PGP-UK Genomics Report for NA12878wxs

1 Summary

This is the genome report was produced using collaborative research tools, including SNPedia and GetEvidence. This section shows an overview of all the small variants which were found in the genome for this individual, when compared with a reference genome. These variants are summarised in Table 1 and the pie-charts in Figures 2, 3 and 4.

This report was generated automatically and is not clinically approved. It is provided for personal and research purposes only.

This document contains hyperlinks, shown in grey, that will take you to external websites where you can find more detailed explanations. Some of the technical terms are also explained in more detail in the Ensembl Glossary. We would welcome your feedback about this report, for example, if you would like more information about anything or if any of the links have become inactive. You can contact us on: pgp-uk@ucl.ac.uk.

This summary shows an overview of all the variants which were found in the genome for this individual. The "variants remaining after filtering" refers to any differences in the DNA identified when compared to the reference genome. Of these, the majority will have already been found in some other sequenced individual and put on a database (existing variants) while others have not yet been annotated (novel variants).

"Overlapped genes" refers to the number of times where a variant was found in a region of the genome containing a gene. The diagram in Figure 1 is a simplification of the usual gene structure. "Exon" refers to the part of the gene which goes on to form a protein, and variants in this part of the gene are more likely to cause changes in the shape of the protein. Upstream, downstream, intronic and intergenic variants are more likely to alter the regulation of that gene but will not change the protein itself.

A transcript for a protein-coding gene can include the exons, introns and other gene features that are transcribed and important for gene function but might not be translated into the final protein. Not all transcripts are for protein-coding genes, with many containing non-coding RNAs that can be overlapping other genes, in introns or in intergenic regions.

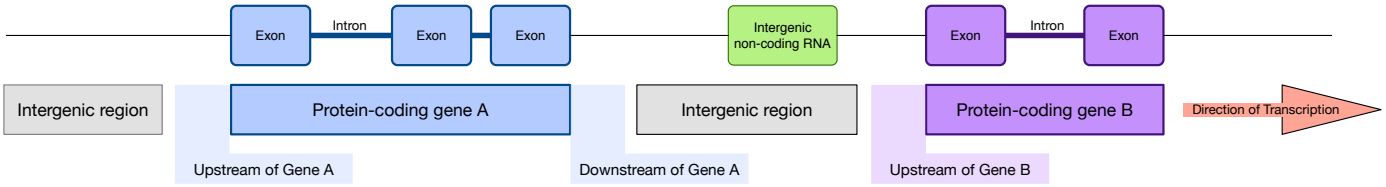

Figure 1: Diagram of gene structure indicating locations of potential variants

2 Ancestry

This plot shows the distribution of the genomes of different populations. Data from several studies which used whole genome sequencing was used to see the relationships between the genomes of the populations. It shows how closely related certain populations are genetically: Groups which cluster closely are more genetically similar than groups which are further apart. The black star symbol shows where this PGP-UK participant sits in relation to other populations, indicating their ancestry and their most closely related populations according to genetic sequence.

Please note that this analysis is limited by the populations available in the 1000 genomes project (1kGP) data. If there are European subpopulations reported, and the ancestry of the participant does not correspond to any of the 1kGP populations, the closest 1kGP sampled subpopulation will be shown (even though it might be different from the participant's actual ancestry).

