PGP-UK Genomics Report for uk9F6EDA

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.

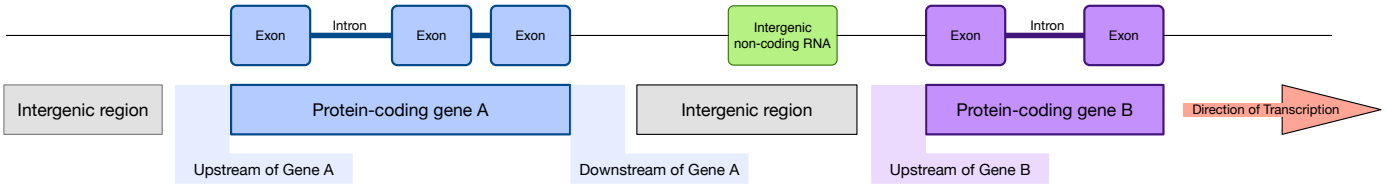

Figure 1: Diagram of gene structure indicating locations of potential variants

Feature	Count
Lines of input read	4641357
Variants filtered out	3689773
Novel / existing variants	0 (0.0) / 951584 (100.0)
Overlapped genes	51645
Overlapped transcripts	59030
Overlapped regulatory features	46191

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.

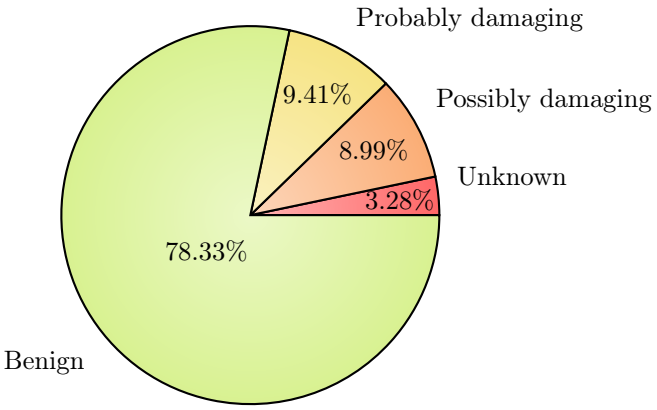

Figure 2: PolyPhen Summary

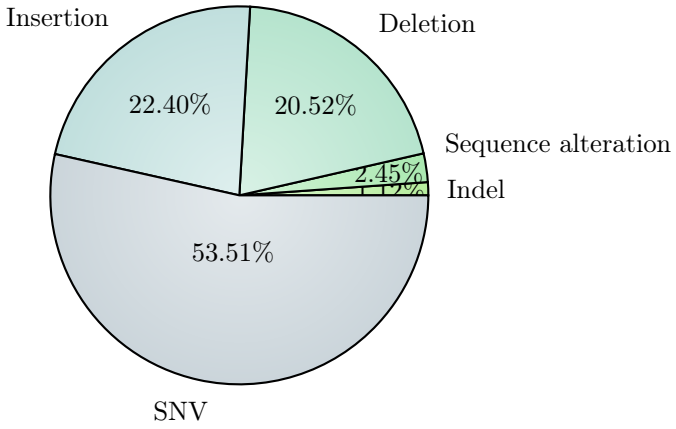

Figure 3: Variant Class

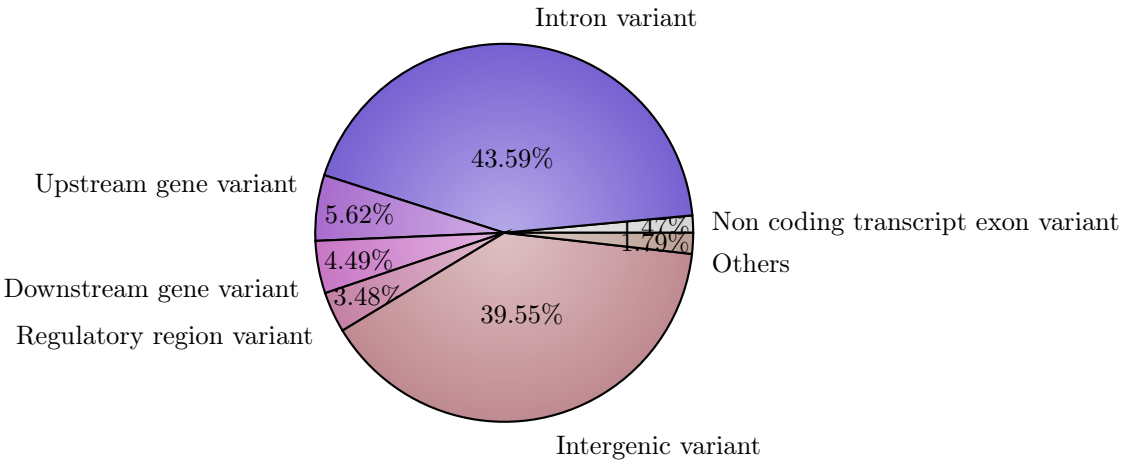

Figure 4: Consequence type

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).

