PGP-UK Genomics Report for ukB4491A

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.

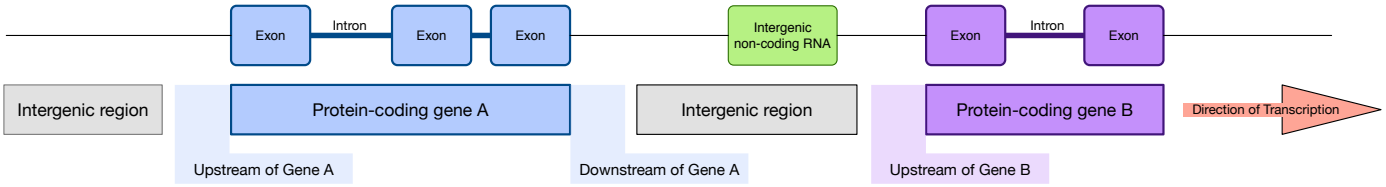

Figure 1: Diagram of gene structure indicating locations of potential variants

Feature	Count
Lines of input read	4801422
Variants filtered out	3261194
Novel / existing variants	0 (0.0) / 1540228 (100.0)
Overlapped genes	55407
Overlapped transcripts	64230
Overlapped regulatory features	143184

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.

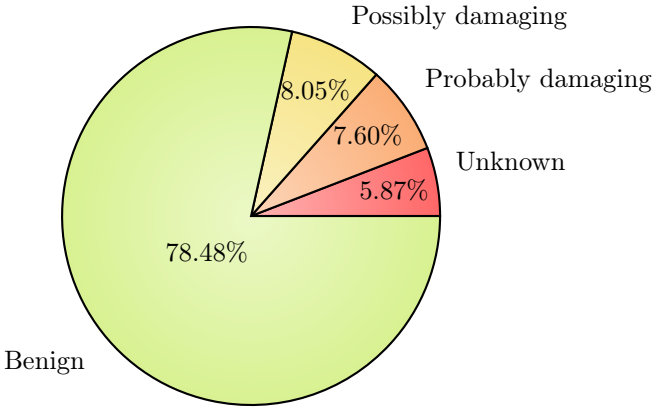

Figure 2: PolyPhen Summary

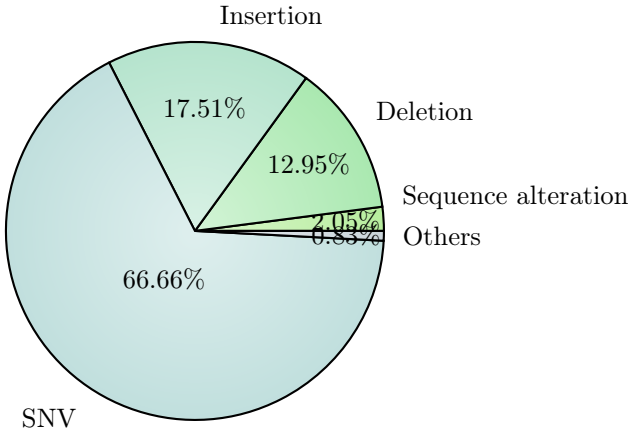

Figure 3: Variant Class

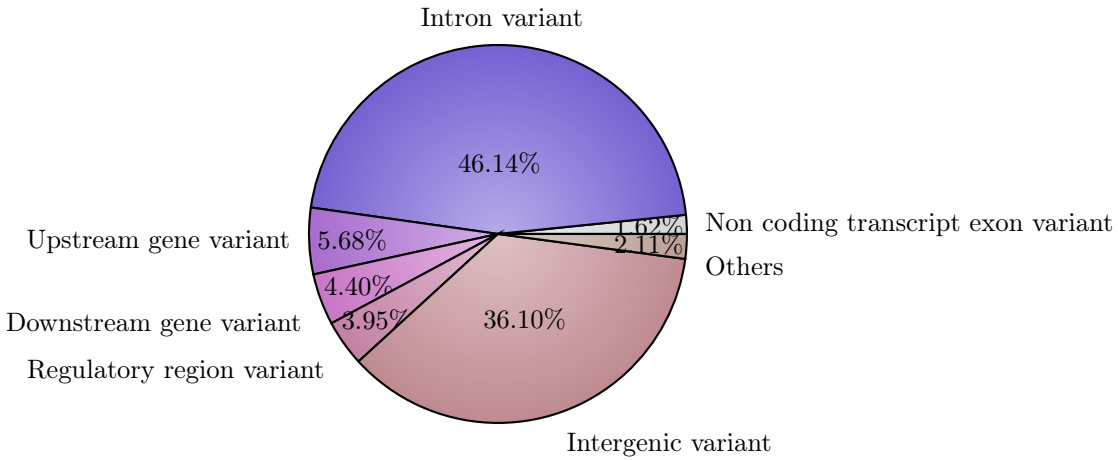

Figure 4: Consequence type

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).