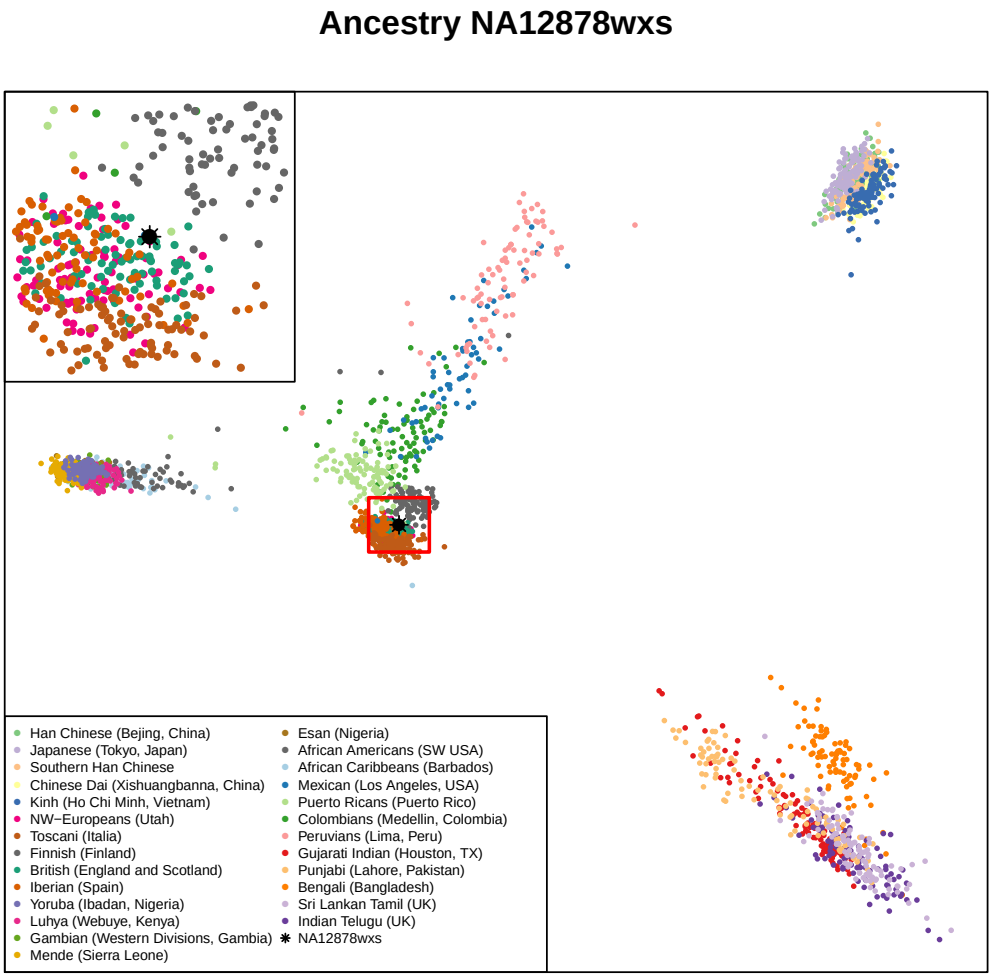

Figure 2: Ancestry Principal Component Analysis

3 Traits (based on SNPedia information)

Existing research has associated many variants with phenotypic traits, some of which can be perceived as beneficial while others appear to have a harmful effect. Some traits are complex and can be affected by several variants. It is likely that some of these would confer a higher risk while others a lower risk of trait manifestation. These can not be combined linearly to produce an actual risk of disease.

It is important to note that in most cases genomic data is probabilistic, not deterministic- i.e. having a genetic predisposition for a disease is not a diagnosis; rather, it shows an increased likelihood of developing that disease. Also, one person can have both potentially beneficial and harmful variants in the same gene, or associated with the same disease.

Some variants can also affect certain populations more, or will only affect a particular gender. For example, a variant for higher risk of endometriosis in the sequence of a male will not directly affect that person, but can be passed on to descendants.

While many traits are the result of a unique variant, many are the combination of several variants throughout the genome. In SNPedia, these are called genosets. These can integrate some of the information already present in the single variant tables, or be the combination of variants that have no phenotypic effect on their own, but contribute to a trait when together.

The variants in the following tables are sorted by magnitude. This is an subjective measure defined in SNPedia to highlight the perceived importance of the genotype described. At the moment this scale goes from 0 to 10. You can read more about it by visiting their explanatory webpage.

As our knowledge grows, the interpretation of the effect of certain variants might change. Clicking on the links in the genome report tables will take you to websites containing more information about each variant.