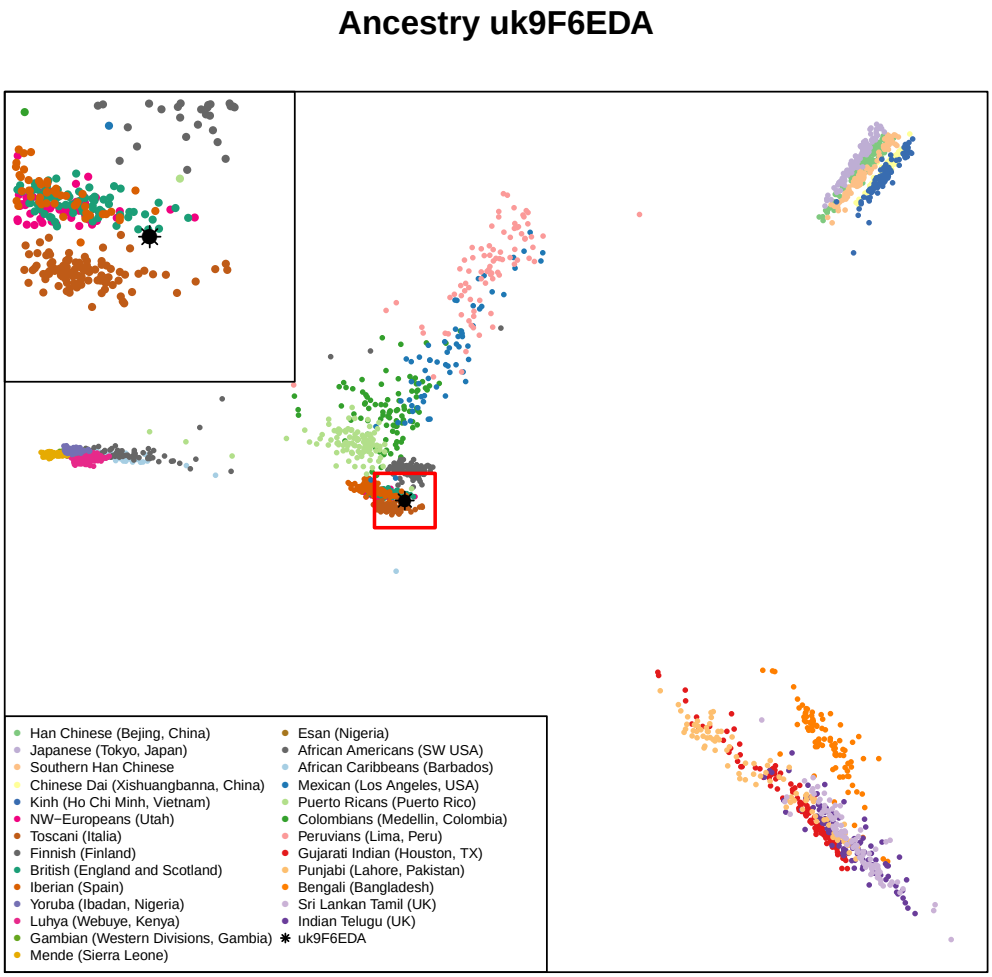

Figure 5: Ancestry Principal Component Analysis

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
2.1	rs2511989	(A;G)	0.63x decreased age-related macular degeneratio...	Link	Link	
2	rs10468017	(C;T)	Associated with higher HDL cholesterol	Link	Link	
2	rs10504861	(A;G)	Reduced risk of migraine without aura	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	rs12979860	(C;C)	~80% of such hepatitis C patients respond to tr...	Link	Link	Link
2	rs1501299	(A;C)	Slightly lower risk of breast cancer	Link		
2	rs174537	(T;T)	Lower LDL-C and total cholesterol	Link		
2	rs1864163	(A;G)	Associated with higher HDL cholesterol	Link	Link	
2	rs2073963	(T;T)	Reduced risk of baldness	Link		
2	rs2241766	(G;T)	Slightly lower risk of breast cancer	Link		
2	rs2542052	(C;C)	Better odds of living to 100	Link		
2	rs261332	(A;A)	Associated with higher HDL cholesterol	Link		
2	rs3738579	(C;T)	0.5x decreased risk for cervical cancer: HNSCC:...	Link		
2	rs3750817	(C;T)	0.78x reduced risk for breast cancer	Link		
2	rs3764261	(G;T)	Associated with higher HDL cholesterol	Link	Link	Link
2	rs3819331	(T;T)	Lower risk of autism	Link		Link
2	rs3914132	(C;T)	Lower otosclerosis risk	Link	Link	
2	rs4149268	(G;G)	Associated with higher HDL cholesterol	Link	Link	
2	rs6505162	(A;C)	0.58x decreased risk for esophageal cancer	Link		
2	rs763110	(C;T)	~0.80x reduced cancer risk	Link		Link
2	rs800292	(T;T)	5% decreased risk of macular degeneration	Link	Link	Link
2	rs801114	(T;T)	0.78x decreased Basal Cell Carcinoma risk.	Link	Link	
2	rs925391	(C;T)	Lower odds of going bald	Link		
1.8	rs1800588	(T;T)	Higher HDL-C levels	Link	Link	