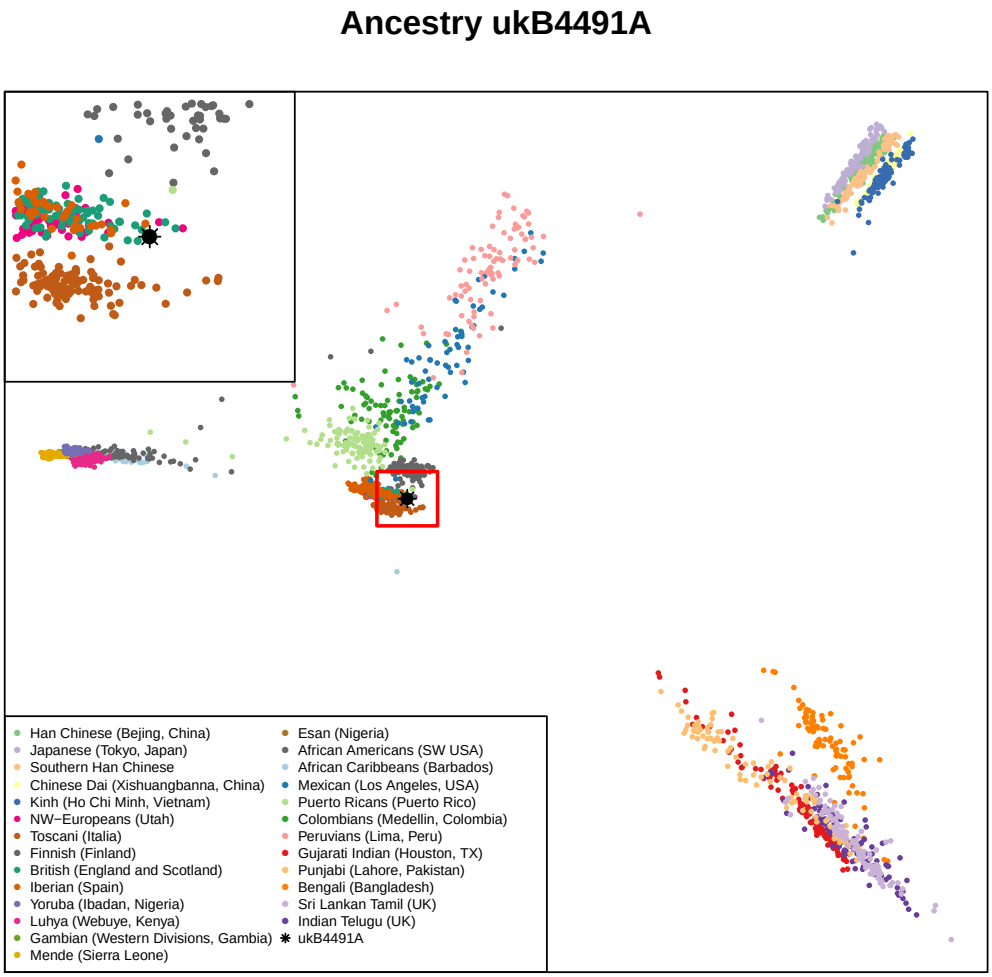

Figure 5: Ancestry Principal Component Analysis

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	rs3764261	(T;T)	Associated with higher HDL cholesterol. HDL	Link	Link	Link
2.4	rs3750817	(T;T)	0.64x reduced risk for breast cancer: and highe...	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	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	rs1229984	(A;G)	0.56x decreased risk of oral/throat cancers	Link	Link	Link
2	rs12979860	(C;C)	~80% of such hepatitis C patients respond to tr...	Link	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	rs2241766	(G;T)	Slightly lower risk of breast cancer	Link		
2	rs2542052	(C;C)	Better odds of living to 100	Link		
2	rs3738579	(C;T)	0.5x decreased risk for cervical cancer: HNSCC:...	Link		
2	rs3782179	(C;T)	3x lower odds of testicular cancer risk for men...	Link		
2	rs3819331	(T;T)	Lower risk of autism	Link		Link
2	rs4149268	(A;G)	Associated with higher HDL cholesterol	Link	Link	
2	rs6855911	(A;G)	0.62x decreased risk for gout	Link	Link	
2	rs7101429	(G;G)	0.70x reduced risk for Alzheimer's risk	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	rs801114	(T;T)	0.78x decreased Basal Cell Carcinoma risk.	Link	Link	
2	rs9525638	(C;C)	Stronger bones	Link		

