Report of HLSA genes (1) listed by SFARI or AutismKB as autism candidate genes, (2) identified as being differentially expressed in autism, (3) identified as being differentially expressed in schizophrenia, (4) identified as being differentially expressed in relevant knockout/knockdowns (e.g. CHD8)

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# HLSA genes according to Fortna, 2004 and my preliminary BLAT analysis

## gene.name  
## 1 ABCB10  
## 2 ABCB10P1  
## 3 ABCB10P3  
## 4 ABCB10P4  
## 5 AC007325.3  
## 6 AC023491.1  
## 7 ACKR4  
## 8 ACTR2  
## 9 ADGRF5  
## 10 ADGRF5P1  
## 11 ADGRF5P2  
## 12 ADGRF5P3  
## 13 AG1  
## 14 AMY1A  
## 15 AMY1B  
## 16 AMY1C  
## 17 AMY2A  
## 18 AMY2B  
## 19 AMYP1  
## 20 ANAPC1  
## 21 AQP7  
## 22 AQP7P1  
## 23 AQP7P2  
## 24 AQP7P3  
## 25 AQP7P4  
## 26 ARHGEF34P  
## 27 ARHGEF35  
## 28 ARHGEF5  
## 29 ARMC10  
## 30 ARMC10P1  
## 31 BIRC1  
## 32 CCRL1  
## 33 CDC27  
## 34 CDH12  
## 35 CELSR2  
## 36 CFC1  
## 37 CFC1B  
## 38 CH17-189H20.1  
## 39 CH17-472G23.2  
## 40 DDX11  
## 41 DDX11L1  
## 42 DDX11L10  
## 43 DDX11L2  
## 44 DDX11L5  
## 45 DDX12P  
## 46 DGCR9  
## 47 DRD5  
## 48 DRD5P1  
## 49 DRD5P2  
## 50 DUXAP10  
## 51 DUXAP10  
## 52 DUXAP8  
## 53 DUXAP8  
## 54 DUXAP9  
## 55 E2F6  
## 56 E2F6P1  
## 57 E2F6P2  
## 58 E2F6P3  
## 59 F379  
## 60 FAM3C  
## 61 FAM3C2  
## 62 FAM72A  
## 63 FAM72B  
## 64 FAM72C  
## 65 FAM72D  
## 66 FCGR1  
## 67 FCGR2  
## 68 FGF7  
## 69 FGF7P1  
## 70 FGF7P2  
## 71 FLJ10520  
## 72 FO538757.2  
## 73 FP325317.1  
## 74 GLIDR  
## 75 GPCR16  
## 76 GTF2H2  
## 77 GTF2H2B  
## 78 GTF2H2C  
## 79 GTF2H2C\_2  
## 80 GTF2I  
## 81 GUSB  
## 82 GUSBP1  
## 83 GUSBP2  
## 84 GUSBP3  
## 85 GUSBP4  
## 86 GUSBP9  
## 87 HIST2H2BB  
## 88 KGFLP1  
## 89 KGFLP2  
## 90 KIAA0738  
## 91 LA16c-60G3.8  
## 92 LINC00623  
## 93 LINC00680-GUSBP4  
## 94 LINC00869  
## 95 LINC01138  
## 96 LOC100233156  
## 97 LOC100507415  
## 98 LOC102724238  
## 99 LOC103091866  
## 100 LOC103908605  
## 101 LOC391793  
## 102 LOC403323  
## 103 LOC441081  
## 104 LOC554249  
## 105 LOC642846  
## 106 LOC645166  
## 107 LOC646214  
## 108 LOC728673  
## 109 MAFIP  
## 110 MST1  
## 111 MST1L  
## 112 MST1P2  
## 113 NAIP  
## 114 NBPF1  
## 115 NBPF10  
## 116 NBPF11  
## 117 NBPF12  
## 118 NBPF13P  
## 119 NBPF14  
## 120 NBPF15  
## 121 NBPF17  
## 122 NBPF18P  
## 123 NBPF19  
## 124 NBPF20  
## 125 NBPF21P  
## 126 NBPF22P  
## 127 NBPF23P  
## 128 NBPF25P  
## 129 NBPF2P  
## 130 NBPF3  
## 131 NBPF4  
## 132 NBPF5P  
## 133 NBPF6  
## 134 NBPF7  
## 135 NBPF8  
## 136 NBPF9  
## 137 NEK2  
## 138 NEK2P2  
## 139 NEK2P4  
## 140 nu30f08  
## 141 NUDT4  
## 142 NUDT4P1  
## 143 NUDT4P2  
## 144 OR2A1  
## 145 OR2A13P  
## 146 OR2A14  
## 147 OR2A15P  
## 148 OR2A2  
## 149 OR2A20P  
## 150 OR2A25  
## 151 OR2A3P  
## 152 OR2A4  
## 153 OR2A42  
## 154 OR2A7  
## 155 OR2A9P  
## 156 OR4F13P  
## 157 OR4F16  
## 158 OR4F1P  
## 159 OR4F21  
## 160 OR4F28P  
## 161 OR4F29  
## 162 OR4F2P  
## 163 OR4F3  
## 164 OR4F7P  
## 165 OR4F8P  
## 166 PAIP1  
## 167 PAK2  
## 168 PARP8  
## 169 PDE4DIP  
## 170 PMP2  
## 171 POM121L9P  
## 172 RAB6C  
## 173 RFWD3  
## 174 ROCK1  
## 175 ROCK1P1  
## 176 RP11-1023L17.1  
## 177 RP11-111F5.4  
## 178 RP11-211N8.2  
## 179 RP11-244H18.4  
## 180 RP11-262H14.4  
## 181 RP11-407P15.2  
## 182 RP11-475124.3  
## 183 RP11-527H14.1  
## 184 RP11-536C10.10  
## 185 RP11-536C10.12  
## 186 RP11-536C10.26  
## 187 RP11-589F5.4  
## 188 RP11-597A11.11  
## 189 RP11-597A11.4  
## 190 RP5-998N21.4  
## 191 RPL23AP25  
## 192 RPL23AP4  
## 193 RPL23AP45  
## 194 RPL23AP54  
## 195 RPL23AP60  
## 196 RPL23AP7  
## 197 RPL23AP82  
## 198 RPL23AP84  
## 199 RPL23AP87  
## 200 RPL23AP88  
## 201 RPL23AP97  
## 202 SLC6A13  
## 203 SMA4  
## 204 SMA5  
## 205 SNORA49A  
## 206 SNORA49B  
## 207 SPTLC2  
## 208 SRGAP2  
## 209 SRGAP2B  
## 210 SRGAP2C  
## 211 SRGAP2D  
## 212 SRP68  
## 213 TCAF1  
## 214 TCAF2  
## 215 TEKT4  
## 216 TEKT4P2  
## 217 USP10  
## 218 WASH1  
## 219 WASH2P  
## 220 WASH3P  
## 221 WASH5P  
## 222 WASH6P  
## 223 WASH7P  
## 224 WDR70  
## 225 XLOC\_007697  
## 226 YBEY