3.1 Possibly Beneficial Traits

Mag.	Identifier	Genotype	Summary	GnomAD	GetEvidence	ClinVar
3	rs1042522	(C;C)	Live 3 years longer. Chemotherapy is more effec...	Link	Link	Link
3	rs925391	(T;T)	Unlikely to go bald	Link		
2.5	rs3764261	(T;T)	Associated with higher HDL cholesterol. HDL	Link	Link	Link
2.4	rs2802288	(A;A)	Longer lifespan	Link		
2.2	rs2511989	(A;A)	0.44x decreased age-related macular degeneratio...	Link	Link	
2.1	rs6505162	(A;A)	0.43x decreased risk for esophageal cancer	Link		
2	rs10468017	(C;T)	Associated with higher HDL cholesterol	Link	Link	
2	rs10936599	(C;C)	Longer telomeres: longer life?	Link		Link
2	rs11045585	(A;A)	24% chance (lower than average) of docetaxel-in...	Link	Link	
2	rs1128535	(G;G)	Reduced risk (0.77x) for Crohn's disease	Link		
2	rs1160312	(G;G)	Reduced risk of Baldness.	Link	Link	
2	rs1501299	(A;C)	Slightly lower risk of breast cancer	Link		
2	rs1544410	(G;G)	Decreased risk of low bone mineral density diso...	Link	Link	
2	rs174537	(T;T)	Lower LDL-C and total cholesterol	Link		
2	rs1864163	(G;G)	Associated with higher HDL cholesterol	Link	Link	
2	rs2073963	(T;T)	Reduced risk of baldness	Link		
2	rs2292813	(T;T)	Decreased risk of autism	Link		
2	rs2542052	(C;C)	Better odds of living to 100	Link		
2	rs261332	(A;A)	Associated with higher HDL cholesterol	Link		
2	rs37973	(A;A)	Possibly better response to inhaled corticoster...	Link		Link
2	rs4149268	(G;G)	Associated with higher HDL cholesterol	Link	Link	
2	rs6511720	(G;T)	Slightly lower odds of developing CHD.	Link	Link	Link
2	rs6807362	(G;G)	Decreased autism risk	Link	Link	
2	rs7216389	(C;C)	0.69x lower risk of Childhood Asthma.	Link	Link	
2	rs763110	(T;T)	~0.80x reduced cancer risk	Link		Link
2	rs7776725	(T;T)	Stronger bones	Link	Link	
2	rs801114	(T;T)	0.78x decreased Basal Cell Carcinoma risk.	Link	Link	

Mag.	Identifier	Genotype	Summary	GnomAD	GetEvidence	ClinVar
2	rs9642880	(G;G)	Slightly lower risk of Bladder Cancer.	Link	Link	
1.8	rs1800588	(C;T)	Higher HDL-C levels	Link	Link	
1.8	rs266729	(C;G)	0.73x decreased risk for colorectal cancer	Link	Link	
1.8	rs4714156	(C;C)	<0.61x risk for restless legs	Link		
1.8	rs6897932	(C;T)	0.91x decreased risk for multiple sclerosis	Link	Link	Link
1.8	rs854560	(T;T)	0.5x lower risk of ovarian cancer	Link	Link	Link
1.6	rs1061170	(T;T)	Lower risk for AMD: generally longer live than ...	Link	Link	Link
1.6	rs10801935	(C;C)	0.3x decreased risk of breast cancer	Link		
1.5	rs1050631	(C;C)	Mean Survival Time of 32 months for esophageal ...	Link		
1.5	rs1063192	(C;C)	0.71x reduced risk of myocardial infarction	Link		
1.5	rs11136000	(T;T)	0.84x decreased risk for Alzheimer's disease	Link	Link	
1.5	rs17367504	(G;G)	Reduction in blood pressure	Link	Link	
1.5	rs2229169	(C;C)	1.5x decreased risk of heart attack and stroke ...	Link		
1.5	rs3790844	(C;T)	Slightly reduced risk (0.77x) for pancreatic ca...	Link		
1.5	rs3851179	(A;G)	0.85x decreased risk for Alzheimer's disease	Link	Link	
1.5	rs4149274	(C;C)	Associated with higher HDL (good) cholesterol.	Link		
1.5	rs4939883	(C;C)	Associated with higher HDL cholesterol	Link	Link	
1.5	rs610932	(A;A)	A allele associated with reduced risk of Alzhei...	Link		
1.4	rs1165205	(A;T)	0.85x decreased gout risk	Link	Link	
1.4	rs6495446	(C;T)	0.8x reduced risk for chronic kidney disease	Link		
1.2	rs4867568	(T;T)	Decreased risk for knee osteoporosis	Link		
1.1	rs2235040	(A;G)	Possibly higher chances of remission only for i...	Link	Link	
1.1	rs2293347	(G;G)	Among NSCLC patients: better Gefitinib response...	Link		Link
1	rs182549	(T;T)	Can digest milk.	Link		Link
1	rs2952768	(C;C)	Less drug dependence: decreased effectiveness o...	Link		Link
1	rs4148739	(G;G)	Possibly: inpatients more likely to remit on ce...	Link	Link	
1	rs4939827	(C;C)	0.73x decreased risk for colorectal cancer	Link	Link	Link
1	rs7850258	(A;G)	Typical odds of developing primary hypothyroidi...	Link		
1.0	rs11246226	(C;C)	Decreased risk of schizophrenia in limited stud...	Link	Link	
0.1	rs1726866	(C;C)	Can taste bitter	Link	Link	Link

