

PGP-UK Genomics Report for uk97975D

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.

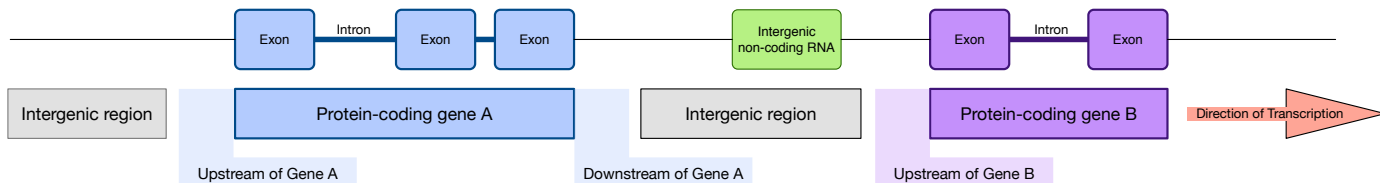


Figure 1: Diagram of gene structure indicating locations of potential variants

Feature	Count
Lines of input read	4787383
Variants filtered out	3262044
Novel / existing variants	0 (0.0) / 1525339 (100.0)
Overlapped genes	55236
Overlapped transcripts	64014
Overlapped regulatory features	142151

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.

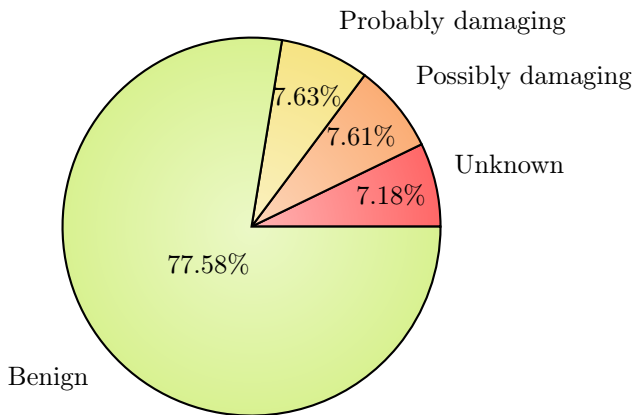


Figure 2: PolyPhen Summary

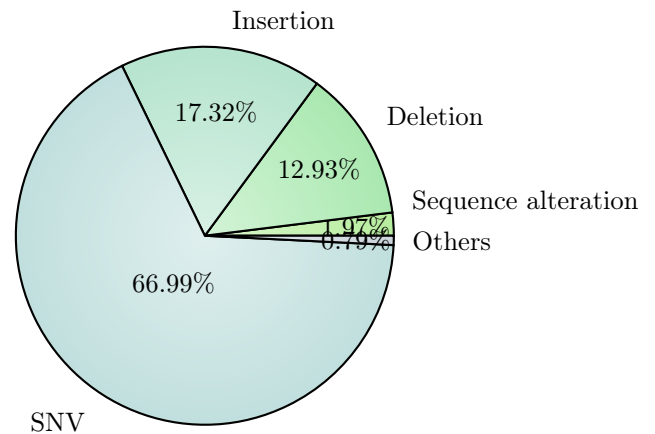


Figure 3: Variant Class

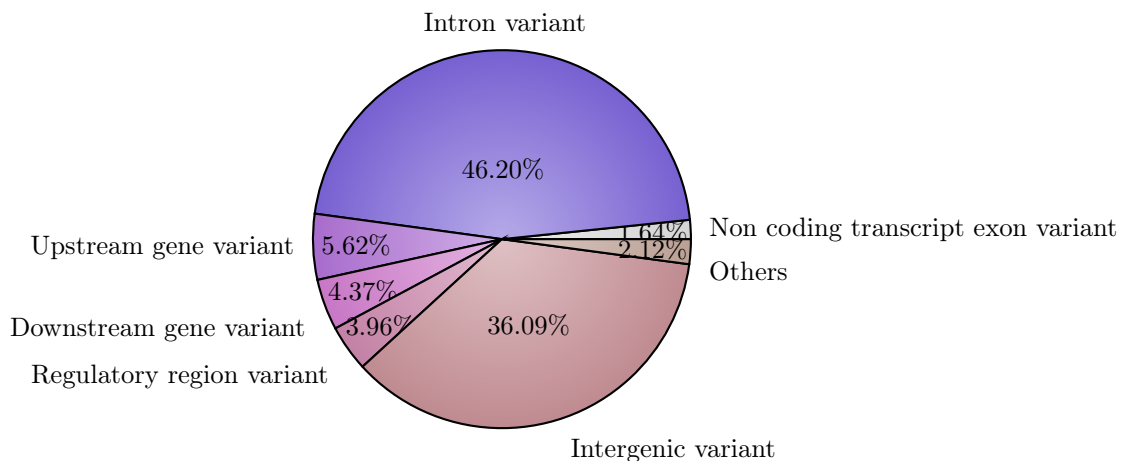


Figure 4: Consequence type

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).