# Identify HLSA genes that are found in the SFARI and AutKB lists of autism candidate genes

## SFARI (downloaded on: )

### HLSA genes in SFARI list

## gene.name  
## 1 DDX11  
## 2 GTF2I

## Autism KB (downloaded on: )

### HLSA genes in Autism KB Core Candidate List

## [1] gene.name  
## <0 rows> (or 0-length row.names)

### HLSA genes in Autism KB Syndromic List

## [1] gene.name  
## <0 rows> (or 0-length row.names)

### HLSA genes in Autism KB Non Syndromic List

## gene.name  
## 1 AMY1C  
## 2 AMY2B  
## 3 ARMC10  
## 4 CDH12  
## 5 DGCR9  
## 6 DRD5  
## 7 FAM3C  
## 8 GTF2I  
## 9 NBPF10  
## 10 NBPF20  
## 11 PAIP1  
## 12 PDE4DIP  
## 13 ROCK1  
## 14 SLC6A13  
## 15 WASH3P  
## 16 WDR70

# Studies of Differential Expression in Post-mortem Brain

## Garbett, 2008

### Samples Size: 6 cases, 6 controls

### RNA Source: Post-mortem temporal cortex

### Analysis method: microarray - no model details given

## gene.name  
## 1 DGCR9

## PROBE GENE.NAME SYMBOL UniGene  
## 1 215003\_at DiGeorge syndrome critical region gene 9 DGCR9 Hs.106311  
## Entrez AUT CTRL ALR Pair.pVal Group.pVal Rank.PVal X X.1 X.2 X.3  
## 1 25787 6.29 7.51 -1.22 0.005733 0.000475 0.001146 NA NA NA NA

## Voineagu, 2011

### Sample size: Discovery set: 19 cases and 17 controls, Replication cohort: 9 ASD cases and 5 controls

### Brain area(s): frontal cortex (BA9), temporal cortex (BA41/42 or BA22) and cerebellum (vermis), with the exception of three controls lacking the cerebellum sample. Replication set only looked at tissue from frontal cortex (BA44/45). Analysis focused on "cortex samples" - as they only found two differentially expressed genes in cerebellum.

