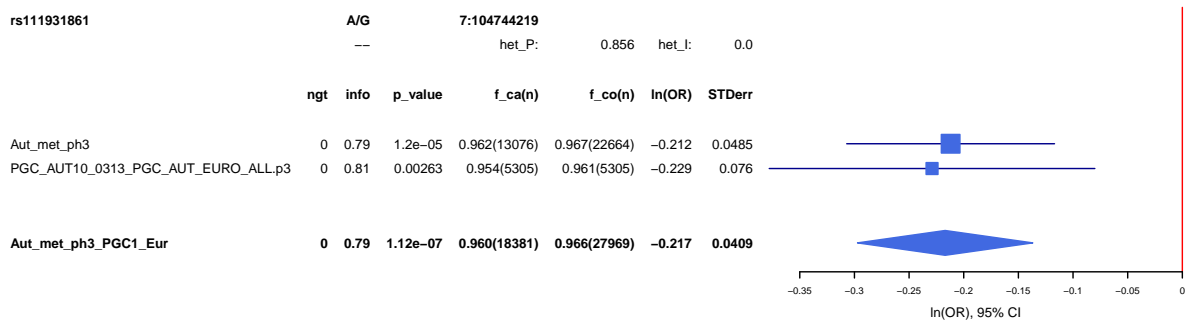
Supplementary Figure 4: Forest plot for rs10099100 for meta analysis of iPSYCH ASD GWAS (13 076 cases and 22 664 controls) and the PGC ASD GWAS (5305 cases and 5305 pseudo-controls). The plot shows the effect estimate with 95%-confidence interval for rs10099100 in the iPSYCH sample (Aut_met_ph3), PGC (PGC_AUT10_0313_PGC_AUT_EURO_ALL.p3) and the the inverse variance weighted meta analysis (Aut_met_ph3_PGC1_Eur).



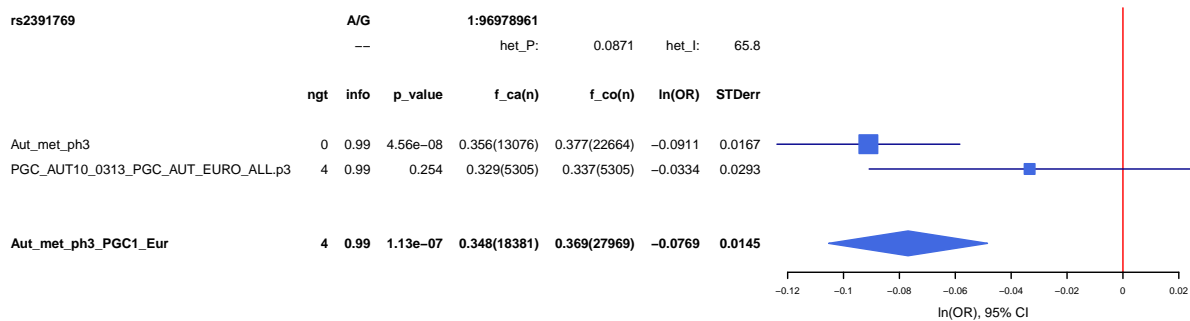
Supplementary Figure 5: Forest plot for rs71190156 for meta analysis of iPSYCH ASD GWAS (13 076 cases and 22 664 controls) and the PGC ASD GWAS (5305 cases and 5305 pseudo-controls). The plot shows the effect estimate with 95%-confidence interval for rs71190156 in the iPSYCH sample (Aut_met_ph3), PGC (PGC_AUT10_0313_PGC_AUT_EURO_ALL.p3) and the the inverse variance weighted meta analysis (Aut_met_ph3_PGC1_Eur).



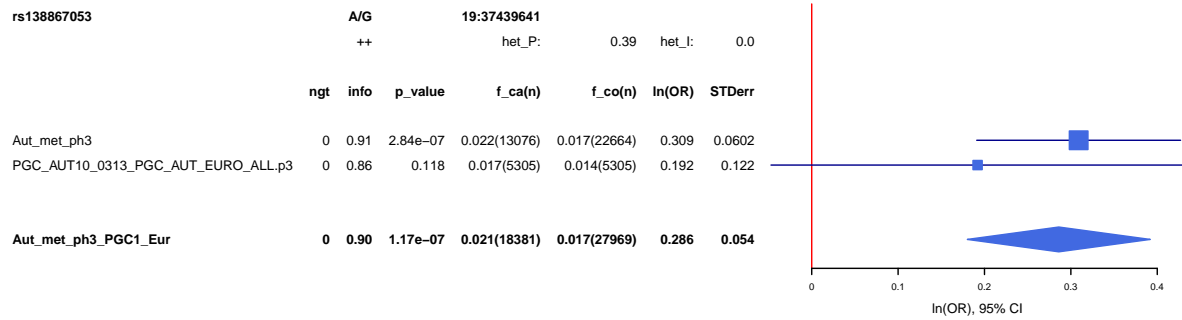
Supplementary Figure 6: Forest plot for rs6047270 for meta analysis of iPSYCH ASD GWAS (13 076 cases and 22 664 controls) and the PGC ASD GWAS (5 305 cases and 5 305 pseudo-controls). The plot shows the effect estimate with 95%-confidence interval for rs6047270 in the iPSYCH sample (Aut_met_ph3), PGC (PGC_AUT10_0313_PGC_AUT_EURO_ALL.p3) and the the inverse variance weighted meta analysis (Aut_met_ph3_PGC1_Eur).



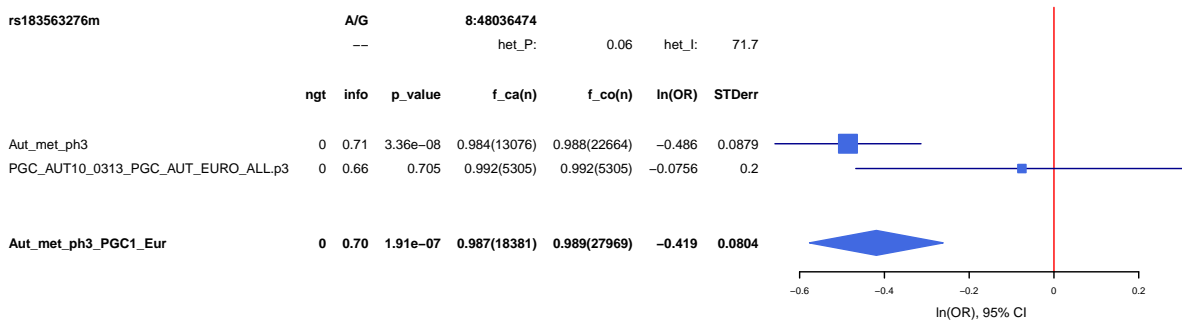
Supplementary Figure 7: Forest plot for rs111931861 for meta analysis of iPSYCH ASD GWAS (13 076 cases and 22 664 controls) and the PGC ASD GWAS (5 305 cases and 5 305 pseudo-controls). The plot shows the effect estimate with 95%-confidence interval for rs111931861 in the iPSYCH sample (Aut_met_ph3), PGC (PGC_AUT10_0313_PGC_AUT_EURO_ALL.p3) and the the inverse variance weighted meta analysis (Aut_met_ph3_PGC1_Eur).



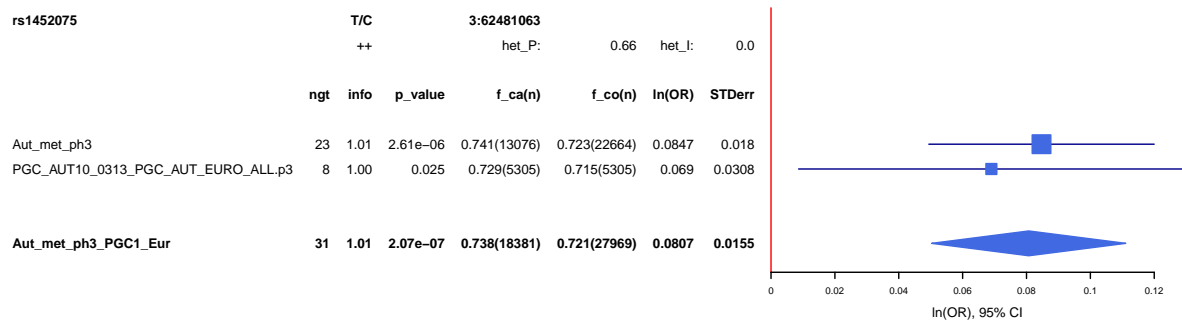
Supplementary Figure 8: Forest plot for rs2391769 for meta analysis of iPSYCH ASD GWAS (13 076 cases and 22 664 controls) and the PGC ASD GWAS (5 305 cases and 5 305 pseudo-controls). The plot shows the effect estimate with 95%-confidence interval for rs2391769 in the iPSYCH sample (Aut_met_ph3), PGC (PGC_AUT10_0313_PGC_AUT_EURO_ALL.p3) and the the inverse variance weighted meta analysis (Aut_met_ph3_PGC1_Eur).



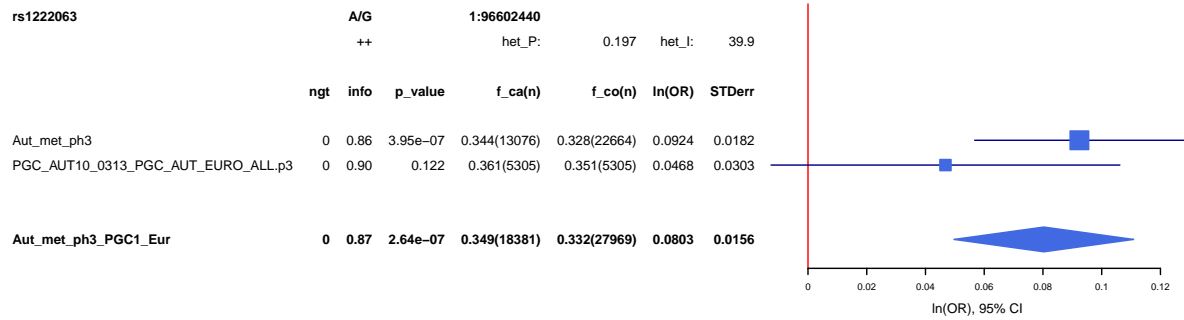
Supplementary Figure 9: Forest plot for rs138867053 for meta analysis of iPSYCH ASD GWAS (13 076 cases and 22 664 controls) and the PGC ASD GWAS (5305 cases and 5305 pseudo-controls). The plot shows the effect estimate with 95%-confidence interval for rs138867053 in the iPSYCH sample (Aut_met_ph3), PGC (PGC_AUT10_0313_PGC_AUT_EURO_ALL.p3) and the the inverse variance weighted meta analysis (Aut_met_ph3_PGC1_Eur).



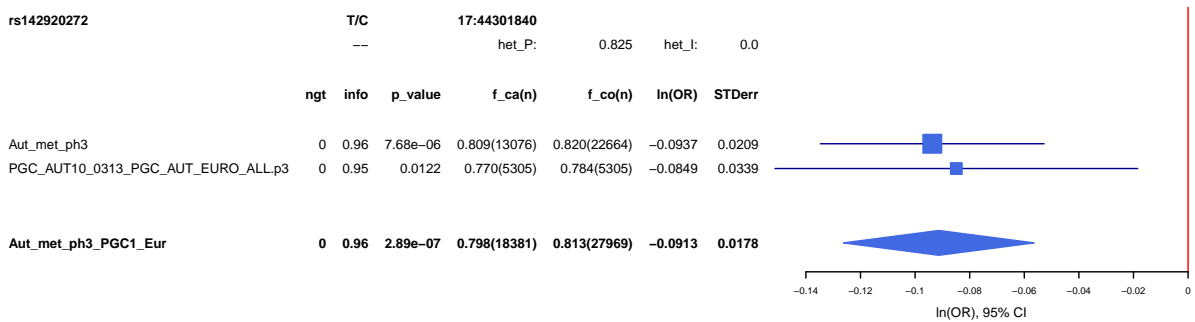
Supplementary Figure 10: Forest plot for rs183563276m for meta analysis of iPSYCH ASD GWAS (13 076 cases and 22 664 controls) and the PGC ASD GWAS (5305 cases and 5305 pseudo-controls). The plot shows the effect estimate with 95%-confidence interval for rs183563276m in the iPSYCH sample (Aut_met_ph3), PGC (PGC_AUT10_0313_PGC_AUT_EURO_ALL.p3) and the the inverse variance weighted meta analysis (Aut_met_ph3_PGC1_Eur).



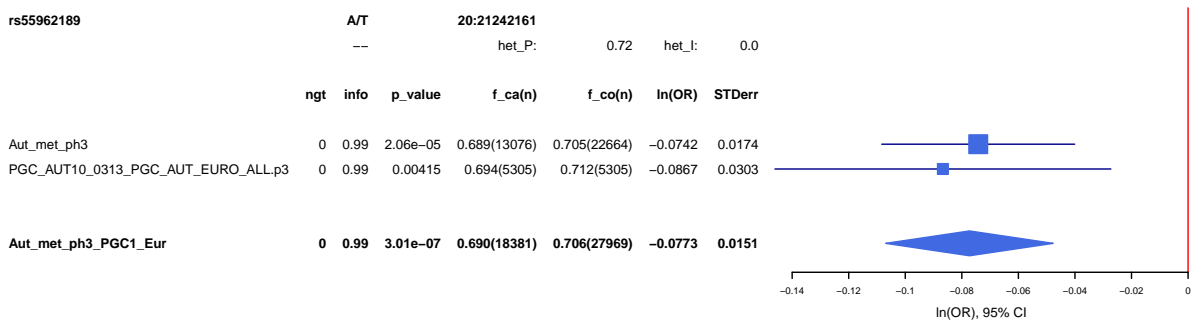
Supplementary Figure 11: Forest plot for rs1452075 for meta analysis of iPSYCH ASD GWAS (13 076 cases and 22 664 controls) and the PGC ASD GWAS (5305 cases and 5305 pseudo-controls). The plot shows the effect estimate with 95%-confidence interval for rs1452075 in the iPSYCH sample (Aut_met_ph3), PGC (PGC_AUT10_0313_PGC_AUT_EURO_ALL.p3) and the the inverse variance weighted meta analysis (Aut_met_ph3_PGC1_Eur).



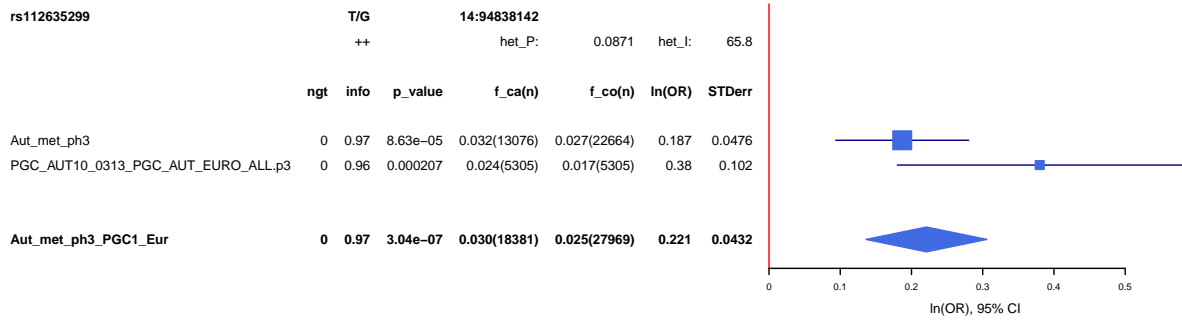
Supplementary Figure 12: Forest plot for rs1222063 for meta analysis of iPSYCH ASD GWAS (13 076 cases and 22 664 controls) and the PGC ASD GWAS (5305 cases and 5305 pseudo-controls). The plot shows the effect estimate with 95%-confidence interval for rs1222063 in the iPSYCH sample (Aut_met_ph3), PGC (PGC_AUT10_0313_PGC_AUT_EURO_ALL.p3) and the the inverse variance weighted meta analysis (Aut_met_ph3_PGC1_Eur).



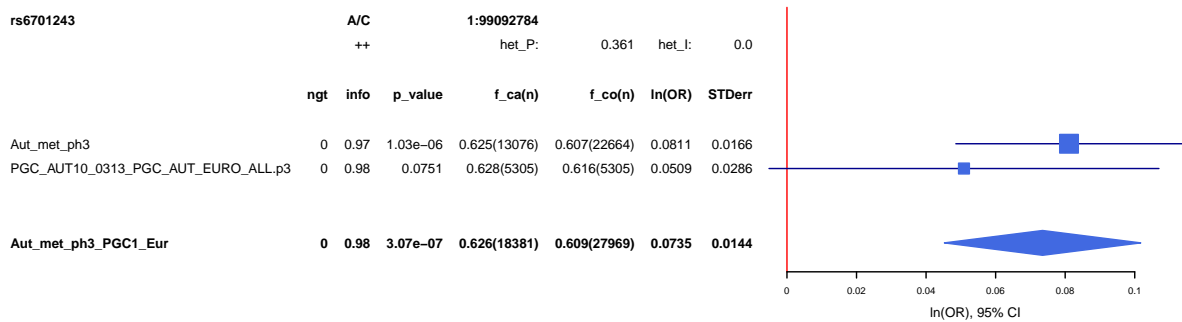
Supplementary Figure 13: Forest plot for rs142920272 for meta analysis of iPSYCH ASD GWAS (13 076 cases and 22 664 controls) and the PGC ASD GWAS (5305 cases and 5305 pseudo-controls). The plot shows the effect estimate with 95%-confidence interval for rs142920272 in the iPSYCH sample (Aut_met_ph3), PGC (PGC_AUT10_0313_PGC_AUT_EURO_ALL.p3) and the the inverse variance weighted meta analysis (Aut_met_ph3_PGC1_Eur).



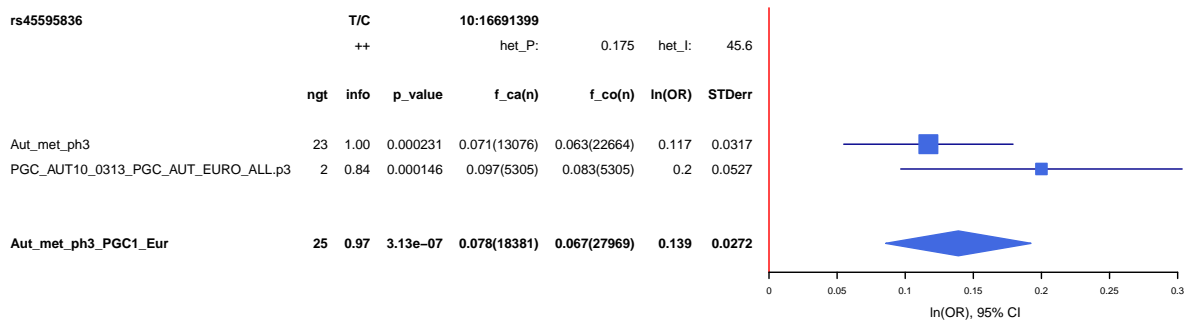
Supplementary Figure 14: Forest plot for rs55962189 for meta analysis of iPSYCH ASD GWAS (13 076 cases and 22 664 controls) and the PGC ASD GWAS (5305 cases and 5305 pseudo-controls). The plot shows the effect estimate with 95%-confidence interval for rs55962189 in the iPSYCH sample (Aut_met_ph3), PGC (PGC_AUT10_0313_PGC_AUT_EURO_ALL.p3) and the the inverse variance weighted meta analysis (Aut_met_ph3_PGC1_Eur).



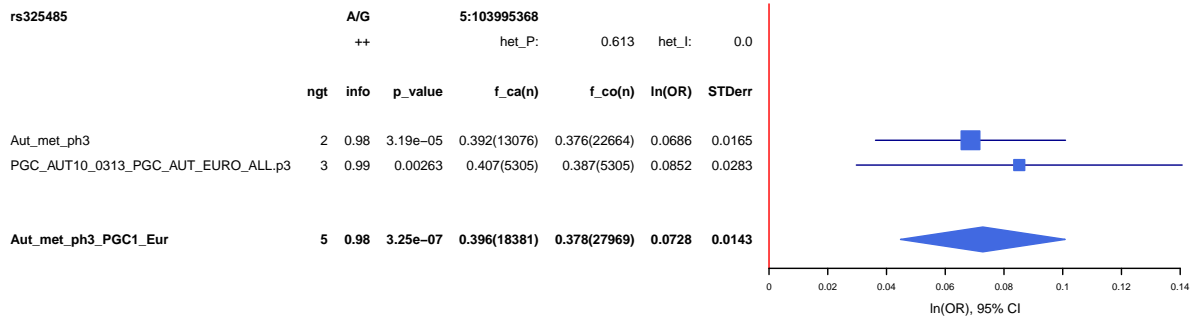
Supplementary Figure 15: Forest plot for rs112635299 for meta analysis of iPSYCH ASD GWAS (13 076 cases and 22 664 controls) and the PGC ASD GWAS (5305 cases and 5305 pseudo-controls). The plot shows the effect estimate with 95%-confidence interval for rs112635299 in the iPSYCH sample (Aut_met_ph3), PGC (PGC_AUT10_0313_PGC_AUT_EURO_ALL.p3) and the the inverse variance weighted meta analysis (Aut_met_ph3_PGC1_Eur).



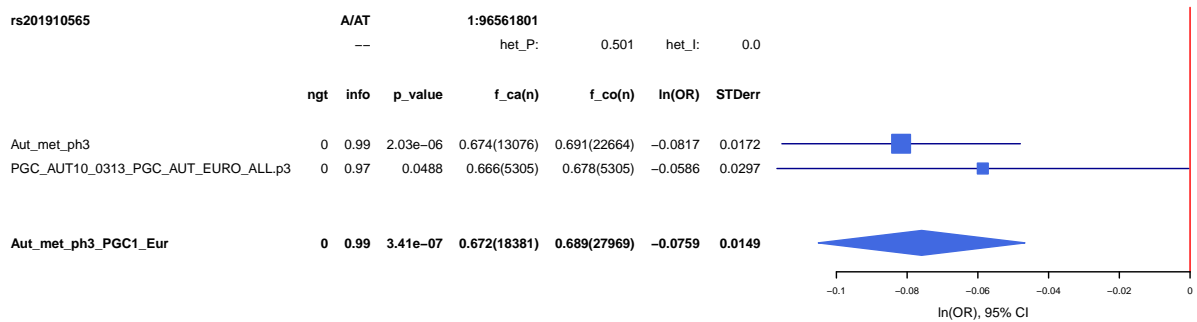
Supplementary Figure 16: Forest plot for rs6701243 for meta analysis of iPSYCH ASD GWAS (13 076 cases and 22 664 controls) and the PGC ASD GWAS (5305 cases and 5305 pseudo-controls). The plot shows the effect estimate with 95%-confidence interval for rs6701243 in the iPSYCH sample (Aut_met_ph3), PGC (PGC_AUT10_0313_PGC_AUT_EURO_ALL.p3) and the the inverse variance weighted meta analysis (Aut_met_ph3_PGC1_Eur).



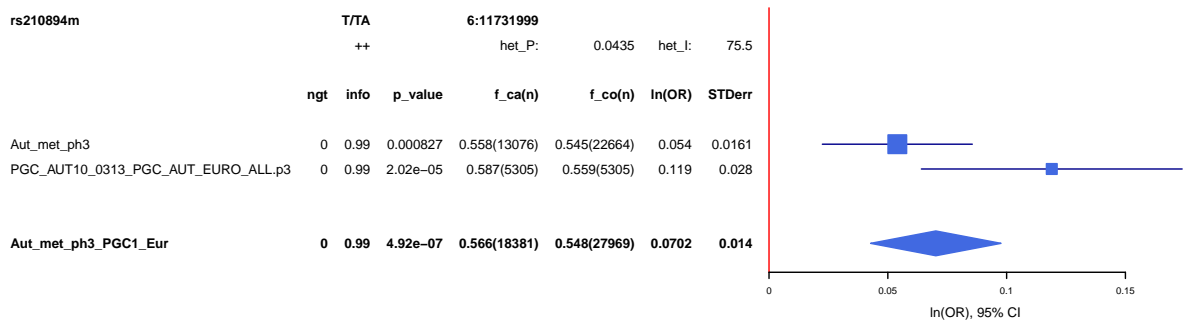
Supplementary Figure 17: Forest plot for rs45595836 for meta analysis of iPSYCH ASD GWAS (13 076 cases and 22 664 controls) and the PGC ASD GWAS (5305 cases and 5305 pseudo-controls). The plot shows the effect estimate with 95%-confidence interval for rs45595836 in the iPSYCH sample (Aut_met_ph3), PGC (PGC_AUT10_0313_PGC_AUT_EURO_ALL.p3) and the the inverse variance weighted meta analysis (Aut_met_ph3_PGC1_Eur).



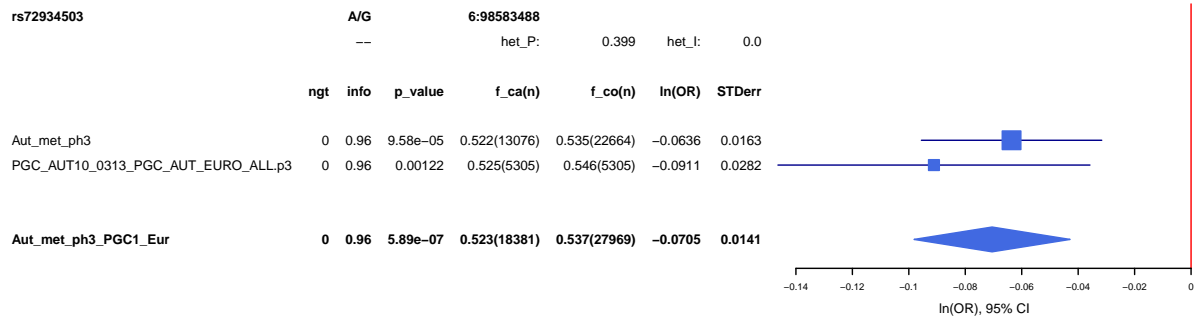
Supplementary Figure 18: Forest plot for rs325485 for meta analysis of iPSYCH ASD GWAS (13 076 cases and 22 664 controls) and the PGC ASD GWAS (5305 cases and 5305 pseudo-controls). The plot shows the effect estimate with 95%-confidence interval for rs325485 in the iPSYCH sample (Aut_met_ph3), PGC (PGC_AUT10_0313_PGC_AUT_EURO_ALL.p3) and the the inverse variance weighted meta analysis (Aut_met_ph3_PGC1_Eur).



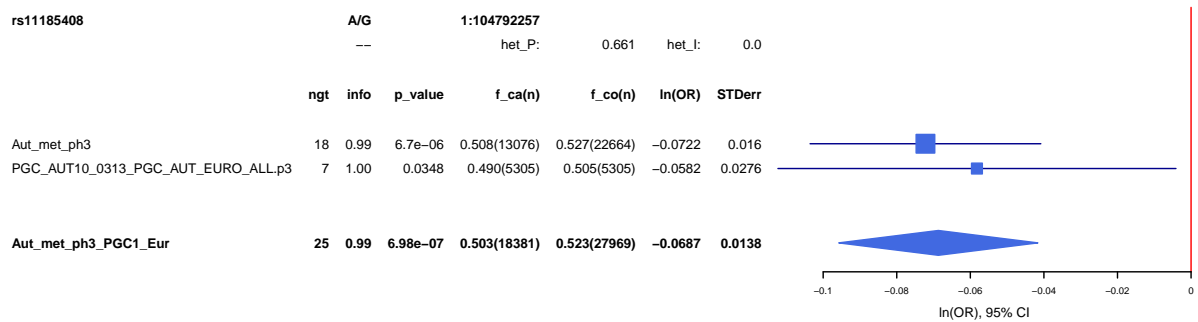
Supplementary Figure 19: Forest plot for rs201910565 for meta analysis of iPSYCH ASD GWAS (13 076 cases and 22 664 controls) and the PGC ASD GWAS (5305 cases and 5305 pseudo-controls). The plot shows the effect estimate with 95%-confidence interval for rs201910565 in the iPSYCH sample (Aut_met_ph3), PGC (PGC_AUT10_0313_PGC_AUT_EURO_ALL.p3) and the the inverse variance weighted meta analysis (Aut_met_ph3_PGC1_Eur).



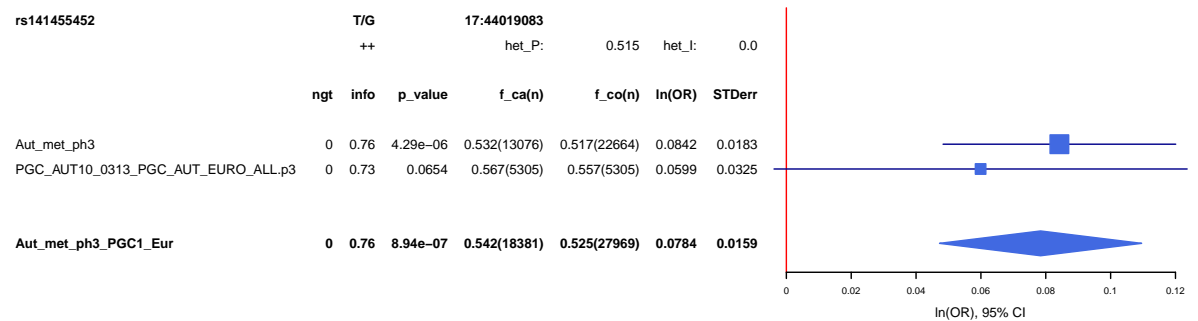
Supplementary Figure 20: Forest plot for rs210894m for meta analysis of iPSYCH ASD GWAS (13 076 cases and 22 664 controls) and the PGC ASD GWAS (5305 cases and 5305 pseudo-controls). The plot shows the effect estimate with 95%-confidence interval for rs210894m in the iPSYCH sample (Aut_met_ph3), PGC (PGC_AUT10_0313_PGC_AUT_EURO_ALL.p3) and the the inverse variance weighted meta analysis (Aut_met_ph3_PGC1_Eur).



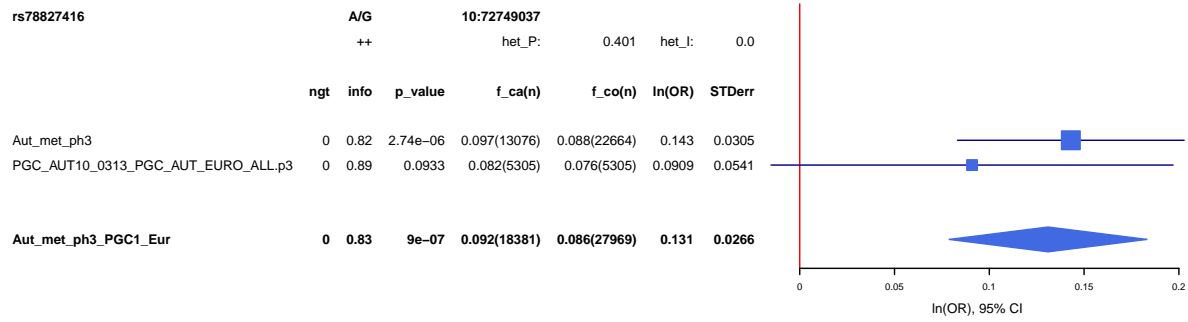
Supplementary Figure 21: Forest plot for rs72934503 for meta analysis of iPSYCH ASD GWAS (13 076 cases and 22 664 controls) and the PGC ASD GWAS (5305 cases and 5305 pseudo-controls). The plot shows the effect estimate with 95%-confidence interval for rs72934503 in the iPSYCH sample (Aut_met_ph3), PGC (PGC_AUT10_0313_PGC_AUT_EURO_ALL.p3) and the the inverse variance weighted meta analysis (Aut_met_ph3_PGC1_Eur).



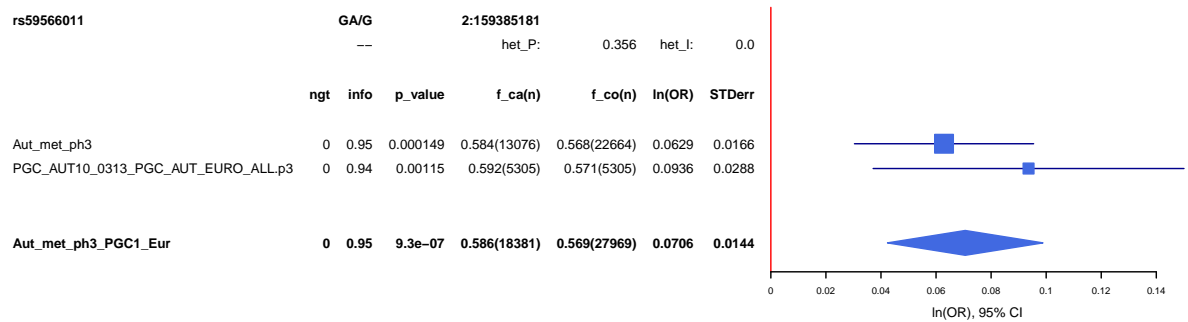
Supplementary Figure 22: Forest plot for rs11185408 for meta analysis of iPSYCH ASD GWAS (13 076 cases and 22 664 controls) and the PGC ASD GWAS (5305 cases and 5305 pseudo-controls). The plot shows the effect estimate with 95%-confidence interval for rs11185408 in the iPSYCH sample (Aut_met_ph3), PGC (PGC_AUT10_0313_PGC_AUT_EURO_ALL.p3) and the the inverse variance weighted meta analysis (Aut_met_ph3_PGC1_Eur).