Mag.	Identifier	Genotype	Summary	GnomAD	GetEvidence	ClinVar
1.8	rs187238	(C;G)	Hypertension not a risk factor for sudden cardi...	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	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
1.5	rs1050631	(C;C)	Mean Survival Time of 32 months for esophageal ...	Link		
1.5	rs1063192	(C;T)	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	rs11212617	(A;C)	Somewhat increased likelihood of treatment succ...	Link		Link
1.5	rs16991615	(A;G)	Slight increase (11 months) in avg age at menop...	Link	Link	
1.5	rs2229169	(C;C)	1.5x decreased risk of heart attack and stroke ...	Link		
1.5	rs309375	(G;G)	Smaller mosquito bites	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	rs6427528	(A;G)	For rheumatoid arthritis patients: better respo...	Link		
1.5	rs729302	(A;C)	0.89x decreased risk of developing rheumatoid a...	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	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	rs4988235	(T;T)	Can digest milk	Link		Link
1	rs182549	(T;T)	Can digest milk.	Link		Link
1	rs2952768	(C;T)	Slightly less drug dependence: decreased effect...	Link		Link
1	rs4939827	(C;T)	0.86x decreased risk for colorectal cancer	Link	Link	Link
1	rs7850258	(A;G)	Typical odds of developing primary hypothyroidi...	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	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	
2.6	rs8034191	(C;C)	1.80x lung cancer risk; decreased response to a...	Link	Link	
2.5	rs10974944	(C;G)	Increased odds (2 - 4 fold?) of V617F-associate...	Link	Link	
2.5	rs11190870	(T;T)	Possibly even more increased risk of scoliosis	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	rs16969968	(A;G)	Slightly higher risk for nicotine dependence: l...	Link	Link	Link
2.5	rs17696736	(G;G)	1.94x risk of type-1 diabetes	Link	Link	
2.5	rs2943634	(C;C)	Slightly higher risk of ischemic stroke	Link	Link	
2.5	rs339331	(T;T)	Prostate cancer risk	Link		
2.5	rs3738919	(C;C)	1.94x risk of developing rheumatoid arthritis	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	rs664143	(C;T)	Higher risk for number of cancers	Link		
2.5	rs795484	(A;A)	Even more increased morphine dose requirement a...	Link		
2.5	rs9934438	(A;A)	Coumadin resistance	Link	Link	Link
2.4	rs1143679	(A;G)	1.78x increased risk for SLE	Link	Link	
2.3	rs7966230	(C;G)	Slightly lower levels of plasma VWF	Link		
2.1	rs10811661	(T;T)	1.2x increased risk for type-2 diabetes	Link	Link	
2.1	rs17070145	(C;C)	Reduced memory abilities	Link		Link
2.1	rs2231142	(A;C)	1.74x increased gout risk; gefinitib takers 4x ...	Link	Link	Link
2.1	rs2306402	(C;C)	1.18x increased risk for late-onset Alzheimer's...	Link		
2.1	rs4149056	(C;T)	Reduced breakdown of some drugs; 5x increased m...	Link	Link	Link
2.1	rs4363657	(C;T)	4.5x increased myopathy risk for statin users	Link	Link	
2.1	rs4430796	(A;A)	1.38x increased risk for prostate cancer	Link	Link	
2.1	rs5751876	(T;T)	Significantly higher anxiety levels after moder...	Link		
2.1	rs646776	(A;A)	1.2x risk of coronary artery disease	Link	Link	
2.1	rs944289	(C;T)	1.3x increased thyroid cancer risk	Link	Link	
2	rs10086908	(C;T)	1.7x increased risk for prostate cancer	Link		
2	rs1045642	(C;T)	Slower metaboliser for some drugs	Link	Link	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	rs1064395	(A;G)	Having any copies of A at this SNP heightens yo...	Link		
2	rs10889677	(C;C)	Baseline (average) risk for certain autoimmune ...	Link	Link	
2	rs10984447	(A;G)	1.17x increased risk for multiple sclerosis	Link	Link	
2	rs110419	(A;A)	1.7x increased risk for neuroblastoma	Link		
2	rs11123857	(A;G)	1.44-fold increased risk of bipolar disorder or...	Link		
2	rs1143699	(C;C)	In men: 2.19x risk of type 2 diabetes	Link		
2	rs11650354	(T;T)	8x risk for allergic asthma	Link		
2	rs12037606	(A;A)	1.52x risk of developing Crohn's disease	Link		
2	rs12431733	(T;T)	Increased risk of developing Parkinson's Diseas...	Link	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	rs1544410	(A;A)	Increased risk of low bone mineral density diso...	Link	Link	
2	rs1585215	(A;G)	2x increased risk for Hodgkin lymphoma	Link		
2	rs1691053	(A;G)	Increased risk of developing prostate cancer	Link		
2	rs16942	(A;G)	Very slightly increased breast cancer risk	Link	Link	Link