### Analysis technique: Microarray - Illumina Ref8 v3 microarrays

### Other Notes: Brain area in replication set different from discovery cohort. Also, not all samples in the replication set were independent from discovery cohorot - they reused cases, and just looked at different brain area.

### Genes differentially expressed in discovery cohort

## gene.name  
## 1 AMY2B  
## 2 ARMC10  
## 3 CDH12  
## 4 FAM3C  
## 5 NBPF10  
## 6 NBPF20

## ProbeID Gene.Symbol Cortex\_AutismvsControls\_FC  
## 1 ILMN\_1709269 CDH12 0.8370447  
## 2 ILMN\_1777261 FAM3C 0.7276693  
## 3 ILMN\_2052598 ARMC10 0.8782015  
## 4 ILMN\_2073157 AMY2B 0.8631691  
## 5 ILMN\_2115490 NBPF20 1.5284685  
## 6 ILMN\_2155719 NBPF10 1.3487623  
## 7 ILMN\_2368773 FAM3C 0.7497569  
## Cortex\_AutismvsControls\_qvalue Temporal\_AutismvsControls\_FC  
## 1 4.1698198 0.8280483  
## 2 0.7096086 0.7240259  
## 3 17.5014686 0.9685903  
## 4 4.1698198 0.8414077  
## 5 0.0000000 1.5586869  
## 6 0.0000000 1.3037540  
## 7 0.9329120 0.7529778  
## Temporal\_AutismvsControls\_qvalue Frontal\_AutismvsControls\_FC  
## 1 31.751193 0.8444261  
## 2 35.088545 0.7306430  
## 3 79.727978 0.8110089  
## 4 33.575113 0.8812641  
## 5 9.898855 1.5043479  
## 6 14.005943 1.3864733  
## 7 36.736471 0.7471500  
## Frontal\_AutismvsControls\_qvalue Controls\_TemporalvsFrontal\_FC  
## 1 26.720024 1.2662659  
## 2 8.601491 0.9866823  
## 3 12.253806 0.7984737  
## 4 28.273537 1.1681708  
## 5 6.467286 1.1065897  
## 6 8.601491 1.0605629  
## 7 9.312892 1.0145796  
## Controls\_TemporalvsFrontal\_qvalue Autism\_TemporalvsFrontal\_FC  
## 1 0.0000000 1.0986129  
## 2 81.8326961 0.9207208  
## 3 0.9112667 0.9208453  
## 4 4.2629751 1.1018553  
## 5 60.0804651 1.2010468  
## 6 64.4555840 1.0368892  
## 7 78.6134113 0.9770911  
## Autism\_TemporalvsFrontal\_qvalue  
## 1 70.80669  
## 2 76.65605  
## 3 57.69769  
## 4 62.15390  
## 5 63.55850  
## 6 81.78632  
## 7 87.71360

### Differentially expressed genes that were replicated

## gene.name  
## 1 NBPF10  
## 2 NBPF20

## Probe.ID Gene.Symbol FC\_AllAutismCasesvsControls  
## 1 ILMN\_2155719 NBPF10 1.302284  
## 2 ILMN\_2115490 NBPF20 1.212793  
## pvalue\_AllAutismCasesvsControls FC\_IndependentAutismCasesvsControls  
## 1 0.03539898 1.381083  
## 2 0.21247376 1.527279  
## pvalue\_IndependentAutismCasesvsControls  
## 1 0.05709377  
## 2 0.02569975

