PGP-UK Genomics Report for uk6F5B69

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.

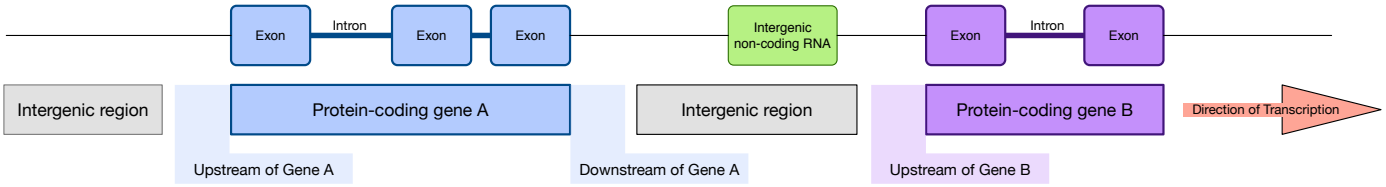

Figure 1: Diagram of gene structure indicating locations of potential variants

Feature	Count
Lines of input read	4690185
Variants filtered out	3721713
Novel / existing variants	0 (0.0) / 968472 (100.0)
Overlapped genes	51781
Overlapped transcripts	59192
Overlapped regulatory features	46967

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.

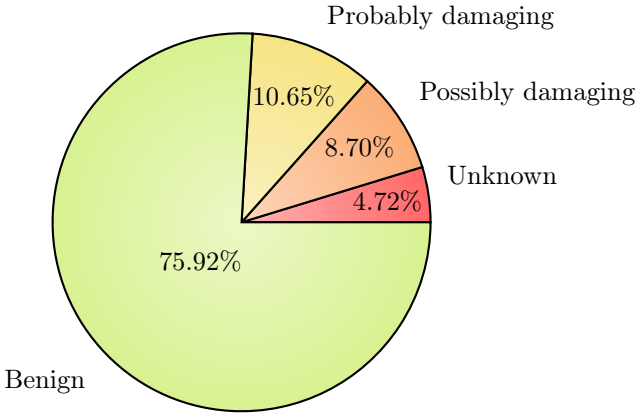

Figure 2: PolyPhen Summary

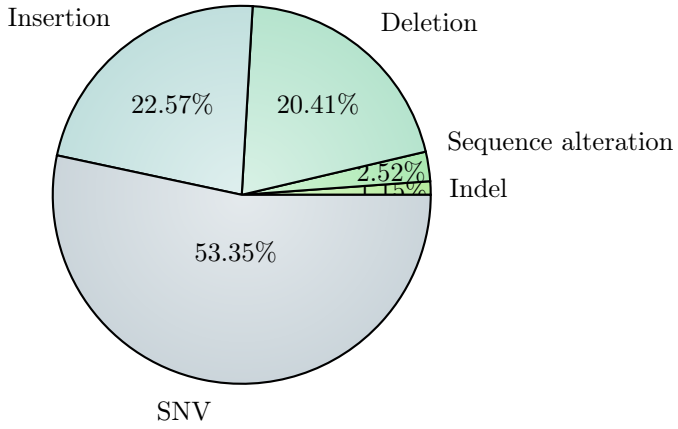

Figure 3: Variant Class

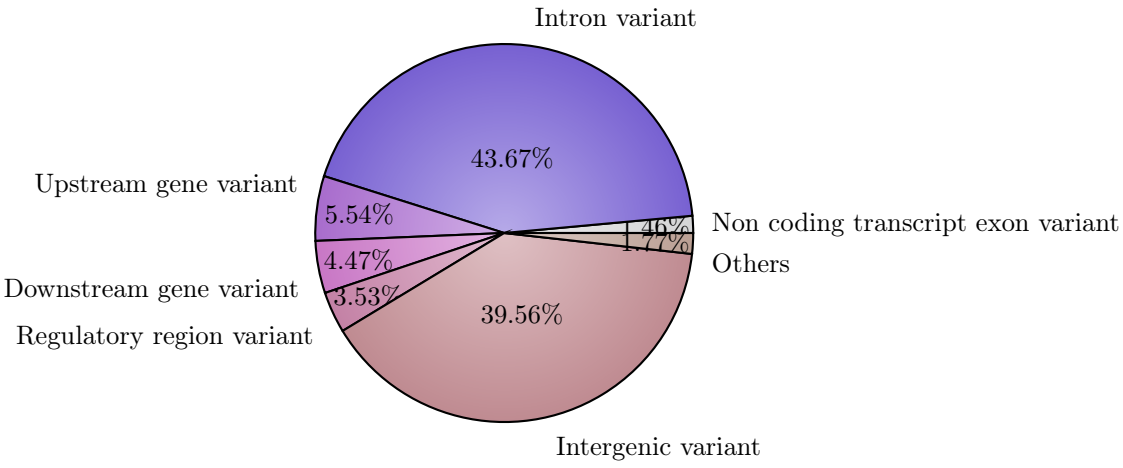

Figure 4: Consequence type

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).