Mag.	Identifier	Genotype	Summary	GnomAD	GetEvidence	ClinVar
2	rs17228212	(C;T)	1.26x increased risk for heart disease	Link	Link	
2	rs1734791	(A;A)	1.4x increased risk for lupus	Link		
2	rs17487223	(C;T)	Higher lung cancer risk?	Link		
2	rs2156921	(A;G)	1.29x increased risk for depression	Link		
2	rs2201841	(T;T)	2.4x increased risk for Graves' disease	Link	Link	
2	rs2230199	(C;G)	1.6x+ risk of ARMD	Link	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	(C;T)	3.5x increase in risk of asthma for Han Chinese...	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		
2	rs2383207	(A;G)	Increased risk for heart disease	Link		
2	rs25487	(A;G)	2x higher risk for skin cancer; possibly other ...	Link	Link	Link
2	rs2736100	(T;T)	Higher risk of Interstitial lung disease: and t...	Link	Link	Link
2	rs27388	(A;A)	Increased risk of developing schizophrenia	Link		
2	rs3025039	(C;T)	2.6x increased risk for ARMD in a Taiwanese pop...	Link		
2	rs3197999	(T;T)	1.2x risk of Crohn's	Link	Link	
2	rs3212227	(A;C)	Significantly increased risk of developing cerv...	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	rs3775948	(G;G)	Slightly higher risk for gout	Link		
2	rs3793784	(C;G)	1.5x risk for ARMD	Link	Link	Link
2	rs4129148	(C;G)	3x risk of schizophrenia.	Link	Link	
2	rs4420638	(A;G)	~3x increased Alzheimer's risk; 1.4x increased ...	Link	Link	Link
2	rs4633	(C;T)	Higher risk for endometrial cancer	Link	Link	Link
2	rs493258	(A;G)	1.15x risk of Age Related Macular Degeneration	Link		
2	rs4968451	(A;C)	1.61x increased risk for meningioma	Link		
2	rs5174	(A;G)	1.3x increased risk for heart disease	Link	Link	Link
2	rs520354	(A;G)	Increased risk in men for biliary conditions	Link		
2	rs5759167	(T;T)	Higher prostate cancer risk	Link	Link	
2	rs629242	(C;T)	Somewhat higher risk for prostate cancer	Link		
2	rs6601764	(C;C)	1.52x increased risk of developing Crohn's dise...	Link	Link	
2	rs663048	(G;T)	Significantly increased risk of developing lung...	Link	Link	
2	rs6807362	(C;C)	Increased autism risk	Link	Link	
2	rs6896702	(T;T)	Increased risk of developing Parkinson's Diseas...	Link		
2	rs6908425	(C;C)	1.95x increased risk of developing Crohn's dise...	Link	Link	
2	rs6997709	(G;T)	1.2x higher risk for hypertension	Link		
2	rs699	(C;T)	Increased risk of hypertension	Link	Link	Link
2	rs7190458	(A;G)	Slightly higher pancreatic cancer risk	Link		
2	rs7442295	(A;A)	~4x higher risk for hyperuracemia	Link	Link	Link
2	rs744373	(C;T)	1.17x risk of Alzheimer's	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	(G;T)	Slightly increased risk of bladder cancer and 2...	Link		
2	rs854560	(A;A)	Higher risk for heart disease: diabetic retinop...	Link	Link	Link
2	rs855913	(G;T)	Reduced survival with ALS	Link	Link	
2	rs9525638	(T;T)	Weaker bones	Link		
2	rs9652490	(A;A)	~2x increased risk for Parkinson's disease: and...	Link	Link	
2	rs965513	(A;G)	1.77x increased thyroid cancer risk	Link	Link	
2	rs9954153	(G;T)	~2.5x higher risk for Fuchs' dystrophy: a corne...	Link		
2.0	rs4911414	(G;T)	2-4x higher risk of sun sensitivity if part of ...	Link	Link	
2.0	rs9642880	(T;T)	1.5x increased bladder cancer risk	Link	Link	
1.75	rs1010	(G;G)	1.75x risk of MI	Link	Link	
1.7	rs4807015	(C;T)	1.74x risk of type 2 diabetes	Link		