### Genes that were determined to have been differentially spliced in autism cases compared to controls

## [1] gene.name  
## <0 rows> (or 0-length row.names)

## [1] GeneId Gene.Symbol Chromosome C1   
## [5] A C2 X.inc.Difference  
## <0 rows> (or 0-length row.names)

## Chow, 2012

### Sample Size: Young = 9 cases, 7 controls; Adult = 6 cases, 11 controls

### RNA Source: Dorsolateral prefrontal cortex

### Analysis method: Microarray - Illumina Human-Ref8 v3 microarray [27,28].

#### HLSA genes differentially expressed in brains of young (2-14 years) autism cases (9 cases, 7 controls)

## [1] gene.name  
## <0 rows> (or 0-length row.names)

## [1] Num ProbeID p.Value Gene Cytoband Fold.Change  
## <0 rows> (or 0-length row.names)

#### HLSA genes differentially expressed in brains of adult (15 - 56 years) autism cases (6 cases, 11 controls)

## [1] gene.name  
## <0 rows> (or 0-length row.names)

#### HLSA genes differentially expressed across both young and adult cases

## gene.name  
## 1 NBPF15

## Num UniqueID P.value Symbol Cytoband FoldChange X X.1  
## 1 333 ILMN\_2147133 0.0048134 NBPF15 1q21.1d -1.580338152

## Ginsberg, 2012

### Sample Size:

### RNA Source: cerebellar & BA19 (occiptal) post-mortem brain

### Analysis method: microarray

#### HLSA genes within the list of top differentially expressed genes after controlling for brain region

## [1] gene.name  
## <0 rows> (or 0-length row.names)

#### HLSA genes within the list of top differentially expressed genes in the cerebellum.

## [1] gene.name  
## <0 rows> (or 0-length row.names)

## Ziats, 2012

### Sample Size: 2 cases and 2 age & sex-matched controls

### RNA Source: prefrontal cortex and cerebellum

### Analysis method: Microarray: ArrayStar, Inc (Rockville, MD, USA) Human lncRNA Microarray V2.0, For mRNA analysis:

#### HLSA genes in list of differentially expressed lncRNAs

## [1] gene.name  
## <0 rows> (or 0-length row.names)

#### HLSA genes in list of differentially expressed mRNAs in prefrontal cortex

## gene.name  
## 1 PDE4DIP

## seqname GeneSymbol unigene ProteinAccession EntrezID FCAbsolute  
## 1 NM\_001002811 PDE4DIP Hs.728768 NP\_071754 9659 3.34  
## regulation chrom strand txStart txEnd source  
## 1 Down in ASD chr1 - 144890590 144932032 RefSeq\_coding

#### HLSA genes in list of differentially expressed mRNAs in cerebellum

## gene.name  
## 1 GUSB

## seqname GeneSymbol EntrezID unigene ProteinAccession FCAbsolute  
## 1 NM\_000181 GUSB 2990 Hs.255230 NP\_000172 2.92  
## regulation chrom strand txStart txEnd source  
## 1 Down in ASD chr7 - 65425674 65447301 RefSeq\_coding

## Gupta, 2014

### Sample Size: 47 (32 unique individuals) autism and 57 (40 unique individuals) controls

### RNA Source: Brain regions;Multiple cortical tissues corresponding to Brodmann Area 19 (BA19), Brodmann Area 10 (BA10) and Brodmann Area 44 (BA44) were sequenced in 62, 14 and 28 samples, respectively

### Analysis method: RNA-Seq

#### HLSA genes in top differentially expressed genes

## gene.name  
## 1 CELSR2  
## 2 SRGAP2

## Gene  
## 1 CELSR2  
## 2 SRGAP2  
## P.value..EDASeq..normalized.gene.estimates.in.a.ISV..age.sex..site.and.brain.region.adjusted.model.  
## 1 0.000669  
## 2 0.001820  
## Mean.Expression..log2. Beta Standard.Error X X.1  
## 1 9.67 0.28 0.09 NA NA  
## 2 9.49 0.23 0.08 NA NA

# Studies of differential expression lymphoblastoid cell lines (LCLs)

## Nishimura, 2007 \*\*\* Note: Used LCL from snydromic cases \* **### Sample Size: 8 males with FMR1-FM, 7 males with dup(15q), 27 controls (confirm num. of controls used in analysis) ### RNA Source: LCLs ### Analysis Method: Microarray - Whole Human Genome Array G4112A (Agilent) ### Other Notes:** Need to incorporate results from body of paper

#### HLSA genes in list of differentially expressed genes from combined analysis of FMR1-FM & dup15q vs controls

## [1] gene.name  
## <0 rows> (or 0-length row.names)

## [1] Symbol GeneName ProbeName RefSeq Gene.locus P.value   
## <0 rows> (or 0-length row.names)

#### HLSA genes in list of differentially expressed genes in FMR1-FM as determined by SAM

## gene.name  
## 1 AG1  
## 2 AMY1C  
## 3 CELSR2  
## 4 GUSB  
## 5 NEK2  
## 6 NUDT4  
## 7 PARP8  
## 8 PDE4DIP  
## 9 RPL23AP7  
## 10 SMA5