Supplementary Figure 23: Forest plot for rs141455452 for meta analysis of iPSYCH ASD GWAS (13 076 cases and 22 664 controls) and the PGC ASD GWAS (5305 cases and 5305 pseudo-controls). The plot shows the effect estimate with 95%-confidence interval for rs141455452 in the iPSYCH sample (Aut_met_ph3), PGC (PGC_AUT10_0313_PGC_AUT_EURO_ALL.p3) and the the inverse variance weighted meta analysis (Aut_met_ph3_PGC1_Eur).



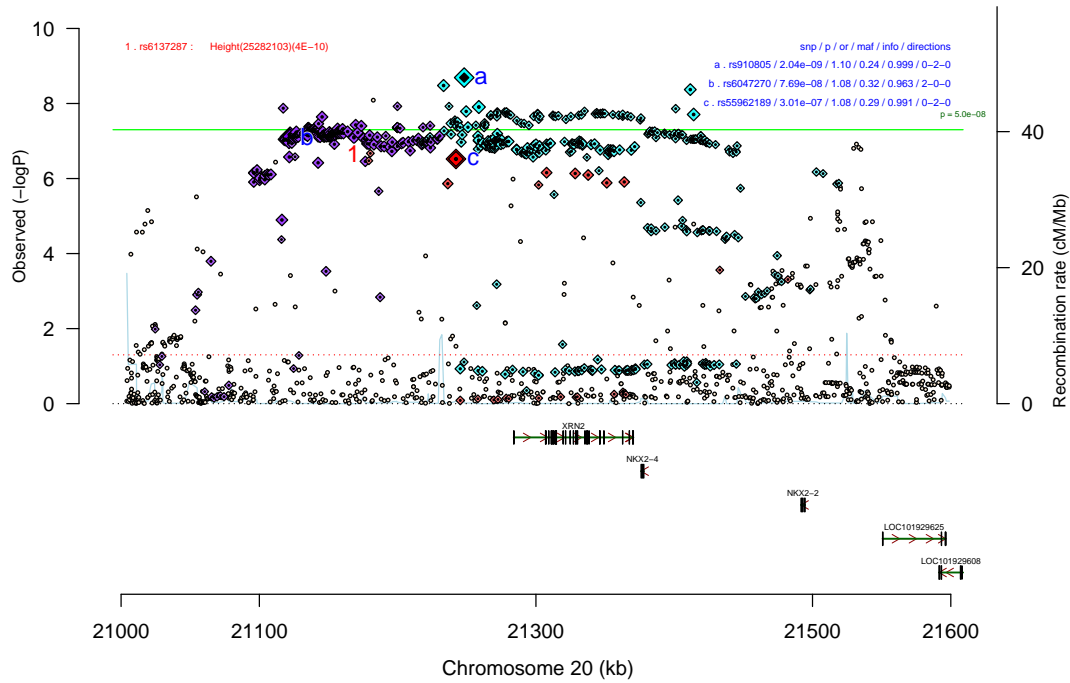
Supplementary Figure 24: Forest plot for rs78827416 for meta analysis of iPSYCH ASD GWAS (13 076 cases and 22 664 controls) and the PGC ASD GWAS (5 305 cases and 5 305 pseudo-controls). The plot shows the effect estimate with 95%-confidence interval for rs78827416 in the iPSYCH sample (Aut_met_ph3), PGC (PGC_AUT10_0313_PGC_AUT_EURO_ALL.p3) and the the inverse variance weighted meta analysis (Aut_met_ph3_PGC1_Eur).



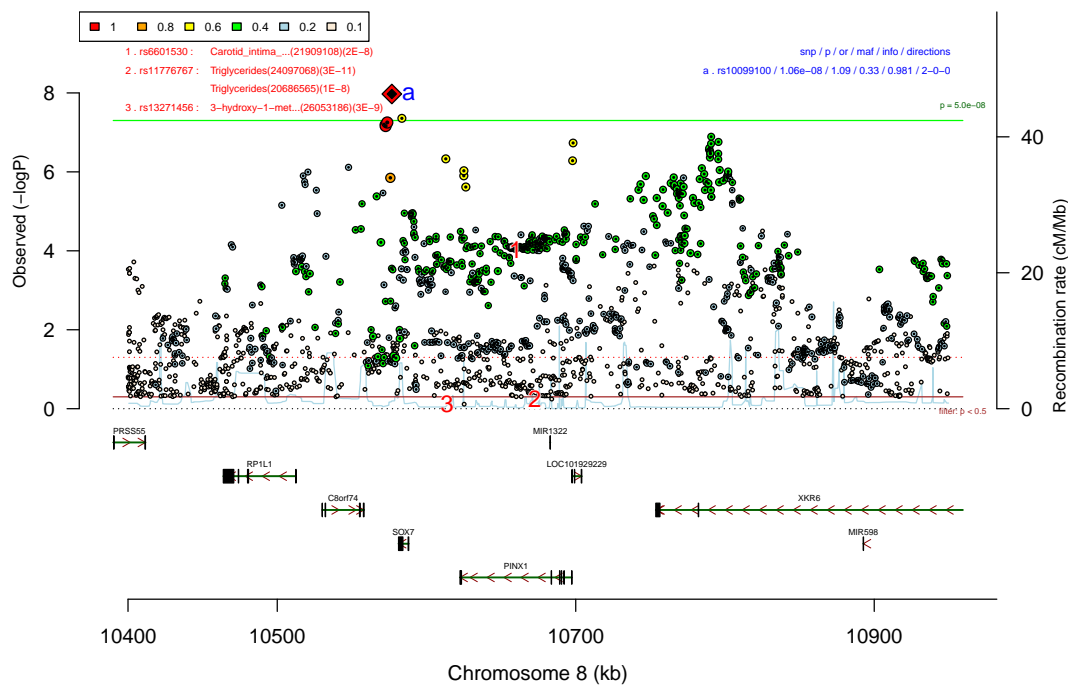
Supplementary Figure 25: Forest plot for rs59566011 for meta analysis of iPSYCH ASD GWAS (13 076 cases and 22 664 controls) and the PGC ASD GWAS (5 305 cases and 5 305 pseudo-controls). The plot shows the effect estimate with 95%-confidence interval for rs59566011 in the iPSYCH sample (Aut_met_ph3), PGC (PGC_AUT10_0313_PGC_AUT_EURO_ALL.p3) and the the inverse variance weighted meta analysis (Aut_met_ph3_PGC1_Eur).

4.1.3 Regional plots

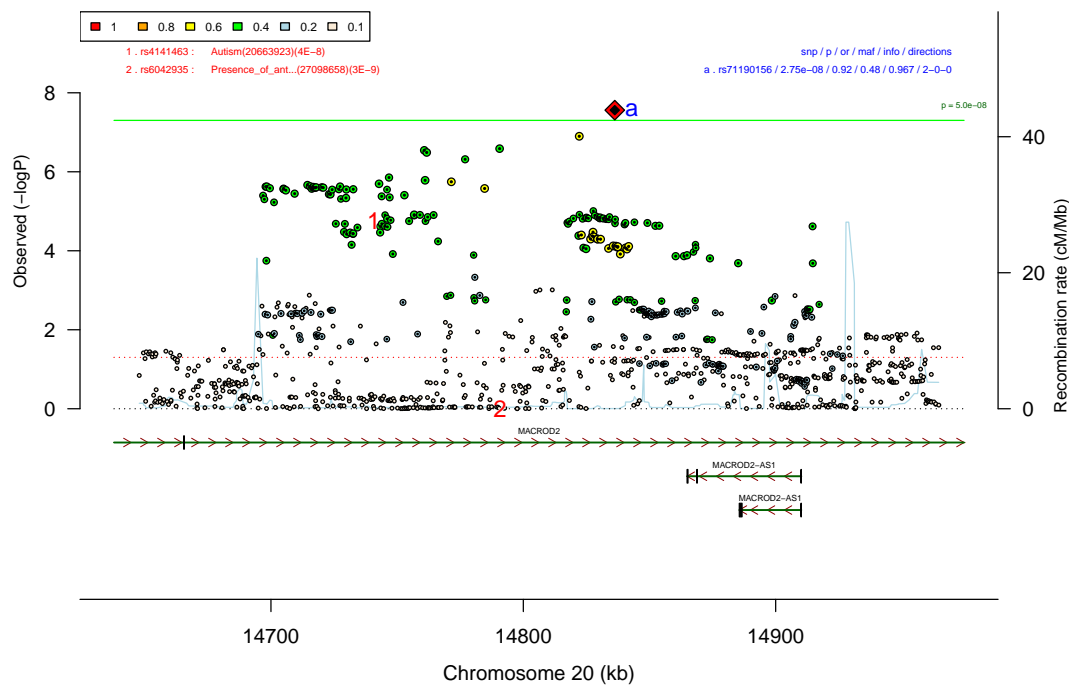
In figures 26–44 we show the regional Manhattan plots for the top signals of the iPSYCH-PGC meta analysis.



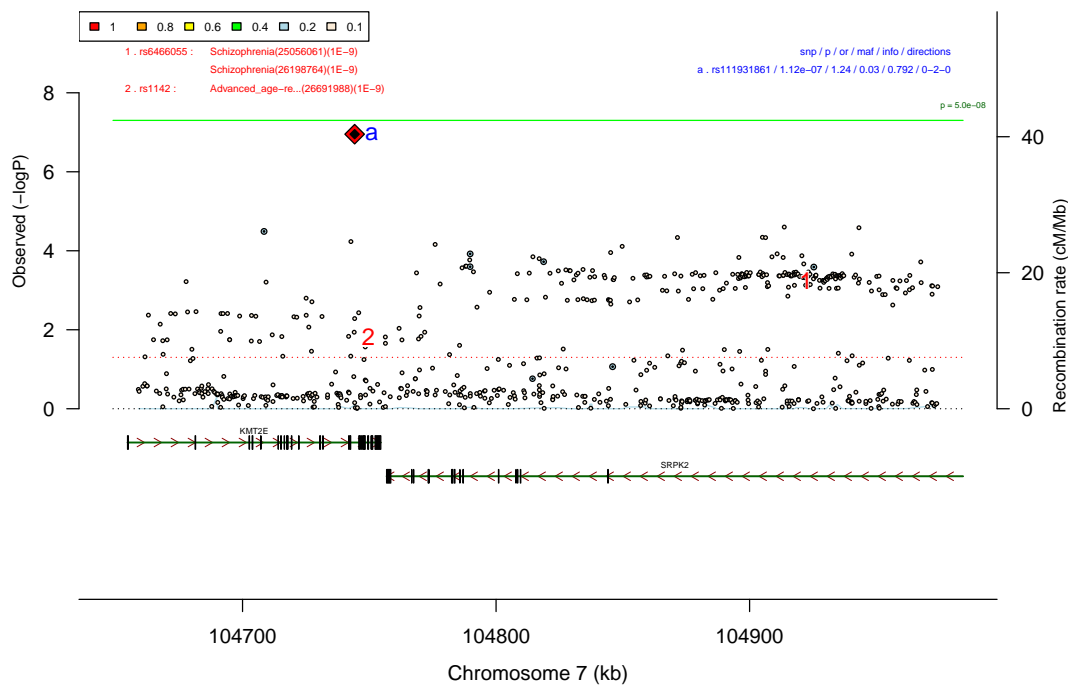
Supplementary Figure 26: Regional association plot around rs910805, rs6047270 & rs55962189 for iPSYCH-PGC meta analysis (18 381 cases and 27 969 controls). Chromosomal position is on the horizontal axis in kb. The significance of the association expressed as $-\log_{10}(P)$ (from z-test) is plotted vertically (left axis) and the recombination rate from HapMap is shown as light blue curve (right axis). The dot size is proportional to LD between the plotted SNP and the index SNP defining the associated region. Index SNPs are marked by diamonds and labelled with letters. As this locus contains multiple independent index SNPs, each index SNP is denoted by a different colour. LD to each index SNP is denoted by the intensity of that colour. A black centre indicates that the markers is present in the 1000 Genomes Project but not in HapMap3. The green line shows the genome-wide significance level ($5 \cdot 10^{-8}$). The upper right corner show details of the index SNPs (marker name/association p-value/odds ratio for the minor allele/minor allele frequency/imputation quality/direction of effect in the two studies (N one direction - N other direction- N missing)). Loci listed in the GWAS Catalogue for other phenotypes are shown in the upper left corner (capped at 10). The green lines in the lower half show genes (based on UCSC) with black vertical lines indicating exons. Arrow heads show the direction of transcription.



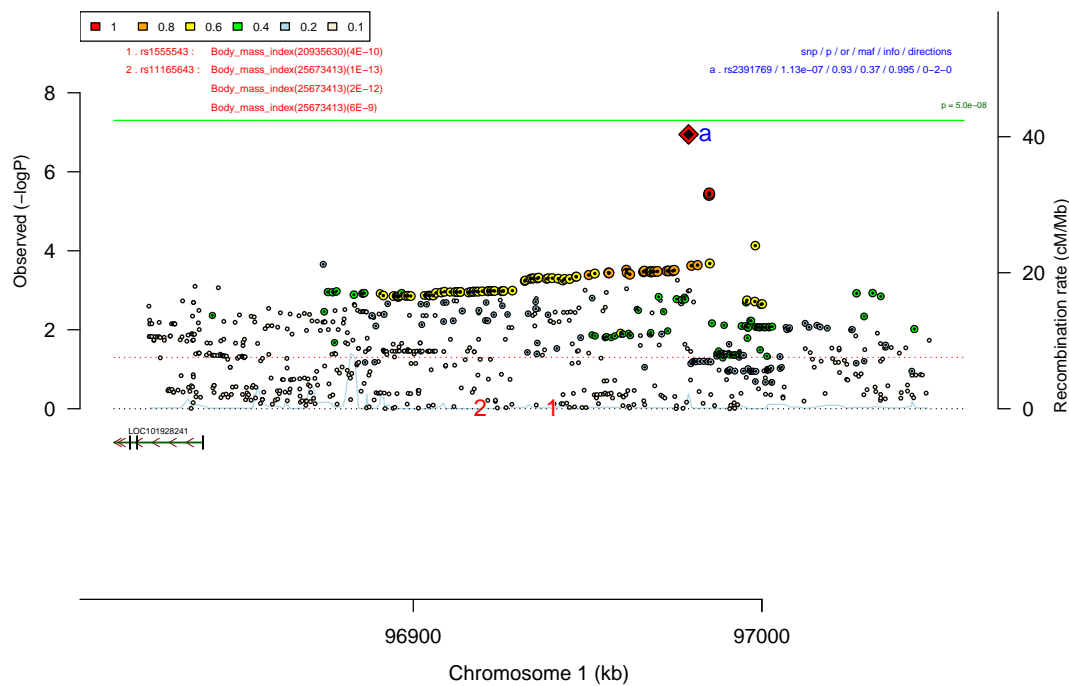
Supplementary Figure 27: Regional association plot around rs10099100 for iPSYCH-PGC meta analysis (18 381 cases and 27 969 controls). Chromosomal position is on the horizontal axis in kb. The significance of the association expressed as $-\log_{10}(P)$ (from z-test) is plotted vertically (left axis) and the recombination rate from HapMap is shown as light blue curve (right axis). The dot size is proportional to LD between the plotted SNP and the index SNP defining the associated region. Index SNPs are marked by diamonds and labelled with letters. As there is only a single index SNP in this region, colouring is based on degree of LD to that SNP as represented by r^2 (legend for r^2 is in the upper left corner). A black centre indicates that the markers is present in the 1000 Genomes Project but not in HapMap3. The green line shows the genome-wide significance level ($5 \cdot 10^{-8}$). The upper right corner show details of the index SNPs (marker name/association p-value/odds ratio for the minor allele/minor allele frequency/imputation quality/direction of effect in the two studies (N one direction - N other direction - N missing)). Loci listed in the GWAS Catalogue for other phenotypes are shown in the upper left corner (capped at 10). The green lines in the lower half show genes (based on UCSC) with black vertical lines indicating exons. Arrow heads show the direction of transcription.



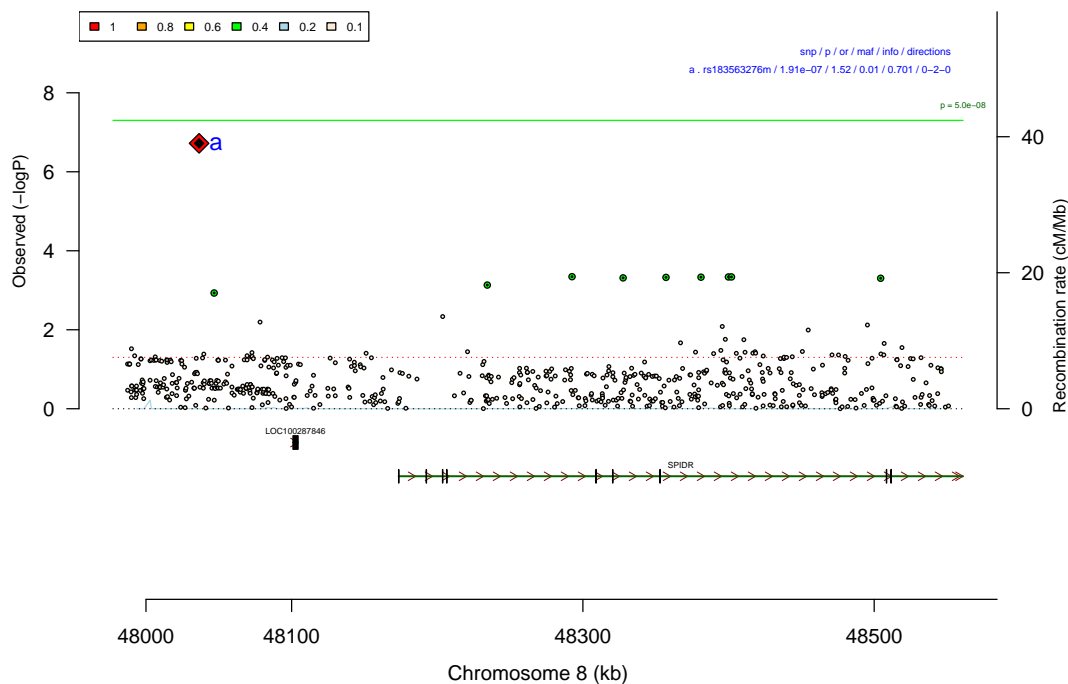
Supplementary Figure 28: Regional association plot around rs71190156 for iPSYCH-PGC meta analysis (18 381 cases and 27 969 controls). Chromosomal position is on the horizontal axis in kb, The significance of the association expressed as $-\log_{10}(P)$ (from z-test) is plotted vertically (left axis) and the recombination rate from HapMap is shown as light blue curve (right axis). The dot size is proportional to LD between the plotted SNP and the index SNP defining the associated region. Index SNPs are marked by diamonds and labelled with letters. As there is only a single index SNP in this region, colouring is based on degree of LD to that SNP as represented by r^2 (legend for r^2 is in the upper left corner). A black centre indicates that the markers is present in the 1000 Genomes Project but not in HapMap3. The green line shows the genome-wide significance level ($5 \cdot 10^{-8}$). The upper right corner show details of the index SNPs (marker name/association p-value/odds ratio for the minor allele/minor allele frequency/imputation quality/direction of effect in the two studies (N one direction - N other direction - N missing)). Loci listed in the GWAS Catalogue for other phenotypes are shown in the upper left corner (capped at 10). The green lines in the lower half show genes (based on UCSC) with black vertical lines indicating exons. Arrow heads show the direction of transcription.



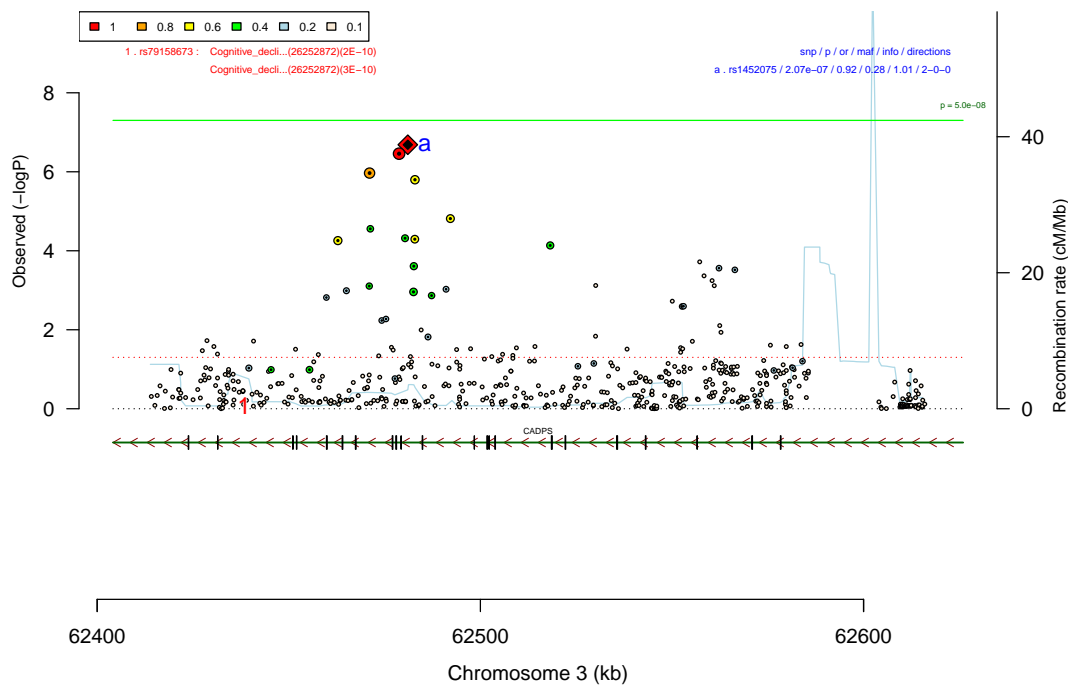
Supplementary Figure 29: Regional association plot around rs11931861 for iPSYCH-PGC meta analysis (18 381 cases and 27 969 controls). Chromosomal position is on the horizontal axis in kb, The significance of the association expressed as $-\log_{10}(P)$ (from z-test) is plotted vertically (left axis) and the recombination rate from HapMap is shown as light blue curve (right axis). The dot size is proportional to LD between the plotted SNP and the index SNP defining the associated region. Index SNPs are marked by diamonds and labelled with letters. As there is only a single index SNP in this region, colouring is based on degree of LD to that SNP as represented by r^2 (legend for r^2 is in the upper left corner). A black centre indicates that the markers is present in the 1000 Genomes Project but not in HapMap3. The green line shows the genome-wide significance level ($5 \cdot 10^{-8}$). The upper right corner show details of the index SNPs (marker name/association p-value/odds ratio for the minor allele/minor allele frequency/imputation quality/direction of effect in the two studies (N one direction - N other direction- N missing)). Loci listed in the GWAS Catalogue for other phenotypes are shown in the upper left corner (capped at 10). The green lines in the lower half show genes (based on UCSC) with black vertical lines indicating exons. Arrow heads show the direction of transcription.



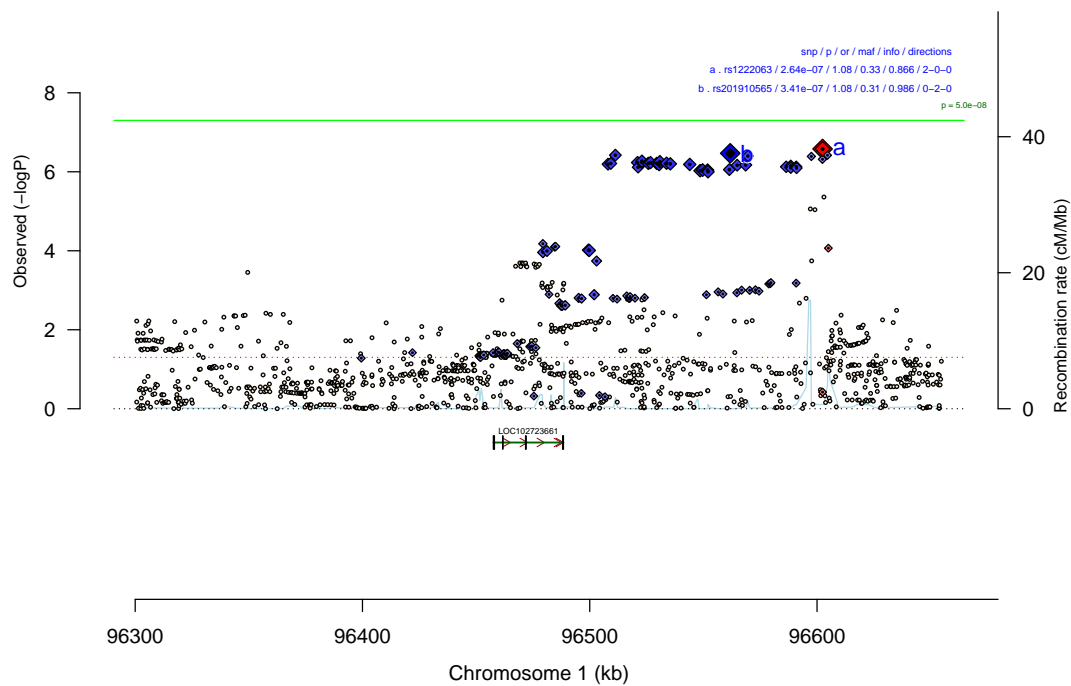
Supplementary Figure 30: Regional association plot around rs2391769 for iPSYCH-PGC meta analysis (18 381 cases and 27 969 controls). Chromosomal position is on the horizontal axis in kb, The significance of the association expressed as $-\log_{10}(P)$ (from z-test) is plotted vertically (left axis) and the recombination rate from HapMap is shown as light blue curve (right axis). The dot size is proportional to LD between the plotted SNP and the index SNP defining the associated region. Index SNPs are marked by diamonds and labelled with letters. As there is only a single index SNP in this region, colouring is based on degree of LD to that SNP as represented by r^2 (legend for r^2 is in the upper left corner). A black centre indicates that the markers is present in the 1000 Genomes Project but not in HapMap3. The green line shows the genome-wide significance level ($5 \cdot 10^{-8}$). The upper right corner show details of the index SNPs (marker name/association p-value/odds ratio for the minor allele/minor allele frequency/imputation quality/direction of effect in the two studies (N one direction - N other direction- N missing)). Loci listed in the GWAS Catalogue for other phenotypes are shown in the upper left corner (capped at 10). The green lines in the lower half show genes (based on UCSC) with black vertical lines indicating exons. Arrow heads show the direction of transcription.



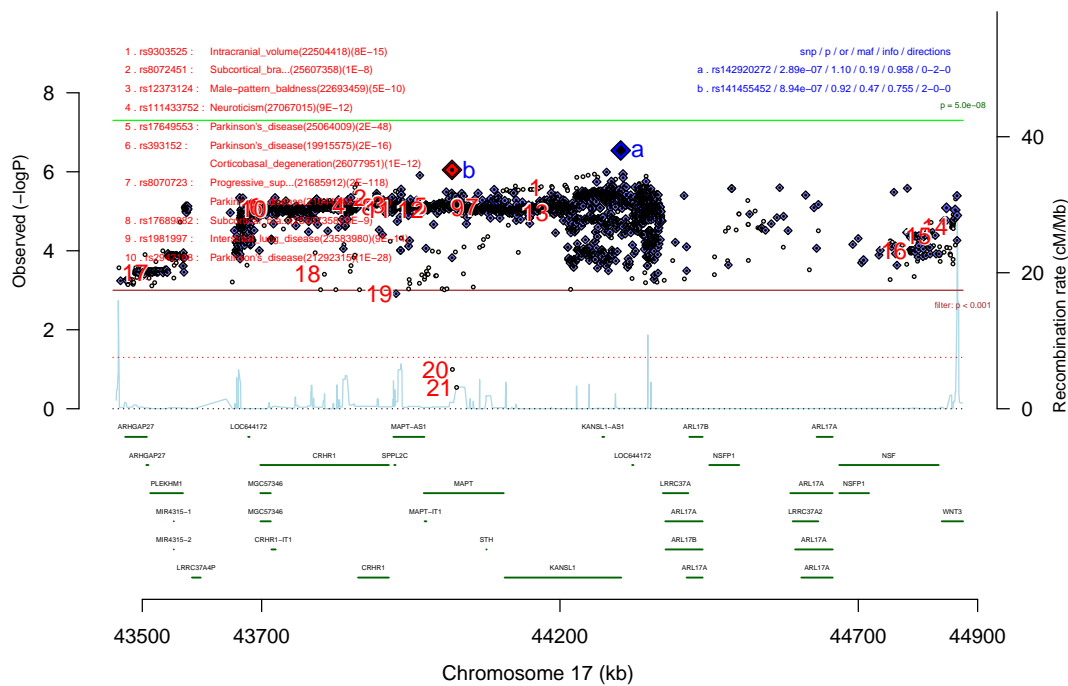
Supplementary Figure 32: Regional association plot around rs183563276m for iPSYCH-PGC meta analysis (18 381 cases and 27 969 controls). Chromosomal position is on the horizontal axis in kb, The significance of the association expressed as $-\log_{10}(P)$ (from z-test) is plotted vertically (left axis) and the recombination rate from HapMap is shown as light blue curve (right axis). The dot size is proportional to LD between the plotted SNP and the index SNP defining the associated region. Index SNPs are marked by diamonds and labelled with letters. As there is only a single index SNP in this region, colouring is based on degree of LD to that SNP as represented by r^2 (legend for r^2 is in the upper left corner). A black centre indicates that the markers is present in the 1000 Genomes Project but not in HapMap3. The green line shows the genome-wide significance level ($5 \cdot 10^{-8}$). The upper right corner show details of the index SNPs (marker name/association p-value/odds ratio for the minor allele/minor allele frequency/imputation quality/direction of effect in the two studies (N one direction - N other direction- N missing)). The green lines in the lower half show genes (based on UCSC) with black vertical lines indicating exons. Arrow heads show the direction of transcription.



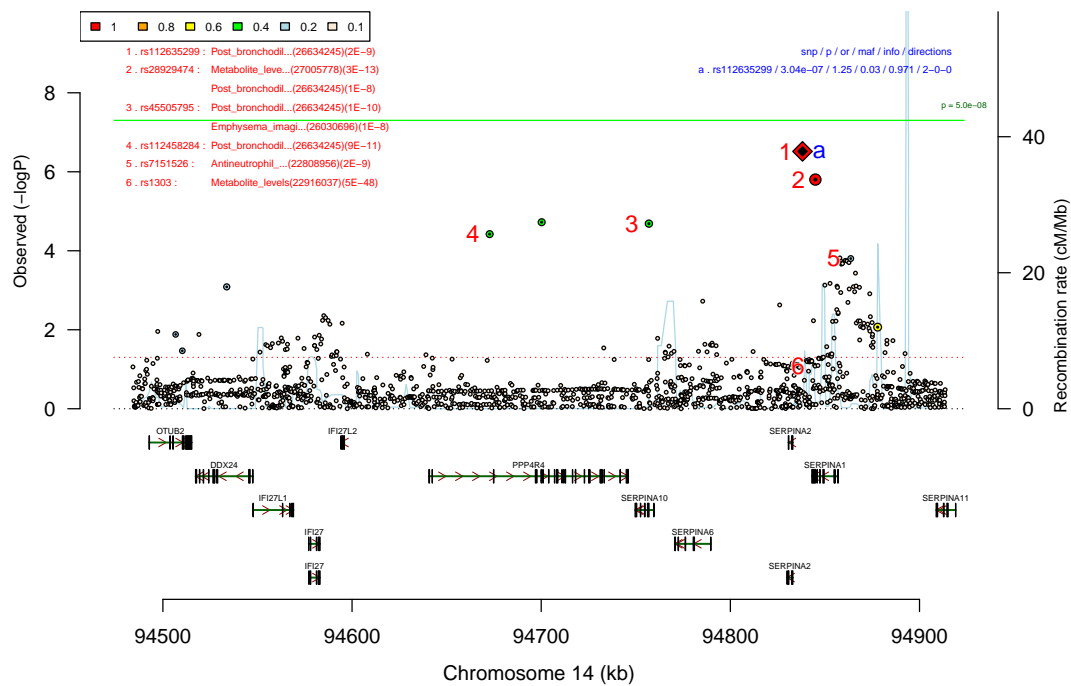
Supplementary Figure 33: Regional association plot around rs1452075 for iPSYCH-PGC meta analysis (18 381 cases and 27 969 controls). Chromosomal position is on the horizontal axis in kb. The significance of the association expressed as $-\log_{10}(P)$ (from z-test) is plotted vertically (left axis) and the recombination rate from HapMap is shown as light blue curve (right axis). The dot size is proportional to LD between the plotted SNP and the index SNP defining the associated region. Index SNPs are marked by diamonds and labelled with letters. As there is only a single index SNP in this region, colouring is based on degree of LD to that SNP as represented by r^2 (legend for r^2 is in the upper left corner). A black centre indicates that the markers is present in the 1000 Genomes Project but not in HapMap3. The green line shows the genome-wide significance level ($5 \cdot 10^{-8}$). The upper right corner show details of the index SNPs (marker name/association p-value/odds ratio for the minor allele/minor allele frequency/imputation quality/direction of effect in the two studies (N one direction - N other direction - N missing)). Loci listed in the GWAS Catalogue for other phenotypes are shown in the upper left corner (capped at 10). The green lines in the lower half show genes (based on UCSC) with black vertical lines indicating exons. Arrow heads show the direction of transcription.



