PGP-UK Genomics Report for uk187685

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.

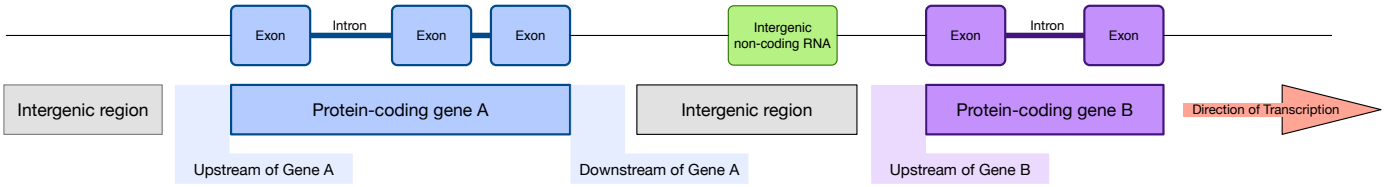

Figure 1: Diagram of gene structure indicating locations of potential variants

Feature	Count
Lines of input read	4677894
Variants filtered out	3713803
Novel / existing variants	0 (0.0) / 964091 (100.0)
Overlapped genes	51815
Overlapped transcripts	59265
Overlapped regulatory features	46752

Table 1: Variant calling summary

There are several different types of genomic variants. The most common change is when one single building block of the DNA (called a nucleotide) is changed, called a single nucleotide variants (SNV). Other variant types include insertions, where the DNA in the individual is longer than the reference sequence due to the insertion of one or more nucleotides; and deletions, where a few nucleotides are missing compared to the reference sequence.

Some of these changes will have no effect on the protein, while some changes may alter the protein function to varying degrees. The PolyPhen analysis software attempts to quantify the effect each mutation will have on the protein function. This ranges from "benign" where no change to the protein function is expected, to "probably damaging" where it is predicted that the mutation will affect protein function. It is nevertheless important to note that what is "damaging" for the protein is not necessarily damaging for the individual.