## Symbol  
## 1 CELSR2  
## 2 SMA5  
## 3 AG1  
## 4 NUDT4  
## 5 PARP8  
## 6 RPL23AP7  
## 7 NUDT4  
## 8 NUDT4  
## 9 NEK2  
## 10 GUSB  
## 11 AMY1C  
## 12 PDE4DIP  
## GeneName  
## 1 cadherin, EGF LAG seven-pass G-type receptor 2 (flamingo homolog, Drosophila)  
## 2 SMA5  
## 3 AG1 protein  
## 4 nudix (nucleoside diphosphate linked moiety X)-type motif 4  
## 5 poly (ADP-ribose) polymerase family, member 8  
## 6 ribosomal protein L23a pseudogene 7  
## 7 nudix (nucleoside diphosphate linked moiety X)-type motif 4  
## 8 nudix (nucleoside diphosphate linked moiety X)-type motif 4  
## 9 NIMA (never in mitosis gene a)-related kinase 2  
## 10 glucuronidase, beta  
## 11 amylase, alpha 1C; salivary  
## 12 phosphodiesterase 4D interacting protein (myomegalin)  
## ProbeName RefSeq SAM.rank  
## 1 A\_23\_P201570 NM\_001408 673  
## 2 A\_23\_P121869 NM\_021036 1587  
## 3 A\_32\_P149492 b 1942  
## 4 A\_23\_P2366 NM\_199040 2085  
## 5 A\_23\_P121898 NM\_024615 2105  
## 6 A\_24\_P153043 NR\_000029 2434  
## 7 A\_24\_P335263 NM\_199040 2858  
## 8 A\_24\_P50753 NM\_199040 3100  
## 9 A\_23\_P35219 NM\_002497 3981  
## 10 A\_23\_P334608 NM\_000181 4276  
## 11 A\_23\_P23611 NM\_001008219 4365  
## 12 A\_23\_P350059 b 4687

#### HLSA genes in list of differentially expressed genes in FMR1-FM as determined by RankPod

## gene.name  
## 1 SMA5

#### HLSA genes in list of differentially expressed genes in dup15q as determined by SAM

## gene.name  
## 1 PDE4DIP

#### HLSA genes in list of differentially expressed genes in dup15q as determined by RankPod

## [1] gene.name  
## <0 rows> (or 0-length row.names)

## Hu, 2009 - Autism Research

### Sample Size: 116 cases, 116 controls

### RNA Source: LCL

### Analysis Technique: Microarry - TIGR 40K human arrays

### Other notes: Completed analysis on all autism cases vs. controls as well as each of three phenotypic sub-groups vs controls

### HLSA Genes differentially expressed when all autism cases compared to controls

## [1] gene.name  
## <0 rows> (or 0-length row.names)

### HLSA genes differentially expressed in each of the three sub-group comparisons (I think)

## gene.name  
## 1 GUSBP1  
## 2 SMA4

### HLSA genes significant in the 4 class analysis (don't know what "4 class analysis means")

## [1] gene.name  
## <0 rows> (or 0-length row.names)

## [1] GENE\_SYMBOL  
## <0 rows> (or 0-length row.names)

### HLSA enes differentially expressed in language impaired sub-group compared to controls at 5% FDR and at 0.1% FDR

#### 0.1% FDR

## gene.name  
## 1 RPL23AP7

#### 5% FDR

## gene.name  
## 1 BIRC1  
## 2 CCRL1  
## 3 MST1  
## 4 NBPF14  
## 5 NBPF3  
## 6 PDE4DIP  
## 7 SLC6A13  
## 8 SPTLC2

## GENE\_SYMBOL  
## 1 BIRC1  
## 2 CCRL1  
## 3 MST1  
## 4 NBPF14  
## 5 NBPF3  
## 6 PDE4DIP  
## 7 SLC6A13  
## 8 SPTLC2

### HLSA genes differentially expressed in mild symptom sub-group compared to controls

## gene.name  
## 1 DGCR9  
## 2 PDE4DIP

### HLSA genes differentially expressed in savant sub-group compared to controls

## [1] gene.name  
## <0 rows> (or 0-length row.names)

## Hu, 2009 - PLOS

### Sample Size: 21 cases and their 21 unaffected siblings. Cases are all males from the author defined "severe language impairement sub-group", all controls are male siblings

### RNA Source: LCL

### Analysis Technique: Microarry - custom array

## gene.name  
## 1 PDE4DIP

## Seno, 2010

### Sample Size: 20 cases and their 22 unaffected siblings

### RNA Source: LCL

### Analysis Technique: Microarry - Illumina HumanRef-8\_V3 gene expression arrays

## [1] gene.name  
## <0 rows> (or 0-length row.names)

## Luo, 2012

### Sample Size:

### RNA Source: LCL

### Analysis method: microarray

### Other Notes: Looked at changes in expression of genes within or near CNVs detected in ASD cases

#### HLSA genes within/nearby ASD case CNVs that are up or down-regulated

## gene.name  
## 1 AMY1A  
## 2 AMY2B  
## 3 DDX11  
## 4 E2F6  
## 5 GTF2I  
## 6 GUSB  
## 7 NBPF10  
## 8 NBPF20  
## 9 PARP8  
## 10 SMA4  
## 11 SRGAP2