Mag.	Identifier	Genotype	Summary	GnomAD	GetEvidence	ClinVar
1.8	rs1128535	(A;G)	0.77x risk for Crohn's disease	Link		
1.8	rs3814113	(C;T)	0.8x decreased risk for ovarian 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
1.6	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;C)	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	rs1165205	(A;A)	0.85x decreased gout risk	Link	Link	
1.5	rs2229169	(C;C)	1.5x decreased risk of heart attack and stroke ...	Link		
1.5	rs3851179	(A;A)	0.85x decreased risk for Alzheimer's disease	Link	Link	
1.5	rs4149274	(C;T)	Associated with higher HDL (good) cholesterol	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		
1.4	rs2294008	(C;C)	Lower risk of gastric and bladder cancer	Link	Link	
1.4	rs4320932	(G;G)	0.74x decreased risk for ovarian cancer	Link		
1.4	rs6495446	(C;T)	0.8x reduced risk for chronic kidney disease	Link		
1.2	rs11172113	(C;C)	0.8x lower risk for migraines	Link		
1.2	rs4867568	(C;T)	Decreased risk of knee osteoporosis	Link		
1.2	rs9306160	(C;T)	0.75x (reduced) risk for metastasis in LN-/ER+ ...	Link	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
1.1	rs4988235	(T;T)	Can digest milk	Link		Link
1.1	rs7568369	(T;T)	0.90x reduced risk of obesity	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	rs33927012	(C;T)	Currently evaluated as benign in ClinVar	Link	Link	Link
1	rs4148739	(A;G)	Possibly: inpatients more likely to remit on ce...	Link	Link	
1	rs4752566	(T;T)	Associated with thicker hair in Asians	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		
1	rs800292	(C;T)	1% decreased risk of macular degeneration	Link	Link	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
3.1	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	rs1983132	(C;T)	2 - 3x higher prostate cancer risk if routinely...	Link		
3	rs2237717	(T;T)	Reduced abilities related to neurocognition and...	Link		
3	rs3803662	(T;T)	1.6x increased risk for breast cancer	Link	Link	
3	rs3903239	(C;C)	Higher frequency of atrial fibrillation	Link		
2.5	rs10484554	(C;T)	2.8x increased risk for psoriasis	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	rs1800255	(A;A)	Increased risk for pelvic organ prolapse	Link	Link	Link
2.5	rs187238	(G;G)	Hypertension increases risk 3.75x for sudden ca...	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	rs5219	(T;T)	2.5x increased risk for type-2 diabetes	Link	Link	Link
2.5	rs6441286	(G;G)	3.08x chance of developing primary biliary cirr...	Link	Link	
2.3	rs7966230	(C;G)	Slightly lower levels of plasma VWF	Link		
2.2	rs283413	(G;T)	3x higher risk for PD	Link	Link	Link
2.1	rs10811661	(T;T)	1.2x increased risk for type-2 diabetes	Link	Link	
2.1	rs1585215	(G;G)	3.5x increased risk for Hodgkin lymphoma	Link		
2.1	rs17070145	(C;C)	Reduced memory abilities	Link		Link
2.1	rs2494732	(C;C)	Greater odds of cannabis-associated psychosis	Link	Link	
2.1	rs5186	(A;C)	~1.4x increased risk of hypertension	Link	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	rs795484	(A;G)	Increased morphine dose requirement and postope...	Link		
2.1	rs944289	(C;T)	1.3x increased thyroid cancer risk	Link	Link	
2	rs10455872	(A;G)	1:51x increased Coronary Heart disease risk	Link		Link
2	rs1045642	(C;T)	Slower metaboliser for some drugs	Link	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;G)	1.17x increased risk for multiple sclerosis	Link	Link	
2	rs11190870	(C;T)	Possibly increased risk of scoliosis	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	rs12431733	(T;T)	Increased risk of developing Parkinson's Diseas...	Link	Link	