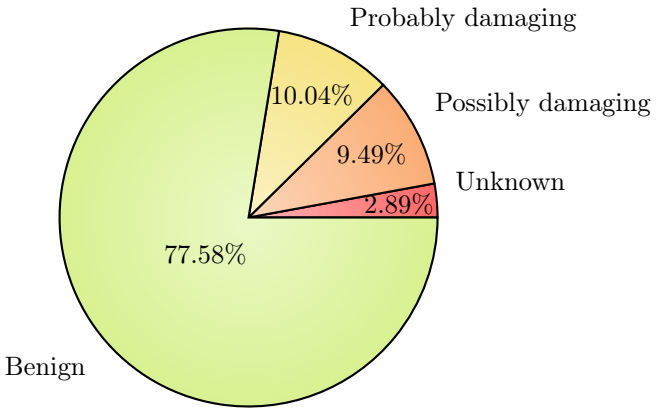

Figure 2: PolyPhen Summary

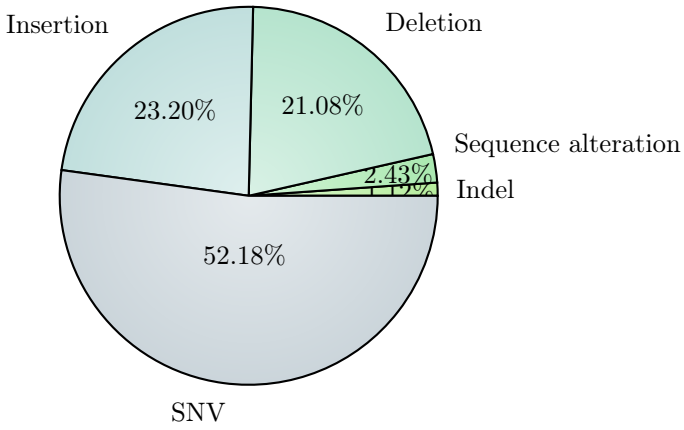

Figure 3: Variant Class

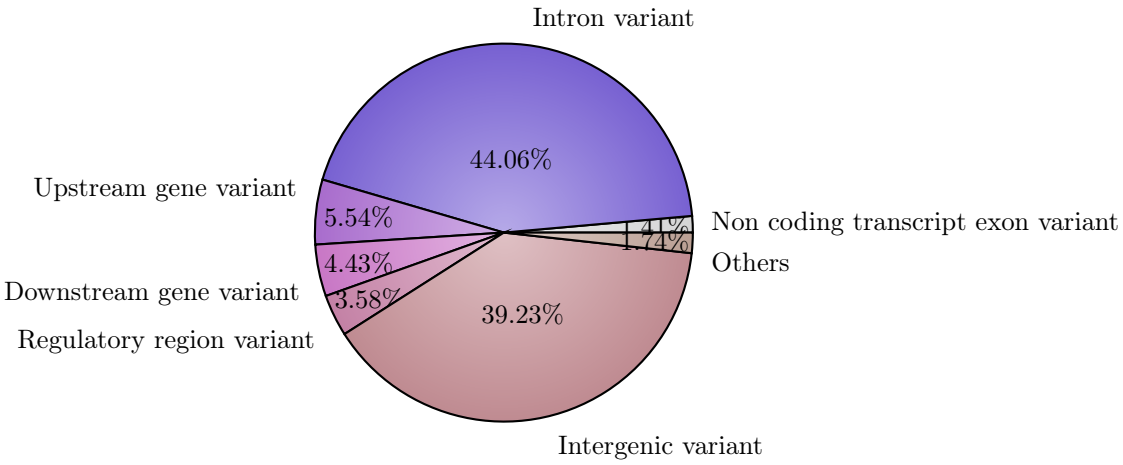

Figure 4: Consequence type

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).