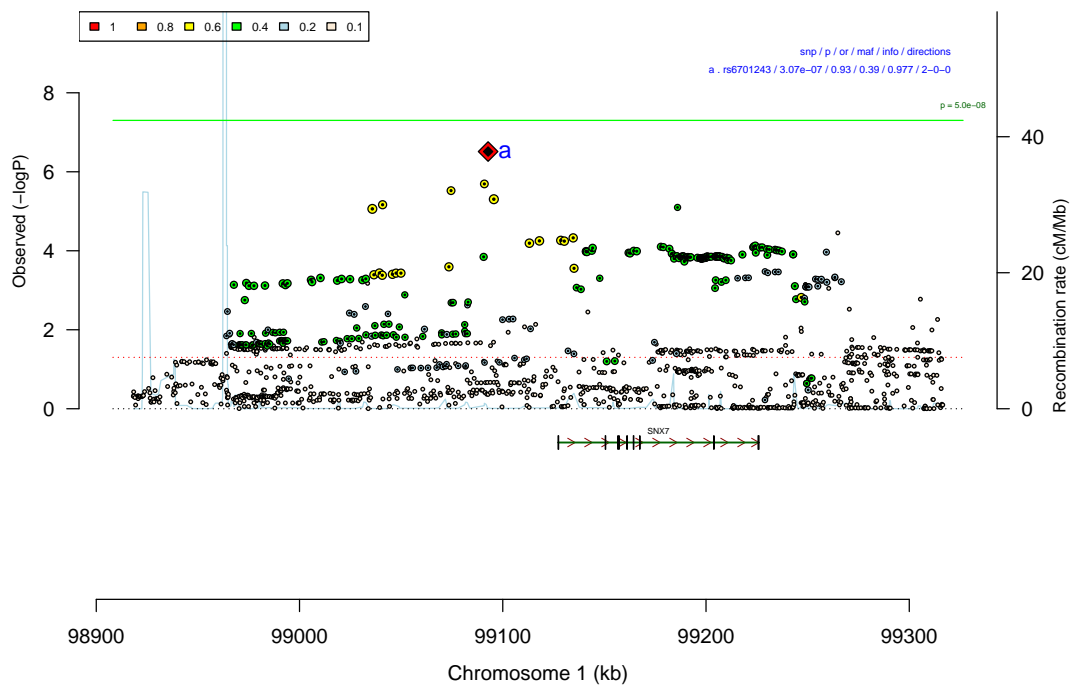
Supplementary Figure 34: Regional association plot around rs1222063 & rs201910565 for iPSYCH-PGC meta analysis (18 381 cases and 27 969 controls). Chromosomal position is on the horizontal axis in kb, The significance of the association expressed as $-\log_{10}(P)$ (from z-test) is plotted vertically (left axis) and the recombination rate from HapMap is shown as light blue curve (right axis). The dot size is proportional to LD between the plotted SNP and the index SNP defining the associated region. Index SNPs are marked by diamonds and labelled with letters. As this locus contains multiple independent index SNPs, each index SNP is denoted by a different colour. LD to each index SNP is denoted by the intensity of that colour. A black centre indicates that the markers is present in the 1000 Genomes Project but not in HapMap3. The green line shows the genome-wide significance level ($5 \cdot 10^{-8}$). The upper right corner show details of the index SNPs (marker name/association p-value/odds ratio for the minor allele/minor allele frequency/imputation quality/direction of effect in the two studies (N one direction - N other direction- N missing)). The green lines in the lower half show genes (based on UCSC) with black vertical lines indicating exons. Arrow heads show the direction of transcription.



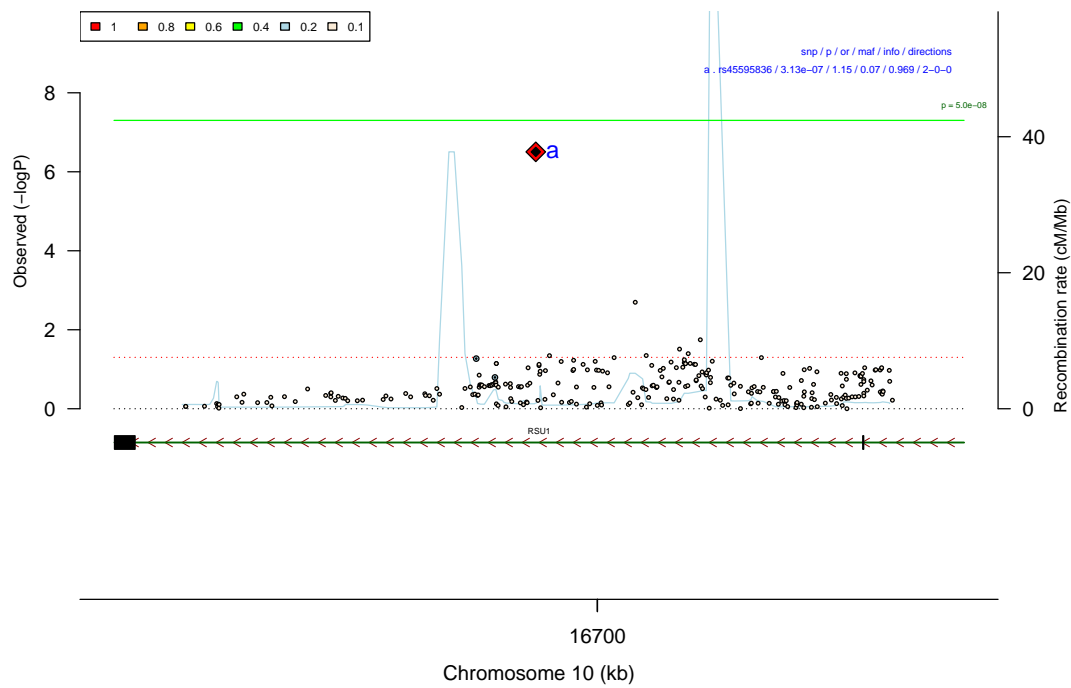
Supplementary Figure 35: Regional association plot around rs142920272 & rs141455452 for iPSYCH-PGC meta analysis (18 381 cases and 27 969 controls). Chromosomal position is on the horizontal axis in kb, The significance of the association expressed as $-\log_{10}(P)$ (from z-test) is plotted vertically (left axis) and the recombination rate from HapMap is shown as light blue curve (right axis). The dot size is proportional to LD between the plotted SNP and the index SNP defining the associated region. Index SNPs are marked by diamonds and labelled with letters. As this locus contains multiple independent index SNPs, each index SNP is denoted by a different colour. LD to each index SNP is denoted by the intensity of that colour. A black centre indicates that the markers is present in the 1000 Genomes Project but not in HapMap3. The green line shows the genome-wide significance level ($5 \cdot 10^{-8}$). The upper right corner show details of the index SNPs (marker name/association p-value/odds ratio for the minor allele/minor allele frequency/imputation quality/direction of effect in the two studies (N one direction - N other direction- N missing)). Loci listed in the GWAS Catalogue for other phenotypes are shown in the upper left corner (capped at 10). The green lines in the lower half show genes (based on UCSC) with black vertical lines indicating exons. Arrow heads show the direction of transcription.



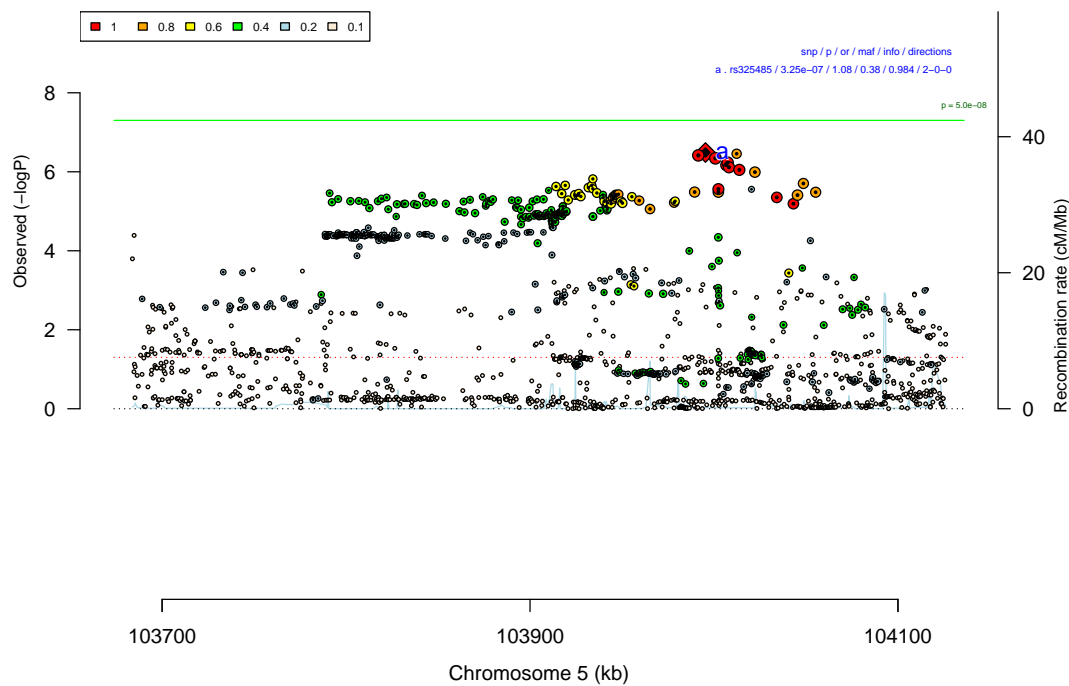
Supplementary Figure 36: Regional association plot around rs112635299 for iPSYCH-PGC meta analysis (18 381 cases and 27 969 controls). Chromosomal position is on the horizontal axis in kb, The significance of the association expressed as $-\log_{10}(P)$ (from z-test) is plotted vertically (left axis) and the recombination rate from HapMap is shown as light blue curve (right axis). The dot size is proportional to LD between the plotted SNP and the index SNP defining the associated region. Index SNPs are marked by diamonds and labelled with letters. As there is only a single index SNP in this region, colouring is based on degree of LD to that SNP as represented by r^2 (legend for r^2 is in the upper left corner). A black centre indicates that the markers is present in the 1000 Genomes Project but not in HapMap3. The green line shows the genome-wide significance level ($5 \cdot 10^{-8}$). The upper right corner show details of the index SNPs (marker name/association p-value/odds ratio for the minor allele/minor allele frequency/imputation quality/direction of effect in the two studies (N one direction - N other direction - N missing)). Loci listed in the GWAS Catalogue for other phenotypes are shown in the upper left corner (capped at 10). The green lines in the lower half show genes (based on UCSC) with black vertical lines indicating exons. Arrow heads show the direction of transcription.



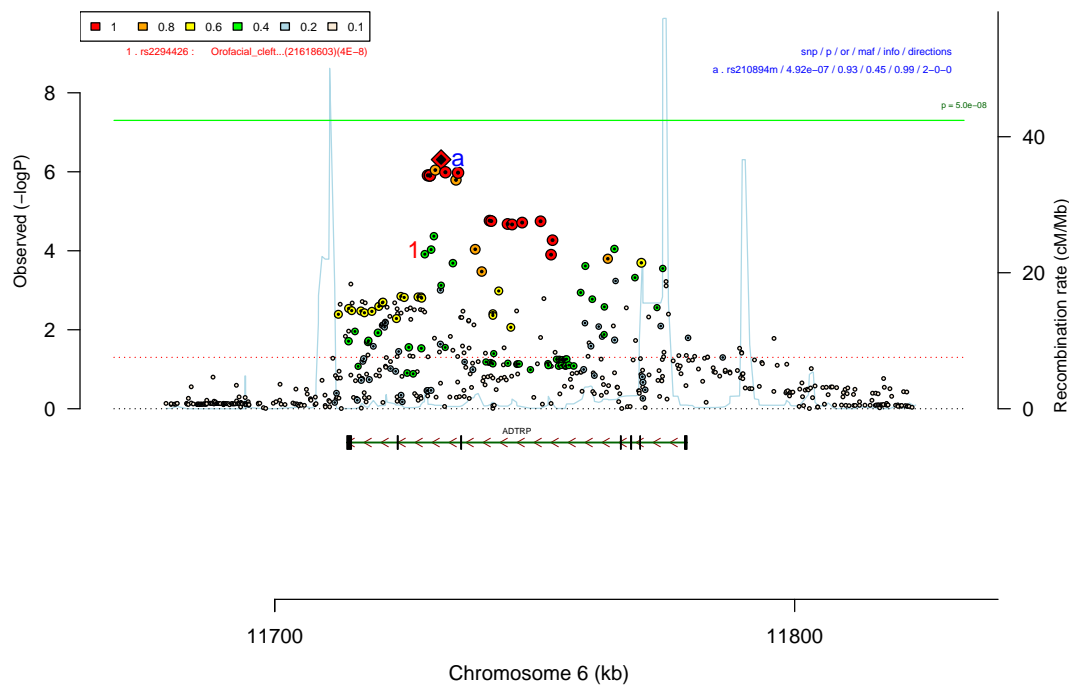
Supplementary Figure 37: Regional association plot around rs6701243 for iPSYCH-PGC meta analysis (18 381 cases and 27 969 controls). Chromosomal position is on the horizontal axis in kb, The significance of the association expressed as $-\log_{10}(P)$ (from z-test) is plotted vertically (left axis) and the recombination rate from HapMap is shown as light blue curve (right axis). The dot size is proportional to LD between the plotted SNP and the index SNP defining the associated region. Index SNPs are marked by diamonds and labelled with letters. As there is only a single index SNP in this region, colouring is based on degree of LD to that SNP as represented by r^2 (legend for r^2 is in the upper left corner). A black centre indicates that the markers is present in the 1000 Genomes Project but not in HapMap3. The green line shows the genome-wide significance level ($5 \cdot 10^{-8}$). The upper right corner show details of the index SNPs (marker name/association p-value/odds ratio for the minor allele/minor allele frequency/imputation quality/direction of effect in the two studies (N one direction - N other direction- N missing)). The green lines in the lower half show genes (based on UCSC) with black vertical lines indicating exons. Arrow heads show the direction of transcription.



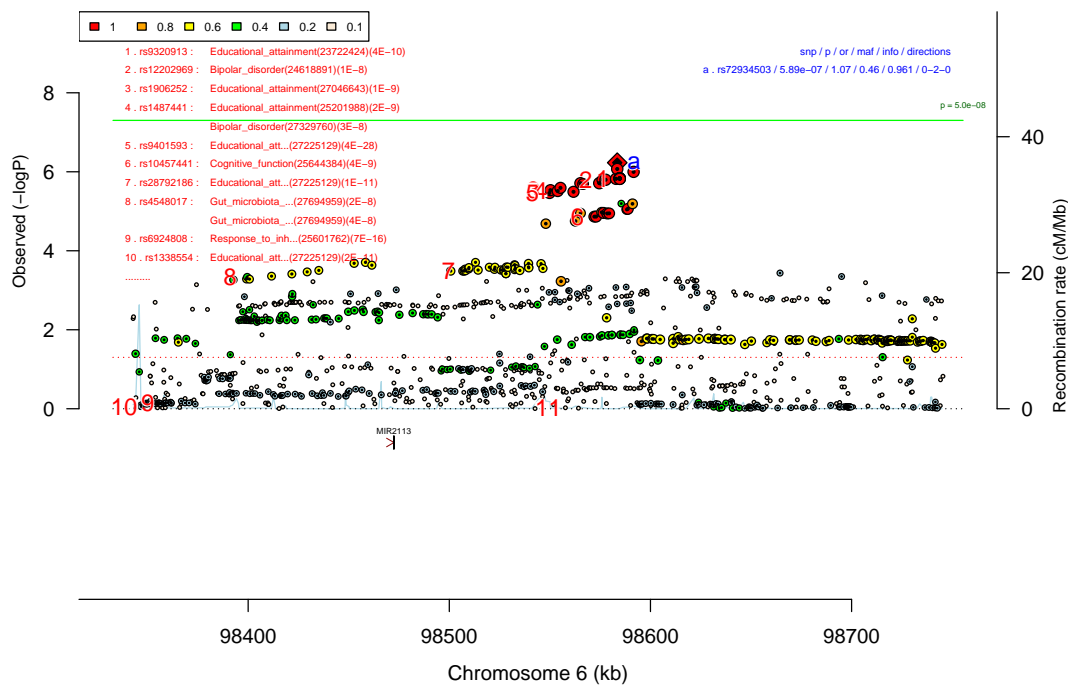
Supplementary Figure 38: Regional association plot around rs45595836 for iPSYCH-PGC meta analysis (18 381 cases and 27 969 controls). Chromosomal position is on the horizontal axis in kb, The significance of the association expressed as $-\log_{10}(P)$ (from z-test) is plotted vertically (left axis) and the recombination rate from HapMap is shown as light blue curve (right axis). The dot size is proportional to LD between the plotted SNP and the index SNP defining the associated region. Index SNPs are marked by diamonds and labelled with letters. As there is only a single index SNP in this region, colouring is based on degree of LD to that SNP as represented by r^2 (legend for r^2 is in the upper left corner). A black centre indicates that the markers is present in the 1000 Genomes Project but not in HapMap3. The green line shows the genome-wide significance level ($5 \cdot 10^{-8}$). The upper right corner show details of the index SNPs (marker name/association p-value/odds ratio for the minor allele/minor allele frequency/imputation quality/direction of effect in the two studies (N one direction - N other direction- N missing)). The green lines in the lower half show genes (based on UCSC) with black vertical lines indicating exons. Arrow heads show the direction of transcription.



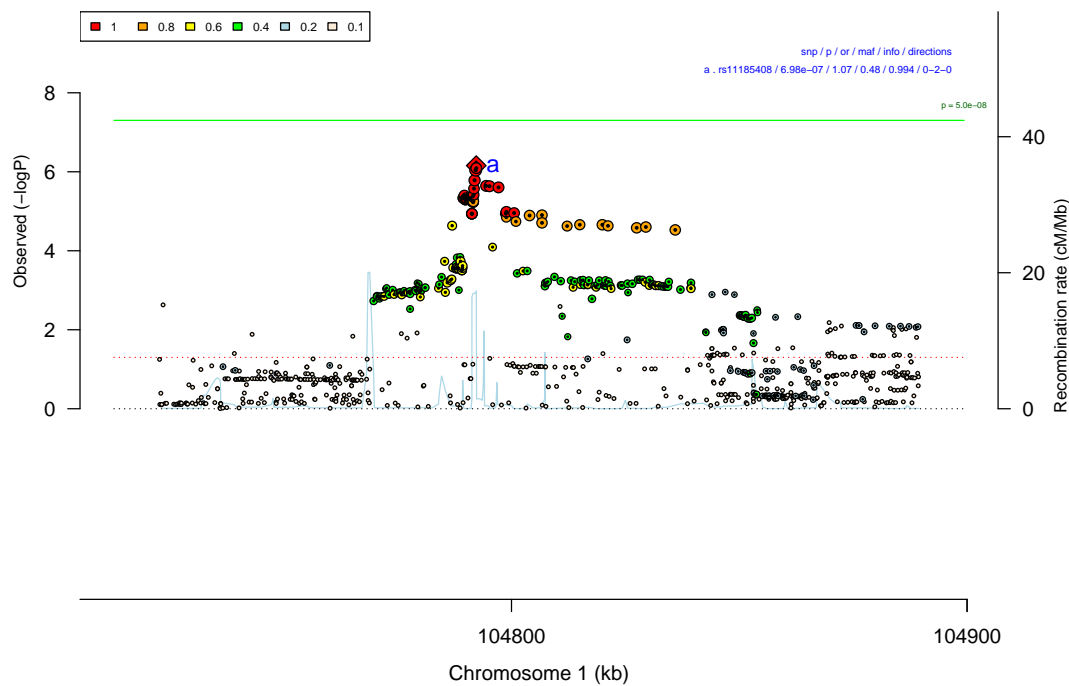
Supplementary Figure 39: Regional association plot around rs325485 for iPSYCH-PGC meta analysis (18 381 cases and 27 969 controls). Chromosomal position is on the horizontal axis in kb, The significance of the association expressed as $-\log_{10}(P)$ (from z-test) is plotted vertically (left axis) and the recombination rate from HapMap is shown as light blue curve (right axis). The dot size is proportional to LD between the plotted SNP and the index SNP defining the associated region. Index SNPs are marked by diamonds and labelled with letters. As there is only a single index SNP in this region, colouring is based on degree of LD to that SNP as represented by r^2 (legend for r^2 is in the upper left corner). A black centre indicates that the markers is present in the 1000 Genomes Project but not in HapMap3. The green line shows the genome-wide significance level ($5 \cdot 10^{-8}$). The upper right corner show details of the index SNPs (marker name/association p-value/odds ratio for the minor allele/minor allele frequency/imputation quality/direction of effect in the two studies (N one direction - N other direction- N missing)).



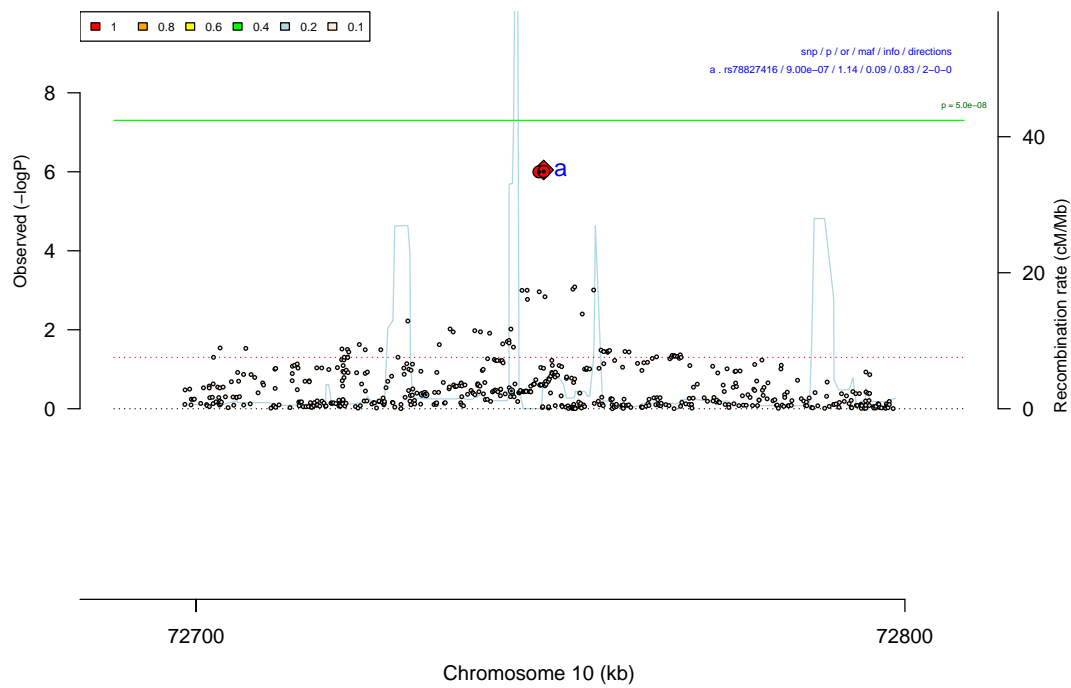
Supplementary Figure 40: Regional association plot around rs210894m for iPSYCH-PGC meta analysis (18 381 cases and 27 969 controls). Chromosomal position is on the horizontal axis in kb, The significance of the association expressed as $-\log_{10}(P)$ (from z-test) is plotted vertically (left axis) and the recombination rate from HapMap is shown as light blue curve (right axis). The dot size is proportional to LD between the plotted SNP and the index SNP defining the associated region. Index SNPs are marked by diamonds and labelled with letters. As there is only a single index SNP in this region, colouring is based on degree of LD to that SNP as represented by r^2 (legend for r^2 is in the upper left corner). A black centre indicates that the markers is present in the 1000 Genomes Project but not in HapMap3. The green line shows the genome-wide significance level ($5 \cdot 10^{-8}$). The upper right corner show details of the index SNPs (marker name/association p-value/odds ratio for the minor allele/minor allele frequency/imputation quality/direction of effect in the two studies (N one direction - N other direction- N missing)). Loci listed in the GWAS Catalogue for other phenotypes are shown in the upper left corner (capped at 10). The green lines in the lower half show genes (based on UCSC) with black vertical lines indicating exons. Arrow heads show the direction of transcription.



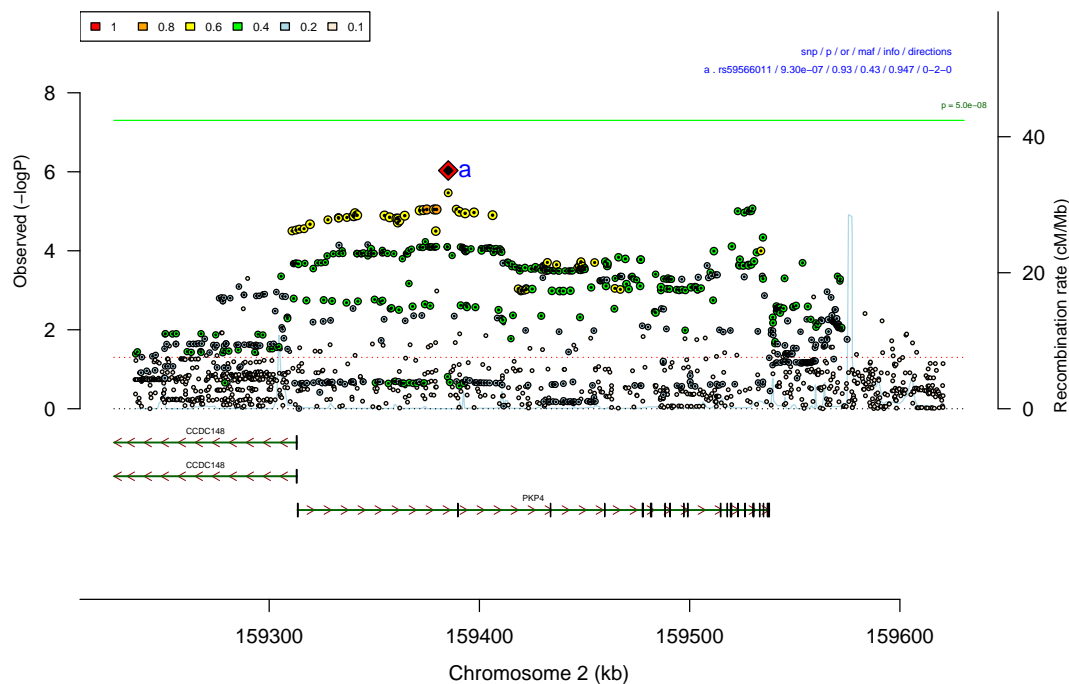
Supplementary Figure 41: Regional association plot around rs72934503 for iPSYCH-PGC meta analysis (18 381 cases and 27 969 controls). Chromosomal position is on the horizontal axis in kb, The significance of the association expressed as $-\log_{10}(P)$ (from z-test) is plotted vertically (left axis) and the recombination rate from HapMap is shown as light blue curve (right axis). The dot size is proportional to LD between the plotted SNP and the index SNP defining the associated region. Index SNPs are marked by diamonds and labelled with letters. As there is only a single index SNP in this region, colouring is based on degree of LD to that SNP as represented by r^2 (legend for r^2 is in the upper left corner). A black centre indicates that the markers is present in the 1000 Genomes Project but not in HapMap3. The green line shows the genome-wide significance level ($5 \cdot 10^{-8}$). The upper right corner show details of the index SNPs (marker name/association p-value/odds ratio for the minor allele/minor allele frequency/imputation quality/direction of effect in the two studies (N one direction - N other direction - N missing)). Loci listed in the GWAS Catalogue for other phenotypes are shown in the upper left corner (capped at 10). The green lines in the lower half show genes (based on UCSC) with black vertical lines indicating exons. Arrow heads show the direction of transcription.



Supplementary Figure 42: Regional association plot around rs11185408 for iPSYCH-PGC meta analysis (18 381 cases and 27 969 controls). Chromosomal position is on the horizontal axis in kb, The significance of the association expressed as $-\log_{10}(P)$ (from z-test) is plotted vertically (left axis) and the recombination rate from HapMap is shown as light blue curve (right axis). The dot size is proportional to LD between the plotted SNP and the index SNP defining the associated region. Index SNPs are marked by diamonds and labelled with letters. As there is only a single index SNP in this region, colouring is based on degree of LD to that SNP as represented by r^2 (legend for r^2 is in the upper left corner). A black centre indicates that the markers is present in the 1000 Genomes Project but not in HapMap3. The green line shows the genome-wide significance level ($5 \cdot 10^{-8}$). The upper right corner show details of the index SNPs (marker name/association p-value/odds ratio for the minor allele/minor allele frequency/imputation quality/direction of effect in the two studies (N one direction - N other direction- N missing)).



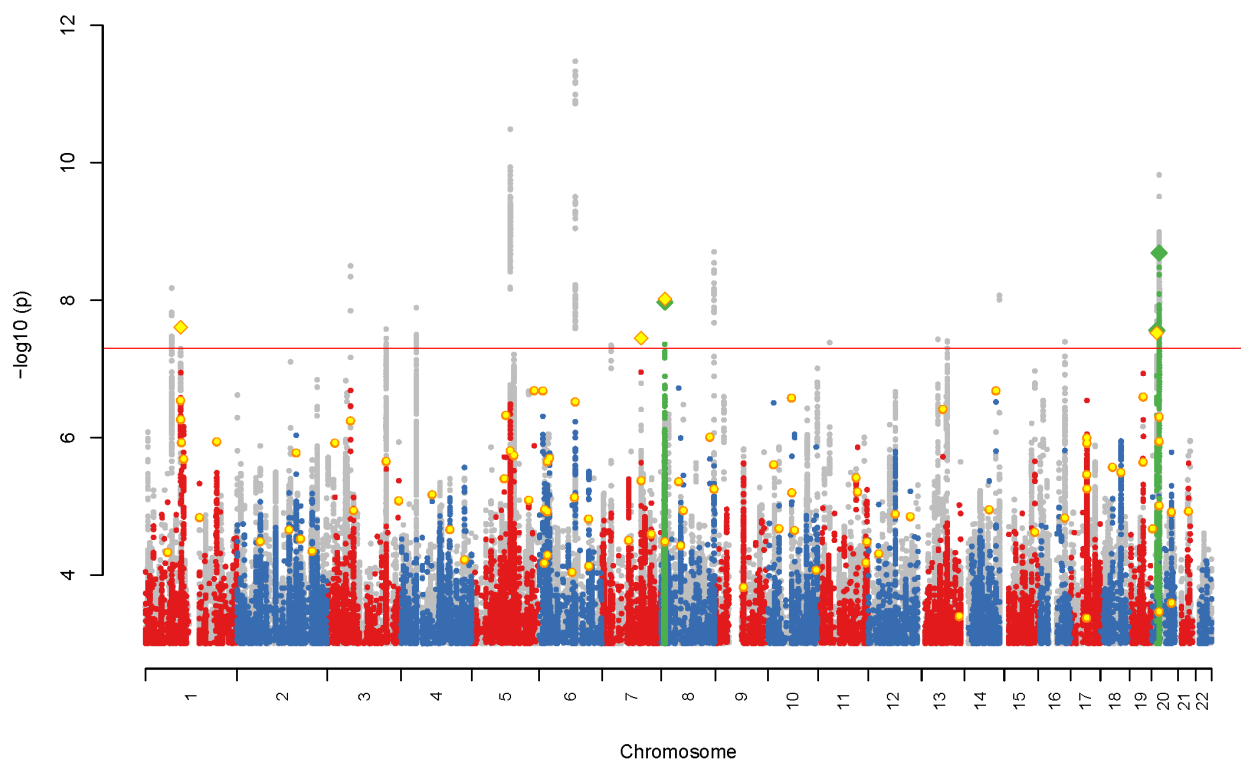
Supplementary Figure 43: Regional association plot around rs78827416 for iPSYCH-PGC meta analysis (18 381 cases and 27 969 controls). Chromosomal position is on the horizontal axis in kb, The significance of the association expressed as $-\log_{10}(P)$ (from z-test) is plotted vertically (left axis) and the recombination rate from HapMap is shown as light blue curve (right axis). The dot size is proportional to LD between the plotted SNP and the index SNP defining the associated region. Index SNPs are marked by diamonds and labelled with letters. As there is only a single index SNP in this region, colouring is based on degree of LD to that SNP as represented by r^2 (legend for r^2 is in the upper left corner). A black centre indicates that the markers is present in the 1000 Genomes Project but not in HapMap3. The green line shows the genome-wide significance level ($5 \cdot 10^{-8}$). The upper right corner show details of the index SNPs (marker name/association p-value/odds ratio for the minor allele/minor allele frequency/imputation quality/direction of effect in the two studies (N one direction - N other direction- N missing)).