2	rs12469063	(G;G)	Increased risk of developing restless legs synd...	Link		
2	rs1265181	(C;G)	Increased risk for psoriasis	Link	Link	
2	rs12696304	(C;G)	Prone to aging faster: at least in European pop...	Link		
2	rs12770228	(A;A)	2x increased risk for meningioma	Link		
2	rs13254738	(C;C)	1.18x prostate cancer risk	Link	Link	
2	rs1333048	(A;C)	1.3x increased coronary artery disease risk	Link		
2	rs1544410	(A;A)	Increased risk of low bone mineral density diso...	Link	Link	
2	rs16942	(A;G)	Very slightly increased breast cancer risk	Link	Link	Link
2	rs16944	(A;A)	Increased risk (~3x) for osteoarthritis	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
2	rs17696736	(A;G)	1.34x risk of type-1 diabetes	Link	Link	
2	rs17782313	(C;T)	Adults likely to be 0.22 BMI units higher	Link	Link	Link
2	rs1799732	(-;C)	1.3x increased adenoma recurrence risk	Link	Link	

Mag.	Identifier	Genotype	Summary	GnomAD	GetEvidence	ClinVar
2	rs1800629	(A;G)	Complex; generally higher risk for certain dise...	Link	Link	Link
2	rs1801516	(A;A)	2.76x odds of pancreatic cancer: but 0.86x redu...	Link	Link	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	rs2305480	(T;T)	If 4 years old or younger: ~3x increased asthma...	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	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	rs2707466	(G;G)	Weaker bones	Link	Link	
2	rs2736100	(T;T)	Higher risk of Interstitial lung disease: and t...	Link	Link	Link
2	rs2736990	(C;C)	Slightly increased risk of developing Parkinson...	Link	Link	
2	rs2908004	(C;C)	Weaker bones	Link	Link	
2	rs326	(A;A)	Lower HDL cholesterol	Link	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	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	rs4825476	(G;G)	1.9x higher risk of suicidal thoughts when taki...	Link	Link	
2	rs4961	(G;T)	1.8x increased risk for high blood pressure	Link	Link	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;A)	Increased risk in men for biliary conditions	Link		
2	rs642961	(A;A)	2.40x increased risk of cleft lip	Link	Link	
2	rs6457617	(C;T)	2.3x risk of rheumatoid arthritis	Link	Link	
2	rs6601764	(C;C)	1.52x increased risk of developing Crohn's dise...	Link	Link	
2	rs6603272	(G;T)	2.74x increased risk of developing schizophreni...	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	rs699	(C;T)	Increased risk of hypertension	Link	Link	Link
2	rs763361	(T;T)	Increased risk for multiple autoimmune diseases...	Link	Link	
2	rs7774434	(C;C)	Increased risk of developing primary biliary ci...	Link		
2	rs7794745	(A;T)	Slightly increased risk for autism	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	rs9303277	(T;T)	1.46x Increased risk of developing primary bili...	Link		
2	rs965513	(A;G)	1.77x increased thyroid cancer risk	Link	Link	
2.0	rs2305795	(A;A)	1.64x higher risk of narcolepsy compared to (G;...	Link		Link
2.0	rs4911414	(G;T)	2-4x higher risk of sun sensitivity if part of ...	Link	Link	
1.9	rs7923837	(A;G)	1.6x risk for T2D	Link		
1.8	rs1136287	(C;T)	1.5x increased risk of wet ARMD in a Taiwanese ...	Link	Link	Link
1.8	rs2278206	(T;T)	1.16x increased risk for asthma	Link	Link	
1.8	rs4474514	(A;G)	3x increased testicular cancer risk for men	Link	Link	
1.7	rs4807015	(C;T)	1.74x risk of type 2 diabetes	Link		
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	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	rs10260404	(C;T)	1.20x risk of developing ALS	Link	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	