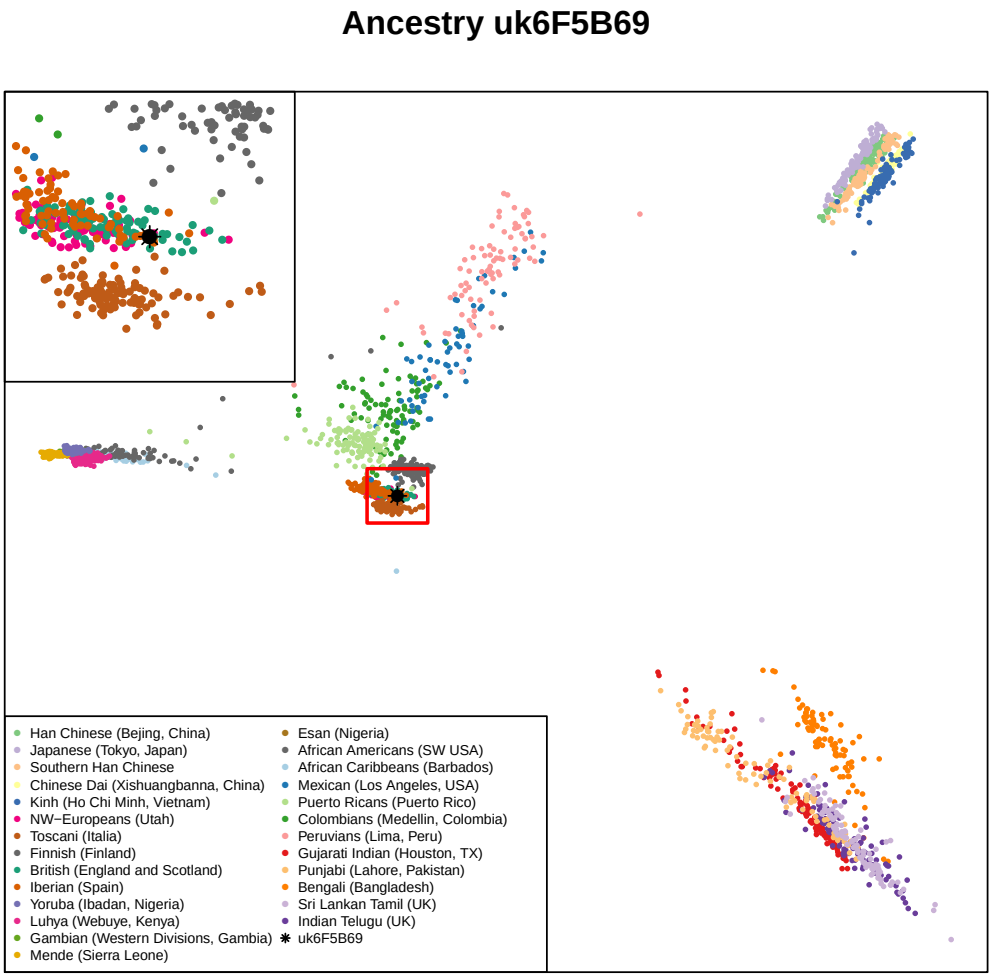

Figure 5: Ancestry Principal Component Analysis

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
3	rs8177374	(C;T)	Resistance to several diseases	Link	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	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	rs17070145	(C;T)	Increased memory performance	Link		Link
2	rs174537	(T;T)	Lower LDL-C and total cholesterol	Link		
2	rs1864163	(A;G)	Associated with higher HDL cholesterol	Link	Link	
2	rs2241423	(A;G)	0.79 decreased risk for obesity	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	rs3782179	(C;T)	3x lower odds of testicular cancer risk for men...	Link		
2	rs37973	(A;A)	Possibly better response to inhaled corticoster...	Link		Link
2	rs3819331	(T;T)	Lower risk of autism	Link		Link
2	rs3914132	(C;T)	Lower otosclerosis risk	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	rs7776725	(T;T)	Stronger bones	Link	Link	
2	rs801114	(T;T)	0.78x decreased Basal Cell Carcinoma risk.	Link	Link	
2	rs9642880	(G;G)	Slightly lower risk of Bladder Cancer.	Link	Link	
1.8	rs1746048	(C;T)	0.94 decreased risk for coronary heart disease	Link	Link	
1.8	rs187238	(C;G)	Hypertension not a risk factor for sudden cardi...	Link		
1.8	rs266729	(C;G)	0.73x decreased risk for colorectal 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		

Mag.	Identifier	Genotype	Summary	GnomAD	GetEvidence	ClinVar
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	rs1063192	(C;T)	0.71x reduced risk of myocardial infarction	Link		
1.5	rs11136000	(C;T)	0.84x decreased risk for Alzheimer's disease	Link	Link	
1.5	rs11465804	(G;T)	0.68x lower risk for spondylitis	Link	Link	