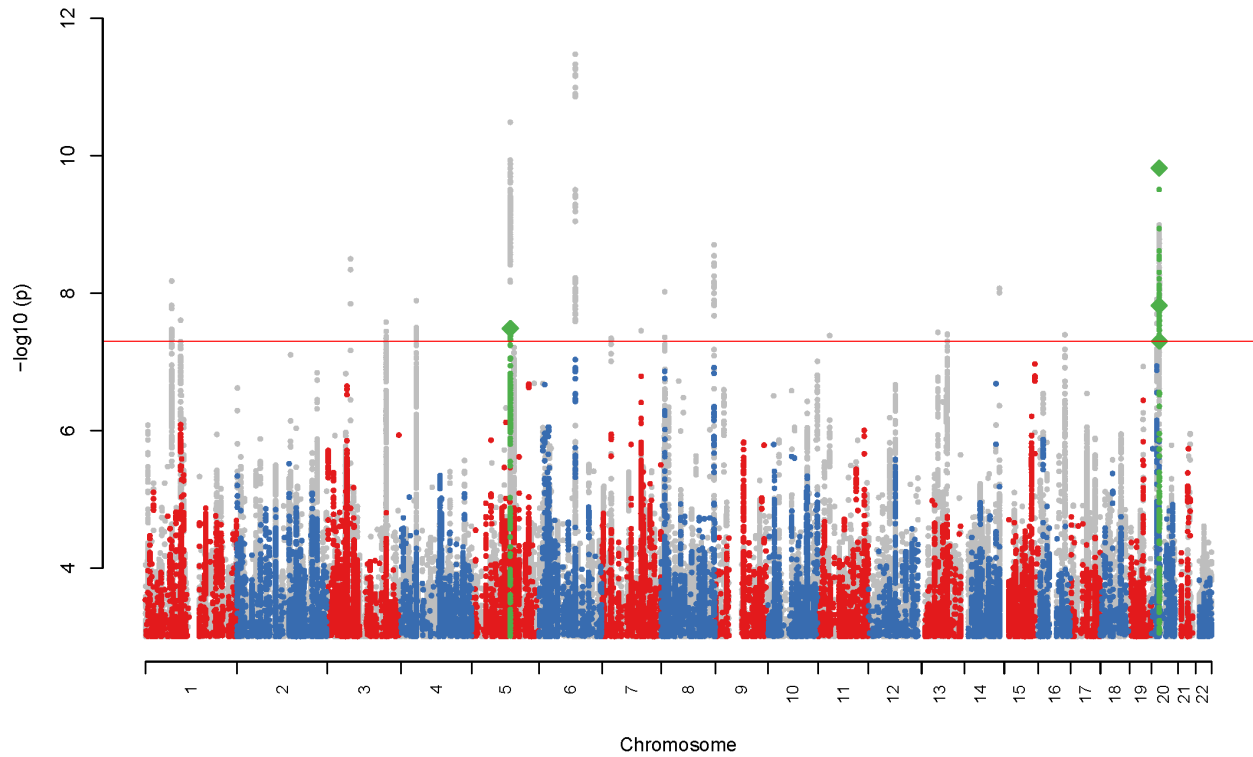
Supplementary Figure 44: Regional association plot around rs59566011 for iPSYCH-PGC meta analysis (18 381 cases and 27 969 controls). Chromosomal position is on the horizontal axis in kb. The significance of the association expressed as $-\log_{10}(P)$ (from z-test) is plotted vertically (left axis) and the recombination rate from HapMap is shown as light blue curve (right axis). The dot size is proportional to LD between the plotted SNP and the index SNP defining the associated region. Index SNPs are marked by diamonds and labelled with letters. As there is only a single index SNP in this region, colouring is based on degree of LD to that SNP as represented by r^2 (legend for r^2 is in the upper left corner). A black centre indicates that the markers is present in the 1000 Genomes Project but not in HapMap3. The green line shows the genome-wide significance level ($5 \cdot 10^{-8}$). The upper right corner shows details of the index SNPs (marker name/association p-value/odds ratio for the minor allele/minor allele frequency/imputation quality/direction of effect in the two studies (N one direction - N other direction - N missing)). The green lines in the lower half show genes (based on UCSC) with black vertical lines indicating exons. Arrow heads show the direction of transcription.

4.1.4 MTAG analyses

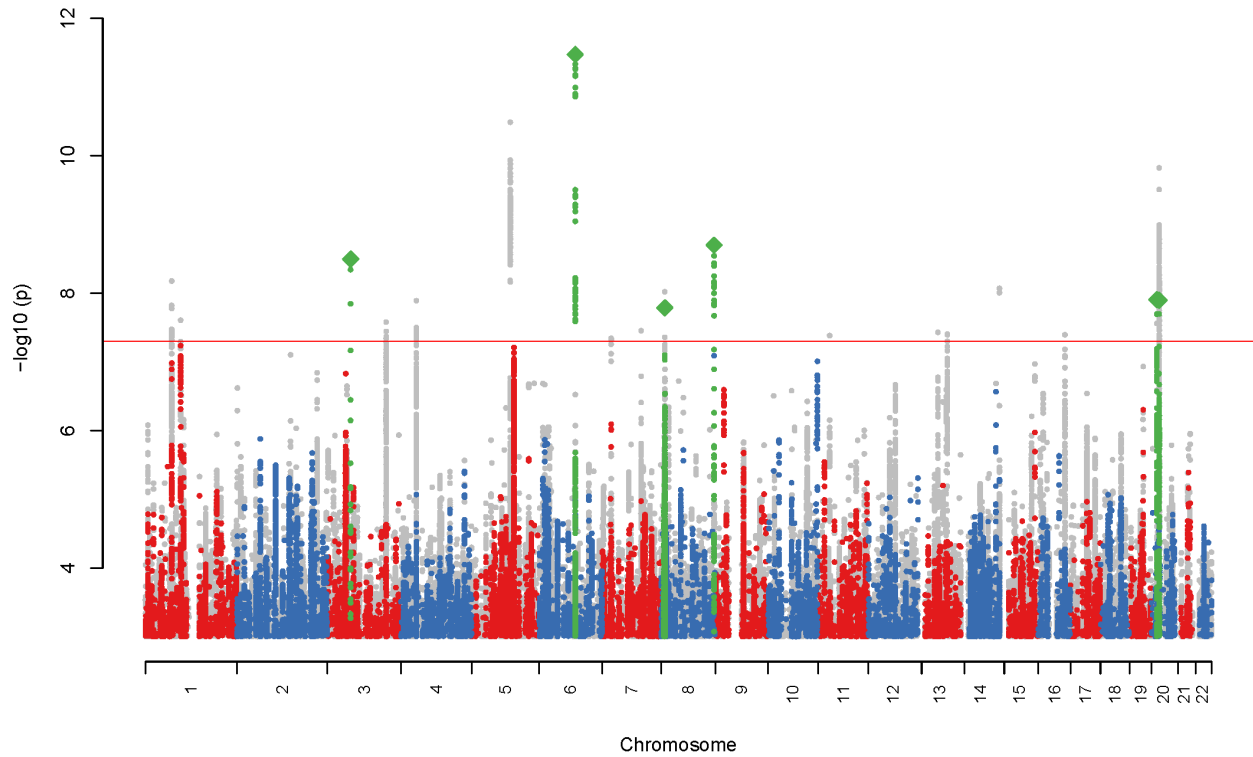
In this section we show the full size version of the Manhattan plots drawn as sub-panels in figure 1 of the main manuscript, Figures 45–48, and regional plots of the 7 top loci, Figures 49–55. Each Manhattan plot is drawn on a background of a composite analysis corresponding to the minimal p-value at each marker achieved in each of the analyses main scan (iPSYCH+PGC), meta-analysis with replication sample, MTAG with schizophrenia[8], MTAG with educational attainment[50], and MTAG of major depression[48]. The regional plot are arranged with three panels showing the local plots for the input GWAS of ASD and the scndary phenotype as well as the resulting MTAG analysis.



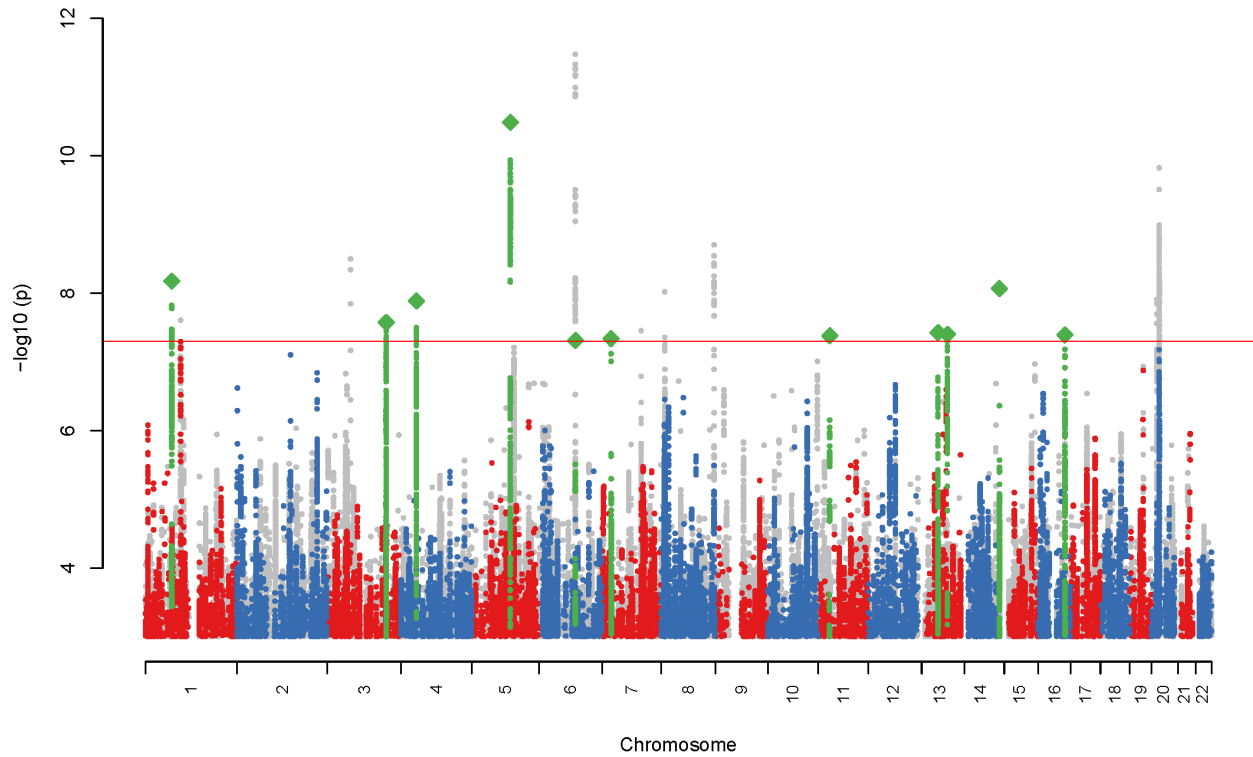
Supplementary Figure 45: Manhattan plot of ASD GWAS (18,381 cases and 27,969 controls) on composite MTAG background. The x axis shows genomic position (chromosomes 1–22) and the y axis the statistical significance as $-\log_{10}(P)$ of z statistics. Yellow marks show results from the meta-analysis with the follow-up samples (2,119 cases and 142,379 controls). Diamonds indicate the index SNP of genome wide significant clumps (at $5 \cdot 10^{-8}$) and genome wide significant clumps are painted green. The grey MTAG composite consists of the maximum at each locus of the results from the main scan, combined with follow-up, and three MTAG analyses.



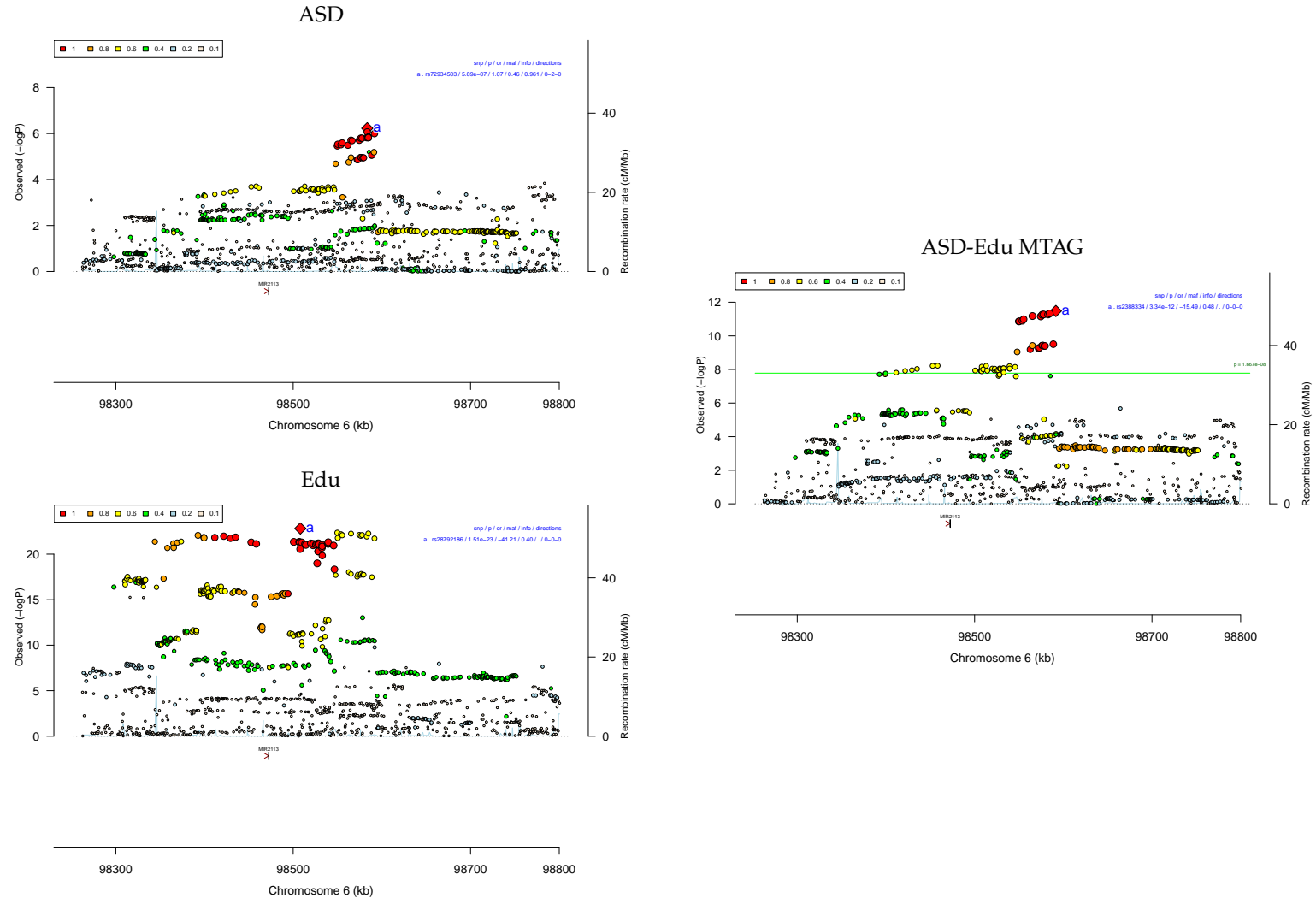
Supplementary Figure 46: Manhattan plot of MTAG analysis of ASD (18381 cases, 27969 controls and mean $\chi^2 = 1.201$) and schizophrenia[8] (without the Danish samples, 34129 cases, 45512 controls, and mean $\chi^2 = 1.804$) drawn on composite MTAG background. MTAG gives schizophrenia a weight of 0.27. The x axis shows genomic position (chromosomes 1–22) and the y axis the statistical significance as $-\log_{10}(P)$ of z statistics. Diamonds indicate the index SNP of genome wide significant clumps (at $5 \cdot 10^{-8}/3$) which in turn are painted green. The grey MTAG composit consists of the maximum at each locus of the results from the main scan, combined with follow-up, and three MTAG analyses.



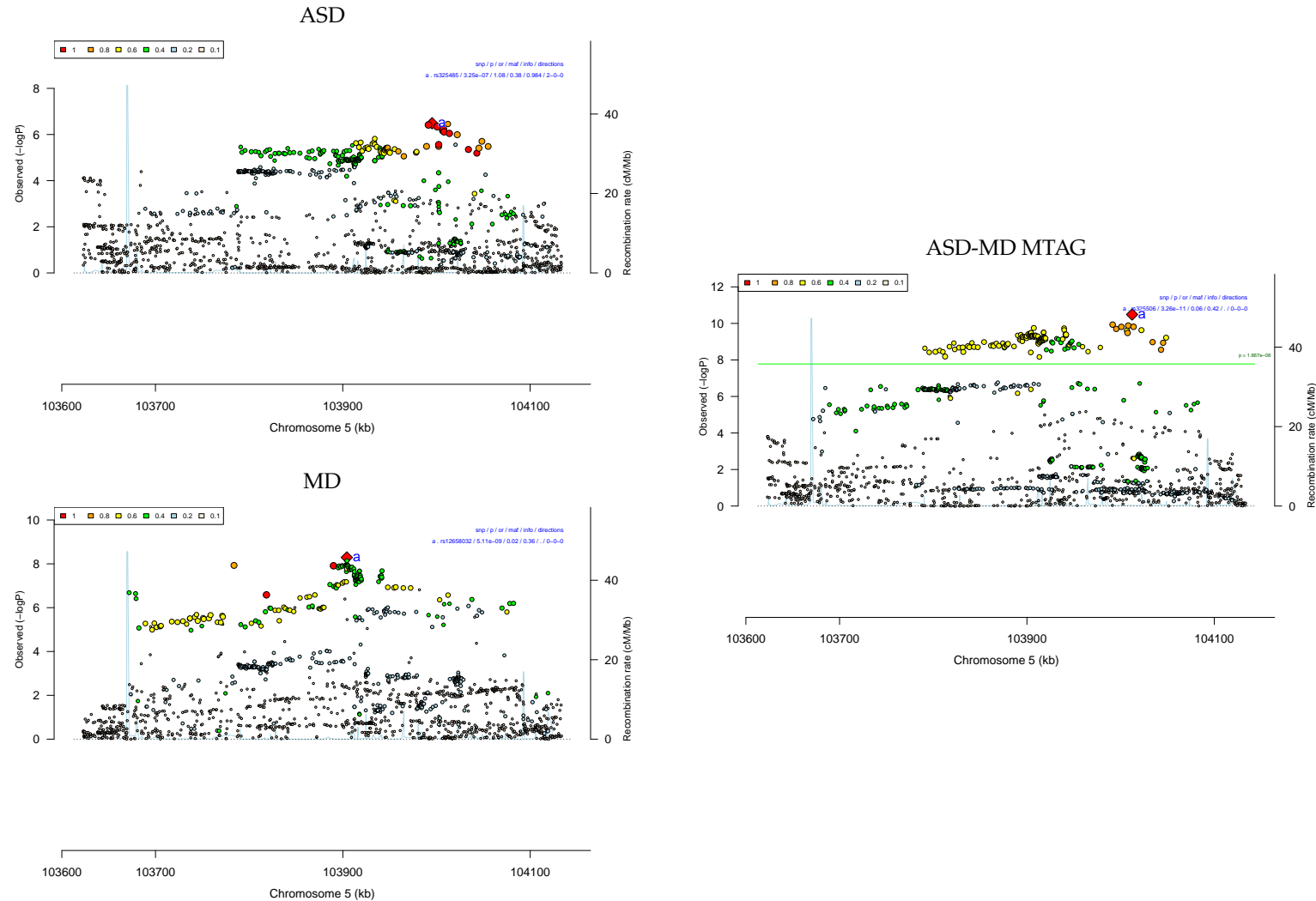
Supplementary Figure 47: Manhattan plot of MTAG analysis of ASD (18381 cases, 27969 controls and mean $\chi^2 = 1.201$) and educational attainment[50] (328917 samples and mean $\chi^2 = 1.648$) drawn on composite MTAG background. MTAG gives educational attainment a weight of 0.11. The x axis shows genomic position (chromosomes 1–22) and the y axis the statistical significance as $-\log_{10}(P)$ of z statistics. Diamonds indicate the index SNP of genome wide significant clumps (at $5 \cdot 10^{-8}/3$) which in turn are painted green. The grey MTAG composite consists of the maximum at each locus of the results from the main scan, combined with follow-up, and three MTAG analyses.



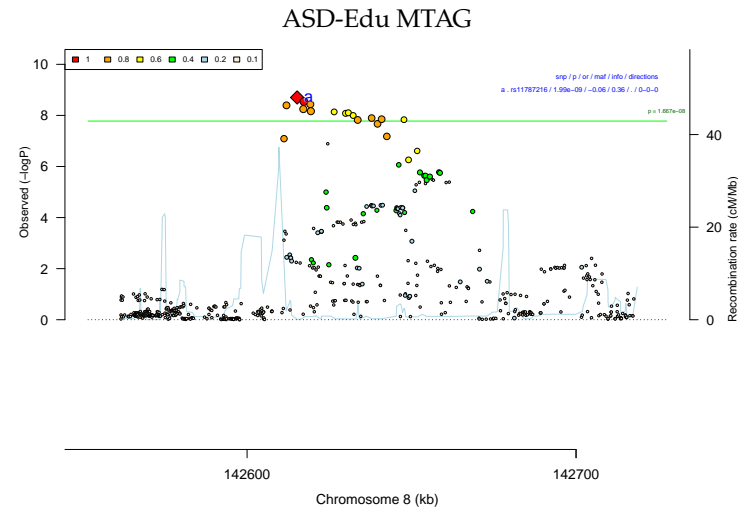
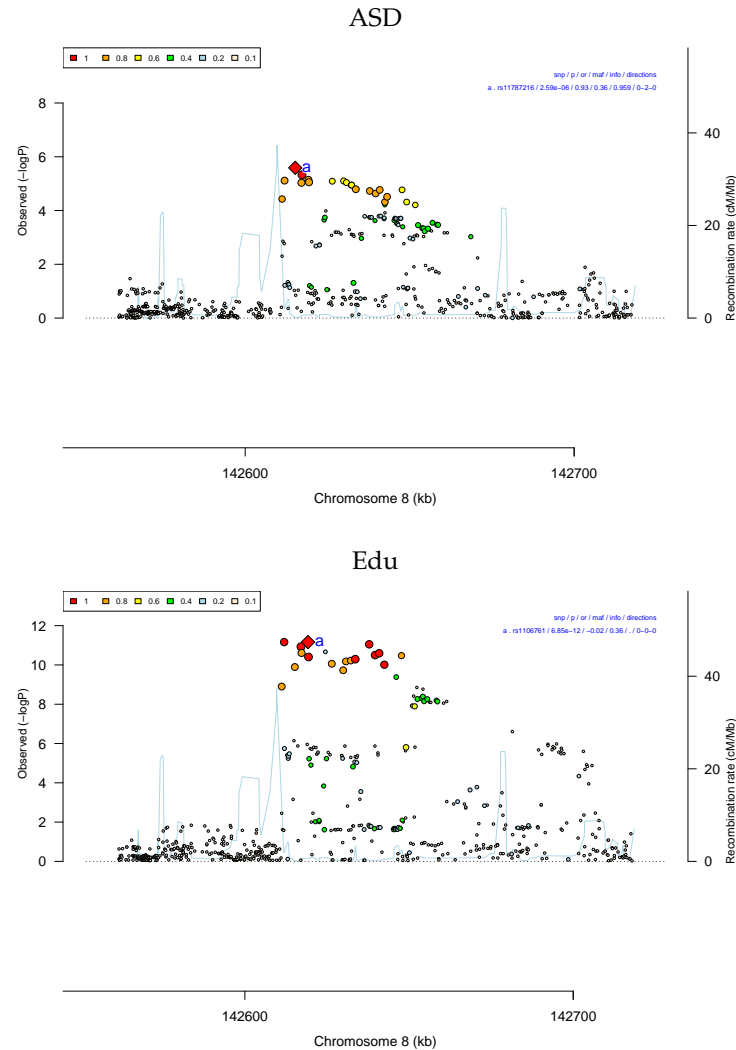
Supplementary Figure 48: Manhattan plot of MTAG analysis of ASD (18381 cases, 27969 controls and mean $\chi^2 = 1.201$) and major depression (including the results from 23andMe[49], but excluding the Danish samples, 111902 cases, 312113 controls, and mean $\chi^2 = 1.477$) drawn on composite MTAG background. MTAG gives major depression a weight of 0.24. The x axis shows genomic position (chromosomes 1–22) and the y axis the statistical significance as $-\log_{10}(P)$ of z statistics. Diamonds indicate the index SNP of genome wide significant clumps (at $5 \cdot 10^{-8}/3$) which in turn are painted green. The grey MTAG composit consists of the maximum at each locus of the results from the main scan, combined with follow-up, and three MTAG analyses.



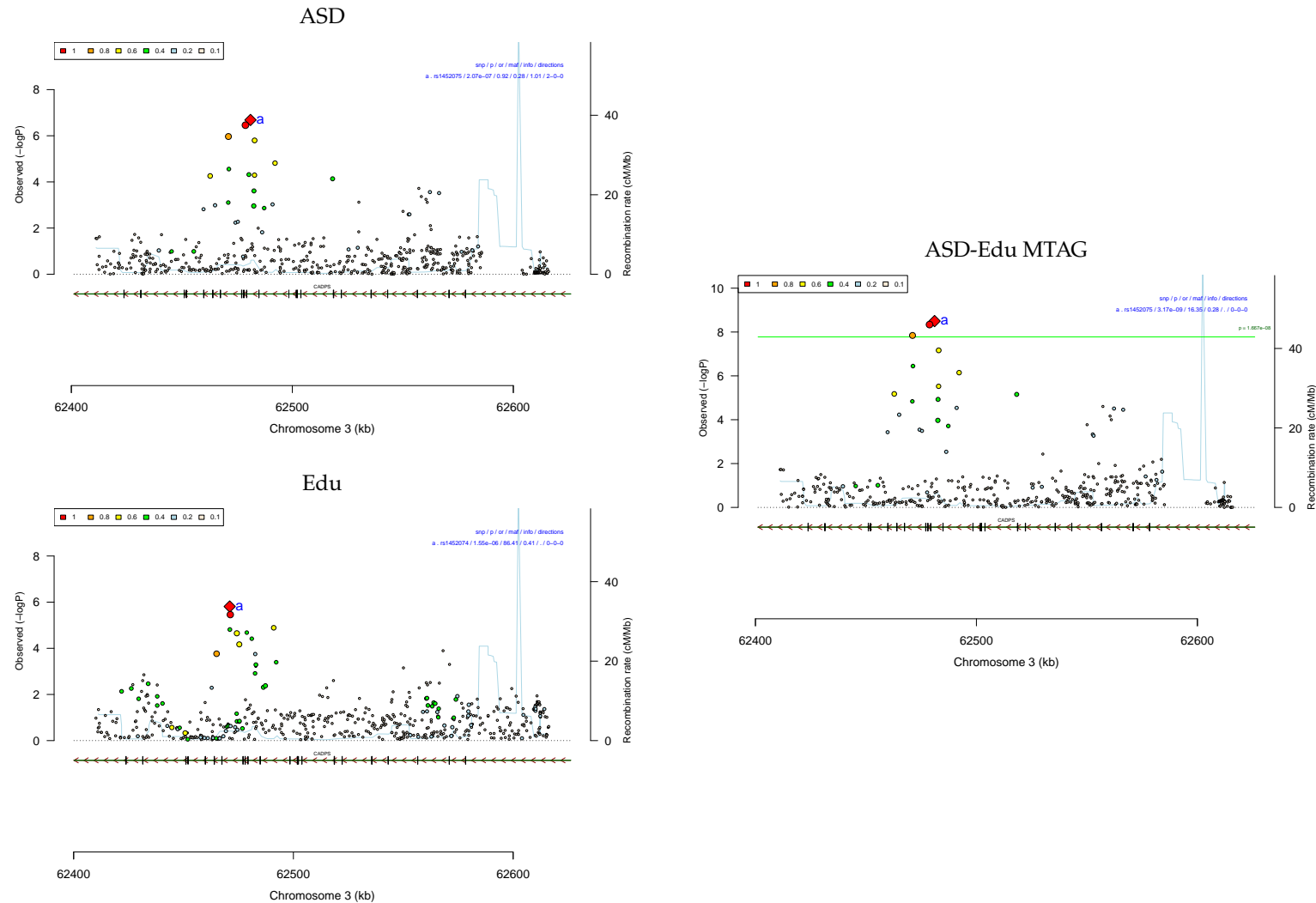
Supplementary Figure 49: Regional association plot around rs2388334 on chr 6 for ASD-Edu MTAG analysis. The left panels show the regional plots for the ASD GWAS (18 381 cases and 27 969 controls) and the Edu GWAS (328 917 samples), and the right panel is the regional plot for the resulting MTAG analysis of ASD and Edu. Plots show very similar patterns with ASD having less power. Horizontal axis shows chromosomal position in kb, Significance of the association expressed as $-\log_{10}(P)$ (from z-test) is plotted vertically (left axis) and the recombination rate from HapMap is shown as light blue curve (right axis). The dot size is proportional to LD between the plotted SNP and the index SNP defining the associated region. Index SNPs are marked by diamonds and labelled with letters. Colouring is based on degree of LD to that SNP as represented by r^2 (legend for r^2 is in the upper left corner). A black centre indicates that the markers are in the 1000 Genomes Project but not in HapMap3. The green line in the right panel shows the MTAG analysis significance level ($1.667 \cdot 10^{-8}$). Details of the index SNPs in the upper right corner show (marker name/association p-value/odds ratio for the minor allele/minor allele frequency/imputation quality/direction of effect in the included studies (info only available for ASD)). The green lines in the lower half show genes (based on UCSC) with black vertical lines indicating exons. Arrow heads show the direction of transcription.



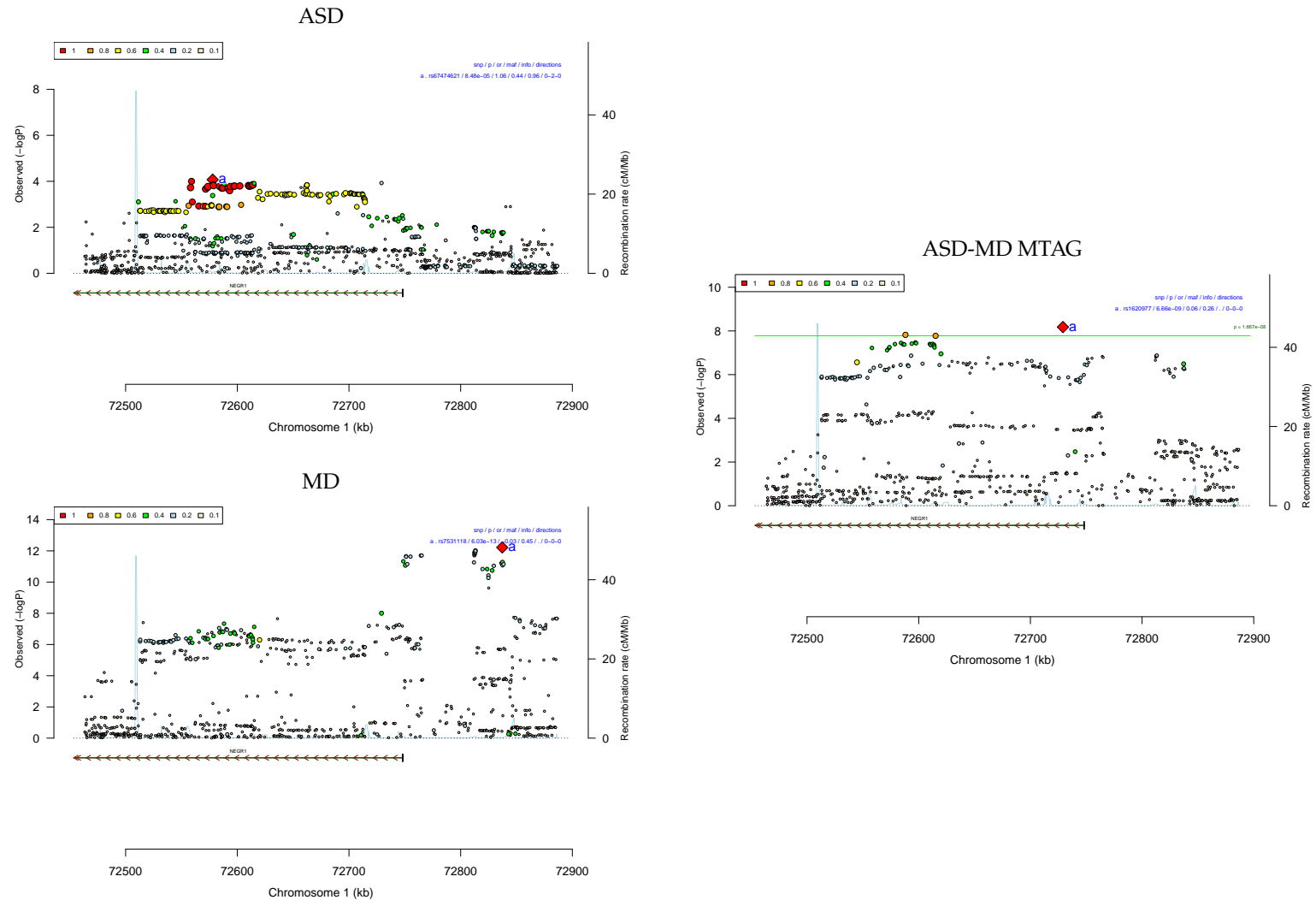
Supplementary Figure 50: Regional association plot around rs325506 on chr 5 for ASD-MD MTAG analysis. The left panels shows the regional plots for the ASD GWAS (18 381 cases and 27 969 controls) and the MD GWAS (111 902 cases and 312 113 controls), and the right panel is the regional plot for the resulting MTAG analysis of ASD and MD. Plots show very similar patterns with ASD having less power. Horizontal axis show chromosomal position in kb, Significance of the association expressed as $-\log_{10}(P)$ (from z-test) is plotted vertically (left axis) and the recombination rate from HapMap is shown as light blue curve (right axis). The dot size is proportional to LD between the plotted SNP and the index SNP defining the associated region. Index SNPs are marked by diamonds and labelled with letters. Colouring is based on degree of LD to that SNP as represented by r^2 (legend for r^2 is in the upper left corner). A black centre indicates that the markers is in the 1000 Genomes Project but not in HapMap3. The green line in the right panel shows the MTAG analysis significance level ($1.667 \cdot 10^{-8}$). Details of the index SNPs is in the upper right corner show (marker name/association p-value/odds ratio for the minor allele/minor allele frequency/imputation quality/direction of effect in the included studies (info only available for ASD)).