3.2 Possibly Harmful Traits

Mag.	Identifier	Genotype	Summary	GnomAD	GetEvidence	ClinVar
3.2	rs2981582	(T;T)	1.7x higher risk of ER+ breast cancer	Link	Link	
3.1	rs1421085	(C;C)	~1.7x increased obesity risk	Link	Link	Link
3.1	rs4143094	(T;T)	Higher (by 39%) risk for colorectal cancer corr...	Link		
3	rs1121980	(T;T)	Moderate increase (2.76x) in risk for obesity	Link	Link	
3	rs13266634	(C;C)	Increased risk for type-2 diabetes	Link	Link	Link
3	rs16969968	(A;A)	Higher risk for nicotine dependence: lower risk...	Link	Link	Link
3	rs2237717	(T;T)	Reduced abilities related to neurocognition and...	Link		
3	rs55705857	(A;G)	6x increased risk of glioma of IDH1/IDH2 subtyp...	Link		
3	rs6920220	(A;G)	1.2x risk Rheumatoid Arthritis	Link	Link	
3	rs891512	(A;A)	Higher blood pressure than G;G	Link		Link
2.5	rs11190870	(T;T)	Possibly even more increased risk of scoliosis	Link		
2.5	rs12536657	(A;A)	Hypermetropia risk - longsightedness	Link		
2.5	rs1800629	(A;A)	Complex; generally higher risk for certain dise...	Link	Link	Link
2.5	rs187238	(G;G)	Hypertension increases risk 3.75x for sudden ca...	Link		
2.5	rs324420	(A;A)	Significantly increased risk for substance use ...	Link	Link	Link
2.5	rs5219	(T;T)	2.5x increased risk for type-2 diabetes	Link	Link	Link
2.5	rs613872	(G;T)	~5 fold higher risk for Fuchs' dystrophy: a cor...	Link		
2.5	rs6441286	(G;G)	3.08x chance of developing primary biliary cirr...	Link	Link	
2.5	rs664143	(C;T)	Higher risk for number of cancers	Link		
2.2	rs944289	(T;T)	1.69x increased thyroid cancer risk	Link	Link	
2.1	rs10811661	(T;T)	1.2x increased risk for type-2 diabetes	Link	Link	
2.1	rs1360780	(T;T)	1.3x increased risk for depression	Link	Link	Link
2.1	rs2306402	(C;C)	1.18x increased risk for late-onset Alzheimer's...	Link		
2.1	rs2572886	(A;A)	1.4x increased risk of HIV infection	Link		
2.1	rs3746444	(C;C)	~1.2x increased risk for cancer	Link		
2.1	rs4363657	(C;T)	4.5x increased myopathy risk for statin users	Link	Link	
2.1	rs5751876	(T;T)	Significantly higher anxiety levels after moder...	Link		
2.1	rs795484	(A;G)	Increased morphine dose requirement and postope...	Link		
2	rs10096097	(G;G)	Increased Anorexia Nervosa risk	Link		
2	rs10492519	(G;G)	Increased risk of developing prostate cancer	Link		
2	rs1050152	(C;T)	2.1x increased risk of Crohn's disease	Link	Link	Link
2	rs1051730	(C;T)	1.3x increased risk of lung cancer	Link	Link	Link
2	rs10883365	(G;G)	1.62x increased risk for developing Crohn's dis...	Link	Link	
2	rs10889677	(C;C)	Baseline (average) risk for certain autoimmune ...	Link	Link	
2	rs10984447	(A;A)	>1.17x increased risk for multiple sclerosis	Link	Link	
2	rs110419	(A;A)	1.7x increased risk for neuroblastoma	Link		
2	rs11123857	(G;G)	2.88-fold risk of bipolar disorder or major dep...	Link		
2	rs1143699	(C;C)	In men: 2.19x risk of type 2 diabetes	Link		
2	rs1219648	(A;G)	1.20x risk for breast cancer	Link	Link	
2	rs1223271	(A;A)	Increased risk of developing Parkinson's Diseas...	Link	Link	
2	rs12431733	(T;T)	Increased risk of developing Parkinson's Diseas...	Link	Link	
2	rs1265181	(C;G)	Increased risk for psoriasis	Link	Link	
2	rs1333048	(A;C)	1.3x increased coronary artery disease risk	Link		
2	rs1361600	(G;G)	~2x increased risk for adult-onset asthma in Ja...	Link		
2	rs16942	(A;G)	Very slightly increased breast cancer risk	Link	Link	Link
2	rs1734791	(A;A)	1.4x increased risk for lupus	Link		
2	rs17782313	(C;T)	Adults likely to be 0.22 BMI units higher	Link	Link	Link
2	rs1800896	(A;G)	1.6x increased prostate cancer risk	Link		
2	rs1994090	(G;G)	Increased risk of developing Parkinson's Diseas...	Link	Link	
2	rs2201841	(T;T)	2.4x increased risk for Graves' disease	Link	Link	
2	rs2230201	(G;G)	>1.4x risk of lupus	Link		Link
2	rs2305795	(A;G)	1.28x higher risk of narcolepsy compared to (G;...	Link		Link
2	rs2383206	(A;G)	1.4x increased risk for heart disease	Link		