1.5	rs11635424	(A;G)	0.70x risk for restless legs	Link	Link	
1.5	rs1165205	(A;A)	0.85x decreased gout risk	Link	Link	
1.5	rs12593813	(A;G)	0.71x risk for restless legs	Link	Link	
1.5	rs3784709	(C;T)	0.71x risk of developing restless legs syndrome...	Link	Link	
1.5	rs3790844	(C;T)	Slightly reduced risk (0.77x) for pancreatic ca...	Link		
1.5	rs4149274	(C;C)	Associated with higher HDL (good) cholesterol.	Link		
1.5	rs4489954	(G;T)	0.69x risk risk of developing restless legs syn...	Link	Link	
1.5	rs4939883	(C;C)	Associated with higher HDL cholesterol	Link	Link	
1.5	rs5888	(C;C)	Higher HDL cholesterol but lower risk for age-r...	Link		
1.5	rs610932	(A;A)	A allele associated with reduced risk of Alzhei...	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		
1.4	rs6495446	(C;T)	0.8x reduced risk for chronic kidney disease	Link		
1.2	rs4686484	(G;G)	Slightly decreased risk for celiac disease	Link		
1.2	rs4867568	(C;T)	Decreased risk of knee osteoporosis	Link		
1.2	rs6048	(A;G)	Slightly lower risk (10-20%) of deep vein throm...	Link	Link	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	rs2293347	(G;G)	Among NSCLC patients: better Gefitinib response...	Link		Link
1.1	rs4988235	(T;T)	Can digest milk	Link		Link
1	rs182549	(T;T)	Can digest milk.	Link		Link
1	rs2351299	(G;T)	Possible reduced risk of Autism	Link		
1	rs2546890	(G;G)	Lower risk of multiple sclerosis	Link		
1	rs2952768	(C;T)	Slightly less drug dependence: decreased effect...	Link		Link
1	rs7850258	(A;A)	Slightly lower odds of developing primary hypot...	Link		
1.0	rs11246226	(C;C)	Decreased risk of schizophrenia in limited stud...	Link	Link	
1.0	rs6583817	(C;T)	~0.80x (lower) risk for late onset Alzheimer's ...	Link		