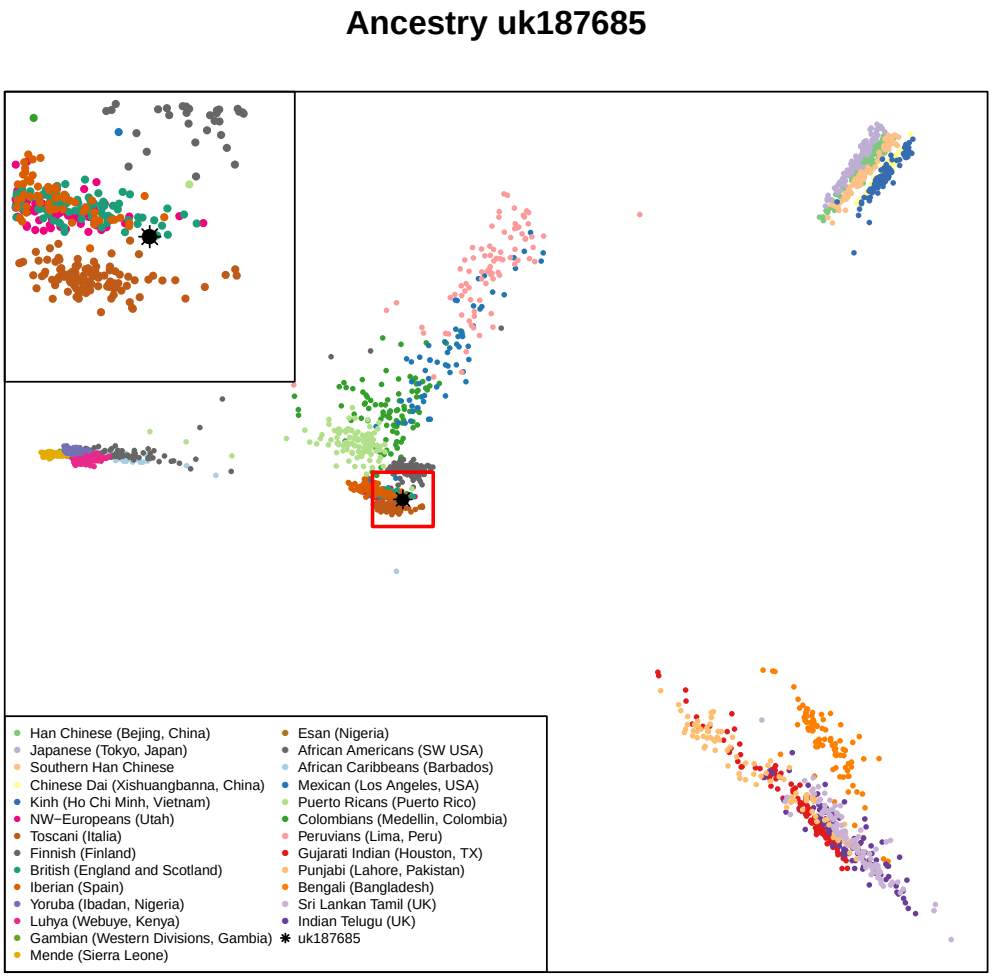

Figure 5: Ancestry Principal Component Analysis

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
2.5	rs11649743	(A;A)	Lower prostate cancer risk?	Link	Link	
2.5	rs2943634	(A;A)	Lower risk of ischemic stroke	Link	Link	
2.5	rs3764261	(T;T)	Associated with higher HDL cholesterol. HDL	Link	Link	Link
2.2	rs2511989	(A;A)	0.44x decreased age-related macular degeneratio...	Link	Link	
2.1	rs3775291	(A;G)	0.71x decreased risk for dry age related macula...	Link	Link	Link
2	rs11045585	(A;A)	24% chance (lower than average) of docetaxel-in...	Link	Link	
2	rs1136410	(C;T)	0.80x reduced risk for glioblastoma	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	rs1799884	(G;G)	Mothers have typical Birth-Weight babies. Sligh...	Link		
2	rs1864163	(G;G)	Associated with higher HDL cholesterol	Link	Link	
2	rs2060793	(A;A)	Lower serum levels of vitamin D	Link		
2	rs2073963	(T;T)	Reduced risk of baldness	Link		
2	rs2241423	(A;G)	0.79 decreased risk for obesity	Link		
2	rs2542052	(C;C)	Better odds of living to 100	Link		
2	rs261332	(A;G)	Associated with higher HDL cholesterol	Link		
2	rs37973	(A;A)	Possibly better response to inhaled corticoster...	Link		Link
2	rs3819331	(T;T)	Lower risk of autism	Link		Link
2	rs4149268	(G;G)	Associated with higher HDL cholesterol	Link	Link	
2	rs4307059	(C;C)	Reduced Autism risk	Link	Link	
2	rs6511720	(G;T)	Slightly lower odds of developing CHD.	Link	Link	Link
2	rs6855911	(A;G)	0.62x decreased risk for gout	Link	Link	
2	rs7216389	(C;C)	0.69x lower risk of Childhood Asthma.	Link	Link	
2	rs763110	(C;T)	~0.80x reduced cancer risk	Link		Link
2	rs7776725	(T;T)	Stronger bones	Link	Link	
2	rs800292	(T;T)	5% decreased risk of macular degeneration	Link	Link	Link