Mag.	Identifier	Genotype	Summary	GnomAD	GetEvidence	ClinVar
2	rs2383207	(A;G)	Increased risk for heart disease	Link		
2	rs2420946	(C;T)	1.20x risk for breast cancer	Link		
2	rs25487	(A;G)	2x higher risk for skin cancer; possibly other ...	Link	Link	Link
2	rs2736990	(C;C)	Slightly increased risk of developing Parkinson...	Link	Link	
2	rs27388	(A;A)	Increased risk of developing schizophrenia	Link		
2	rs2908004	(C;C)	Weaker bones	Link	Link	
2	rs3197999	(T;T)	1.2x risk of Crohn's	Link	Link	
2	rs326	(A;A)	Lower HDL cholesterol	Link	Link	Link
2	rs358806	(C;C)	1.78x increased risk of developing Type-2 diabe...	Link	Link	
2	rs3738579	(C;C)	0.6x decreased risk for cervical cancer: but 1....	Link		
2	rs3738919	(A;C)	1.94x risk of developing rheumatoid arthritis	Link		
2	rs3745516	(A;A)	Increased risk of developing primary biliary ci...	Link		
2	rs3775948	(G;G)	Slightly higher risk for gout	Link		
2	rs3793784	(C;G)	1.5x risk for ARMD	Link	Link	Link
2	rs4027132	(A;A)	1.51x increased risk of developing bipolar diso...	Link		
2	rs4420638	(A;G)	~3x increased Alzheimer's risk; 1.4x increased ...	Link	Link	Link
2	rs4825476	(G;G)	1.9x higher risk of suicidal thoughts when taki...	Link	Link	
2	rs493258	(A;G)	1.15x risk of Age Related Macular Degeneration	Link		
2	rs5174	(A;A)	1.3x increased risk for heart disease	Link	Link	Link
2	rs6457617	(C;T)	2.3x risk of rheumatoid arthritis	Link	Link	
2	rs6896702	(T;T)	Increased risk of developing Parkinson's Diseas...	Link		
2	rs6997709	(G;T)	1.2x higher risk for hypertension	Link		
2	rs7442295	(A;A)	~4x higher risk for hyperuracemia	Link	Link	Link
2	rs7794745	(A;T)	Slightly increased risk for autism	Link	Link	Link
2	rs7807268	(C;G)	1.3x risk for Crohn's disease	Link	Link	
2	rs800292	(C;C)	5% higher risk of Age related macular degenerat...	Link	Link	Link
2	rs828907	(T;T)	Increased risk of bladder cancer and 2x risk of...	Link		
2	rs9525638	(T;T)	Weaker bones	Link		
2	rs965513	(A;G)	1.77x increased thyroid cancer risk	Link	Link	
2.0	rs2156921	(G;G)	1.29x increased risk for depression	Link		
1.9	rs7923837	(A;G)	1.6x risk for T2D	Link		
1.8	rs2278206	(T;T)	1.16x increased risk for asthma	Link	Link	
1.7	rs10181656	(C;G)	1.7x increased SLE risk	Link		
1.6	rs1260326	(T;T)	Slightly higher risk for gout	Link	Link	Link
1.6	rs1978237	(C;G)	1.59x risk of Type 2 diabetes	Link		
1.6	rs2736100	(G;G)	1.6x higher risk for glioma development	Link	Link	Link
1.6	rs356219	(G;G)	1.6x increased risk for Parkinson's disease	Link		
1.6	rs3764880	(A;A)	1.2 - 1.8x increased tuberculosis risk	Link	Link	
1.6	rs7234029	(G;G)	Slightly increased (1.6x) risk for Crohn's dise...	Link		
1.5	rs10509681	(C;T)	Increased risk of GI bleeding with NSAIDs	Link	Link	Link
1.5	rs10859871	(A;C)	Slight (~1.2x) increase in endometriosis risk	Link		
1.5	rs1154155	(G;T)	1.94x increased risk for narcolepsy	Link	Link	
1.5	rs12037606	(A;G)	1.22x risk of developing Crohn's disease	Link		
1.5	rs12469063	(A;G)	Slightly increased risk of developing restless ...	Link		
1.5	rs12498742	(A;A)	1.25 increased risk for gout	Link		
1.5	rs13149290	(C;C)	Slightly increased risk of developing prostate ...	Link		
1.5	rs1375144	(C;C)	1.59x increased risk of developing bipolar diso...	Link		
1.5	rs140701	(A;A)	Increased risk for anxiety disorders	Link		
1.5	rs165599	(G;G)	May indicate increased susceptibility to schizo...	Link	Link	
1.5	rs16944	(A;G)	Minorly increased risk of mental illness and os...	Link	Link	
1.5	rs1801020	(T;T)	1.31x increased risk of heart disease	Link		Link
1.5	rs1801274	(C;T)	Complex; generally greater risk for cancer prog...	Link	Link	Link
1.5	rs2007153	(G;G)	Increased risk of schizophrenia in limited stud...	Link		
1.5	rs2076295	(G;G)	Slightly increased risk for pulmonary fibrosis ...	Link		
1.5	rs2240340	(A;A)	Slightly increased (1.5x) risk for RA	Link		