3.2 Possibly Harmful Traits

Mag.	Identifier	Genotype	Summary	GnomAD	GetEvidence	ClinVar
3.1	rs1421085	(C;C)	~1.7x increased obesity risk	Link	Link	Link
3	rs1121980	(T;T)	Moderate increase (2.76x) in risk for obesity	Link	Link	
3	rs16969968	(A;A)	Higher risk for nicotine dependence: lower risk...	Link	Link	Link
3	rs2981582	(C;T)	1.3x higher risk of ER+ breast cancer	Link	Link	
3	rs3903239	(C;C)	Higher frequency of atrial fibrillation	Link		
3	rs6920220	(A;G)	1.2x risk Rheumatoid Arthritis	Link	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	
2.6	rs8034191	(C;C)	1.80x lung cancer risk; decreased response to a...	Link	Link	
2.5	rs1051730	(T;T)	1.8x increased risk of lung cancer; reduced res...	Link	Link	Link
2.5	rs12803066	(A;G)	Increased risk of myopia	Link		
2.5	rs13266634	(C;T)	Increased risk for type-2 diabetes	Link	Link	Link
2.5	rs17487223	(T;T)	1.28x lung cancer risk	Link		
2.5	rs17696736	(G;G)	1.94x risk of type-1 diabetes	Link	Link	
2.5	rs339331	(T;T)	Prostate cancer risk	Link		
2.5	rs4143094	(G;T)	Slightly (17%) higher risk of colorectal cancer...	Link		
2.5	rs6441286	(G;G)	3.08x chance of developing primary biliary cirr...	Link	Link	
2.5	rs795484	(A;A)	Even more increased morphine dose requirement a...	Link		
2.5	rs891512	(A;G)	Higher blood pressure than G;G	Link		Link
2.3	rs7966230	(C;G)	Slightly lower levels of plasma VWF	Link		
2.2	rs2004640	(G;T)	1.4x increased risk for SLE	Link	Link	
2.2	rs944289	(T;T)	1.69x increased thyroid cancer risk	Link	Link	
2.1	rs2306402	(C;C)	1.18x increased risk for late-onset Alzheimer's...	Link		
2.1	rs2494732	(C;C)	Greater odds of cannabis-associated psychosis	Link	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	rs5186	(A;C)	~1.4x increased risk of hypertension	Link	Link	Link
2	rs1050631	(C;T)	Mean Survival Time of 25 months for esophageal ...	Link		
2	rs10811661	(C;T)	1.2x increased risk for type-2 diabetes	Link	Link	
2	rs10871777	(A;G)	Adults likely to be 0.22 BMI units higher	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	rs11123857	(A;G)	1.44-fold increased risk of bipolar disorder or...	Link		
2	rs1219648	(A;G)	1.20x risk for breast cancer	Link	Link	
2	rs12431733	(T;T)	Increased risk of developing Parkinson's Diseas...	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	rs16942	(G;G)	Very slightly increased breast cancer risk	Link	Link	Link
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	rs17782313	(C;T)	Adults likely to be 0.22 BMI units higher	Link	Link	Link
2	rs1799853	(T;T)	~ 40% reduction in warfarin metabolism: greater...	Link	Link	Link
2	rs1867277	(A;A)	2x increased risk for thyroid cancer	Link		
2	rs2073963	(G;T)	Increased risk of baldness	Link		
2	rs2201841	(T;T)	2.4x increased risk for Graves' disease	Link	Link	
2	rs2230199	(G;G)	2.5x+ 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	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	rs2420946	(C;T)	1.20x risk for breast cancer	Link		