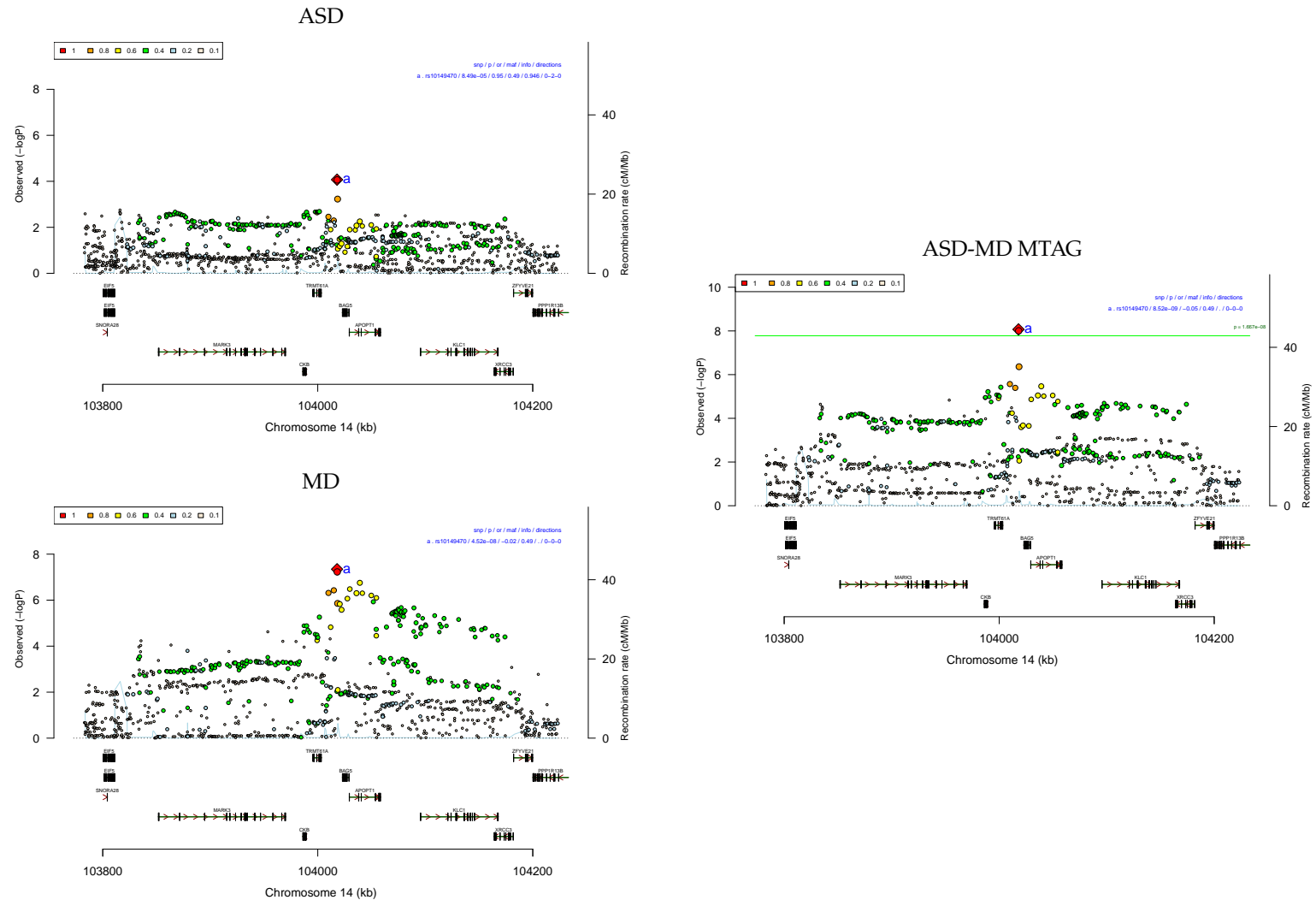
Supplementary Figure 51: Regional association plot around rs11787216 on chr 8 for ASD-Edu MTAG analysis. The left panels shows the regional plots for the ASD GWAS (18 381 cases and 27 969 controls) and the Edu GWAS (328 917 samples), and the right panel is the regional plot for the resulting MTAG analysis of ASD and Edu. Plots show very similar patterns with ASD having less power. Horizontal axis show chromosomal position in kb, Significance of the association expressed as $-\log_{10}(P)$ (from z-test) is plotted vertically (left axis) and the recombination rate from HapMap is shown as light blue curve (right axis). The dot size is proportional to LD between the plotted SNP and the index SNP defining the associated region. Index SNPs are marked by diamonds and labelled with letters. Colouring is based on degree of LD to that SNP as represented by r^2 (legend for r^2 is in the upper left corner). A black centre indicates that the markers is in the 1000 Genomes Project but not in HapMap3. The green line in the right panel shows the MTAG analysis significance level ($1.667 \cdot 10^{-8}$). Details of the index SNPs is in the upper right corner show (marker name/association p-value/odds ratio for the minor allele/minor allele frequency/imputation quality/direction of effect in the included studies (info only available for ASD)).



Supplementary Figure 52: Regional association plot around rs1452075 on chr 3 for ASD-Edu MTAG analysis. The left panels shows the regional plots for the ASD GWAS (18 381 cases and 27 969 controls) and the Edu GWAS (328 917 samples), and the right panel is the regional plot for the resulting MTAG analysis of ASD and Edu. Plots show very similar patterns with ASD having less power. Horizontal axis show chromosomal position in kb, Significance of the association expressed as $-\log_{10}(P)$ (from z-test) is plotted vertically (left axis) and the recombination rate from HapMap is shown as light blue curve (right axis). The dot size is proportional to LD between the plotted SNP and the index SNP defining the associated region. Index SNPs are marked by diamonds and labelled with letters. Colouring is based on degree of LD to that SNP as represented by r^2 (legend for r^2 is in the upper left corner). A black centre indicates that the markers is in the 1000 Genomes Project but not in HapMap3. The green line in the right panel shows the MTAG analysis significance level ($1.667 \cdot 10^{-8}$). Details of the index SNPs is in the upper right corner show (marker name/association p-value/odds ratio for the minor allele/minor allele frequency/imputation quality/direction of effect in the included studies (info only available for ASD)). The green lines in the lower half show genes (based on UCSC) with black vertical lines indicating exons. Arrow heads show the direction of transcription.

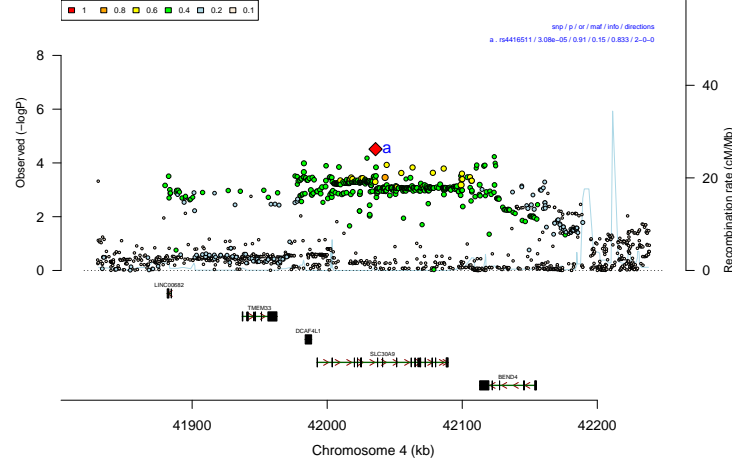


Supplementary Figure 53: Regional association plot around rs1620977 on chr 1 for ASD-MD MTAG analysis. The left panels show the regional plots for the ASD GWAS (18 381 cases and 27 969 controls) and the MD GWAS (111 902 cases and 312 113 controls), and the right panel is the regional plot for the resulting MTAG analysis of ASD and MD. Plots show very similar patterns with ASD having less power and the index SNP shifted. Horizontal axis shows chromosomal position in kb. Significance of the association expressed as $-\log_{10}(P)$ (from z-test) is plotted vertically (left axis) and the recombination rate from HapMap is shown as light blue curve (right axis). The dot size is proportional to LD between the plotted SNP and the index SNP defining the associated region. Index SNPs are marked by diamonds and labelled with letters. Colouring is based on degree of LD to that SNP as represented by r^2 (legend for r^2 is in the upper left corner). A black centre indicates that the markers are in the 1000 Genomes Project but not in HapMap3. The green line in the right panel shows the MTAG analysis significance level ($1.667 \cdot 10^{-8}$). Details of the index SNPs are in the upper right corner shown (marker name/association p-value/odds ratio for the minor allele/minor allele frequency/imputation quality/direction of effect in the included studies (info only available for ASD)). The green lines in the lower half show genes (based on UCSC) with black vertical lines indicating exons. Arrow heads show the direction of transcription.

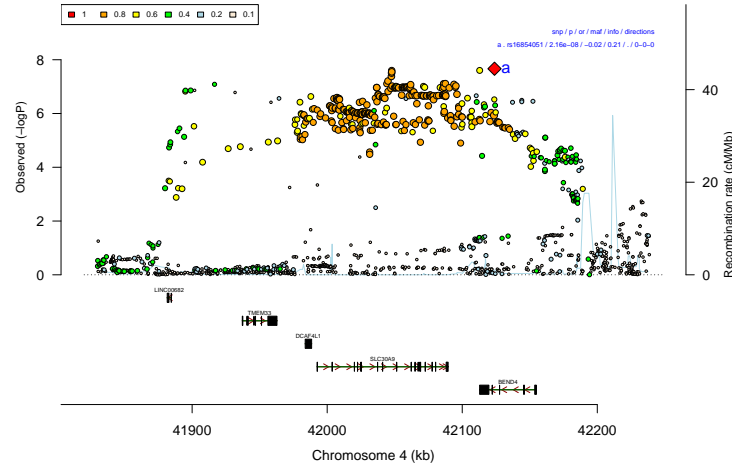


Supplementary Figure 54: Regional association plot around rs10149470 on chr 14 for ASD-MD MTAG analysis. The left panels shows the regional plots for the ASD GWAS (18 381 cases and 27 969 controls) and the MD GWAS (111 902 cases and 312 113 controls), and the right panel is the regional plot for the resulting MTAG analysis of ASD and MD. Plots show very similar patterns with ASD having less power. Horizontal axis show chromosomal position in kb, Significance of the association expressed as $-\log_{10}(P)$ (from z-test) is plotted vertically (left axis) and the recombination rate from HapMap is shown as light blue curve (right axis). The dot size is proportional to LD between the plotted SNP and the index SNP defining the associated region. Index SNPs are marked by diamonds and labelled with letters. Colouring is based on degree of LD to that SNP as represented by r^2 (legend for r^2 is in the upper left corner). A black centre indicates that the markers is in the 1000 Genomes Project but not in HapMap3. The green line in the right panel shows the MTAG analysis significance level ($1.667 \cdot 10^{-8}$). Details of the index SNPs is in the upper right corner show (marker name/association p-value/odds ratio for the minor allele/minor allele frequency/imputation quality/direction of effect in the included studies (info only available for ASD)). The green lines in the lower half show genes (based on UCSC) with black vertical lines indicating exons. Arrow heads show the direction of transcription.

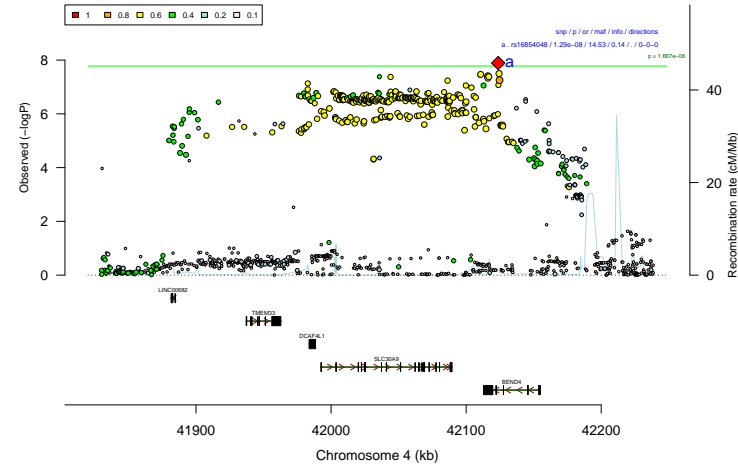
ASD



MD

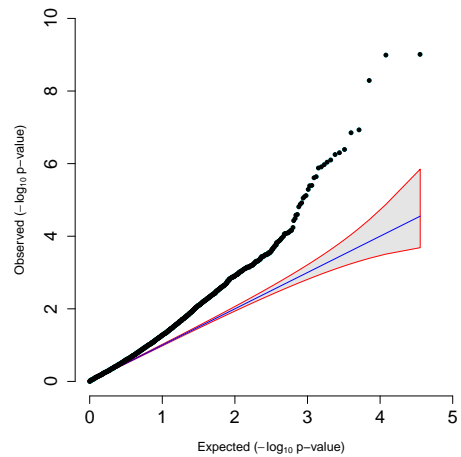


ASD-MD MTAG



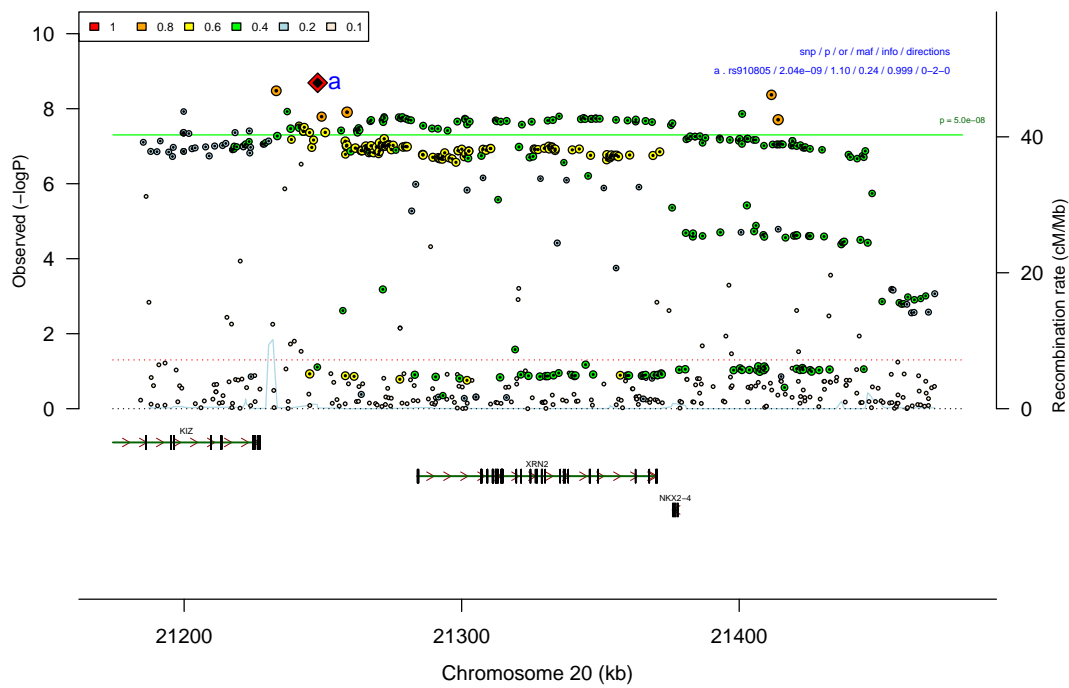
Supplementary Figure 55: Regional association plot around rs16854048 on chr 4 for ASD-MD MTAG analysis. The left panels shows the regional plots for the ASD GWAS (18 381 cases and 27 969 controls) and the MD GWAS (111 902 cases and 312 113 controls), and the right panel is the regional plot for the resulting MTAG analysis of ASD and MD. Plots show very similar patterns with ASD having less power and the index SNP shifted. Horizontal axis show chromosomal position in kb, Significance of the association expressed as $-\log_{10}(P)$ (from z-test) is plotted vertically (left axis) and the recombination rate from HapMap is shown as light blue curve (right axis). The dot size is proportional to LD between the plotted SNP and the index SNP defining the associated region. Index SNPs are marked by diamonds and labelled with letters. Colouring is based on degree of LD to that SNP as represented by r^2 (legend for r^2 is in the upper left corner). A black centre indicates that the markers is in the 1000 Genomes Project but not in HapMap3. The green line in the right panel shows the MTAG analysis significance level ($1.667 \cdot 10^{-8}$). Details of the index SNPs is in the upper right corner show (marker name/association p-value/odds ratio for the minor allele/minor allele frequency/imputation quality/direction of effect in the included studies (info only available for ASD)). The green lines in the lower half show genes (based on UCSC) with black vertical lines indicating exons. Arrow heads show the direction of transcription.

4.1.5 Gene-based association

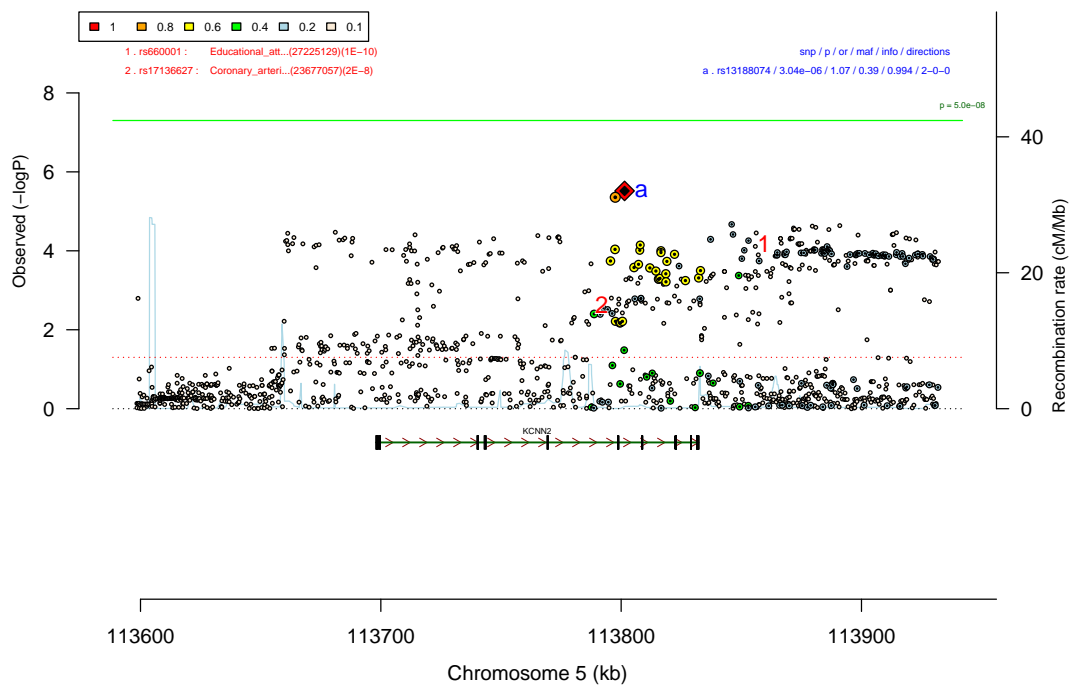


Supplementary Figure 56: Qq plot for gene-based analysis by MAGMA on the combined iPSYCH-PGC GWAS composed of 18 381 cases and 27 969 controls. Association quantiles of the p-values (achieved from z-score) are plotted against the quantiles expected under the null. The shading indicates 95%-confidence region under the null. The genomic inflation factor, lambda, is the observed median χ^2 test statistic divided by the median expected χ^2 test statistic under the null hypothesis, lambda1000 estimates what lambda would be equivalent to in a sample of 500 cases and 500 controls..

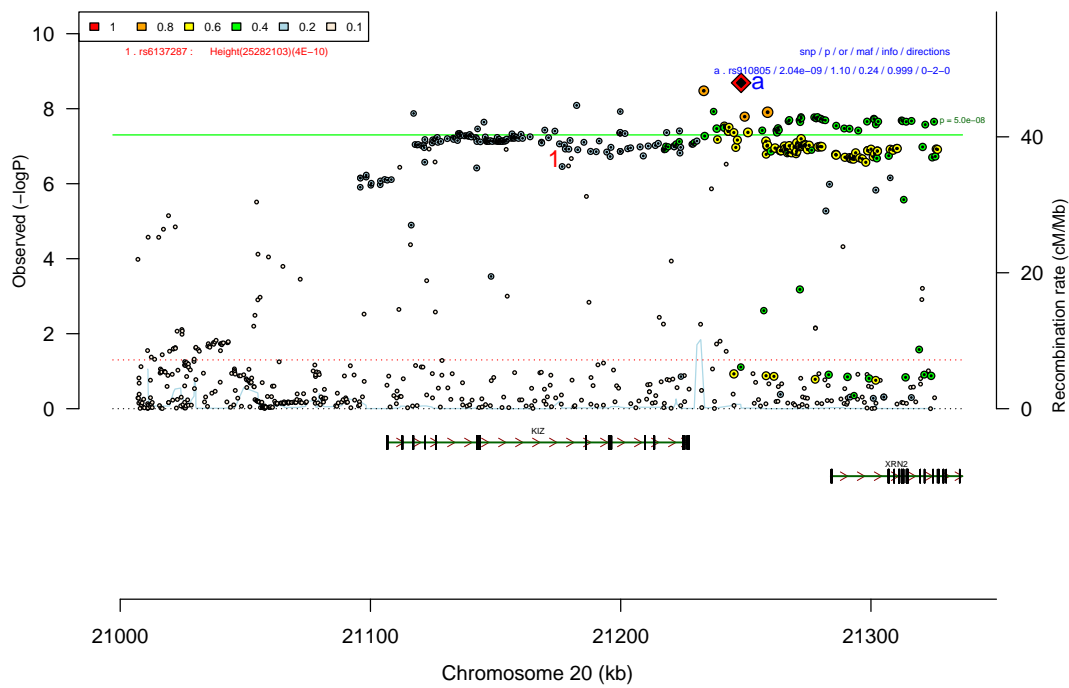
The figures [57–81](#) we show the regional Manhattan plots the individual markers around the areas of the top signals of the iPSYCH-PGC meta MAGMA analysis.



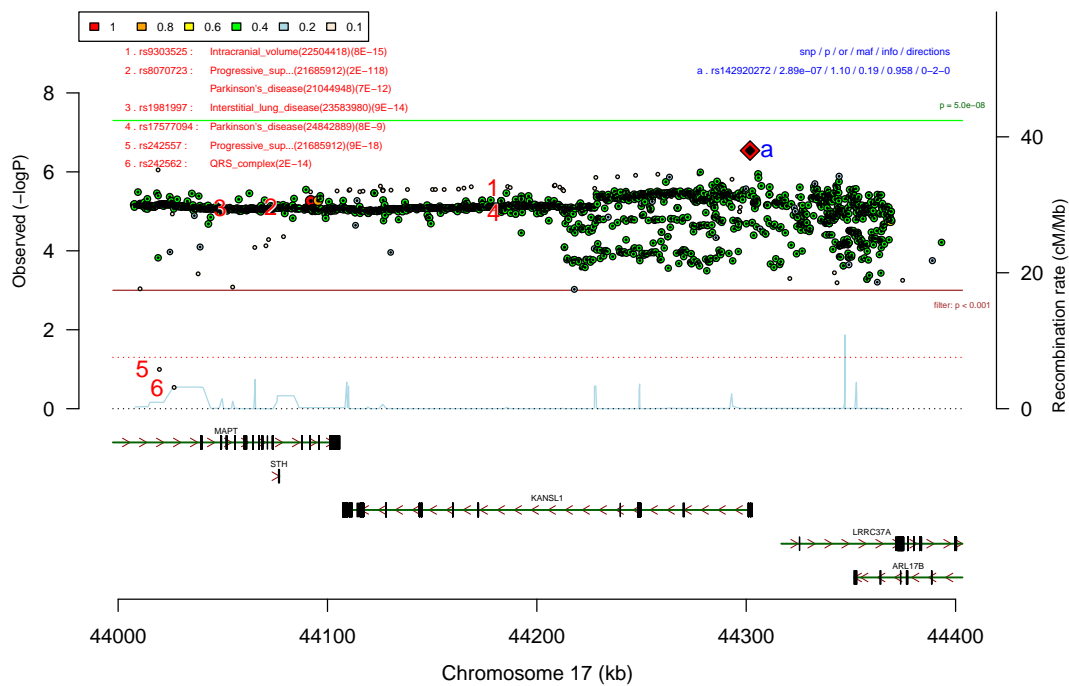
Supplementary Figure 57: Regional plot around *XRN2* for gene-based MAGMA analysis of iPSYCH-PGC meta (18 381 cases and 27 969 controls). Chromosomal position is on the horizontal axis in kb, The significance of the association expressed as $-\log_{10}(P)$ (from z-test) is plotted vertically (left axis) and the recombination rate from HapMap is shown as light blue curve (right axis). The dot size is proportional to LD between the plotted SNP and the index SNP defining the associated region. Index SNPs are marked by diamonds and labelled with letters. Colouring is based on degree of LD to that SNP as represented by r^2 (legend for r^2 is in the upper left corner). A black centre indicates that the markers is present in the 1000 Genomes Project but not in HapMap3. The green line shows the genome-wide significance level ($5 \cdot 10^{-8}$). The upper right corner show details of the index SNPs (marker name/association p-value/odds ratio for the minor allele/minor allele frequency/imputation quality/direction of effect in the two studies (N one direction - N other direction- N missing)). The green lines in the lower half show genes (based on UCSC) with black vertical lines indicating exons. Arrow heads show the direction of transcription.



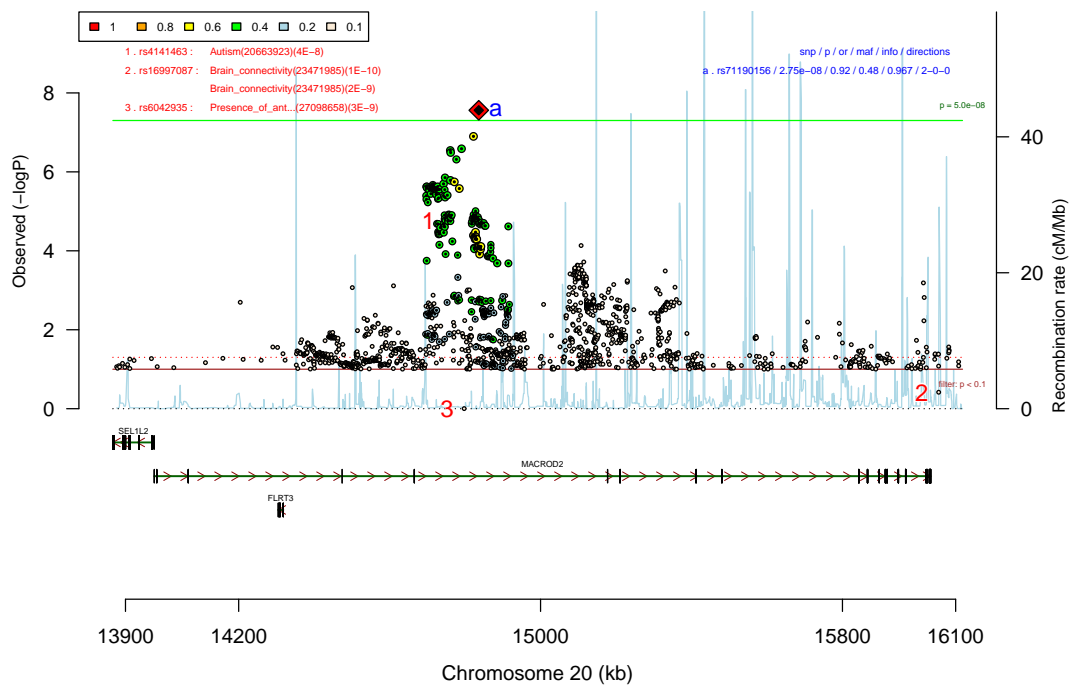
Supplementary Figure 58: Regional plot around *KCNN2* for gene-based MAGMA analysis of iPSYCH-PGC meta (18 381 cases and 27 969 controls). Chromosomal position is on the horizontal axis in kb, The significance of the association expressed as $-\log_{10}(P)$ (from z-test) is plotted vertically (left axis) and the recombination rate from HapMap is shown as light blue curve (right axis). The dot size is proportional to LD between the plotted SNP and the index SNP defining the associated region. Index SNPs are marked by diamonds and labelled with letters. Colouring is based on degree of LD to that SNP as represented by r^2 (legend for r^2 is in the upper left corner). A black centre indicates that the markers is present in the 1000 Genomes Project but not in HapMap3. The green line shows the genome-wide significance level ($5 \cdot 10^{-8}$). The upper right corner show details of the index SNPs (marker name/association p-value/odds ratio for the minor allele/minor allele frequency/imputation quality/direction of effect in the two studies (N one direction - N other direction- N missing)). Loci listed in the GWAS Catalogue for other phenotypes are shown in the upper left corner (capped at 10). The green lines in the lower half show genes (based on UCSC) with black vertical lines indicating exons. Arrow heads show the direction of transcription.



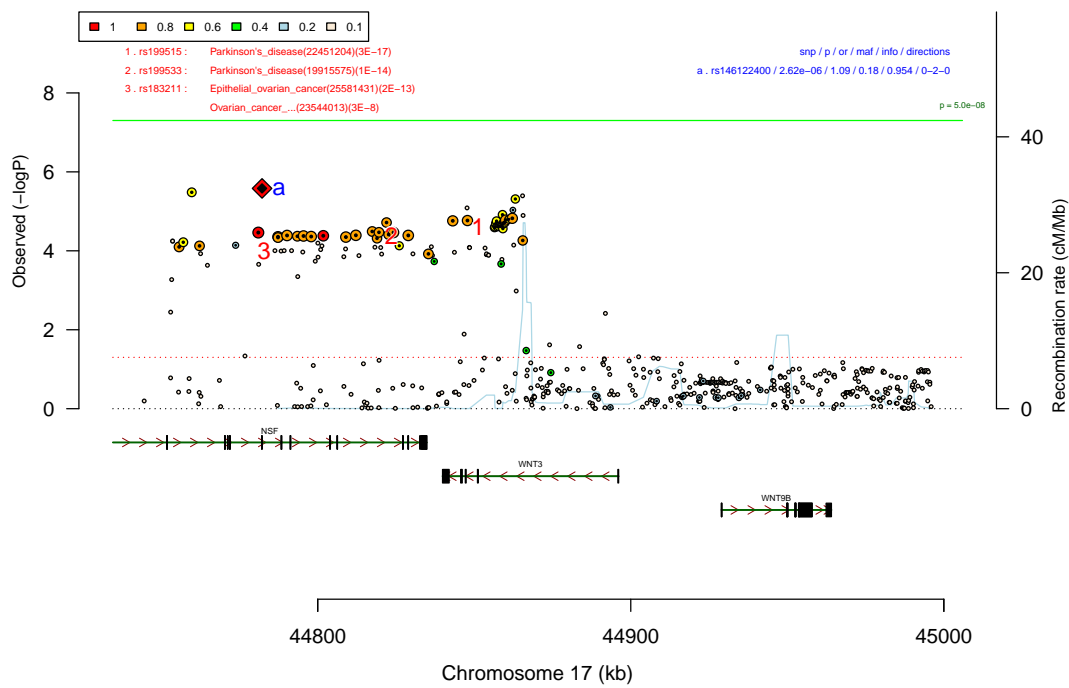
Supplementary Figure 59: Regional plot around *KIZ* for gene-based MAGMA analysis of iPSYCH-PGC meta (18 381 cases and 27 969 controls). Chromosomal position is on the horizontal axis in kb, The significance of the association expressed as $-\log_{10}(P)$ (from z-test) is plotted vertically (left axis) and the recombination rate from HapMap is shown as light blue curve (right axis). The dot size is proportional to LD between the plotted SNP and the index SNP defining the associated region. Index SNPs are marked by diamonds and labelled with letters. Colouring is based on degree of LD to that SNP as represented by r^2 (legend for r^2 is in the upper left corner). A black centre indicates that the markers is present in the 1000 Genomes Project but not in HapMap3. The green line shows the genome-wide significance level ($5 \cdot 10^{-8}$). The upper right corner show details of the index SNPs (marker name/association p-value/odds ratio for the minor allele/minor allele frequency/imputation quality/direction of effect in the two studies (N one direction - N other direction- N missing)). Loci listed in the GWAS Catalogue for other phenotypes are shown in the upper left corner (capped at 10). The green lines in the lower half show genes (based on UCSC) with black vertical lines indicating exons. Arrow heads show the direction of transcription.