Mag.	Identifier	Genotype	Summary	GnomAD	GetEvidence	ClinVar
2	rs801114	(T;T)	0.78x decreased Basal Cell Carcinoma risk.	Link	Link	
2.0	rs3790844	(C;C)	Reduced risk (0.59x) of pancreatic cancer	Link		
1.8	rs1800588	(C;T)	Higher HDL-C levels	Link	Link	
1.8	rs3814113	(C;T)	0.8x decreased risk for ovarian cancer	Link	Link	
1.8	rs6897932	(C;T)	0.91x decreased risk for multiple sclerosis	Link	Link	Link
1.8	rs7101429	(A;G)	0.70x reduced risk for Alzheimer's risk	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	rs1026732	(A;G)	0.70x risk for restless legs	Link	Link	
1.5	rs1050631	(C;C)	Mean Survival Time of 32 months for esophageal ...	Link		
1.5	rs11136000	(C;T)	0.84x decreased risk for Alzheimer's disease	Link	Link	
1.5	rs11635424	(A;G)	0.70x risk for restless legs	Link	Link	
1.5	rs12593813	(A;G)	0.71x risk for restless legs	Link	Link	
1.5	rs2229169	(C;C)	1.5x decreased risk of heart attack and stroke ...	Link		
1.5	rs3784709	(C;T)	0.71x risk of developing restless legs syndrome...	Link	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;T)	Associated with higher HDL cholesterol	Link	Link	
1.5	rs9939609	(T;T)	Lower risk of obesity and Type-2 diabetes	Link	Link	
1.4	rs10513789	(G;T)	0.8x decreased risk of Parkinson's disease	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		
1.25	rs10088218	(A;G)	0.76x decreased risk for ovarian cancer	Link		
1.2	rs4686484	(G;G)	Slightly decreased risk for celiac disease	Link		
1.2	rs4867568	(T;T)	Decreased risk for knee osteoporosis	Link		
1.2	rs9306160	(C;T)	0.75x (reduced) risk for metastasis in LN-/ER+ ...	Link	Link	
1.1	rs10166942	(C;T)	0.85x lower risk for migraines	Link		
1.1	rs11172113	(C;T)	0.9x lower risk for migraines	Link		
1.1	rs13333226	(A;G)	Slightly lower risk for hypertension	Link		Link
1.1	rs2293347	(G;G)	Among NSCLC patients: better Gefitinib response...	Link		Link
1.1	rs4988235	(T;T)	Can digest milk	Link		Link
1	rs10784502	(C;T)	Slightly higher intracranial volume	Link		
1	rs182549	(T;T)	Can digest milk.	Link		Link
1	rs2952768	(C;T)	Slightly less drug dependence: decreased effect...	Link		Link
1	rs4752566	(G;T)	Associated with thicker hair in Asians	Link		
1	rs4939827	(C;C)	0.73x decreased risk for colorectal cancer	Link	Link	Link
1.0	rs6583817	(C;T)	~0.80x (lower) risk for late onset Alzheimer's ...	Link		
0.1	rs1726866	(C;C)	Can taste bitter	Link	Link	Link
0.1	rs891512	(G;G)	Lower blood pressure than those with an A allel...	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	rs1983132	(C;T)	2 - 3x higher prostate cancer risk if routinely...	Link		
3	rs3738579	(T;T)	1.5x - 2x increased risk for cervical cancer: H...	Link		
3	rs7754840	(C;G)	1.3x increased risk for type-2 diabetes	Link	Link	
2.7	rs10830963	(C;G)	Increased type-2 diabetes risk; higher gestatio...	Link	Link	
2.5	rs10490924	(G;T)	2.7x risk for age related macular degeneration	Link	Link	Link
2.5	rs10974944	(C;G)	Increased odds (2 - 4 fold?) of V617F-associate...	Link	Link	
2.5	rs12340895	(C;G)	Increased odds (2 fold?) of developing V617F-po...	Link		
2.5	rs12343867	(C;T)	Increased odds (2 fold?) of V617F-associated MP...	Link		