Mag.	Identifier	Genotype	Summary	GnomAD	GetEvidence	ClinVar
2	rs25487	(A;G)	2x higher risk for skin cancer; possibly other ...	Link	Link	Link
2	rs27388	(A;A)	Increased risk of developing schizophrenia	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	rs3746444	(C;T)	~1.2x increased risk for cancer	Link		
2	rs3775948	(G;G)	Slightly higher risk for gout	Link		
2	rs3793784	(C;G)	1.5x risk for ARMD	Link	Link	Link
2	rs3825776	(G;G)	>1.3x increased risk for ALS	Link	Link	
2	rs4129148	(C;G)	3x risk of schizophrenia.	Link	Link	
2	rs4402960	(G;T)	1.2x increased risk for type-2 diabetes: ~1x ri...	Link	Link	Link
2	rs4464148	(C;C)	1.35x increased risk for colorectal cancer	Link		
2	rs4792311	(A;G)	Increased risk of prostate cancer	Link	Link	Link
2	rs493258	(A;G)	1.15x risk of Age Related Macular Degeneration	Link		
2	rs4961	(G;T)	1.8x increased risk for high blood pressure	Link	Link	Link
2	rs5174	(A;A)	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	rs6498169	(A;A)	>1.14x risk of multiple sclerosis	Link	Link	
2	rs6603272	(G;T)	2.74x increased risk of developing schizophreni...	Link		
2	rs662799	(A;G)	1.4x higher early heart attack risk; less weigh...	Link	Link	Link
2	rs663048	(G;T)	Significantly increased risk of developing lung...	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	rs7190458	(A;G)	Slightly higher pancreatic cancer risk	Link		
2	rs7216389	(T;T)	1.5x increased risk for Childhood Asthma.	Link	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;C)	1.4x risk for Crohn's disease	Link	Link	
2	rs7961152	(A;A)	1.5x higher risk for hypertension	Link		
2	rs800292	(C;C)	5% higher risk of Age related macular degenerat...	Link	Link	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	rs9543325	(C;C)	1.37x Slightly higher pancreatic cancer risk	Link		
2	rs9652490	(A;A)	~2x increased risk for Parkinson's disease: and...	Link	Link	
2	rs965513	(A;A)	3.1x increased thyroid cancer risk	Link	Link	
2.0	rs2156921	(G;G)	1.29x increased risk for depression	Link		
1.8	rs1136287	(C;T)	1.5x increased risk of wet ARMD in a Taiwanese ...	Link	Link	Link
1.8	rs1800587	(C;T)	Slightly higher risk for lumbar disc disease	Link		
1.8	rs4474514	(A;G)	3x increased testicular cancer risk for men	Link	Link	
1.5	rs10492519	(A;G)	Slightly increased risk of developing prostate ...	Link		
1.5	rs10509681	(C;T)	Increased risk of GI bleeding with NSAIDs	Link	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	rs11572080	(A;G)	1.81x risk of GI bleeding with NSAID drugs	Link	Link	Link
1.5	rs12037606	(A;G)	1.22x risk of developing Crohn's disease	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	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	rs140701	(A;G)	Increased risk for anxiety disorders	Link		
1.5	rs144848	(G;T)	Very slightly increased breast cancer risk	Link	Link	Link
1.5	rs165599	(G;G)	May indicate increased susceptibility to schizo...	Link	Link	

Mag.	Identifier	Genotype	Summary	GnomAD	GetEvidence	ClinVar
1.5	rs16944	(A;G)	Minorly increased risk of mental illness and os...	Link	Link	
1.5	rs17221417	(C;G)	1.3x higher risk for Crohn's 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	rs2241880	(C;T)	1.4x increased risk for Crohn's disease in Cauc...	Link	Link	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	rs2282679	(C;C)	Lower vitamin D levels	Link		
1.5	rs2736990	(C;T)	Slightly increased risk of developing Parkinson...	Link	Link	
1.5	rs28694718	(A;A)	>2x higher risk for schizophrenia	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	rs3764880	(A;G)	Possible 1.2 - 1.8x increased tuberculosis susc...	Link	Link	
1.5	rs3790565	(C;T)	Slightly increased risk of developing primary b...	Link		
1.5	rs4506565	(A;T)	1.4x increased risk for type-2 diabetes	Link	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	rs4785763	(A;C)	1.5x higher risk for melanoma	Link	Link	
1.5	rs4845618	(G;T)	1.7x increased melanoma risk	Link		
1.5	rs486907	(A;G)	1.5x increased prostate cancer risk	Link	Link	Link
1.5	rs4939827	(T;T)	1x risk for colorectal cancer	Link	Link	Link
1.5	rs5746059	(A;A)	Slightly higher fat mass	Link		
1.5	rs6532197	(A;G)	Slightly increased risk of developing Parkinson...	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	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	rs807701	(C;T)	Slightly increased dyslexia risk	Link		
1.5	rs872071	(G;G)	~1.5x increased risk for chronic lymphocytic le...	Link	Link	
1.5	rs966221	(C;C)	1.5x increased stroke risk certain populations	Link		
1.4	rs10134944	(C;T)	1.4x risk of bipolar disorder.	Link	Link	
1.4	rs10865331	(A;A)	1.4x higher risk for ankylosing spondylitis	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	rs2228314	(C;G)	1.48x risk of osteoarthritis	Link	Link	
1.4	rs3184504	(C;T)	Slightly increased risk for celiac disease	Link	Link	
1.4	rs6010620	(G;G)	1.4x higher risk for glioma development; but th...	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	rs1047286	(C;T)	1.3x increased risk for age-related macular deg...	Link	Link	Link
1.3	rs1375144	(C;T)	1.32x increased risk of developing bipolar diso...	Link		
1.3	rs1434536	(A;G)	1.29x increased breast cancer risk	Link		Link
1.3	rs2059693	(C;T)	1.3x increased risk for testicular cancer	Link		
1.3	rs2736100	(G;T)	1.3x higher risk for glioma development: 2.1x r...	Link	Link	Link
1.3	rs356219	(A;G)	1.3x increased risk for Parkinson's disease	Link		
1.3	rs501120	(A;G)	1.3x increased risk for heart disease	Link	Link	
1.3	rs7234029	(A;G)	Slightly increased (1.36x) risk for Crohn's dis...	Link		
1.2	rs10210302	(C;T)	1.2x increased risk for Crohn's disease	Link	Link	
1.2	rs11037909	(T;T)	1.47x type II diabetes risk	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	rs143383	(C;T)	1.1x increased risk for osteoarthritis	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		