Mag.	Identifier	Genotype	Summary	GnomAD	GetEvidence	ClinVar
1.5	rs10980705	(C;T)	2.3x increased risk for knee osteoarthritis	Link		
1.5	rs11171739	(C;T)	1.34x risk of developing Type-1 diabetes	Link	Link	
1.5	rs13149290	(C;T)	Slightly increased risk of developing prostate ...	Link		
1.5	rs13376333	(C;T)	1.5x higher risk of atrial fibrillation	Link	Link	
1.5	rs17221417	(C;G)	1.3x higher risk for Crohn's disease	Link	Link	
1.5	rs1867277	(A;G)	1.5x increased risk for thyroid cancer	Link		
1.5	rs1975197	(C;T)	1.3x increased risk of developing restless legs...	Link	Link	
1.5	rs2007153	(G;G)	Increased risk of schizophrenia in limited stud...	Link		
1.5	rs2286812	(C;T)	~2x higher risk for Fuchs' dystrophy: a corneal...	Link		
1.5	rs2697962	(A;G)	Slightly increased risk of developing Parkinson...	Link		
1.5	rs27388	(A;G)	Slightly increased risk of developing schizophr...	Link		
1.5	rs28694718	(A;A)	>2x higher risk for schizophrenia	Link		
1.5	rs2881766	(G;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	rs309375	(T;T)	Larger mosquito bites	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	rs358806	(A;C)	0.86x increased risk of developing Type-2 diabe...	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	rs393152	(A;A)	Increased risk of both PD and AD	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	rs4585	(T;T)	Slightly poorer (0.75x) response to metformin i...	Link		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	rs4785763	(A;C)	1.5x higher risk for melanoma	Link	Link	
1.5	rs486907	(A;G)	1.5x increased prostate cancer risk	Link	Link	Link
1.5	rs5746059	(A;G)	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	rs6710341	(A;G)	Slightly increased risk of developing restless ...	Link		
1.5	rs6908425	(C;T)	1.63x increased risk of developing Crohn's dise...	Link	Link	
1.5	rs7341475	(G;G)	1.58x increased schizophrenia risk for women	Link	Link	
1.5	rs7454108	(C;T)	Single HLA-DQ8 haplotype	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	rs9642880	(G;T)	1.2x increased bladder cancer risk	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	rs1545843	(A;A)	1.4x increased risk for depression (for those u...	Link		
1.4	rs1800693	(G;G)	Slight (1.4x) increase in risk for multiple scl...	Link	Link	Link
1.4	rs2046210	(C;T)	1.4x increased breast cancer risk	Link	Link	Link
1.4	rs3131296	(G;G)	1.4x increased risk for schizophrenia	Link	Link	
1.4	rs3184504	(C;T)	Slightly increased risk for celiac disease	Link	Link	
1.4	rs8050136	(A;A)	1.4x increased risk for T2D in some populations...	Link	Link	
1.3	rs1042713	(A;G)	1.3x increased risk that pediatric inhaler use ...	Link	Link	Link
1.3	rs1047286	(C;T)	1.3x increased risk for age-related macular deg...	Link	Link	Link
1.3	rs10947262	(C;C)	1.3x increased risk for osteoarthritis	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	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	rs2059693	(C;T)	1.3x increased risk for testicular cancer	Link		