2.5	rs13266634	(C;T)	Increased risk for type-2 diabetes	Link	Link	Link
2.5	rs187238	(G;G)	Hypertension increases risk 3.75x for sudden ca...	Link		
2.5	rs2004640	(T;T)	1.4x increased risk for SLE	Link	Link	
2.5	rs2241880	(C;C)	2x-3x increased risk for Crohn's disease in Cau...	Link	Link	Link
2.5	rs339331	(T;T)	Prostate cancer risk	Link		
2.5	rs3780374	(A;G)	Substantially increased odds of developing V617...	Link		
2.5	rs4143094	(G;T)	Slightly (17%) higher risk of colorectal cancer...	Link		
2.5	rs4495487	(C;T)	Increased odds (2 fold?) of developing V617F-as...	Link		
2.5	rs5888	(C;T)	3x higher risk for age-related macular degenera...	Link		
2.5	rs613872	(G;G)	~20-30x higher risk for Fuchs' dystrophy: a cor...	Link		
2.5	rs7574865	(G;T)	1.3x risk of rheumatoid arthritis; 1.55x risk o...	Link	Link	Link
2.5	rs9934438	(A;A)	Coumadin resistance	Link	Link	Link
2.4	rs7966230	(G;G)	Slightly lower levels of plasma VWF	Link		
2.2	rs3129934	(T;T)	3.3x increased risk for multiple sclerosis	Link	Link	
2.1	rs1050152	(T;T)	2.1x increased risk of Crohn's disease	Link	Link	Link
2.1	rs10811661	(T;T)	1.2x increased risk for type-2 diabetes	Link	Link	
2.1	rs1219648	(G;G)	1.64x risk for breast cancer	Link	Link	
2.1	rs17070145	(C;C)	Reduced memory abilities	Link		Link
2.1	rs17563	(C;C)	Risk for otosclerosis	Link	Link	Link
2.1	rs2294008	(T;T)	Increased risk of gastric and bladder cancer	Link	Link	
2.1	rs2383207	(G;G)	Increased risk for heart disease	Link		
2.1	rs2420946	(T;T)	1.64x risk for breast cancer	Link		
2.1	rs4430796	(A;A)	1.38x increased risk for prostate cancer	Link	Link	
2.1	rs5186	(A;C)	~1.4x increased risk of hypertension	Link	Link	Link
2.1	rs629242	(T;T)	Somewhat higher risk for prostate cancer	Link		
2.1	rs646776	(A;A)	1.2x risk of coronary artery disease	Link	Link	
2.1	rs795484	(A;G)	Increased morphine dose requirement and postope...	Link		
2.1	rs944289	(C;T)	1.3x increased thyroid cancer risk	Link	Link	
2	rs1045642	(C;T)	Slower metaboliser for some drugs	Link	Link	Link
2	rs10488631	(C;T)	2x increased risk of developing SLE; 1.6x incre...	Link	Link	
2	rs10889677	(A;C)	1.5x increased risk for certain autoimmune dise...	Link	Link	
2	rs10936599	(T;T)	Shorter telomeres: shorter life?	Link		Link
2	rs10984447	(A;A)	>1.17x increased risk for multiple sclerosis	Link	Link	
2	rs11171739	(C;C)	1.75x risk of developing Type-1 diabetes	Link	Link	
2	rs1143699	(C;C)	In men: 2.19x risk of type 2 diabetes	Link		
2	rs1160312	(A;G)	1.6x increased risk of Male Pattern Baldness.	Link	Link	
2	rs12567232	(A;G)	Increased risk for Crohn's Disease	Link	Link	
2	rs12696304	(G;G)	Prone to aging faster: at least in European pop...	Link		
2	rs13254738	(A;C)	1.18x prostate cancer risk	Link	Link	
2	rs1333048	(A;C)	1.3x increased coronary artery disease risk	Link		
2	rs1360780	(C;T)	1.3x increased risk for depression	Link	Link	Link
2	rs144848	(G;G)	Very slightly increased breast cancer risk	Link	Link	Link
2	rs1734791	(A;A)	1.4x increased risk for lupus	Link		
2	rs17576	(A;G)	Higher risk for MI and lung cancer: and COPD in...	Link	Link	Link