Mag.	Identifier	Genotype	Summary	GnomAD	GetEvidence	ClinVar
1.6	rs11523871	(A;C)	1.6x increased breast cancer risk for women ove...	Link	Link	
1.6	rs1260326	(T;T)	Slightly higher risk for gout	Link	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	rs2981745	(C;T)	1.6x increased risk for breast cancer in female...	Link		
1.6	rs3764880	(A;A)	1.2 - 1.8x increased tuberculosis risk	Link	Link	
1.5	rs10757272	(C;T)	1.30x increased risk for Coronary artery diseas...	Link		
1.5	rs10784502	(T;T)	Less intracranial volume?	Link		
1.5	rs10883365	(A;G)	1.2x increased risk for developing Crohn's dise...	Link	Link	
1.5	rs10895068	(A;G)	2.5x increased odds of breast cancer among horm...	Link		
1.5	rs10980705	(C;T)	2.3x increased risk for knee osteoarthritis	Link		
1.5	rs1223271	(A;G)	Slightly increased risk of developing Parkinson...	Link	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	rs13181	(G;T)	1.12x increased risk for cutaneous melanoma	Link	Link	Link
1.5	rs13376333	(C;T)	1.5x higher risk of atrial fibrillation	Link	Link	
1.5	rs144848	(G;T)	Very slightly increased breast cancer risk	Link	Link	Link
1.5	rs16944	(A;G)	Minorly increased risk of mental illness and os...	Link	Link	
1.5	rs1801020	(C;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	rs1867277	(A;G)	1.5x increased risk for thyroid cancer	Link		
1.5	rs2007153	(G;G)	Increased risk of schizophrenia in limited stud...	Link		
1.5	rs2240340	(A;G)	Slightly increased (1.5x) risk for RA	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	rs2736990	(C;T)	Slightly increased risk of developing Parkinson...	Link	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	rs3745516	(A;G)	Slightly increased risk of developing primary b...	Link		
1.5	rs3825776	(A;G)	1.3x increased risk for ALS	Link	Link	
1.5	rs4027132	(A;G)	1.39x increased risk of developing bipolar diso...	Link		
1.5	rs4464148	(C;T)	1.10x increased risk for colorectal cancer	Link		
1.5	rs4626664	(A;G)	1.44x increased risk of developing restless leg...	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	rs486907	(A;G)	1.5x increased prostate cancer risk	Link	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	rs642961	(A;G)	1.68x increased risk of cleft lip	Link	Link	
1.5	rs699473	(C;T)	~1.5x increased brain tumor risk	Link		
1.5	rs7536563	(A;G)	1.12x risk of multiple sclerosis	Link	Link	
1.5	rs807701	(C;T)	Slightly increased dyslexia risk	Link		
1.5	rs872071	(A;G)	~1.5x increased risk for chronic lymphocytic le...	Link	Link	
1.5	rs9303277	(C;T)	1.46x Slightly increased risk of developing pri...	Link		
1.5	rs966221	(C;C)	1.5x increased stroke risk certain populations	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	rs12770228	(A;G)	1.4x increased risk for meningioma	Link		
1.4	rs1801157	(A;G)	1.4x higher risk for breast cancer	Link		
1.4	rs1893217	(C;T)	Slightly increased (1.4x) risk for Crohn's dise...	Link	Link	
1.4	rs6010620	(G;G)	1.4x higher risk for glioma development; but th...	Link	Link	
1.3	rs1047286	(C;T)	1.3x increased risk for age-related macular deg...	Link	Link	Link
1.3	rs1434536	(A;G)	1.29x increased breast cancer risk	Link		Link