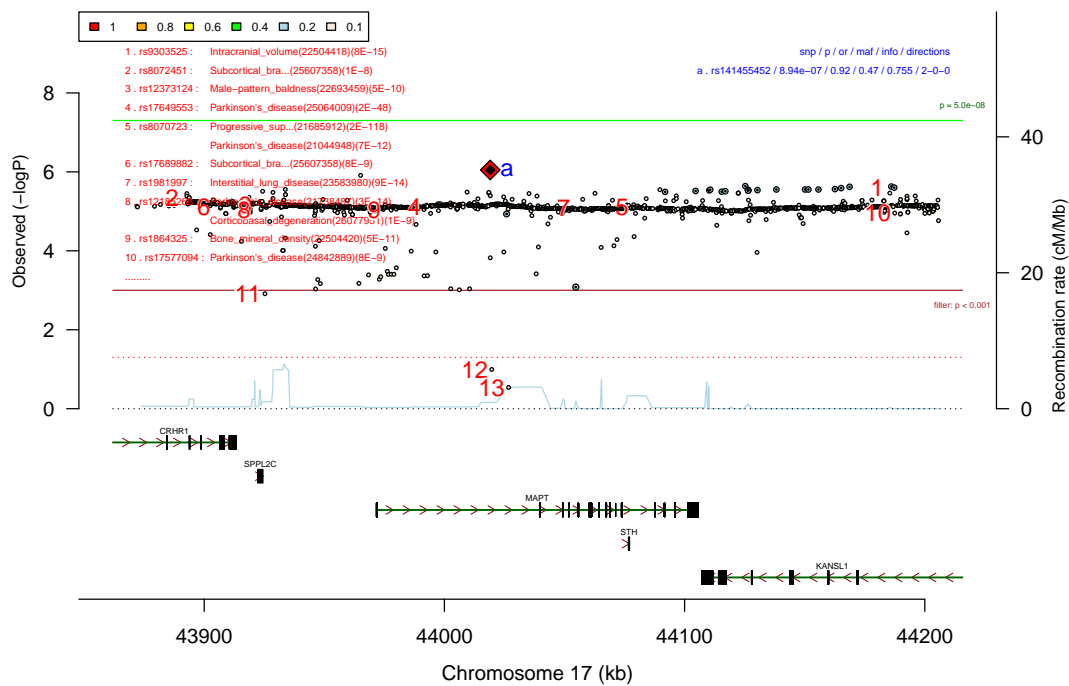
Supplementary Figure 60: Regional plot around *KANSL1* for gene-based MAGMA analysis of iPSYCH-PGC meta (18 381 cases and 27 969 controls). Chromosomal position is on the horizontal axis in kb, The significance of the association expressed as $-\log_{10}(P)$ (from z-test) is plotted vertically (left axis) and the recombination rate from HapMap is shown as light blue curve (right axis). The dot size is proportional to LD between the plotted SNP and the index SNP defining the associated region. Index SNPs are marked by diamonds and labelled with letters. Colouring is based on degree of LD to that SNP as represented by r^2 (legend for r^2 is in the upper left corner). A black centre indicates that the markers is present in the 1000 Genomes Project but not in HapMap3. The green line shows the genome-wide significance level ($5 \cdot 10^{-8}$). The upper right corner show details of the index SNPs (marker name/association p-value/odds ratio for the minor allele/minor allele frequency/imputation quality/direction of effect in the two studies (N one direction - N other direction- N missing)). Loci listed in the GWAS Catalogue for other phenotypes are shown in the upper left corner (capped at 10). The green lines in the lower half show genes (based on UCSC) with black vertical lines indicating exons. Arrow heads show the direction of transcription.



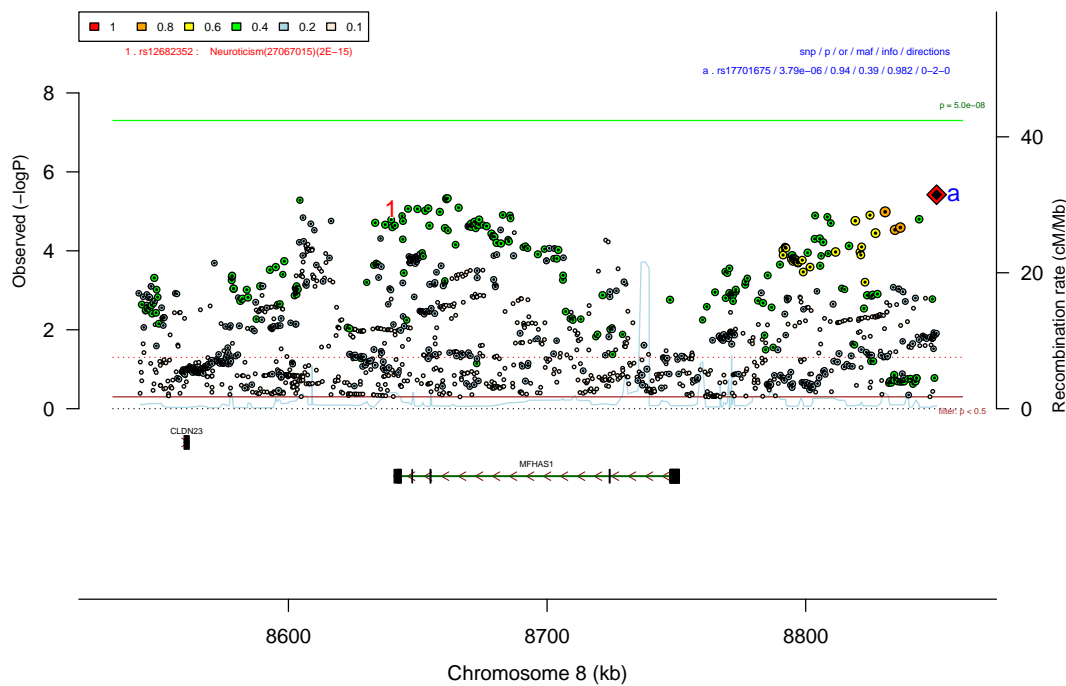
Supplementary Figure 61: Regional plot around *MACROD2* for gene-based MAGMA analysis of iPSYCH-PGC meta (18381 cases and 27969 controls). Chromosomal position is on the horizontal axis in kb, The significance of the association expressed as $-\log_{10}(P)$ (from z-test) is plotted vertically (left axis) and the recombination rate from HapMap is shown as light blue curve (right axis). The dot size is proportional to LD between the plotted SNP and the index SNP defining the associated region. Index SNPs are marked by diamonds and labelled with letters. Colouring is based on degree of LD to that SNP as represented by r^2 (legend for r^2 is in the upper left corner). A black centre indicates that the markers is present in the 1000 Genomes Project but not in HapMap3. The green line shows the genome-wide significance level ($5 \cdot 10^{-8}$). The upper right corner show details of the index SNPs (marker name/association p-value/odds ratio for the minor allele/minor allele frequency/imputation quality/direction of effect in the two studies (N one direction - N other direction- N missing)). Loci listed in the GWAS Catalogue for other phenotypes are shown in the upper left corner (capped at 10). The green lines in the lower half show genes (based on UCSC) with black vertical lines indicating exons. Arrow heads show the direction of transcription.



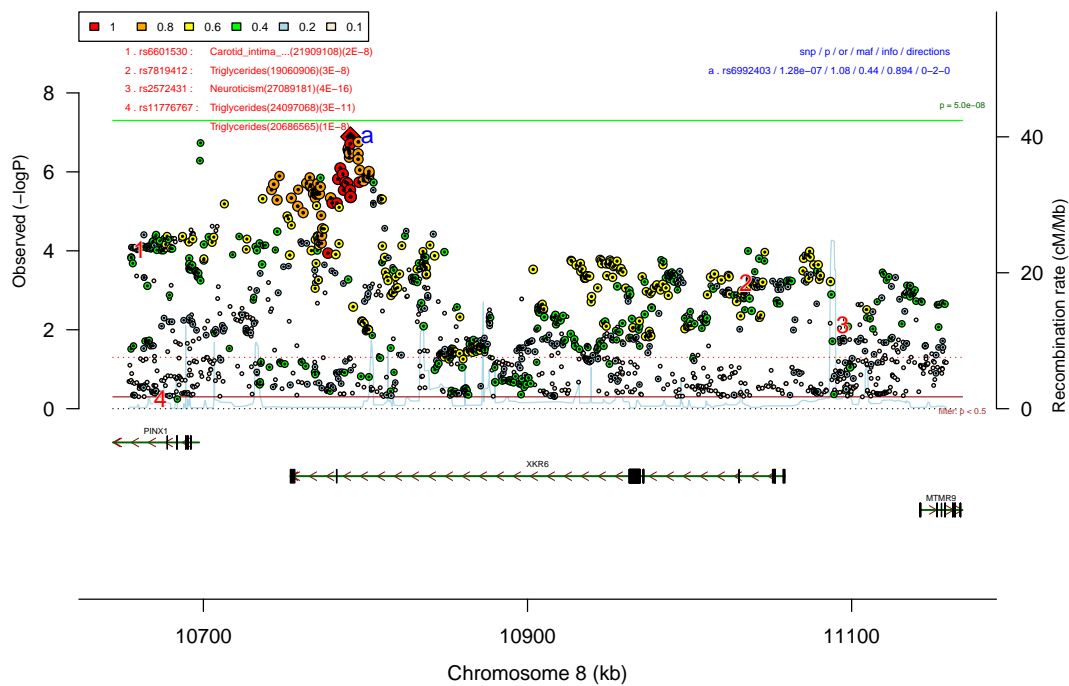
Supplementary Figure 62: Regional plot around *WNT3* for gene-based MAGMA analysis of iPSYCH-PGC meta (18 381 cases and 27 969 controls). Chromosomal position is on the horizontal axis in kb, The significance of the association expressed as $-\log_{10}(P)$ (from z-test) is plotted vertically (left axis) and the recombination rate from HapMap is shown as light blue curve (right axis). The dot size is proportional to LD between the plotted SNP and the index SNP defining the associated region. Index SNPs are marked by diamonds and labelled with letters. Colouring is based on degree of LD to that SNP as represented by r^2 (legend for r^2 is in the upper left corner). A black centre indicates that the markers is present in the 1000 Genomes Project but not in HapMap3. The green line shows the genome-wide significance level ($5 \cdot 10^{-8}$). The upper right corner show details of the index SNPs (marker name/association p-value/odds ratio for the minor allele/minor allele frequency/imputation quality/direction of effect in the two studies (N one direction - N other direction- N missing)). Loci listed in the GWAS Catalogue for other phenotypes are shown in the upper left corner (capped at 10). The green lines in the lower half show genes (based on UCSC) with black vertical lines indicating exons. Arrow heads show the direction of transcription.



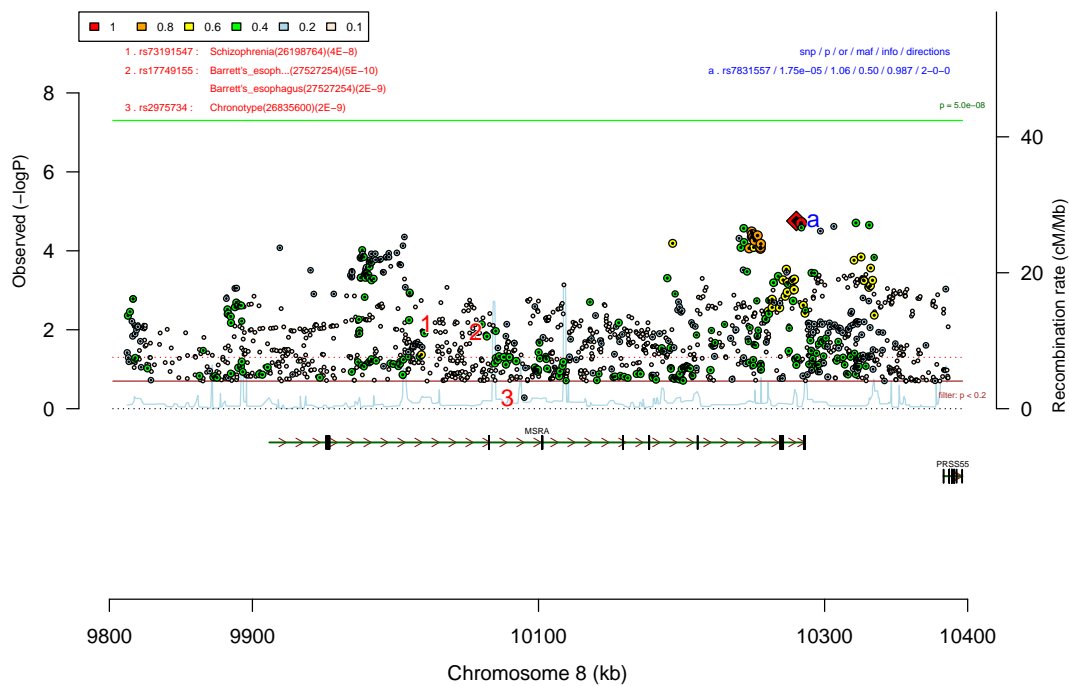
Supplementary Figure 63: Regional plot around *MAPT* for gene-based MAGMA analysis of iPSYCH-PGC meta (18 381 cases and 27 969 controls). Chromosomal position is on the horizontal axis in kb, The significance of the association expressed as $-\log_{10}(P)$ (from z-test) is plotted vertically (left axis) and the recombination rate from HapMap is shown as light blue curve (right axis). The dot size is proportional to LD between the plotted SNP and the index SNP defining the associated region. Index SNPs are marked by diamonds and labelled with letters. Colouring is based on degree of LD to that SNP as represented by r^2 (legend for r^2 is in the upper left corner). A black centre indicates that the markers is present in the 1000 Genomes Project but not in HapMap3. The green line shows the genome-wide significance level ($5 \cdot 10^{-8}$). The upper right corner show details of the index SNPs (marker name/association p-value/odds ratio for the minor allele/minor allele frequency/imputation quality/direction of effect in the two studies (N one direction - N other direction- N missing)). Loci listed in the GWAS Catalogue for other phenotypes are shown in the upper left corner (capped at 10). The green lines in the lower half show genes (based on UCSC) with black vertical lines indicating exons. Arrow heads show the direction of transcription.



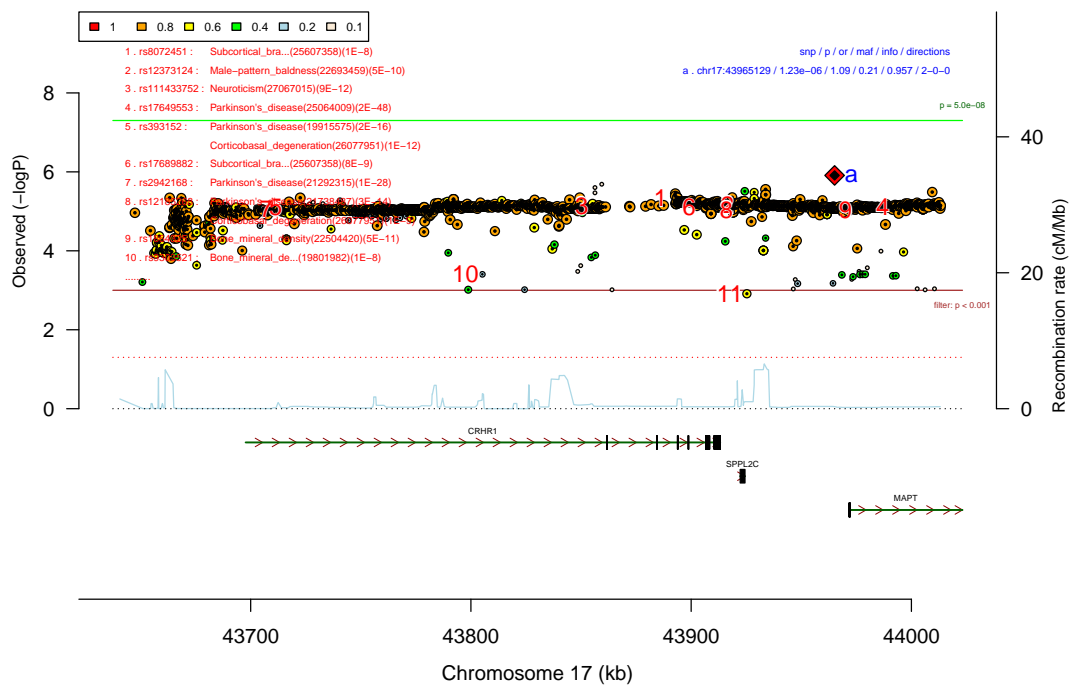
Supplementary Figure 64: Regional plot around *MFHAS1* for gene-based MAGMA analysis of iPSYCH-PGC meta (18 381 cases and 27 969 controls). Chromosomal position is on the horizontal axis in kb, The significance of the association expressed as $-\log_{10}(P)$ (from z-test) is plotted vertically (left axis) and the recombination rate from HapMap is shown as light blue curve (right axis). The dot size is proportional to LD between the plotted SNP and the index SNP defining the associated region. Index SNPs are marked by diamonds and labelled with letters. Colouring is based on degree of LD to that SNP as represented by r^2 (legend for r^2 is in the upper left corner). A black centre indicates that the markers is present in the 1000 Genomes Project but not in HapMap3. The green line shows the genome-wide significance level ($5 \cdot 10^{-8}$). The upper right corner show details of the index SNPs (marker name/association p-value/odds ratio for the minor allele/minor allele frequency/imputation quality/direction of effect in the two studies (N one direction - N other direction- N missing)). Loci listed in the GWAS Catalogue for other phenotypes are shown in the upper left corner (capped at 10). The green lines in the lower half show genes (based on UCSC) with black vertical lines indicating exons. Arrow heads show the direction of transcription.



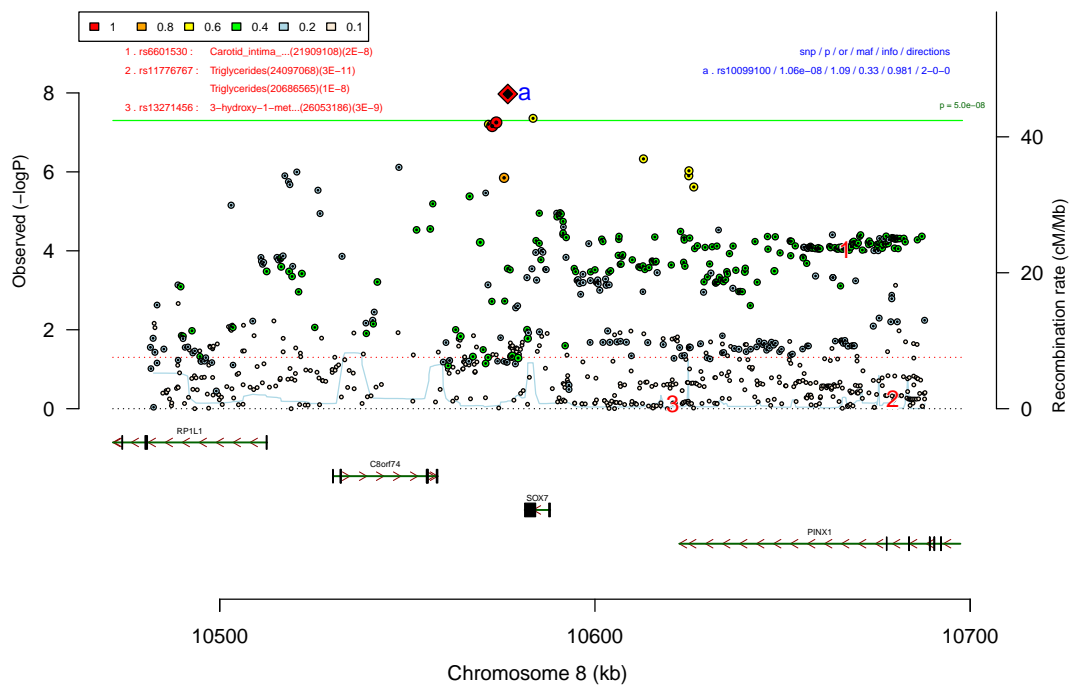
Supplementary Figure 65: Regional plot around *XKR6* for gene-based MAGMA analysis of iPSYCH-PGC meta (18 381 cases and 27 969 controls). Chromosomal position is on the horizontal axis in kb, The significance of the association expressed as $-\log_{10}(P)$ (from z-test) is plotted vertically (left axis) and the recombination rate from HapMap is shown as light blue curve (right axis). The dot size is proportional to LD between the plotted SNP and the index SNP defining the associated region. Index SNPs are marked by diamonds and labelled with letters. Colouring is based on degree of LD to that SNP as represented by r^2 (legend for r^2 is in the upper left corner). A black centre indicates that the markers is present in the 1000 Genomes Project but not in HapMap3. The green line shows the genome-wide significance level ($5 \cdot 10^{-8}$). The upper right corner show details of the index SNPs (marker name/association p-value/odds ratio for the minor allele/minor allele frequency/imputation quality/direction of effect in the two studies (N one direction - N other direction- N missing)). Loci listed in the GWAS Catalogue for other phenotypes are shown in the upper left corner (capped at 10). The green lines in the lower half show genes (based on UCSC) with black vertical lines indicating exons. Arrow heads show the direction of transcription.



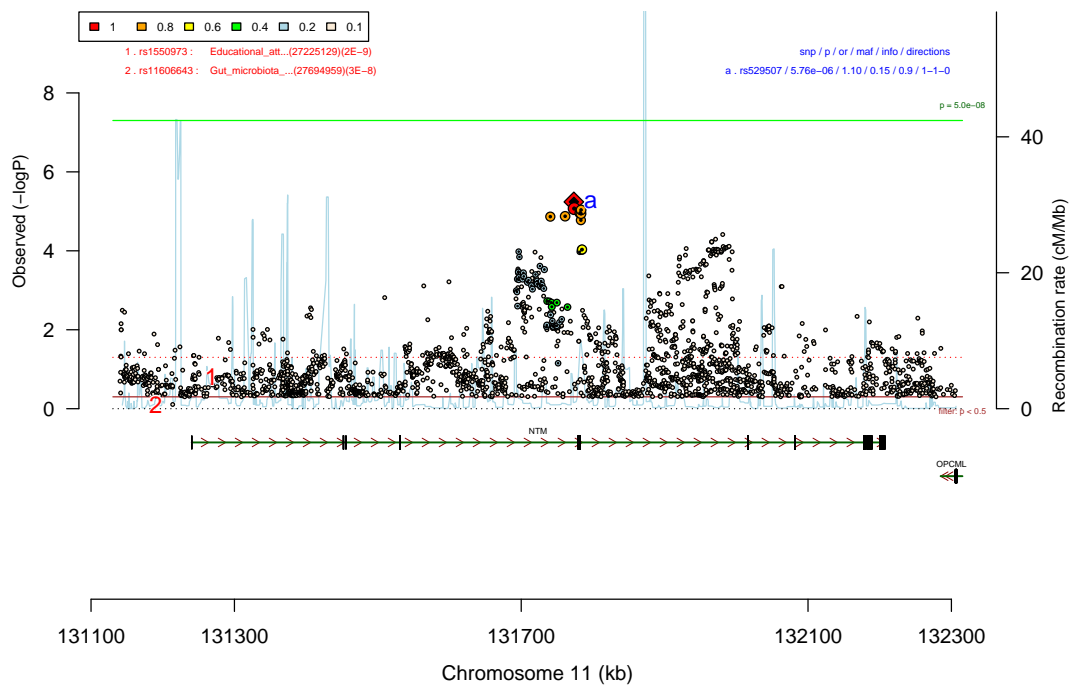
Supplementary Figure 66: Regional plot around *MSRA* for gene-based MAGMA analysis of iPSYCH-PGC meta (18 381 cases and 27 969 controls). Chromosomal position is on the horizontal axis in kb, The significance of the association expressed as $-\log_{10}(P)$ (from z-test) is plotted vertically (left axis) and the recombination rate from HapMap is shown as light blue curve (right axis). The dot size is proportional to LD between the plotted SNP and the index SNP defining the associated region. Index SNPs are marked by diamonds and labelled with letters. Colouring is based on degree of LD to that SNP as represented by r^2 (legend for r^2 is in the upper left corner). A black centre indicates that the markers is present in the 1000 Genomes Project but not in HapMap3. The green line shows the genome-wide significance level ($5 \cdot 10^{-8}$). The upper right corner show details of the index SNPs (marker name/association p-value/odds ratio for the minor allele/minor allele frequency/imputation quality/direction of effect in the two studies (N one direction - N other direction- N missing)). Loci listed in the GWAS Catalogue for other phenotypes are shown in the upper left corner (capped at 10). The green lines in the lower half show genes (based on UCSC) with black vertical lines indicating exons. Arrow heads show the direction of transcription.



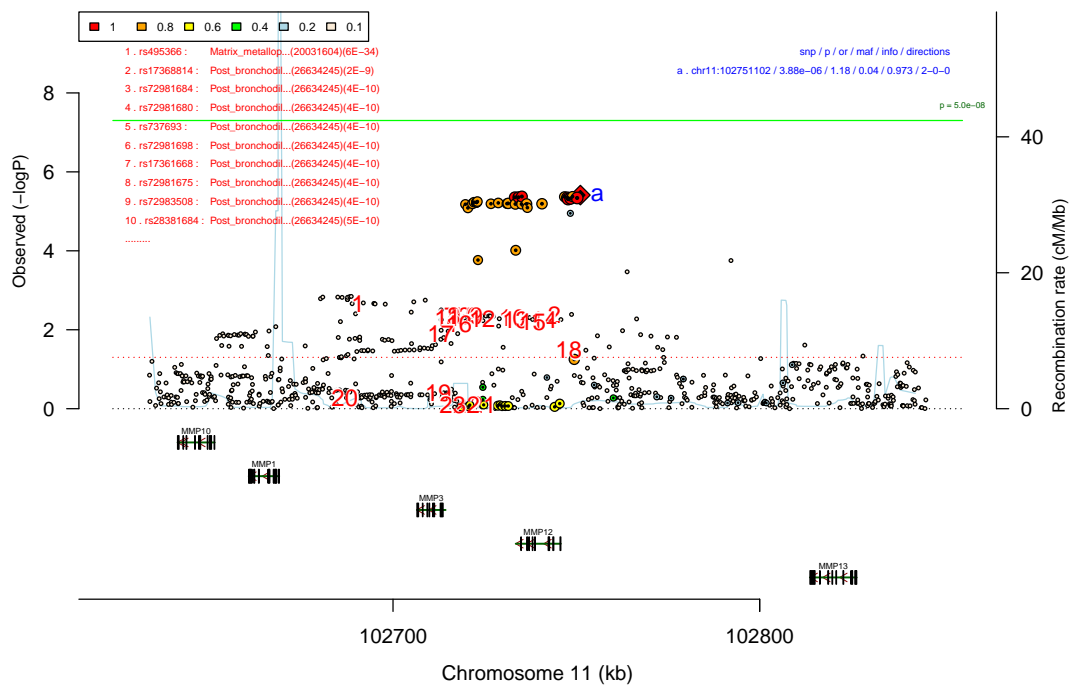
Supplementary Figure 67: Regional plot around *CRHR1* for gene-based MAGMA analysis of iPSYCH-PGC meta (18 381 cases and 27 969 controls). Chromosomal position is on the horizontal axis in kb, The significance of the association expressed as $-\log_{10}(P)$ (from z-test) is plotted vertically (left axis) and the recombination rate from HapMap is shown as light blue curve (right axis). The dot size is proportional to LD between the plotted SNP and the index SNP defining the associated region. Index SNPs are marked by diamonds and labelled with letters. Colouring is based on degree of LD to that SNP as represented by r^2 (legend for r^2 is in the upper left corner). A black centre indicates that the markers is present in the 1000 Genomes Project but not in HapMap3. The green line shows the genome-wide significance level ($5 \cdot 10^{-8}$). The upper right corner show details of the index SNPs (marker name/association p-value/odds ratio for the minor allele/minor allele frequency/imputation quality/direction of effect in the two studies (N one direction - N other direction- N missing)). Loci listed in the GWAS Catalogue for other phenotypes are shown in the upper left corner (capped at 10). The green lines in the lower half show genes (based on UCSC) with black vertical lines indicating exons. Arrow heads show the direction of transcription.



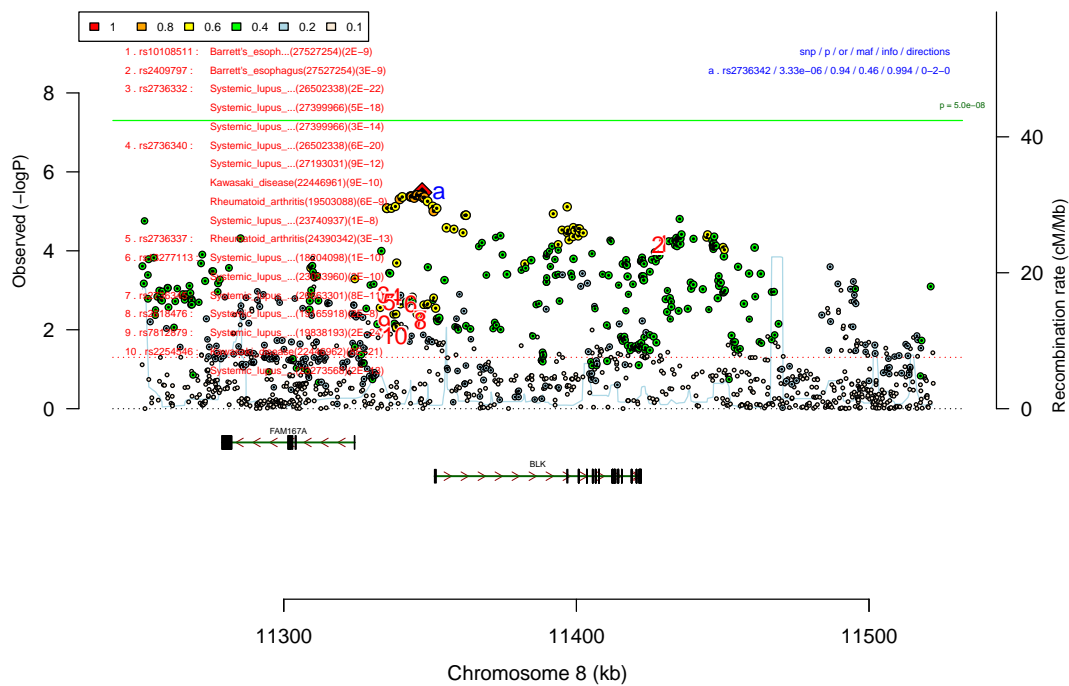
Supplementary Figure 68: Regional plot around *SOX7* for gene-based MAGMA analysis of iPSYCH-PGC meta (18 381 cases and 27 969 controls). Chromosomal position is on the horizontal axis in kb, The significance of the association expressed as $-\log_{10}(P)$ (from z-test) is plotted vertically (left axis) and the recombination rate from HapMap is shown as light blue curve (right axis). The dot size is proportional to LD between the plotted SNP and the index SNP defining the associated region. Index SNPs are marked by diamonds and labelled with letters. Colouring is based on degree of LD to that SNP as represented by r^2 (legend for r^2 is in the upper left corner). A black centre indicates that the markers is present in the 1000 Genomes Project but not in HapMap3. The green line shows the genome-wide significance level ($5 \cdot 10^{-8}$). The upper right corner show details of the index SNPs (marker name/association p-value/odds ratio for the minor allele/minor allele frequency/imputation quality/direction of effect in the two studies (N one direction - N other direction- N missing)). Loci listed in the GWAS Catalogue for other phenotypes are shown in the upper left corner (capped at 10). The green lines in the lower half show genes (based on UCSC) with black vertical lines indicating exons. Arrow heads show the direction of transcription.



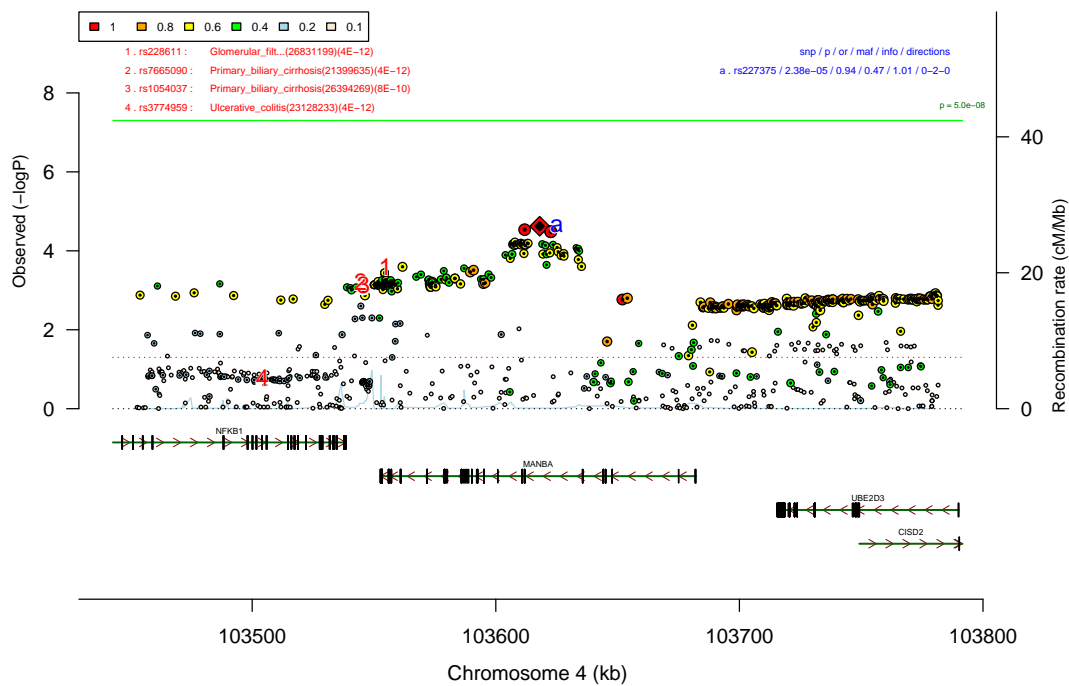
Supplementary Figure 69: Regional plot around *NTM* for gene-based MAGMA analysis of iPSYCH-PGC meta (18 381 cases and 27 969 controls). Chromosomal position is on the horizontal axis in kb, The significance of the association expressed as $-\log_{10}(P)$ (from z-test) is plotted vertically (left axis) and the recombination rate from HapMap is shown as light blue curve (right axis). The dot size is proportional to LD between the plotted SNP and the index SNP defining the associated region. Index SNPs are marked by diamonds and labelled with letters. Colouring is based on degree of LD to that SNP as represented by r^2 (legend for r^2 is in the upper left corner). A black centre indicates that the markers is present in the 1000 Genomes Project but not in HapMap3. The green line shows the genome-wide significance level ($5 \cdot 10^{-8}$). The upper right corner show details of the index SNPs (marker name/association p-value/odds ratio for the minor allele/minor allele frequency/imputation quality/direction of effect in the two studies (N one direction - N other direction- N missing)). Loci listed in the GWAS Catalogue for other phenotypes are shown in the upper left corner (capped at 10). The green lines in the lower half show genes (based on UCSC) with black vertical lines indicating exons. Arrow heads show the direction of transcription.