## cnv.chrom cnv.start cnv.end gene  
## 1 chr1 120872731 121162446 SRGAP2  
## 2 chr1 104012569 104063231 AMY2B  
## 3 chr1 103941535 104094454 AMY1A  
## 4 chr12 9526879 9587954 DDX11  
## 5 chr12 9559647 9611502 DDX11  
## 6 chr12 31157554 31298174 DDX11  
## 7 chr5 49770866 49778751 PARP8  
## 8 chr2 11936601 11938595 E2F6  
## 9 chr7 65053082 65104000 GUSB  
## 10 chr7 65053082 65104000 GUSB  
## 11 chr1 147305744 147478120 NBPF20  
## 12 chr1 103907158 103961238 AMY1A  
## 13 chr1 103907158 103961238 AMY1A  
## 14 chr1 103941535 104094454 AMY1A  
## 15 chr5 68871012 70672298 SMA4  
## 16 chr1 17112560 17135964 NBPF10  
## 17 chr1 147305744 147849072 NBPF20  
## 18 chr12 9535285 9611502 DDX11  
## 19 chr1 103954623 103961238 AMY1A  
## 20 chr1 103955388 103961238 AMY2B  
## 21 chr5 70222045 70415222 SMA4  
## 22 chr7 73617066 73782113 GTF2I  
## 23 chr1 103954623 103961238 AMY1A

## Blumenthal, 2014

### Sample Size: 34 individuals from seven multiplex ASD families (4 deletion, 10 duplication, 20 controls)

### RNASource: LCLs from multiplex ASD cases

### Analysis method: RNA-Seq

## GeneInfo GeneID Chr Start Stop  
## 1 OR4F16/chr1:367658-180795226 OR4F16 chr1 367658 180795226  
## 2 OR4F3/chr1:367658-180795226 OR4F3 chr1 367658 180795226  
## 3 OR4F29/chr1:367658-180795226 OR4F29 chr1 367658 180795226  
## 4 SPTLC2/chr14:77972339-78083110 SPTLC2 chr14 77972339 78083110  
## 5 ARHGEF5/chr7:144052488-144077725 ARHGEF5 chr7 144052488 144077725  
## permutedPval\_Reciprocal NaturalLogBeta\_Reciprocal  
## 1 0.01360000 0.1702969  
## 2 0.01420000 0.1702969  
## 3 0.01500000 0.1702969  
## 4 0.01775873 0.1392901  
## 5 0.04565217 -0.2465684

## GeneInfo GeneID Chr Start Stop  
## 1 NUDT4/chr12:93771700-93797024 NUDT4 chr12 93771700 93797024  
## 2 OR4F16/chr1:367658-180795226 OR4F16 chr1 367658 180795226  
## 3 OR4F3/chr1:367658-180795226 OR4F3 chr1 367658 180795226  
## 4 NUDT4P1/chr12:93771745-93796052 NUDT4P1 chr12 93771745 93796052  
## 5 OR4F29/chr1:367658-180795226 OR4F29 chr1 367658 180795226  
## 6 OR2A9P/chr7:143947563-143997598 OR2A9P chr7 143947563 143997598  
## 7 NAIP/chr5:70264309-70320941 NAIP chr5 70264309 70320941  
## 8 OR2A20P/chr7:143947766-143997395 OR2A20P chr7 143947766 143997395  
## 9 ARHGEF5/chr7:144052488-144077725 ARHGEF5 chr7 144052488 144077725  
## 10 PDE4DIP/chr1:144851423-145076186 PDE4DIP chr1 144851423 145076186  
## 11 MST1P2/chr1:16972068-16976915 MST1P2 chr1 16972068 16976915  
## 12 DDX11L10/chr16:61554-64090 DDX11L10 chr16 61554 64090  
## permPval\_Dup FoldChange  
## 1 0.00360000 0.7454901  
## 2 0.00400000 1.3416684  
## 3 0.00400000 1.3416684  
## 4 0.00420000 0.7482999  
## 5 0.00460000 1.3416684  
## 6 0.01593027 0.5143211  
## 7 0.01617754 0.6250568  
## 8 0.01767677 0.5310899  
## 9 0.01800000 0.5869483  
## 10 0.03684210 1.3394626  
## 11 0.03783784 0.6157589  
## 12 0.04783599 0.5565200