Mag.	Identifier	Genotype	Summary	GnomAD	GetEvidence	ClinVar
1.3	rs2295490	(A;G)	1.32x increased risk of early-onset type-2 diab...	Link	Link	
1.3	rs356219	(A;G)	1.3x increased risk for Parkinson's disease	Link		
1.25	rs748404	(T;T)	Slightly increased risk (1.25) for lung cancer...	Link	Link	
1.2	rs11037909	(T;T)	1.47x type II diabetes risk	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	rs2254958	(C;T)	1.24x reported increased risk for Alzheimer's; ...	Link		
1.2	rs2814707	(A;G)	1.2x increased risk for ALS	Link	Link	
1.2	rs35677470	(A;G)	2x higher risk for scleroderma	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	rs4686484	(A;A)	Slightly increased risk for celiac disease	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	rs6897876	(C;C)	Slight increase in testicular cancer risk for m...	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		
1.2	rs9960767	(A;C)	1.2x increased risk for schizophrenia	Link	Link	
1.17	rs17465637	(A;C)	1.17x higher risk for myocardial infarction	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	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	rs34516635	(G;G)	Less longevity for Ashkenazi Jewish women.	Link		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	rs7171755	(A;A)	Very slight descrease in cortical thickness and...	Link		
1.1	rs889312	(A;C)	Very slightly higher risk for breast cancer	Link	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	rs11206244	(C;T)	Slight risk of decreased thyroid hormone metabo...	Link		
1	rs1143674	(A;G)	1.3x increased autism risk	Link		
1	rs2546890	(A;A)	Higher risk of multiple sclerosis	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	rs6976	(C;T)	Slight risk of osteoarthritis	Link		
1	rs7453920	(G;G)	Slight increase in risk for chronic hepatitis B...	Link		
1	rs761100	(G;G)	Higher risk for dyslexia	Link		
1.0	rs11246226	(A;A)	Increased risk of schizophrenia in limited stud...	Link	Link	
0.1	rs11110912	(C;G)	Maybe some quite minor increase in high blood p...	Link		
0.1	rs3095870	(A;G)	1.7x 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
4	gs144	Male
3.5	gs112	Haplogroup I2 (Y-DNA)
3.1	gs122	7x risk of male baldness
3	gs241	Lighter green: brown or hazel eye color
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	gs242	Increased risk of individuals with prostate can...
2.5	gs259	Homozygous for eye color haplotype #3
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
2.3	gs255	Homozygous eye color haplotype #1
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	gs173	CYP2D6*10
2	gs211	Ethanol biodisposition
2	gs221	Autoimmune disorder risk in Europeans
2	gs244	2x increased risk for esophageal squamous cell ...
2	gs313	Normal DPYD activity and thus 5-FU metabolism p...
2	gs317	Parkinson's risk might be decreased depending u...
1.7	gs233	Normal pain sensitivity; APS/APS: LPS/APS: and ...
1.6	gs196	Haplogroup I (Y-DNA); 50% higher age-adjusted r...
1.5	gs230	Possible Alzheimer's disease-related haplotype
1.5	gs247	Parkinson's Disease Risk
1.2	gs184	Able to taste bitterness.
1.1	gs138	NAT2 rapid metabolizer
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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