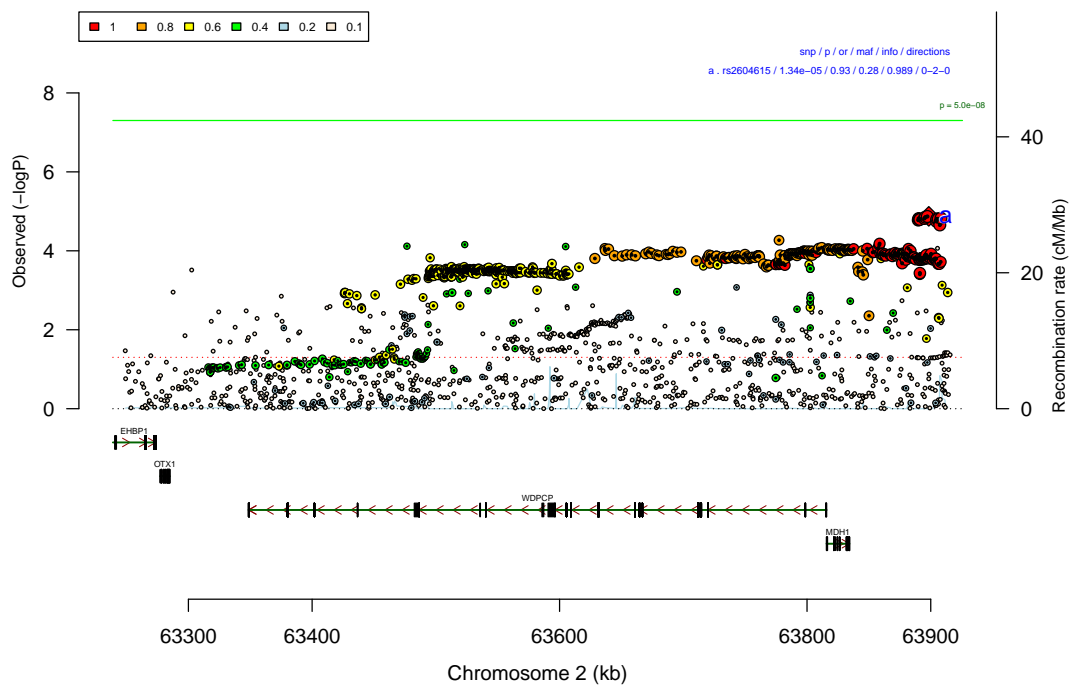
Supplementary Figure 70: Regional plot around *MMP12* for gene-based MAGMA analysis of iPSYCH-PGC meta (18 381 cases and 27 969 controls). Chromosomal position is on the horizontal axis in kb, The significance of the association expressed as $-\log_{10}(P)$ (from z-test) is plotted vertically (left axis) and the recombination rate from HapMap is shown as light blue curve (right axis). The dot size is proportional to LD between the plotted SNP and the index SNP defining the associated region. Index SNPs are marked by diamonds and labelled with letters. Colouring is based on degree of LD to that SNP as represented by r^2 (legend for r^2 is in the upper left corner). A black centre indicates that the markers is present in the 1000 Genomes Project but not in HapMap3. The green line shows the genome-wide significance level ($5 \cdot 10^{-8}$). The upper right corner show details of the index SNPs (marker name/association p-value/odds ratio for the minor allele/minor allele frequency/imputation quality/direction of effect in the two studies (N one direction - N other direction- N missing)). Loci listed in the GWAS Catalogue for other phenotypes are shown in the upper left corner (capped at 10). The green lines in the lower half show genes (based on UCSC) with black vertical lines indicating exons. Arrow heads show the direction of transcription.



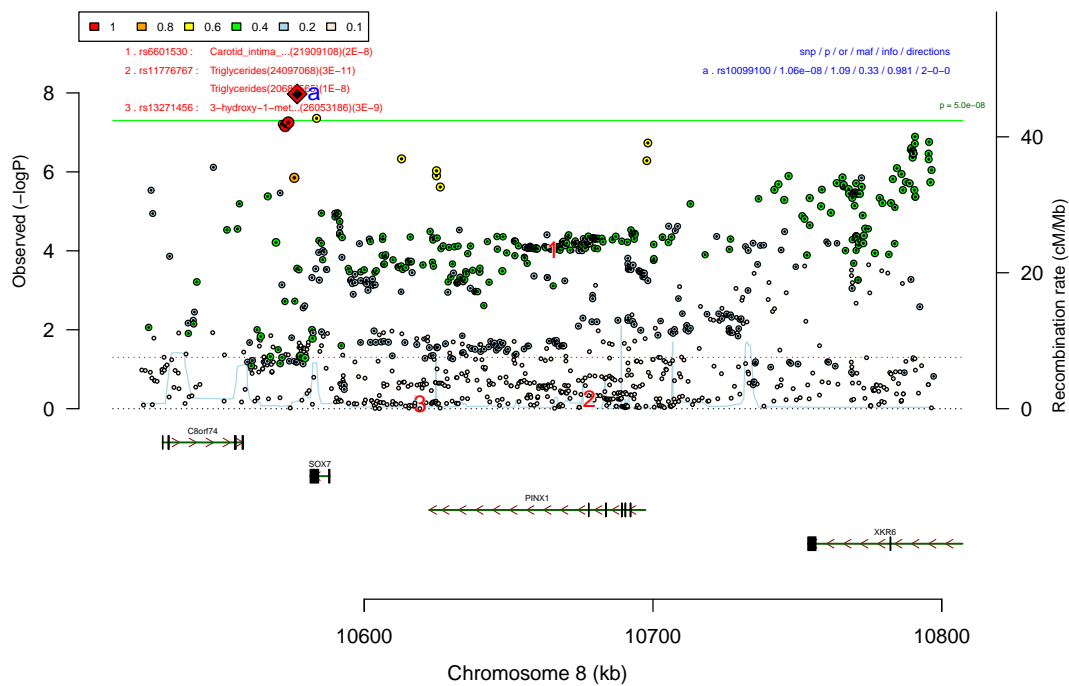
Supplementary Figure 71: Regional plot around *BLK* for gene-based MAGMA analysis of iPSYCH-PGC meta (18 381 cases and 27 969 controls). Chromosomal position is on the horizontal axis in kb, The significance of the association expressed as $-\log_{10}(P)$ (from z-test) is plotted vertically (left axis) and the recombination rate from HapMap is shown as light blue curve (right axis). The dot size is proportional to LD between the plotted SNP and the index SNP defining the associated region. Index SNPs are marked by diamonds and labelled with letters. Colouring is based on degree of LD to that SNP as represented by r^2 (legend for r^2 is in the upper left corner). A black centre indicates that the markers is present in the 1000 Genomes Project but not in HapMap3. The green line shows the genome-wide significance level ($5 \cdot 10^{-8}$). The upper right corner show details of the index SNPs (marker name/association p-value/odds ratio for the minor allele/minor allele frequency/imputation quality/direction of effect in the two studies (N one direction - N other direction- N missing)). Loci listed in the GWAS Catalogue for other phenotypes are shown in the upper left corner (capped at 10). The green lines in the lower half show genes (based on UCSC) with black vertical lines indicating exons. Arrow heads show the direction of transcription.



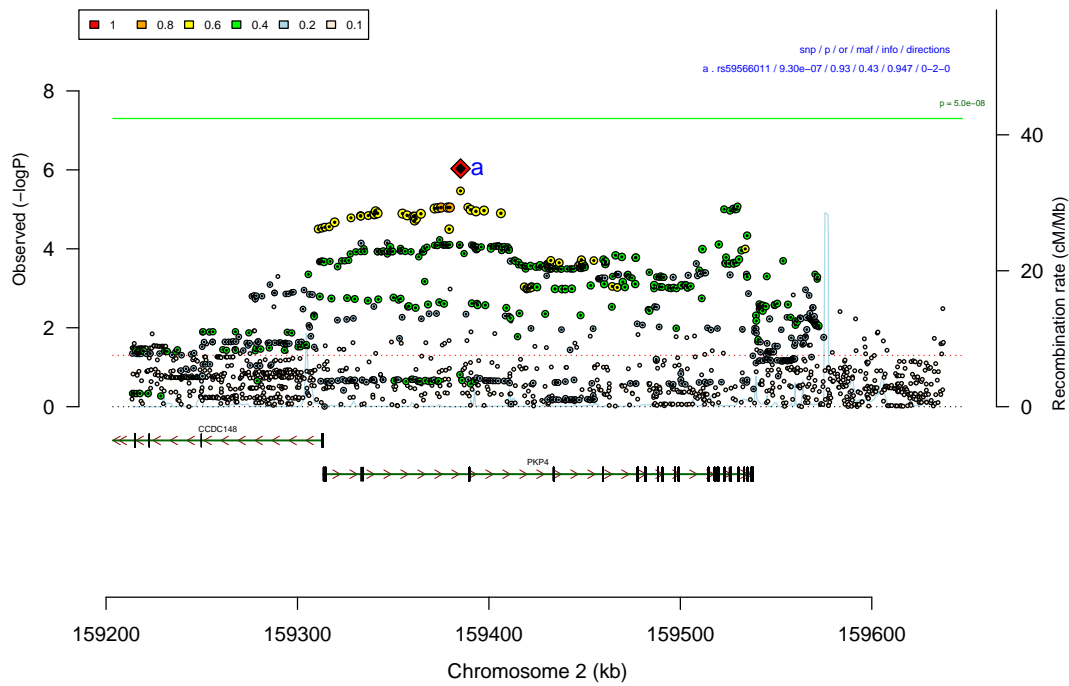
Supplementary Figure 72: Regional plot around *MANBA* for gene-based MAGMA analysis of iPSYCH-PGC meta (18 381 cases and 27 969 controls). Chromosomal position is on the horizontal axis in kb, The significance of the association expressed as $-\log_{10}(P)$ (from z-test) is plotted vertically (left axis) and the recombination rate from HapMap is shown as light blue curve (right axis). The dot size is proportional to LD between the plotted SNP and the index SNP defining the associated region. Index SNPs are marked by diamonds and labelled with letters. Colouring is based on degree of LD to that SNP as represented by r^2 (legend for r^2 is in the upper left corner). A black centre indicates that the markers is present in the 1000 Genomes Project but not in HapMap3. The green line shows the genome-wide significance level ($5 \cdot 10^{-8}$). The upper right corner show details of the index SNPs (marker name/association p-value/odds ratio for the minor allele/minor allele frequency/imputation quality/direction of effect in the two studies (N one direction - N other direction- N missing)). Loci listed in the GWAS Catalogue for other phenotypes are shown in the upper left corner (capped at 10). The green lines in the lower half show genes (based on UCSC) with black vertical lines indicating exons. Arrow heads show the direction of transcription.



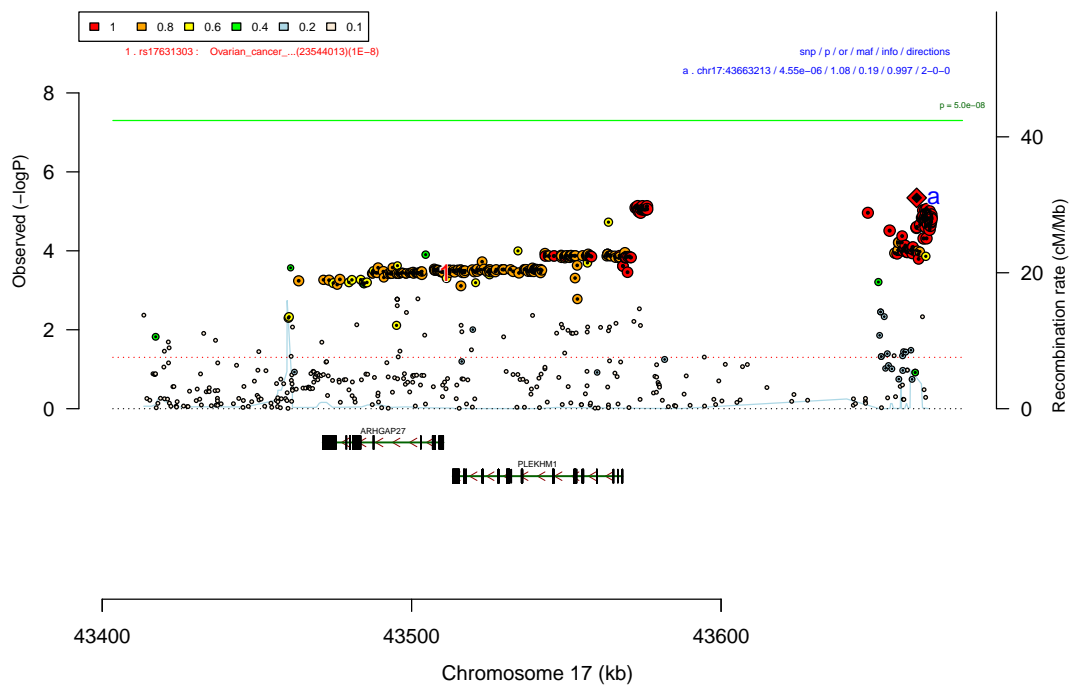
Supplementary Figure 74: Regional plot around *WDPCP* for gene-based MAGMA analysis of iPSYCH-PGC meta (18 381 cases and 27 969 controls). Chromosomal position is on the horizontal axis in kb, The significance of the association expressed as $-\log_{10}(P)$ (from z-test) is plotted vertically (left axis) and the recombination rate from HapMap is shown as light blue curve (right axis). The dot size is proportional to LD between the plotted SNP and the index SNP defining the associated region. Index SNPs are marked by diamonds and labelled with letters. Colouring is based on degree of LD to that SNP as represented by r^2 (legend for r^2 is in the upper left corner). A black centre indicates that the markers is present in the 1000 Genomes Project but not in HapMap3. The green line shows the genome-wide significance level ($5 \cdot 10^{-8}$). The upper right corner show details of the index SNPs (marker name/association p-value/odds ratio for the minor allele/minor allele frequency/imputation quality/direction of effect in the two studies (N one direction - N other direction- N missing)). The green lines in the lower half show genes (based on UCSC) with black vertical lines indicating exons. Arrow heads show the direction of transcription.



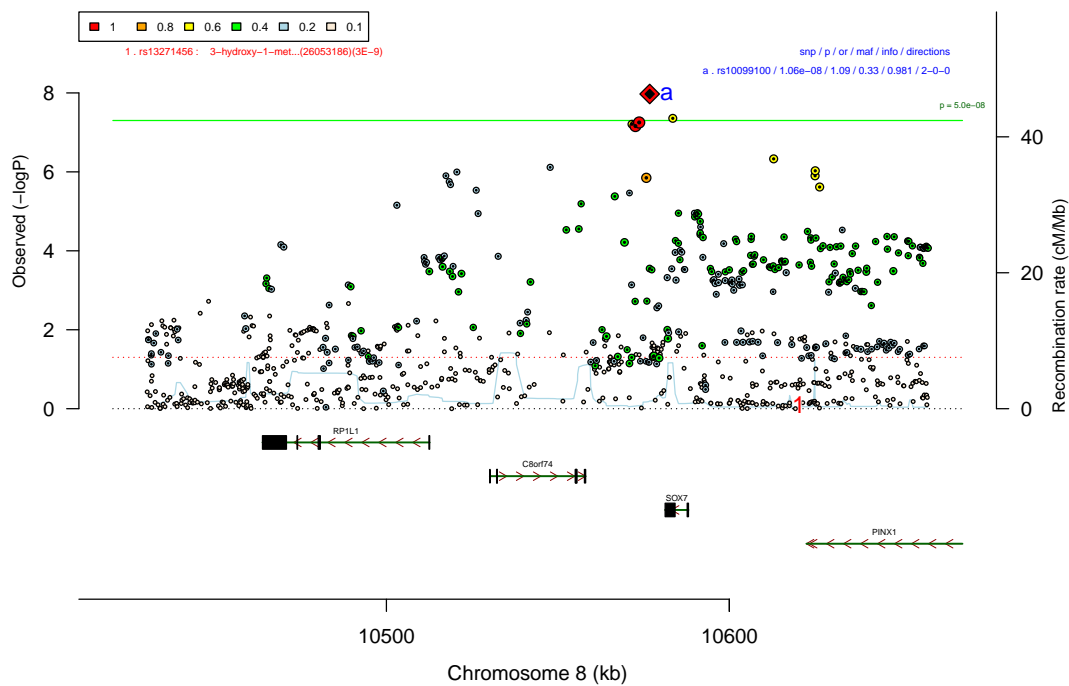
Supplementary Figure 75: Regional plot around *PINX1* for gene-based MAGMA analysis of iPSYCH-PGC meta (18 381 cases and 27 969 controls). Chromosomal position is on the horizontal axis in kb, The significance of the association expressed as $-\log_{10}(P)$ (from z-test) is plotted vertically (left axis) and the recombination rate from HapMap is shown as light blue curve (right axis). The dot size is proportional to LD between the plotted SNP and the index SNP defining the associated region. Index SNPs are marked by diamonds and labelled with letters. Colouring is based on degree of LD to that SNP as represented by r^2 (legend for r^2 is in the upper left corner). A black centre indicates that the markers is present in the 1000 Genomes Project but not in HapMap3. The green line shows the genome-wide significance level ($5 \cdot 10^{-8}$). The upper right corner show details of the index SNPs (marker name/association p-value/odds ratio for the minor allele/minor allele frequency/imputation quality/direction of effect in the two studies (N one direction - N other direction- N missing)). Loci listed in the GWAS Catalogue for other phenotypes are shown in the upper left corner (capped at 10). The green lines in the lower half show genes (based on UCSC) with black vertical lines indicating exons. Arrow heads show the direction of transcription.



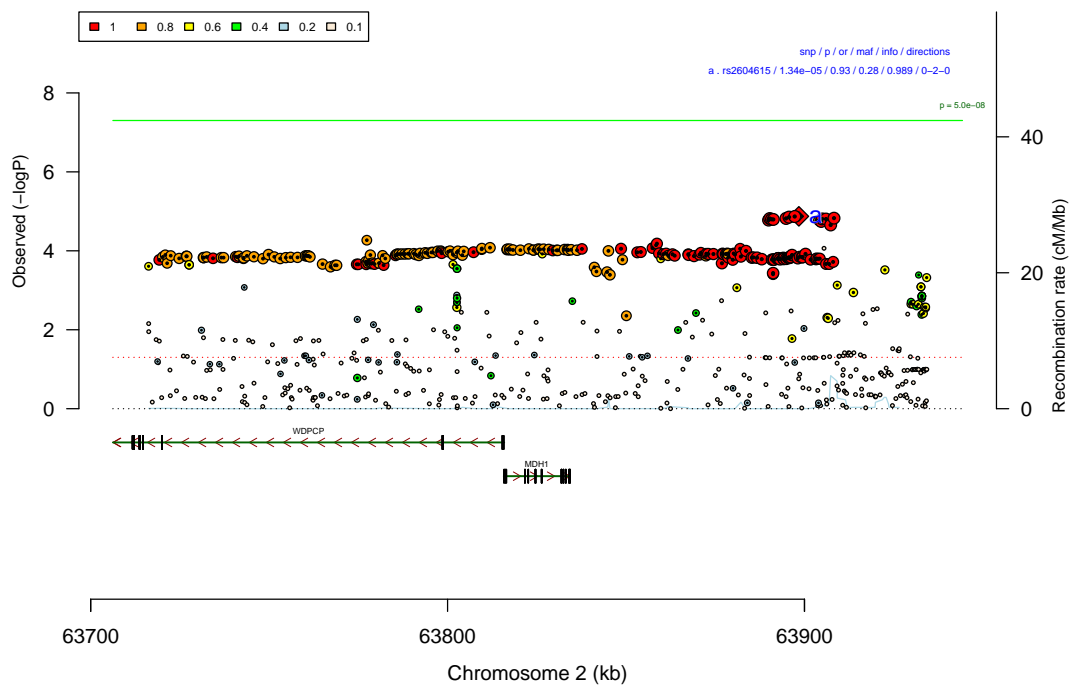
Supplementary Figure 76: Regional plot around *PKP4* for gene-based MAGMA analysis of iPSYCH-PGC meta (18 381 cases and 27 969 controls). Chromosomal position is on the horizontal axis in kb, The significance of the association expressed as $-\log_{10}(P)$ (from z-test) is plotted vertically (left axis) and the recombination rate from HapMap is shown as light blue curve (right axis). The dot size is proportional to LD between the plotted SNP and the index SNP defining the associated region. Index SNPs are marked by diamonds and labelled with letters. Colouring is based on degree of LD to that SNP as represented by r^2 (legend for r^2 is in the upper left corner). A black centre indicates that the markers is present in the 1000 Genomes Project but not in HapMap3. The green line shows the genome-wide significance level ($5 \cdot 10^{-8}$). The upper right corner show details of the index SNPs (marker name/association p-value/odds ratio for the minor allele/minor allele frequency/imputation quality/direction of effect in the two studies (N one direction - N other direction- N missing)). The green lines in the lower half show genes (based on UCSC) with black vertical lines indicating exons. Arrow heads show the direction of transcription.



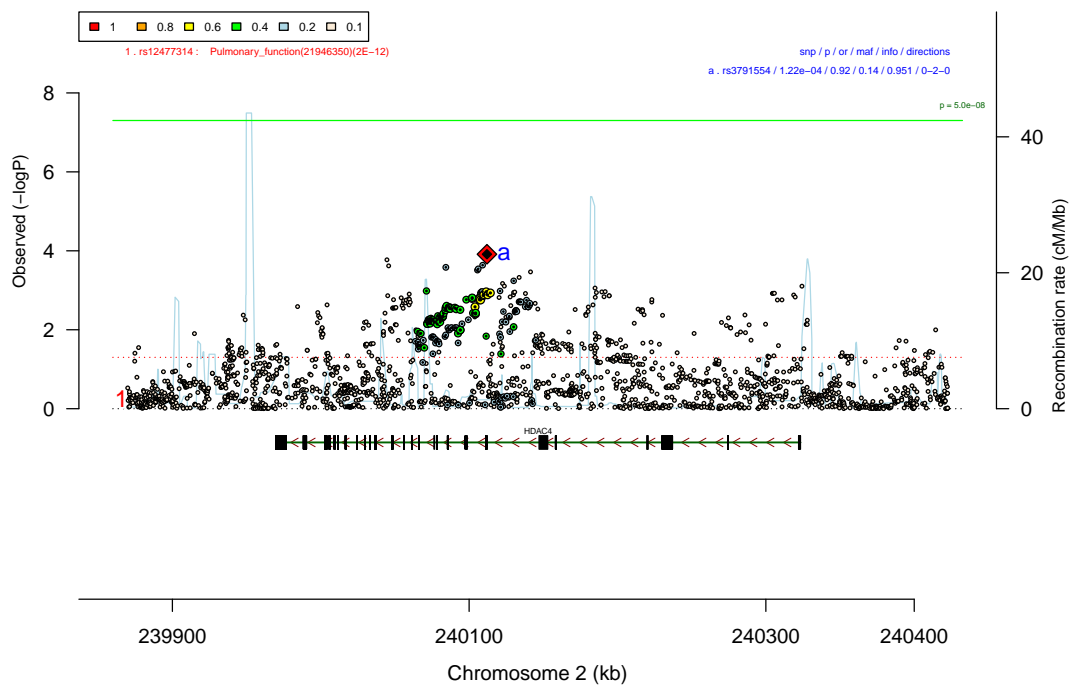
Supplementary Figure 77: Regional plot around *PLEKHM1* for gene-based MAGMA analysis of iPSYCH-PGC meta (18381 cases and 27969 controls). Chromosomal position is on the horizontal axis in kb, The significance of the association expressed as $-\log_{10}(P)$ (from z-test) is plotted vertically (left axis) and the recombination rate from HapMap is shown as light blue curve (right axis). The dot size is proportional to LD between the plotted SNP and the index SNP defining the associated region. Index SNPs are marked by diamonds and labelled with letters. Colouring is based on degree of LD to that SNP as represented by r^2 (legend for r^2 is in the upper left corner). A black centre indicates that the markers is present in the 1000 Genomes Project but not in HapMap3. The green line shows the genome-wide significance level ($5 \cdot 10^{-8}$). The upper right corner show details of the index SNPs (marker name/association p-value/odds ratio for the minor allele/minor allele frequency/imputation quality/direction of effect in the two studies (N one direction - N other direction- N missing)). Loci listed in the GWAS Catalogue for other phenotypes are shown in the upper left corner (capped at 10). The green lines in the lower half show genes (based on UCSC) with black vertical lines indicating exons. Arrow heads show the direction of transcription.



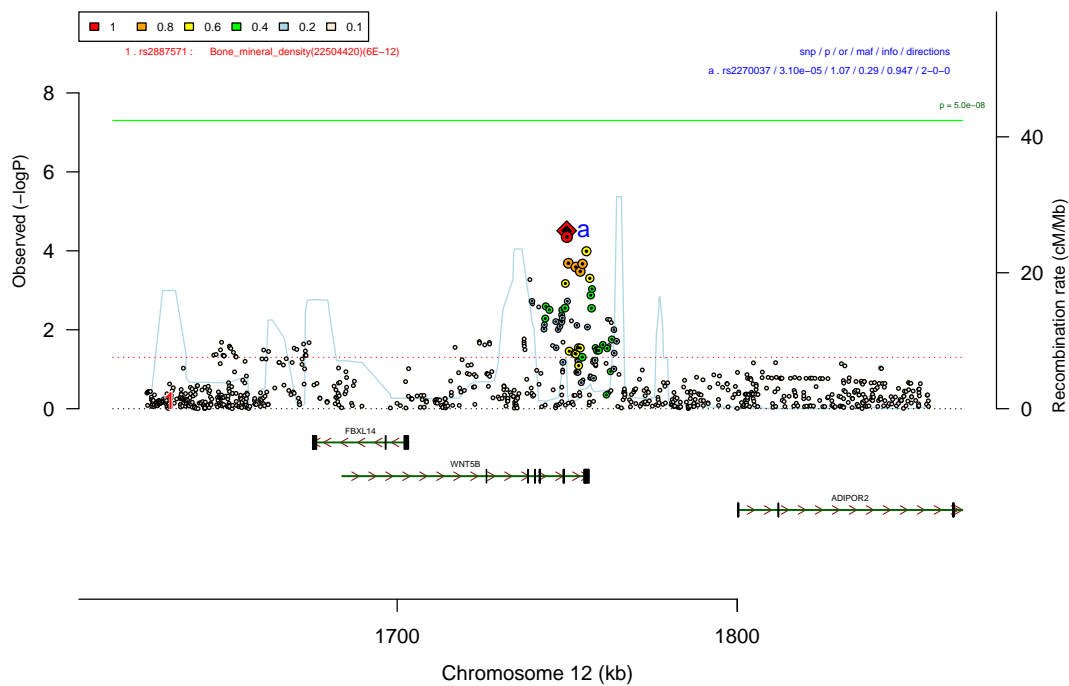
Supplementary Figure 78: Regional plot around *C8orf74* for gene-based MAGMA analysis of iPSYCH-PGC meta (18 381 cases and 27 969 controls). Chromosomal position is on the horizontal axis in kb, The significance of the association expressed as $-\log_{10}(P)$ (from z-test) is plotted vertically (left axis) and the recombination rate from HapMap is shown as light blue curve (right axis). The dot size is proportional to LD between the plotted SNP and the index SNP defining the associated region. Index SNPs are marked by diamonds and labelled with letters. Colouring is based on degree of LD to that SNP as represented by r^2 (legend for r^2 is in the upper left corner). A black centre indicates that the markers is present in the 1000 Genomes Project but not in HapMap3. The green line shows the genome-wide significance level ($5 \cdot 10^{-8}$). The upper right corner show details of the index SNPs (marker name/association p-value/odds ratio for the minor allele/minor allele frequency/imputation quality/direction of effect in the two studies (N one direction - N other direction- N missing)). Loci listed in the GWAS Catalogue for other phenotypes are shown in the upper left corner (capped at 10). The green lines in the lower half show genes (based on UCSC) with black vertical lines indicating exons. Arrow heads show the direction of transcription.



Supplementary Figure 79: Regional plot around *MDH1* for gene-based MAGMA analysis of iPSYCH-PGC meta (18 381 cases and 27 969 controls). Chromosomal position is on the horizontal axis in kb, The significance of the association expressed as $-\log_{10}(P)$ (from z-test) is plotted vertically (left axis) and the recombination rate from HapMap is shown as light blue curve (right axis). The dot size is proportional to LD between the plotted SNP and the index SNP defining the associated region. Index SNPs are marked by diamonds and labelled with letters. Colouring is based on degree of LD to that SNP as represented by r^2 (legend for r^2 is in the upper left corner). A black centre indicates that the markers is present in the 1000 Genomes Project but not in HapMap3. The green line shows the genome-wide significance level ($5 \cdot 10^{-8}$). The upper right corner show details of the index SNPs (marker name/association p-value/odds ratio for the minor allele/minor allele frequency/imputation quality/direction of effect in the two studies (N one direction - N other direction- N missing)). The green lines in the lower half show genes (based on UCSC) with black vertical lines indicating exons. Arrow heads show the direction of transcription.



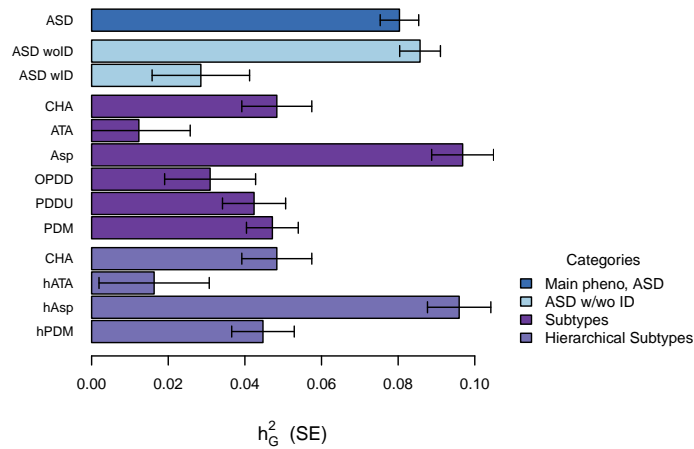
Supplementary Figure 80: Regional plot around *HDAC4* for gene-based MAGMA analysis of iPSYCH-PGC meta (18 381 cases and 27 969 controls). Chromosomal position is on the horizontal axis in kb, The significance of the association expressed as $-\log_{10}(P)$ (from z-test) is plotted vertically (left axis) and the recombination rate from HapMap is shown as light blue curve (right axis). The dot size is proportional to LD between the plotted SNP and the index SNP defining the associated region. Index SNPs are marked by diamonds and labelled with letters. Colouring is based on degree of LD to that SNP as represented by r^2 (legend for r^2 is in the upper left corner). A black centre indicates that the markers is present in the 1000 Genomes Project but not in HapMap3. The green line shows the genome-wide significance level ($5 \cdot 10^{-8}$). The upper right corner show details of the index SNPs (marker name/association p-value/odds ratio for the minor allele/minor allele frequency/imputation quality/direction of effect in the two studies (N one direction - N other direction- N missing)). Loci listed in the GWAS Catalogue for other phenotypes are shown in the upper left corner (capped at 10). The green lines in the lower half show genes (based on UCSC) with black vertical lines indicating exons. Arrow heads show the direction of transcription.



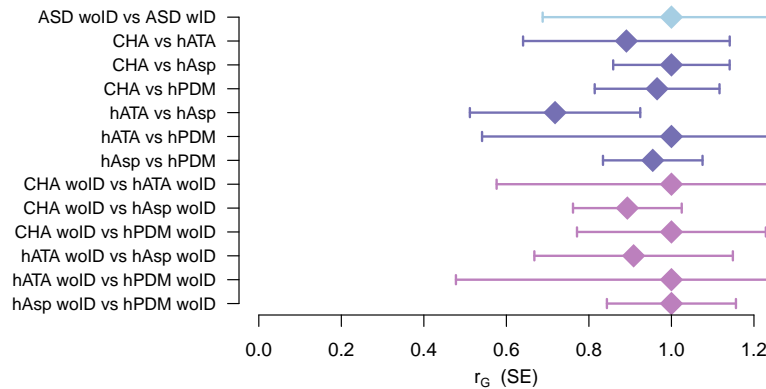
Supplementary Figure 81: Regional plot around *WNT5B* for gene-based MAGMA analysis of iPSYCH-PGC meta (18 381 cases and 27 969 controls). Chromosomal position is on the horizontal axis in kb, The significance of the association expressed as $-\log_{10}(P)$ (from z-test) is plotted vertically (left axis) and the recombination rate from HapMap is shown as light blue curve (right axis). The dot size is proportional to LD between the plotted SNP and the index SNP defining the associated region. Index SNPs are marked by diamonds and labelled with letters. Colouring is based on degree of LD to that SNP as represented by r^2 (legend for r^2 is in the upper left corner). A black centre indicates that the markers is present in the 1000 Genomes Project but not in HapMap3. The green line shows the genome-wide significance level ($5 \cdot 10^{-8}$). The upper right corner show details of the index SNPs (marker name/association p-value/odds ratio for the minor allele/minor allele frequency/imputation quality/direction of effect in the two studies (N one direction - N other direction- N missing)). Loci listed in the GWAS Catalogue for other phenotypes are shown in the upper left corner (capped at 10). The green lines in the lower half show genes (based on UCSC) with black vertical lines indicating exons. Arrow heads show the direction of transcription.