Mag.	Identifier	Genotype	Summary	GnomAD	GetEvidence	ClinVar
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		
1.2	rs2252586	(A;G)	1.2x higher risk for glioma development	Link		
1.2	rs2254958	(C;T)	1.24x reported increased risk for Alzheimer's; ...	Link		
1.2	rs2651899	(G;G)	1.2x higher risk for migraines	Link		
1.2	rs2665390	(C;T)	1.2x increased risk for ovarian cancer	Link		
1.2	rs2814707	(A;G)	1.2x increased risk for ALS	Link	Link	
1.2	rs3740878	(A;A)	1.46x type II diabetes risk; common	Link		Link
1.2	rs3849942	(A;G)	1.2x increased risk for ALS	Link	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	rs7514229	(G;G)	Associated with early-onset autoimmune thyroid ...	Link		
1.2	rs7528684	(G;G)	1.2x risk of Rheumatoid Arthritis; various risk...	Link		
1.17	rs3802842	(A;C)	1.17x increased risk of colorectal cancer	Link	Link	
1.1	rs10248420	(A;A)	Possibly less likely to remit on certain antide...	Link	Link	
1.1	rs13387042	(A;G)	1.12x increased risk for breast cancer	Link	Link	
1.1	rs1800450	(A;G)	Carrier of mannose binding deficiency but of lo...	Link	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	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	rs5030737	(C;T)	Carrier of mannose binding deficiency but of lo...	Link	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	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	rs1010	(A;G)	1.75x risk of MI	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	rs3194051	(A;A)	>1.1x risk of type-1 diabetes	Link	Link	Link
1	rs6932590	(C;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	
0.5	rs1566734	(G;T)	Somatic mutation: cancer associated	Link	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		
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
6	gs216	2 copies of the APOE-Î¼4*allele*
4	gs145	Female
3.5	gs126	Poor warfarin metabolizer
3.3	gs162	CYP2C9 Poor Metabolizers
3.1	gs191	Impaired NSAID drug metabolism
3	gs137	5x risk of thyroid cancer
2.7	gs311	Slow metabolizer of certain substances
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	gs154	NAT2 Slow metabolizer
2	gs211	Ethanol biodisposition
2	gs244	2x increased risk for esophageal squamous cell ...
2	gs290	You might have two short form 5-HTTLPR.
2	gs313	Normal DPYD activity and thus 5-FU metabolism p...
1.7	gs232	Possible low pain sensitivity; LPS/LPS
1.5	gs185	The beta blocker metoprolol is effective: with ...
1.5	gs230	Possible Alzheimer's disease-related haplotype
1.5	gs247	Parkinson's Disease Risk

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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