Mag.	Identifier	Genotype	Summary	GnomAD	GetEvidence	ClinVar
1.5	rs2241880	(C;T)	1.4x increased risk for Crohn's disease in Cauc...	Link	Link	Link
1.5	rs2254958	(C;C)	1.61x reported increased risk for Alzheimer's; ...	Link		
1.5	rs2272127	(C;C)	Associated with herpes and schizophrenia	Link		
1.5	rs2305089	(T;T)	Higher risk for chordoma reported in one study;...	Link	Link	
1.5	rs2881766	(T;T)	Slightly increased risk for pregnancy-induced h...	Link		
1.5	rs3087243	(G;G)	Increased risk for autoimmune diseases	Link	Link	Link
1.5	rs3212227	(A;A)	1.43x increased risk of developing psoriasis an...	Link		Link
1.5	rs356220	(T;T)	Increased risk of Parkinson's Disease	Link		
1.5	rs3790565	(C;T)	Slightly increased risk of developing primary b...	Link		
1.5	rs3814570	(C;T)	1.3x increased risk for Crohn's disease with il...	Link		
1.5	rs3825776	(A;G)	1.3x increased risk for ALS	Link	Link	
1.5	rs393152	(A;A)	Increased risk of both PD and AD	Link	Link	
1.5	rs401681	(C;T)	~1.2x increased risk for several types of cance...	Link	Link	
1.5	rs464049	(T;T)	Increased risk of schizophrenia in limited stud...	Link		
1.5	rs4785763	(A;A)	2x higher risk for melanoma	Link	Link	
1.5	rs486907	(A;G)	1.5x increased prostate cancer risk	Link	Link	Link
1.5	rs4982731	(C;C)	Possible higher risk of childhood acute lymphob...	Link		
1.5	rs5746059	(A;G)	Slightly higher fat mass	Link		
1.5	rs6498169	(A;G)	1.14x risk of multiple sclerosis	Link	Link	
1.5	rs6710341	(A;G)	Slightly increased risk of developing restless ...	Link		
1.5	rs7341475	(G;G)	1.58x increased schizophrenia risk for women	Link	Link	
1.5	rs7774434	(C;T)	Slightly increased risk of developing primary b...	Link		
1.5	rs9561778	(G;T)	~2x increased risk of adverse drug reactions fr...	Link	Link	
1.5	rs9652490	(A;G)	Slightly increased risk of developing Parkinson...	Link	Link	
1.5	rs995030	(G;G)	Non-protective against testicular cancer	Link	Link	
1.4	rs1545843	(A;A)	1.4x increased risk for depression (for those u...	Link		
1.4	rs2228314	(C;G)	1.48x risk of osteoarthritis	Link	Link	
1.4	rs4977756	(G;G)	1.93x higher risk for glioma development	Link	Link	
1.4	rs8050136	(A;A)	1.4x increased risk for T2D in some populations...	Link	Link	
1.34	rs17465637	(C;C)	1.34x higher risk for myocardial infarction	Link	Link	
1.3	rs10947262	(C;C)	1.3x increased risk for osteoarthritis	Link		
1.3	rs1434536	(A;G)	1.29x increased breast cancer risk	Link		Link
1.3	rs1746048	(C;C)	1.03 increased risk for coronary heart disease	Link	Link	
1.3	rs2024513	(A;G)	1.3x higher risk for schizophrenia (among Han C...	Link		
1.3	rs34330	(T;T)	1.2x higher breast cancer risk; 1.3x higher ris...	Link		Link
1.3	rs4295627	(G;T)	1.36x higher risk for glioma development	Link	Link	
1.3	rs4712653	(C;T)	Very slightly (~1.3x) increased risk for neurob...	Link		
1.3	rs9858542	(A;A)	1.8x risk of Crohn's disease	Link	Link	
1.25	rs748404	(T;T)	Slightly increased risk (1.25) for lung cancer...	Link	Link	
1.2	rs10210302	(C;T)	1.2x increased risk for Crohn's disease	Link	Link	
1.2	rs12050604	(A;A)	Slightly increased risk for lung cancer	Link		
1.2	rs1344706	(T;T)	1.2x increased risk for schizophrenia	Link	Link	
1.2	rs3131296	(A;G)	1.2x increased risk for schizophrenia	Link	Link	
1.2	rs3176336	(T;T)	Slightly higher (1.25x) higher risk for breast ...	Link		
1.2	rs35677470	(A;G)	2x higher risk for scleroderma	Link	Link	
1.2	rs3818361	(T;T)	1.2x increased risk for late-onset Alzheimer's	Link		
1.2	rs4795067	(A;G)	Slight increase in risk for psoriatic arthritis...	Link		
1.2	rs7514229	(G;G)	Associated with early-onset autoimmune thyroid ...	Link		
1.2	rs851715	(A;A)	Risk of nonsense-word repetition problems if sp...	Link		
1.1	rs11650354	(C;T)	Possible risk for allergic asthma	Link		
1.1	rs2295190	(G;T)	Slightly increased risk for ovarian cancer in w...	Link	Link	Link
1.1	rs249954	(C;T)	Potentially increased risk of Breast Cancer	Link		Link
1.1	rs34516635	(G;G)	Less longevity for Ashkenazi Jewish women.	Link		Link
1.1	rs6707530	(G;G)	In colorectal cancer: may allow cancer cells to...	Link		
1.1	rs7171755	(A;A)	Very slight descrease in cortical thickness and...	Link		