Mag.	Identifier	Genotype	Summary	GnomAD	GetEvidence	ClinVar
2	rs1799950	(G;G)	Very slightly increased breast cancer risk	Link	Link	Link
2	rs2201841	(C;T)	1.5x increased risk for Crohn's disease; 2x inc...	Link	Link	
2	rs2230201	(G;G)	>1.4x risk of lupus	Link		Link
2	rs2274223	(A;G)	1.5x increased risk for stomach and esophageal ...	Link	Link	Link
2	rs2305480	(T;T)	If 4 years old or younger: ~3x increased asthma...	Link	Link	
2	rs2305795	(A;G)	1.28x higher risk of narcolepsy compared to (G;...	Link		Link
2	rs2306402	(C;T)	1.18x increased risk for late-onset Alzheimer's...	Link		
2	rs2383206	(A;G)	1.4x increased risk for heart disease	Link		
2	rs25487	(G;G)	2x higher risk for skin cancer; possibly other ...	Link	Link	Link
2	rs2707466	(G;G)	Weaker bones	Link	Link	
2	rs27388	(A;A)	Increased risk of developing schizophrenia	Link		
2	rs2908004	(C;C)	Weaker bones	Link	Link	
2	rs326	(A;A)	Lower HDL cholesterol	Link	Link	Link
2	rs351855	(C;T)	1.2x increased risk for prostate cancer	Link	Link	Link
2	rs358806	(C;C)	1.78x increased risk of developing Type-2 diabe...	Link	Link	
2	rs3738919	(A;C)	1.94x risk of developing rheumatoid arthritis	Link		
2	rs4129148	(C;G)	3x risk of schizophrenia.	Link	Link	
2	rs4242382	(A;G)	1.7x increased risk for prostate cancer	Link	Link	
2	rs4402960	(G;T)	1.2x increased risk for type-2 diabetes: ~1x ri...	Link	Link	Link
2	rs4420638	(A;G)	~3x increased Alzheimer's risk; 1.4x increased ...	Link	Link	Link
2	rs493258	(G;G)	1.15x risk of Age Related Macular Degeneration	Link		
2	rs6441286	(G;T)	1.54x chance of developing primary biliary cirr...	Link	Link	
2	rs6603272	(G;T)	2.74x increased risk of developing schizophreni...	Link		
2	rs6908425	(C;C)	1.95x increased risk of developing Crohn's dise...	Link	Link	
2	rs699	(C;C)	Increased risk of hypertension	Link	Link	Link
2	rs7190458	(A;G)	Slightly higher pancreatic cancer risk	Link		
2	rs738409	(C;G)	Increased liver fat: odds of alcoholic liver di...	Link	Link	Link
2	rs7639618	(T;T)	1.45x increased osteoarthritis risk	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	rs7923837	(G;G)	3.2x risk for T2D	Link		
2	rs828907	(T;T)	Increased risk of bladder cancer and 2x risk of...	Link		
2	rs854560	(A;A)	Higher risk for heart disease: diabetic retinop...	Link	Link	Link
2	rs9303277	(T;T)	1.46x Increased risk of developing primary bili...	Link		
2	rs9652490	(A;A)	~2x increased risk for Parkinson's disease: and...	Link	Link	
2	rs9954153	(G;T)	~2.5x higher risk for Fuchs' dystrophy: a corne...	Link		
1.8	rs10210302	(T;T)	1.8x increased risk for Crohn's disease	Link	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		
1.7	rs4807015	(C;T)	1.74x risk of type 2 diabetes	Link		
1.7	rs8055236	(G;T)	1.9x risk for heart disease	Link	Link	
1.6	rs11523871	(A;C)	1.6x increased breast cancer risk for women ove...	Link	Link	
1.6	rs1537415	(C;G)	1.6x increased risk for periodontitis	Link	Link	
1.6	rs1978237	(C;G)	1.59x risk of Type 2 diabetes	Link		
1.6	rs2059693	(T;T)	1.6x increased risk for testicular cancer	Link		
1.6	rs2981745	(C;T)	1.6x increased risk for breast cancer in female...	Link		
1.6	rs3775948	(C;G)	Slightly higher risk for gout	Link		
1.5	rs10492519	(A;G)	Slightly increased risk of developing prostate ...	Link		
1.5	rs10757272	(C;T)	1.30x increased risk for Coronary artery diseas...	Link		
1.5	rs10883365	(A;G)	1.2x increased risk for developing Crohn's dise...	Link	Link	
1.5	rs10980705	(C;T)	2.3x increased risk for knee osteoarthritis	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	rs1223271	(A;G)	Slightly increased risk of developing Parkinson...	Link	Link	
1.5	rs12431733	(C;T)	Slightly increased risk of developing Parkinson...	Link	Link	