4.2 Polygenic qualities of ASD and its subtypes

4.2.1 Heritability and genetic correlation across subtypes

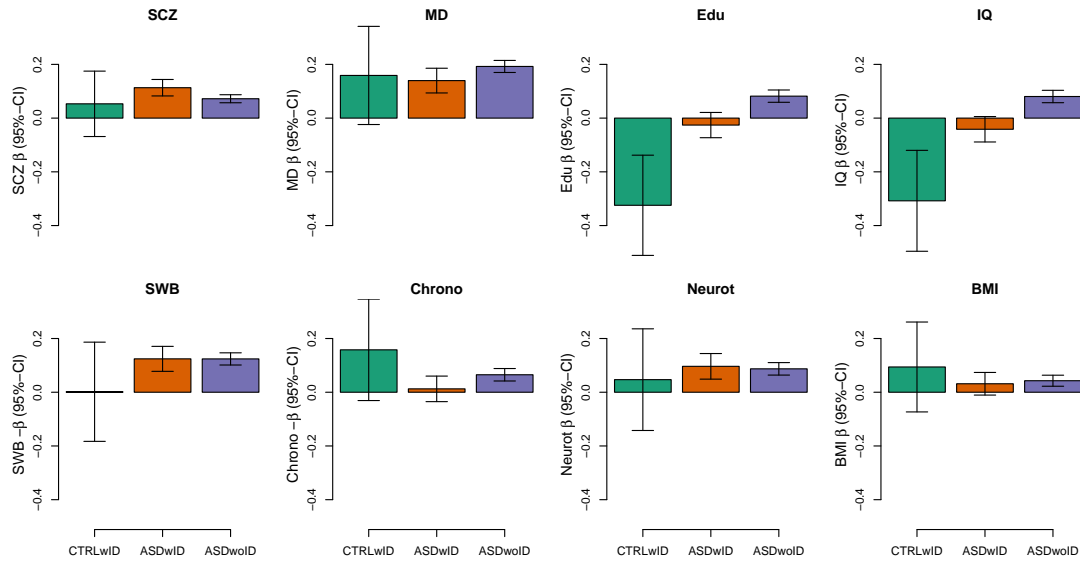


Supplementary Figure 82: Heritability estimates on the liability scale for subtypes and substrata of ASD as estimated by GCTA. Error bars show standard errors. The number of samples are: ASD 13 076, ASD w/ID 1 873, ASD w/o ID 11 203, CHA and hCHA 3 310, ATA 1 607, Asp 4 622, OPDD 2 042, PDDU 3 753, PDM 5 460, hATA 1 494, hAsp 4 417, hPDM 3 855. For tabulated details, look up Table 14.

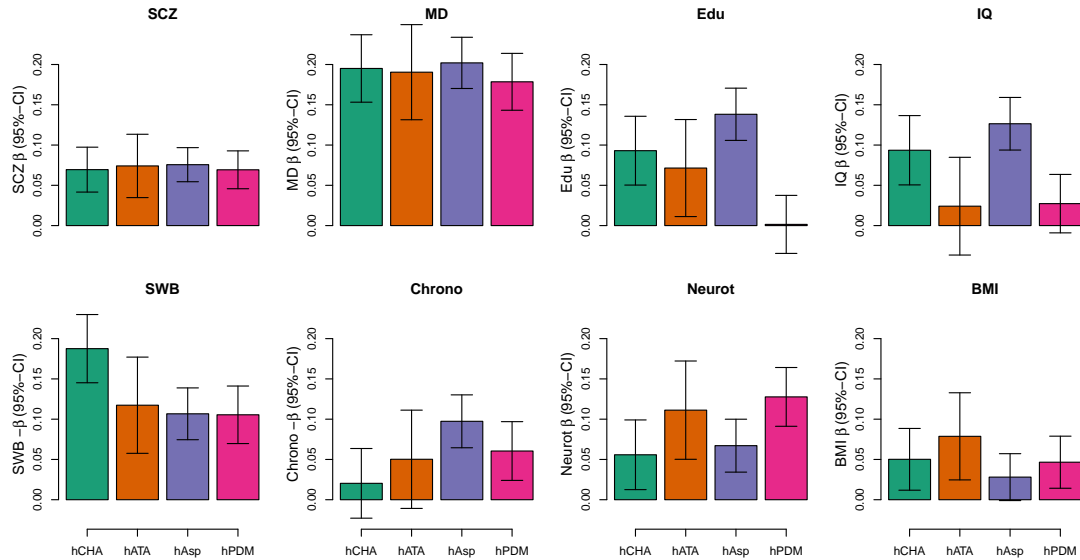


Supplementary Figure 83: GCTA based estimates of genetic correlation, r_G , between ASD subtypes internally in iPSYCH ASD GWAS. Error bars designate standard errors. The number of samples are: ASD 13 076, ASD w/ID 1 873, ASD w/o ID 11 203, CHA and hCHA 3 310, ATA 1 607, Asp 4 622, OPDD 2 042, PDDU 3 753, PDM 5 460, hATA 1 494, hAsp 4 417, hPDM 3 855. For tabulated details see Table 15.

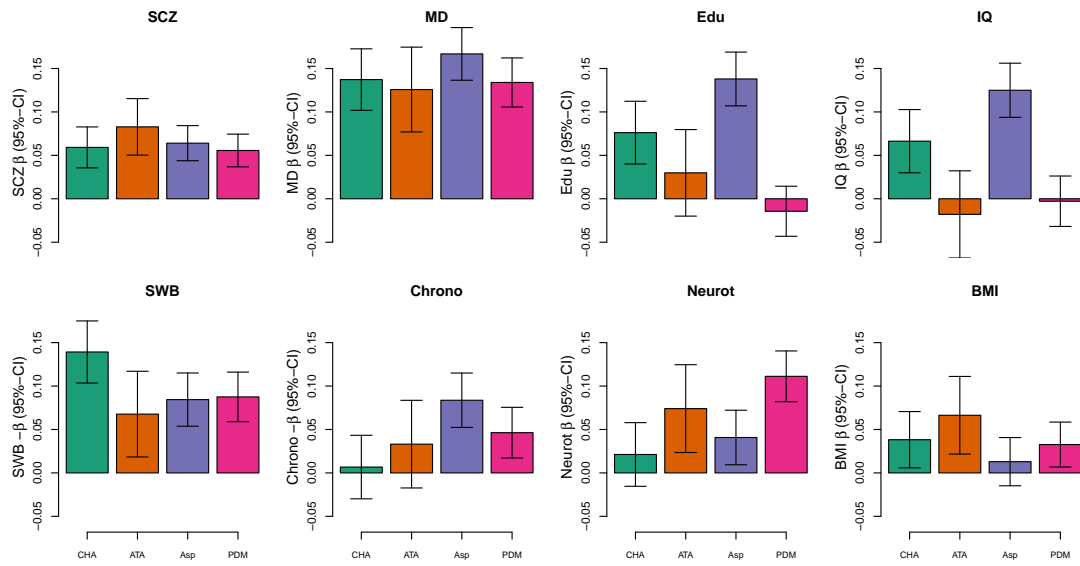
4.2.2 Multivariate regression



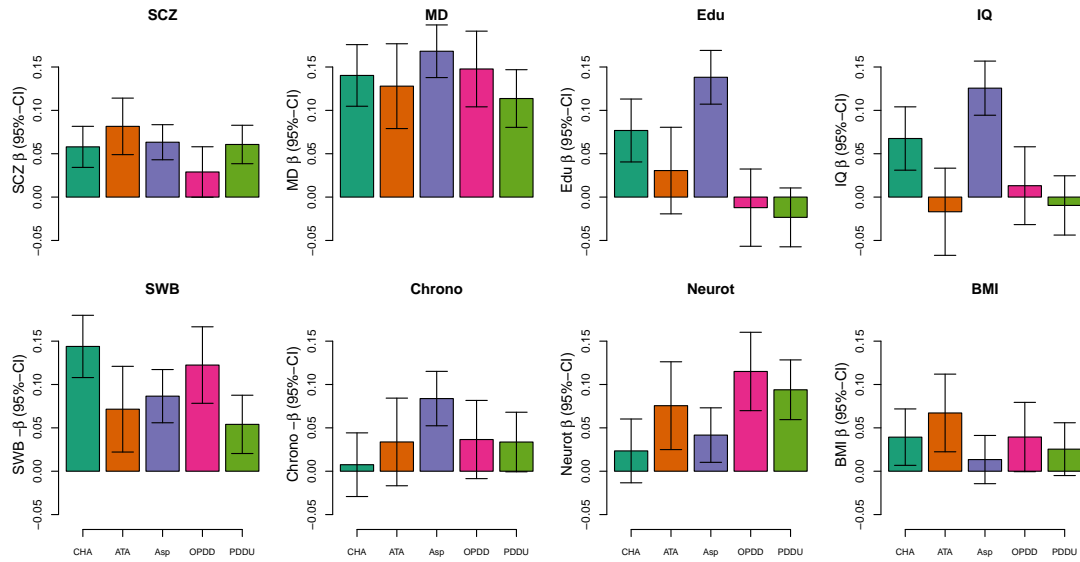
Supplementary Figure 84: Multivariate regression of normalized PRS on ID and ASD subtypes. Controls without ID (22 555) is reference group and the other groups are controls with ID (109), ASD with ID (2 358) and ASD without ID (11 203). The regression is adjusted for waves and relevant PCs, whiskers indicate 95%-CIs). The phenotypes are schizophrenia[8], major depression[48] (both with the Danish samples removed), educational attainment[50], IQ[83], subjective well-being[62] (–1·score), chronotype[63] (–1·score), neuroticism[62], and BMI[236] (–1·score). Beware that the orientation of the scores for subjective well-being, chronotype and BMI have been switched to improve graphical presentation.



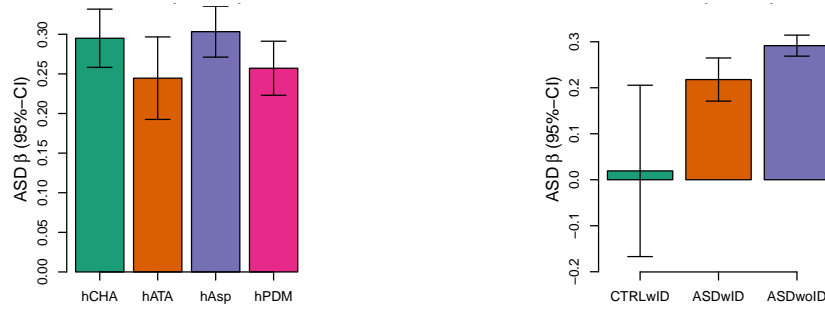
Supplementary Figure 85: Multivariate regression of normalized PRS on hierarchical ASD subtypes without ID (controls is reference group (22 555) and the regression is adjusted for waves and relevant PCs, whiskers indicate 95%-CIs, hCHA 2 358, hATA 1,099, hAsp 4,343, and hPDM 3,403). The plot shows what happens to Figure 3 in the main manuscript when restricting to individuals without ID. Analogous to the analysis including also ID, we find that there is significant heterogeneity across certain subtypes albeit slightly weaker: educational attainment $P = 5.3 \cdot 10^{-8}$, IQ $P = 4.7 \cdot 10^{-5}$, neuroticism $P = 0.017$, chronotype $P = 0.025$ and subjective well-being $P = 0.0064$ (linear hypotheses tested using the Pillai test). The phenotypes are schizophrenia[8], major depression[48] (both with the Danish samples removed), educational attainment[50], IQ[83], subjective well-being[62] (–1·score), chronotype[63] (–1·score), neuroticism[62], and BMI[236] (–1·score). Beware that the orientation of the scores for subjective well-being, chronotype and BMI have been switched to improve graphical presentation.



Supplementary Figure 86: Multivariate regression of normalized PRS on the original (non-hierarchical) ASD subtypes. Controls constitute reference group (22 664) and the subtypes are childhood autism (3 310), atypical autism (1 607), Asperger's (4 622), and the two pervasive disorder types comined (5 460). The regression is adjusted for waves and relevant PCs, whiskers indicate 95%-CIs. The plot shows what happens to Figure 3 in the main manuscript when regressing on the original subtypes that includes overlaps. We see that construction of the hierarchical subtypes (and the order in which it is done) does little to change the overall pattern. The phenotypes are schizophrenia[8], major depression[48] (both with the Danish samples removed), educational attainment[50], IQ[83], subjective well-being[62] (−1·score), chronotype[63] (−1·score), neuroticism[62], and BMI[236] (−1·score). Beware that the orientation of the scores for subjective well-being, chronotype and BMI have been switched to improve graphical presentation.

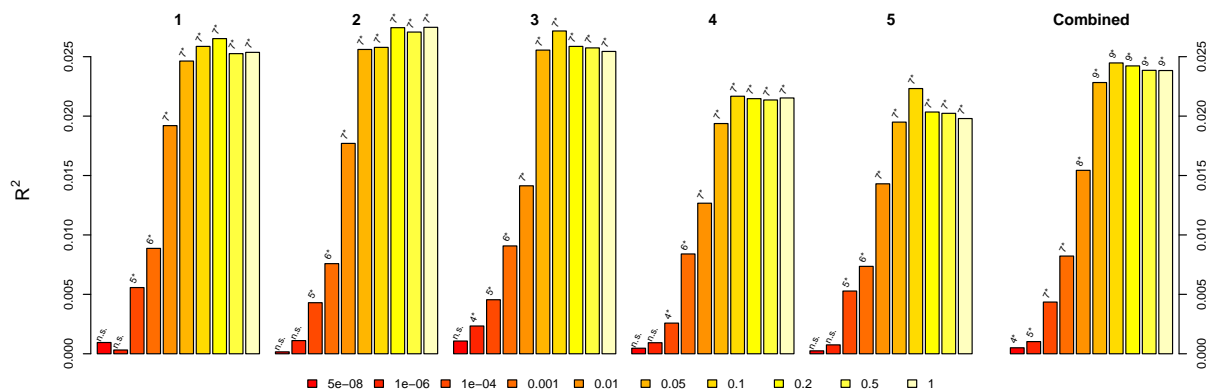


Supplementary Figure 87: Multivariate regression of normalized PRS on the original non-hierarchical ASD subtypes before lumping the two pervasive disorder subtypes Controls constitute reference group (22 664) and the subtypes are childhood autism (3 310), atypical autism (1 607), Asperger's (4 622), other pervasive devlopmental disorder (2 042), and pervasive developmental disorder, unspecified (3 753). The regression is adjusted for waves and relevant PCs, whiskers indicate 95%-CIs. The plot shows what happens to Figure 86 when separating the two pervasive disorder subtypes. The overall pattern remains the same, and analogous to the analysis on grouped hierarchical subtypes. The phenotypes are schizophrenia[8], major depression[48] (both with the Danish samples removed), educational attainment[50], IQ[83], subjective well-being[62] (−1·score), chronotype[63] (−1·score), neuroticism[62], and BMI[236] (−1·score). Beware that the orientation of the scores for subjective well-being, chronotype and BMI have been switched to improve graphical presentation.

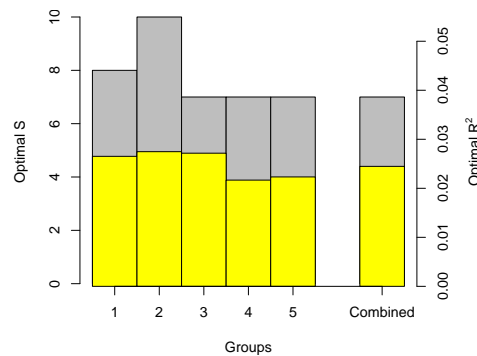


Supplementary Figure 88: Regression of the internally trained ASD PRS on hierarchical subtypes (left) and ID/ASD subtypes (right). In both panels the regression is adjusted for waves and relevant PCs, and the whiskers indicate 95%-CIs. In the left panel, the reference group consists of all controls (22,664) and subtypes are the hierarchically defined subtypes for childhood autism (hCHA, N = 3,310), atypical autism (hATA, N = 1,494), Asperger's (hAsp, N = 4,417), and the lumped pervasive disorders developmental group (hPDM, N = 3,855). In the right panel, the reference groups is restricted to controls without ID (22 555), and the subtypes are controls with ID (109), ASD with ID (2 358) and ASD without ID (11 203).

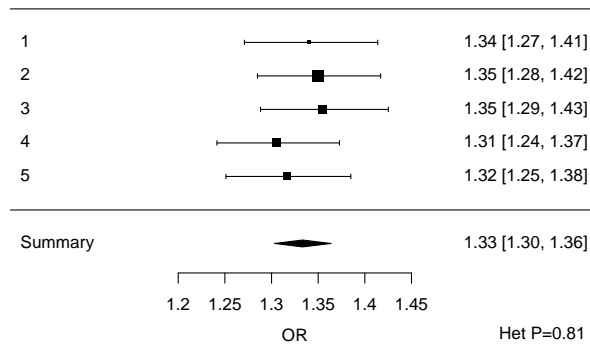
4.2.3 Internally generated PRS



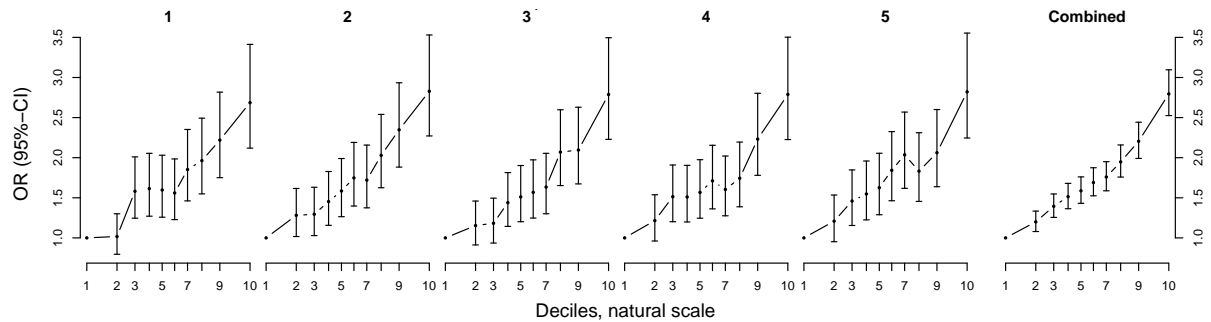
Supplementary Figure 89: Nagelkerke R^2 of PRS trained internally on leave-one-group-out and the PGC ASD shown here when estimated on each of the five groups left out when training as well as on the combined sample (cases/controls in groups 1: 2 624/3 694, 2: 2 622/5 432, 3: 2 611/4 666, 4: 2 583/4 360, 5: 2 636/4 512, and in total 13 076/22 664). Colouring is as shown in the legend signifying the 10 different p-value cut-off in the training set.



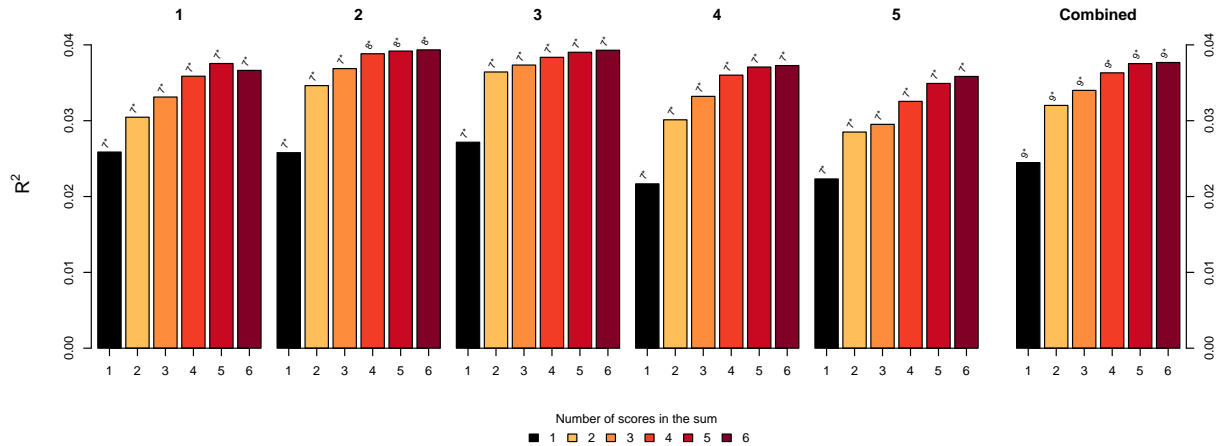
Supplementary Figure 90: Summarizing optimal Nagelkerke R^2 in target set (yellow, right axis) and p-value threshold in training set (grey, left axis) for internally trained leave-one-group-out ASD score across the groups as well as on the combined sample (cases/controls in groups 1: 2 624/3 694, 2: 2 622/5 432, 3: 2 611/4 666, 4: 2 583/4 360, 5: 2 636/4 512, and in total 13 076/22 664). In for most groups as well as for the combined analysis on the full data set, the 7th p-value threshold of 0.1 is the optimal cut point.



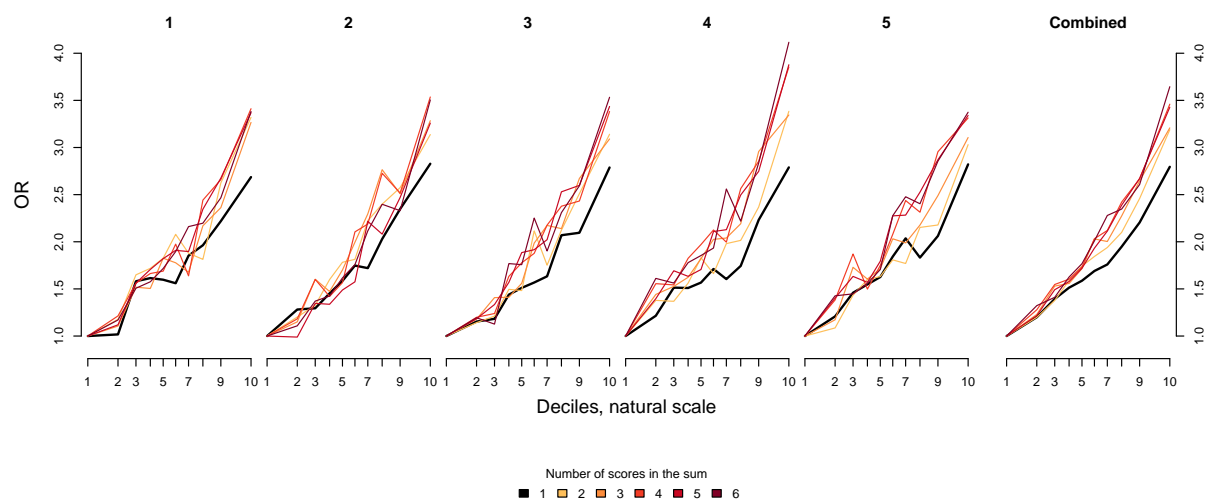
Supplementary Figure 91: Forest plot of effect of the internally trained leave-one-group-out ASD scores as continuous measure with 95%-confidence intervals (cases/controls in groups 1: 2 624/3 694, 2: 2 622/5 432, 3: 2 611/4 666, 4: 2 583/4 360, 5: 2 636/4 512, and in total 13 076/22 664). The score used here are for the p-value threshold $S=7$, $p=0.1$.



Supplementary Figure 92: Decile plots of internal ASD score over groups. The plot show odds ratio at each score decile relative to the first in the first five panels for the results in each of the 5 groups, and in the last panel the results of regressing the combined score in the full sample (right axis is a identical the left, cases/controls in groups 1: 2 624/3 694, 2: 2 622/5 432, 3: 2 611/4 666, 4: 2 583/4 360, 5: 2 636/4 512, and in total 13 076/22 664). The score used here is for the p-value threshold $S=7$, $p=0.1$.

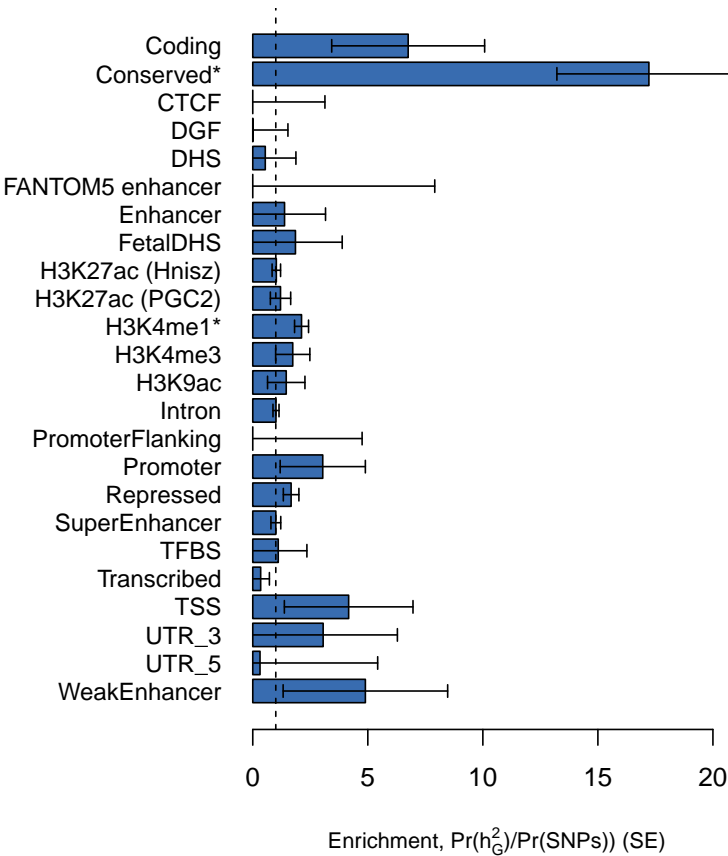


Supplementary Figure 93: Nagelkerke R^2 for multi-phenotype scores presented across leave-out-groups and on the full sample (cases/controls in groups 1: 2 624/3 694, 2: 2 622/5 432, 3: 2 611/4 666, 4: 2 583/4 360, 5: 2 636/4 512, and in total 13 076/22 664). Starting with the internally trained ASD score ($S=7$, $p=0.1$) new scores are successively generated by adding terms in a weighted sums of scores for 2 Major depression[48], 3 Subjective Well-Being[62], 4 Schizophrenia[8], 5 Educational attainment[50], 6 Chronotyp[63]. Each colour coded bar give the Nagelkerke R^2 for the weighted sum of that many scores as a predictor for ASD in our sample.

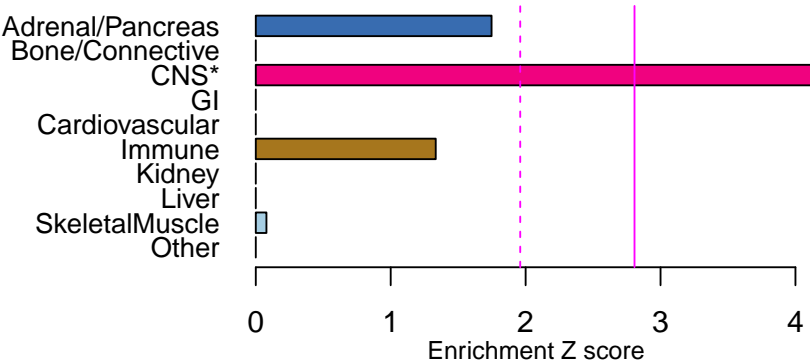


Supplementary Figure 94: Decile plots for multi-phenotype scores over groups on the complete sample (cases/controls in groups 1: 2624/3694, 2: 2622/5432, 3: 2611/4666, 4: 2583/4360, 5: 2636/4512, and in total 13076/22664). The plot show for each group and combined to the full sample, the odds ratio at each score decile relative to the first (right axis is identical to the left). Starting with the internally trained ASD score ($S=7, p=0.1$) new scores are successively generated by adding terms in a weighted sums of scores for 2 Major depression[48], 3 Subjective Well-Being[62], 4 Schizophrenia[8], 5 Educational attainment[50], 6 Chronotyp[63].

4.2.4 Functional and tissue specific enrichment



Supplementary Figure 95: Enrichment estimates by LDSC for the 24 main annotations from [58] and based on the main ASD scan of 18 381 cases and 27 969 controls. The error bars represent jackknife standard errors around the estimates of enrichment, the dashed line designate the neutral scenario or no enrichment, and significance (from z-scores as described in [58]) at α -level 0.05 after Bonferroni correction for the 24 hypotheses tested is indicated by an asterisk at the name.

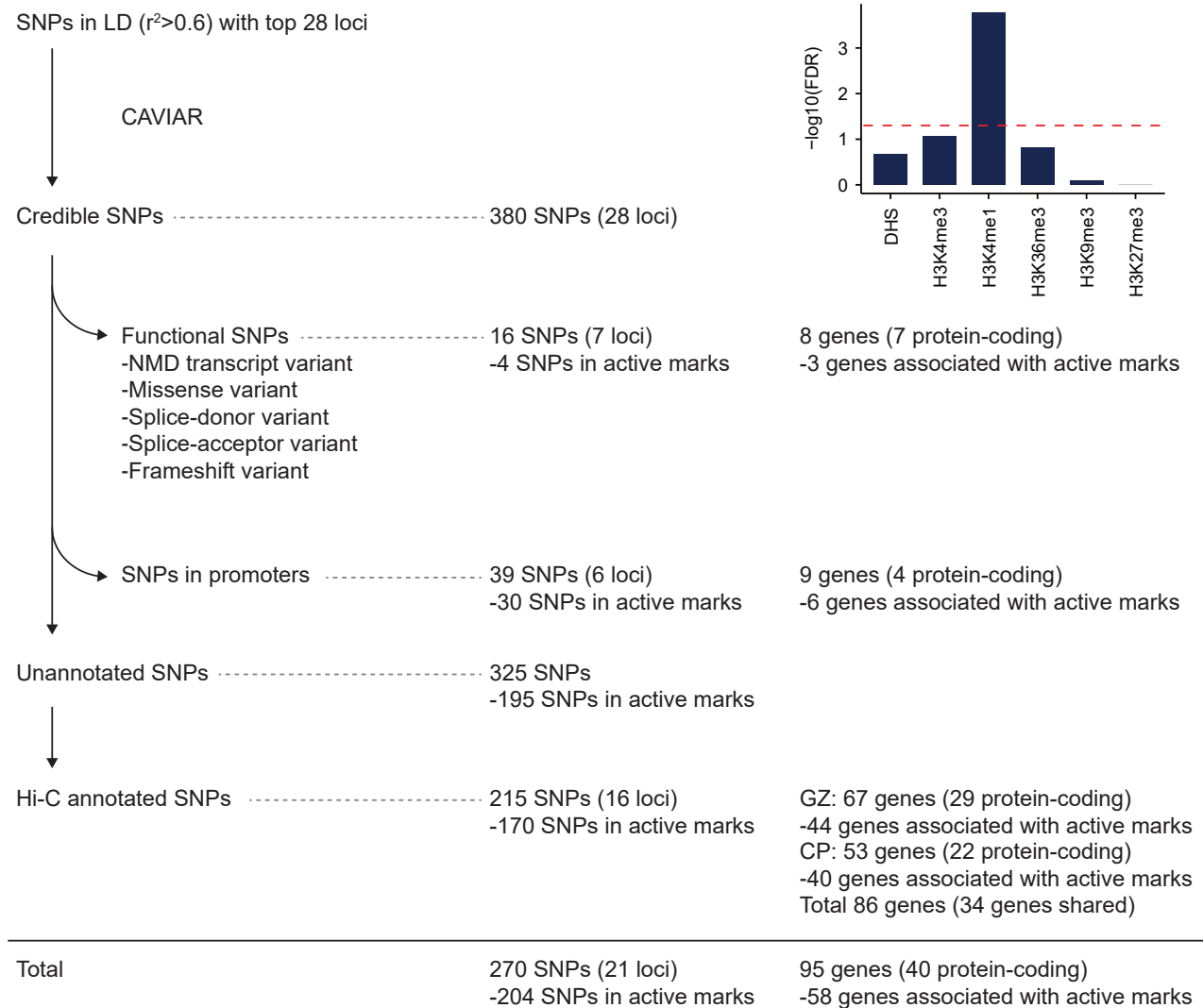


Supplementary Figure 96: Tissue enrichment based on LDSC, using the original cell type annotation of LDSC. The dashed magenta line indicates statistical significance at α -level 0.05 and the solid line significance after Bonferroni correction for the number of tissues tested (significance from z-test). Tissues achieving global statistical significance are marked by an asterisk.



Supplementary Figure 97: Tissue enrichment based on LDSC and roadmap h3k4me1 cell type annotation[59, 60]. The dashed magenta line indicates statistical significance at α -level 0.05 and the solid line significance after Bonferroni correction for the number of tissues tested (significance from z-test). Tissues achieving global statistical significance are marked by an asterisk.

4.3 Hi-C analysis



Supplementary Figure 98: Defining putative target genes of ASD loci. Credible SNPs were derived from top 28 ASD loci by CAVIAR, resulting in 380 SNPs that are enriched in H3K4me1 marks in fetal brain. Credible SNPs were subsequently categorized into functional SNPs (SNPs that cause nonsynonymous variants and SNPs located within promoters) and unannotated SNPs. Functional SNPs were directly assigned to their target genes, while unannotated SNPs were assigned to their target genes based on chromatin contact profiles in fetal brain.