## GeneInfo GeneID Chr Start Stop  
## 1 LOC646214/chr15:21932513-21940739 LOC646214 chr15 21932513 21940739  
## 2 SMA4/chr5:69434688-69586004 SMA4 chr5 69434688 69586004  
## 3 GTF2I/chr7:74072029-74175022 GTF2I chr7 74072029 74175022  
## 4 GUSBP3/chr5:68935289-69006272 GUSBP3 chr5 68935289 69006272  
## 5 DDX11/chr12:31226778-31257725 DDX11 chr12 31226778 31257725  
## 6 ARMC10/chr7:102715327-102740210 ARMC10 chr7 102715327 102740210  
## 7 SMA5/chr5:69776869-69881549 SMA5 chr5 69776869 69881549  
## 8 GUSBP2/chr6:26839265-26924333 GUSBP2 chr6 26839265 26924333  
## 9 GUSBP9/chr5:69812078-70555122 GUSBP9 chr5 69812078 70555122  
## 10 FAM72A/chr1:206138910-206155074 FAM72A chr1 206138910 206155074  
## 11 FAM72B/chr1:120839004-120855681 FAM72B chr1 120839004 120855681  
## 12 GTF2H2C/chr5:68856050-68888729 GTF2H2C chr5 68856050 68888729  
## permPval\_Del FoldChange  
## 1 0.02655203 0.7775047  
## 2 0.03100089 0.6888396  
## 3 0.03143713 0.8757981  
## 4 0.03270009 0.6978500  
## 5 0.03382187 0.7388957  
## 6 0.03438677 0.8506611  
## 7 0.03697183 0.6846112  
## 8 0.03728693 0.7395858  
## 9 0.04132231 0.6745483  
## 10 0.04200000 0.6682687  
## 11 0.04677060 0.6753670  
## 12 0.04761905 0.8001148

## Migliavacca, 2015

### Sample Size: 16p11.2 deletion & duplication carriers compared to controls

### RNA Source: LCLs

### Analysis method: microarray - Affymetrix GeneChips Human Genome U133+ PM 24 array plates

### Other notes: analyzied 16p11.2 duplication and deletion carriers and controls. **Note, some of their results were non-gene specifici (e.g could map to multiple NBPF genes). In order to perform the analysis in R, I had to split up each of these individual genes, so for example, multiple NBPF genes may show up as "hits" when in fact the hit only applied to one or a subset of the genes - but the mircroarray probe couldn't distinguish between them** \*

# Studies of differential expression in whole blood

## Glatt, 2012

### Sample Size:

### RNA Source: peripheal blood mononculear cells isolated from whole blood

### Analysis method: mircoarray

## gene.name  
## 1 USP10

## Kong, 2012 - PLOS One

### Sample Size: 170 ASD cases and 115 age/sex-matched controls

### RNA Source: whole blood

### Analysis method: Affymetrix HG-U133 Plus 2.0 and Affymetrix Gene 1.0 ST

### Other notes: two different cohorts analyzed; P1 (66 ASD and 33 controls) and P2 (104 ASD and 82 controls)

### HLSA genes in list of differentially expressed genes in "P1" cohort

## gene.name  
## 1 ACTR2  
## 2 PARP8  
## 3 ROCK1

#### HLSA genes in list of differentially expressed genes in "P2" cohort

## [1] gene.name  
## <0 rows> (or 0-length row.names)

#### HLSA genes in list of genes used as prediction model

## gene.name  
## 1 ROCK1

## Kong, 2013 - Neurogenetics

### Sample Size: 20 probands and their unaffected siblings

### RNA Source: whole blood

### Analysis method: Microarray - Affymetrix Gene 1.0 ST

## gene.name  
## 1 ANAPC1

## Campbell, 2013 \*\* Need to double check details of RNASource, and other details \*\*\* ### Sample Size: ### RNA Source: Blood (I think) ### Analysis method: microarray ### Other notes: I think this study used the same expression data as in Luo, 2012 - resutls may be slightly different because they sub-divided full data set into a "neurogenesis group", but results of this study may not be independent from others (Luo, 2012)

# Analyses of differentially expressed genes in iPS cells from ASD cases and controls that were coaxed into being neuronal-like structures

## Mariani, 2015

### Sample Size:

### RNA Source: iPSC generated neural structures

### Analysis method: RNA-seq

### Other notes: analyzed differentially expressed genes at two time-points in the development of their neural structures

#### HLSA genes in list of diferrentially expressed genes at Day 11

## gene.name study RNAsource  
## 1 GUSBP4 mariani.15.iPSC.neural.structures iPSC.neural  
## 2 TEKT4 mariani.15.iPSC.neural.structures iPSC.neural

#### HLSA genes in list of differentially expressed genest at Day 31

## gene.name study RNAsource  
## 1 ARHGEF35 mariani.15.iPSC.neural.structures iPSC.neural  
## 2 ARHGEF5 mariani.15.iPSC.neural.structures iPSC.neural  
## 3 CELSR2 mariani.15.iPSC.neural.structures iPSC.neural  
## 4 GUSBP4 mariani.15.iPSC.neural.structures iPSC.neural