Mag.	Identifier	Genotype	Summary	GnomAD	GetEvidence	ClinVar
1.3	rs16847548	(C;T)	1.3x increased risk for sudden cardiac death in...	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	rs2295490	(A;G)	1.32x increased risk of early-onset type-2 diab...	Link	Link	
1.3	rs2542151	(G;T)	1.3x risk for Crohn's; 1.3x for T1D	Link	Link	
1.3	rs34330	(C;T)	1.3x higher risk for endometrial cancer (in Chi...	Link		Link
1.3	rs7234029	(A;G)	Slightly increased (1.36x) risk for Crohn's dis...	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	rs10865331	(A;G)	1.2x higher risk for ankylosing spondylitis	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	rs2076295	(G;T)	One copy of the risk allele (G): slightly incre...	Link		
1.2	rs2665390	(C;T)	1.2x increased risk for ovarian cancer	Link		
1.2	rs4977756	(A;G)	1.39x higher risk for glioma development	Link	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		
1.17	rs17465637	(A;C)	1.17x higher risk for myocardial infarction	Link	Link	
1.1	rs10248420	(A;A)	Possibly less likely to remit on certain antide...	Link	Link	
1.1	rs11037909	(C;T)	1.27x type II diabetes risk	Link		
1.1	rs13387042	(A;G)	1.12x increased risk for breast cancer	Link	Link	
1.1	rs1344706	(G;T)	1.1x increased risk for schizophrenia	Link	Link	
1.1	rs2235040	(G;G)	Possibly lesser chances of remission only for i...	Link	Link	
1.1	rs2295190	(G;T)	Slightly increased risk for ovarian cancer in w...	Link	Link	Link
1.1	rs2651899	(A;G)	1.1x higher risk for migraines	Link		
1.1	rs2653349	(G;G)	2-6x increased risk for cluster headaches	Link	Link	
1.1	rs2828520	(G;G)	1.35x major depressive disorder risk	Link		
1.1	rs34516635	(G;G)	Less longevity for Ashkenazi Jewish women.	Link		Link
1.1	rs3740878	(A;G)	1.26x type II diabetes risk	Link		Link
1.1	rs4324715	(C;T)	1.5x increased testicular cancer risk for men	Link		
1.1	rs4977574	(A;G)	Some studies - but not others - report a slight...	Link	Link	
1.1	rs688034	(C;T)	1.1x risk higher risk for coronary artery disea...	Link	Link	
1.1	rs6897876	(C;T)	Slight increase in testicular cancer risk for m...	Link		
1.1	rs7531806	(A;G)	Very slightly increased risk of acne occurrence...	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;C)	1.5x risk of Crohn's disease: 1.2 for developin...	Link	Link	
1	rs1143674	(A;G)	1.3x increased autism risk	Link		
1	rs17300539	(G;G)	Increased risk of insulin resistance	Link		
1	rs2546890	(A;G)	Higher risk of multiple sclerosis	Link		
1	rs3194051	(A;A)	>1.1x risk of type-1 diabetes	Link	Link	Link
1	rs6932590	(T;T)	1.1x increased risk for schizophrenia	Link	Link	
1	rs761100	(G;G)	Higher risk for dyslexia	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	rs2304256	(C;C)	1.6x increased risk for SLE	Link	Link	Link
0.1	rs3095870	(A;G)	1.7x increased risk for SLE (lupus)	Link		

3.3 Genosets (Multi-variant Phenotypes)

Magnitude	Identifier	Summary
4	gs145	Female
3	gs273	Lowest risk (13% of white women) of Atrial Fibr...
2.5	gs155	CYP3A5 non-expressor
2.5	gs157	More stimulated by coffee
2.5	gs281	Part of the 88% of the population claimed not t...
2.5	gs285	Claimed to lose 2.5x as much weight on a low fa...
2.4	gs297	Lower heart attack risk than average
2	gs101	Probably able to digest milk
2	gs110	Higher allergic asthma risk
2	gs156	NAT2 Rapid metabolizer.
2	gs211	Ethanol biodisposition
2	gs244	2x increased risk for esophageal squamous cell ...
2	gs246	APOE E3/E3
2	gs249	Parkinson's Disease Risk
2	gs288	You have two long form 5-HTTLPR.
2	gs313	Normal DPYD activity and thus 5-FU metabolism p...
1.7	gs233	Normal pain sensitivity; APS/APS: LPS/APS: and ...
1.5	gs139	NAT2 intermediate metabolizer
1.5	gs185	The beta blocker metoprolol is effective: with ...
1.5	gs230	Possible Alzheimer's disease-related haplotype
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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