Mag.	Identifier	Genotype	Summary	GnomAD	GetEvidence	ClinVar
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	rs1994090	(G;T)	Slightly increased risk of developing Parkinson...	Link	Link	
1.5	rs2007153	(G;G)	Increased risk of schizophrenia in limited stud...	Link		
1.5	rs2240340	(A;A)	Slightly increased (1.5x) risk for RA	Link		
1.5	rs2272127	(C;C)	Associated with herpes and schizophrenia	Link		
1.5	rs2280714	(A;A)	1.4x increased risk of SLE	Link		
1.5	rs28694718	(A;G)	2x higher risk for schizophrenia	Link		
1.5	rs2881766	(T;T)	Slightly increased risk for pregnancy-induced h...	Link		
1.5	rs3087243	(A;G)	Increased risk for auto-immune diseases	Link	Link	Link
1.5	rs3212227	(A;A)	1.43x increased risk of developing psoriasis an...	Link		Link
1.5	rs3745516	(A;G)	Slightly increased risk of developing primary b...	Link		
1.5	rs3764880	(A;G)	Possible 1.2 - 1.8x increased tuberculosis susc...	Link	Link	
1.5	rs4027132	(A;G)	1.39x increased risk of developing bipolar diso...	Link		
1.5	rs4585	(T;T)	Slightly poorer (0.75x) response to metformin i...	Link		Link
1.5	rs464049	(T;T)	Increased risk of schizophrenia in limited stud...	Link		
1.5	rs4656461	(A;G)	1.5x increased risk for open angle glaucoma	Link		
1.5	rs4785763	(A;A)	2x higher risk for melanoma	Link	Link	
1.5	rs5219	(C;T)	1.3x increased risk for type-2 diabetes	Link	Link	Link
1.5	rs5746059	(A;A)	Slightly higher fat mass	Link		
1.5	rs619203	(C;G)	Increases susceptibility to Myocardial Infarcti...	Link	Link	
1.5	rs6498169	(A;G)	1.14x risk of multiple sclerosis	Link	Link	
1.5	rs6601764	(C;T)	1.16x increased risk of developing Crohn's dise...	Link	Link	
1.5	rs6656401	(A;G)	1.18x increased risk for late-onset Alzheimer...	Link		
1.5	rs6896702	(C;T)	Slightly increased risk of developing Parkinson...	Link		
1.5	rs699473	(C;T)	~1.5x increased brain tumor risk	Link		
1.5	rs7341475	(G;G)	1.58x increased schizophrenia risk for women	Link	Link	
1.5	rs7850258	(G;G)	Slightly higher odds of developing primary hypo...	Link		
1.5	rs872071	(G;G)	~1.5x increased risk for chronic lymphocytic le...	Link	Link	
1.5	rs9642880	(G;T)	1.2x increased bladder cancer risk	Link	Link	
1.5	rs995030	(G;G)	Non-protective against testicular cancer	Link	Link	
1.4	rs1126497	(C;T)	1.4x increased risk for breast cancer	Link	Link	Link
1.4	rs1801157	(A;G)	1.4x higher risk for breast cancer	Link		
1.4	rs2046210	(C;T)	1.4x increased breast cancer risk	Link	Link	Link
1.4	rs2228314	(C;G)	1.48x risk of osteoarthritis	Link	Link	
1.4	rs3184504	(C;T)	Slightly increased risk for celiac disease	Link	Link	
1.3	rs1042713	(A;G)	1.3x increased risk that pediatric inhaler use ...	Link	Link	Link
1.3	rs110419	(A;G)	1.3x increased risk for neuroblastoma	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	rs2736100	(G;T)	1.3x higher risk for glioma development: 2.1x r...	Link	Link	Link
1.3	rs34330	(C;T)	1.3x higher risk for endometrial cancer (in Chi...	Link		Link
1.3	rs4712653	(C;T)	Very slightly (~1.3x) increased risk for neurob...	Link		
1.3	rs7234029	(A;G)	Slightly increased (1.36x) risk for Crohn's dis...	Link		
1.25	rs13387042	(A;A)	1.24x increased risk for breast cancer	Link	Link	
1.2	rs10865331	(A;G)	1.2x higher risk for ankylosing spondylitis	Link		
1.2	rs1344706	(T;T)	1.2x increased risk for schizophrenia	Link	Link	
1.2	rs1800693	(A;G)	Slight (1.2x) increase in risk for multiple scl...	Link	Link	Link
1.2	rs2056116	(A;G)	1.18x risk for breast cancer	Link		
1.2	rs2072590	(G;T)	1.2x increased risk for ovarian cancer	Link		
1.2	rs2076295	(G;T)	One copy of the risk allele (G): slightly incre...	Link		