# Analyses of differential expression in cells with knock-out/knock-down of known autism genes

# CHD8 knockdown/knockout

## Wang, 2015

### Sample Size:

### RNA Source: Neurons and neural progenitor cells

### Analysis method:

### Other notes: Analyzed changes in gene expression in CDH8 hemizygous knockout neurons and neural progenitor cells

#### HLSA genes in list of differentially expressed genes in neurons with p-value <= 0.1

## gene.name  
## 1 NBPF10  
## 2 FGF7  
## 3 PAK2  
## 4 PARP8  
## 5 GTF2I  
## 6 NBPF14  
## 7 NBPF9  
## 8 USP10  
## 9 NBPF20  
## 10 PAIP1  
## 11 CELSR2  
## 12 NUDT4

#### HLSA genes in list of differentially expressed genes in neural progenitor cells with p-value <= 0.1

## gene.name  
## 1 CELSR2  
## 2 FGF7  
## 3 NBPF14  
## 4 PARP8  
## 5 NBPF10  
## 6 NBPF9  
## 7 NBPF3  
## 8 DDX11  
## 9 NUDT4

## Cotney, 2014

### Sample Size:

### RNA Source:

### Analysis method:

### Other Notes: Analyzed gene expression following knockdown of CHD8 with two different shRNAs ("C" and "G")

#### HLSA genes in list of differentially expressed genes when CHD8 knocked down with shRNA "C"

## gene.name  
## 1 NBPF10

#### HLSA genes in list of differentially expressed genes when CHD8 knocked down with shRNA "G"

## gene.name  
## 1 WASH2P

## Wilkinson, 2015

### Sample Size:

### RNA Source:

### Analysis method:

### Other Notes: Knocked down CHD8 expression with siRNA

#### HLSA genes in list of coding genes differentially expressed after CHD8 knockdown with siRNA

## gene.name  
## 1 ABCB10  
## 2 E2F6  
## 3 FAM3C  
## 4 GTF2I  
## 5 PAIP1  
## 6 WASH1

#### HLSA genes in list of non-coding genes differentially expressed after CHD8 knockdown with siRNA

## gene.name  
## 1 LOC645166  
## 2 NUDT4P1  
## 3 TEKT4P2  
## 4 WASH5P

# Analysis of FMRP targets/differential expression

## Darnell, 2011

### In MOUSE cells, they performed sequencing of RNAs bound to FMRP (cross-linking immunoprecipitation - CLIP)

## gene.name  
## 1 ANAPC1  
## 2 CELSR2  
## 3 PDE4DIP

## Ascano, 2012

### What they did: determined RNAs bount to FMRP in human HEK293 cells

## Gene Reads.FMR1.iso1 Reads.FMR1.iso.7 Reads.FXR1 Reads.FXR2  
## 1 SPTLC2 1444 330 31 55  
## 2 CDC27 916 139 13 85  
## 3 PDE4DIP 728 249 6 135  
## 4 CELSR2 630 1280 NA 49  
## 5 RFWD3 565 358 47 184  
## 6 ABCB10 289 183 24 43  
## 7 SRP68 236 132 7 75  
## 8 PAK2 149 40 NA 48  
## 9 ROCK1 102 78 NA 34  
## 10 SRGAP2 90 150 NA 9  
## 11 ACTR2 85 20 NA 5  
## 12 USP10 81 71 5 26  
## 13 ANAPC1 69 7 5 23  
## 14 NEK2 60 5 5 22  
## 15 NBPF14 35 7 NA 13  
## 16 ARMC10 30 32 NA NA  
## 17 NBPF3 30 10 NA NA  
## 18 NBPF9 24 5 NA NA  
## 19 E2F6 17 NA NA NA  
## 20 PAIP1 16 24 13 11  
## 21 NBPF20 13 NA NA NA  
## 22 GTF2I 11 5 NA 10  
## 23 FAM3C 7 NA NA NA  
## 24 NBPF10 7 14 NA 6  
## 25 NBPF15 6 7 NA NA  
## 26 DDX11 5 40 NA NA  
## 27 TEKT4P2 5 22 NA NA  
## 28 ARHGEF35 NA NA NA NA  
## 29 ARHGEF5 NA 22 NA 5  
## 30 CDH12 NA NA NA NA  
## 31 FAM72B NA NA NA NA  
## 32 GUSB NA 7 NA NA  
## 33 NUDT4 NA NA NA NA  
## 34 PARP8 NA 33 NA NA  
## 35 WDR70 NA 57 NA NA

## Summary of all gene expression hits