Mag.	Identifier	Genotype	Summary	GnomAD	GetEvidence	ClinVar
1.1	rs7531806	(A;G)	Very slightly increased risk of acne occurrence...	Link		
1.1	rs889312	(A;C)	Very slightly higher risk for breast cancer	Link	Link	
1.07	rs2291834	(C;C)	Very slightly higher risk for myocardial infarc...	Link		
1	rs1004819	(C;C)	1.5x risk of Crohn's disease: 1.2 for developin...	Link	Link	
1	rs11206244	(C;T)	Slight risk of decreased thyroid hormone metabo...	Link		
1	rs1143674	(A;A)	1.3x increased autism risk	Link		
1	rs2282679	(A;C)	Somewhat lower vitamin D levels	Link		
1	rs2435357	(A;A)	Slightly higher (2x?) risk for Hirschsprung dis...	Link		Link
1	rs2546890	(A;A)	Higher risk of multiple sclerosis	Link		
1	rs6166	(G;G)	Females slightly more likely to be sterile	Link	Link	Link
1	rs6932590	(T;T)	1.1x increased risk for schizophrenia	Link	Link	
1	rs6974491	(A;G)	Higher risk of coeliac and/or inflammatory bowe...	Link		
1	rs987525	(A;C)	2.5x increased risk for cleft lip	Link	Link	
0.1	rs11110912	(C;G)	Maybe some quite minor increase in high blood p...	Link		
0.1	rs2070744	(C;C)	Increased prostate cancer risk	Link	Link	Link
0.1	rs2304256	(C;C)	1.6x increased risk for SLE	Link	Link	Link
0.1	rs3095870	(G;G)	1.7x increased risk for SLE (lupus)	Link		
0.1	rs3748079	(G;G)	1.9x increased risk for SLE (lupus)	Link		