Mag.	Identifier	Genotype	Summary	GnomAD	GetEvidence	ClinVar
1.2	rs2254958	(C;T)	1.24x reported increased risk for Alzheimer's; ...	Link		
1.2	rs4324715	(C;C)	>1.5x increased testicular cancer risk for men	Link		
1.2	rs4977756	(A;G)	1.39x higher risk for glioma development	Link	Link	
1.2	rs498872	(C;T)	1.2x higher risk for glioma development	Link	Link	
1.2	rs6010620	(A;G)	1.2x higher risk for glioma development: 1.17x ...	Link	Link	
1.2	rs7514229	(G;G)	Associated with early-onset autoimmune thyroid ...	Link		
1.17	rs17465637	(A;C)	1.17x higher risk for myocardial infarction	Link	Link	
1.17	rs3802842	(A;C)	1.17x increased risk of colorectal cancer	Link	Link	
1.15	rs748404	(C;T)	Very slightly increased risk (1.15) for lung ca...	Link	Link	
1.1	rs10248420	(A;A)	Possibly less likely to remit on certain antide...	Link	Link	
1.1	rs2235040	(G;G)	Possibly lesser chances of remission only for i...	Link	Link	
1.1	rs34516635	(G;G)	Less longevity for Ashkenazi Jewish women.	Link		Link
1.1	rs3818361	(C;T)	1.15x increased risk for late-onset Alzheimer's...	Link		
1.1	rs4977574	(A;G)	Some studies - but not others - report a slight...	Link	Link	
1.1	rs5030737	(C;T)	Carrier of mannose binding deficiency but of lo...	Link	Link	Link
1.1	rs6800901	(T;T)	1.3x multiple myeloma risk	Link		
1.1	rs6897876	(C;T)	Slight increase in testicular cancer risk for m...	Link		
1.1	rs7171755	(A;G)	Very slight decrease in cortical thickness and ...	Link		
1.1	rs889312	(A;C)	Very slightly higher risk for breast cancer	Link	Link	
1.1	rs997669	(G;G)	Very slightly increased (1.18x) increased breas...	Link		
1.09	rs12050604	(A;C)	Very slightly increased risk for lung cancer	Link		
1.05	rs2291834	(C;T)	Very slightly higher risk for myocardial infarc...	Link		
1	rs1004819	(C;T)	1.5x risk of Crohn's disease: 1.2 for developin...	Link	Link	
1	rs1010	(A;G)	1.75x risk of MI	Link	Link	
1	rs10761659	(A;G)	1.2x risk of Crohn's disease	Link	Link	
1	rs1143674	(A;G)	1.3x increased autism risk	Link		
1	rs1417066	(C;T)	Slightly increased risk of osteoarthritis	Link		
1	rs17300539	(G;G)	Increased risk of insulin resistance	Link		
1	rs2273697	(A;G)	Adverse reaction more likely to carbamazepine i...	Link	Link	Link
1	rs2546890	(A;A)	Higher risk of multiple sclerosis	Link		
1	rs3194051	(A;G)	1.12x risk of type-1 diabetes	Link	Link	Link
1	rs6932590	(C;T)	1.1x increased risk for schizophrenia	Link	Link	
1	rs6974491	(A;G)	Higher risk of coeliac and/or inflammatory bowe...	Link		
0.1	rs3095870	(A;G)	1.7x increased risk for SLE (lupus)	Link		
0.1	rs3748079	(G;G)	1.9x increased risk for SLE (lupus)	Link		
0.1	rs601338	(A;G)	Susceptible to Norovirus infections	Link	Link	Link

3.3 Genosets (Multi-variant Phenotypes)

Magnitude	Identifier	Summary
3.1	gs122	7x risk of male baldness
3	gs273	Lowest risk (13% of white women) of Atrial Fibr...
2.7	gs311	Slow metabolizer of certain substances
2.5	gs155	CYP3A5 non-expressor
2.5	gs281	Part of the 88% of the population claimed not t...
2.5	gs284	Any diet works for you
2.4	gs297	Lower heart attack risk than average
2	gs101	Probably able to digest milk
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	gs244	2x increased risk for esophageal squamous cell ...
2	gs249	Parkinson's Disease Risk
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	gs232	Possible low pain sensitivity; LPS/LPS
1.5	gs139	NAT2 intermediate metabolizer
1.2	gs184	Able to taste bitterness.
1	gs182	CYP2D6*39

4 Report Metadata

Resource	Version	Website
Genome	GRCh37	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 5: Analysis Pipeline Versions

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