3.3 Genosets (Multi-variant Phenotypes)

Magnitude	Identifier	Summary
4	gs145	Female
2.7	gs311	Slow metabolizer of certain substances
2.5	gs155	CYP3A5 non-expressor
2.5	gs197	Increased type-2 diabetes risk
2.5	gs242	Increased risk of individuals with prostate can...
2.5	gs259	Homozygous for eye color haplotype #3
2.4	gs297	Lower heart attack risk than average
2.3	gs255	Homozygous eye color haplotype #1
2.2	gs280	Light hair color for europeans
2	gs101	Probably able to digest milk
2	gs104	Restless legs syndrome risk
2	gs129	Unable to classify the ABO blood type
2	gs156	NAT2 Rapid metabolizer.
2	gs159	CYP1A2 fast metabolizer
2	gs194	Myocardial Infarction Risk
2	gs239	Reduced conversion of beta-carotene to retinol
2	gs290	You might have two short form 5-HTTLPR.
2	gs313	Normal DPYD activity and thus 5-FU metabolism p...
2	gs317	Parkinson's risk might be decreased depending u...
1.7	gs234	Possible high pain sensitivity; HPS/HPS or HPS/...
1.5	gs1001	Mitochondrial Haplogroup H
1.5	gs186	HLA-B*5801ââ heterozygosity is possible: un...
1.5	gs247	Parkinson's Disease Risk
1.2	gs184	Able to taste bitterness.
1	gs182	CYP2D6*39

4 Report Metadata

Resource	Version	Website
Genome	GRCh38	Link
BWA	0.7.12	Link
SAMtools	1.3	Link
GATK	3.4-46	Link
PLINK	v1.90b3.35	Link
SNPedia	02-May-2019	Link
GnomAD	v2.1.1	Link
GetEvidence	10-May-2019	Link
ClinVar	10-May-2019	Link

Table 4: Analysis Pipeline Versions

Report generated on February 18, 2023.