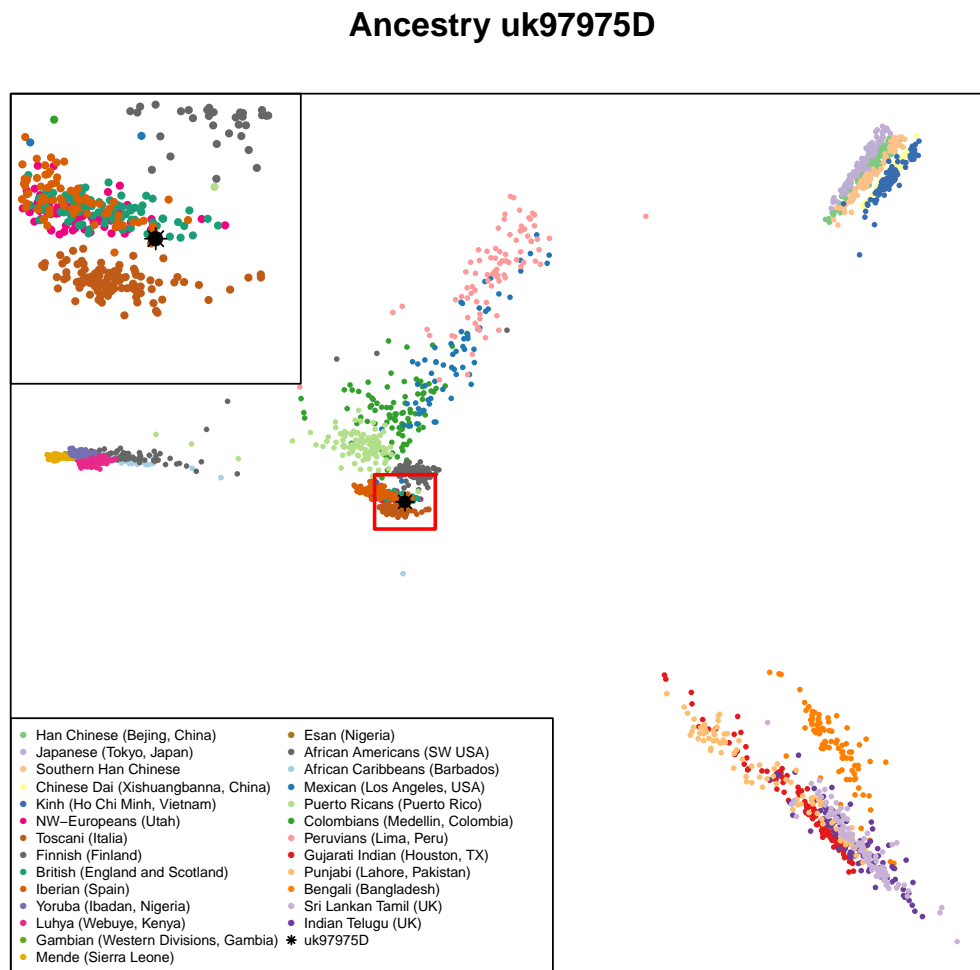


Figure 5: Ancestry Principal Component Analysis

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	
2.1	rs3775291	(A;G)	0.71x decreased risk for dry age related macula...	Link	Link	Link
2	rs10468017	(C;T)	Associated with higher HDL cholesterol	Link	Link	
2	rs10784502	(C;C)	Better intracranial volume?	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	rs1544410	(G;G)	Decreased risk of low bone mineral density diso...	Link	Link	
2	rs17070145	(C;T)	Increased memory performance	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	(A;G)	Associated with higher HDL cholesterol	Link	Link	
2	rs261332	(A;G)	Associated with higher HDL cholesterol	Link		
2	rs3736309	(A;G)	0.44x decreased risk for chronic obstructive pu...	Link		
2	rs3738579	(C;T)	0.5x decreased risk for cervical cancer: HNSCC:...	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	rs4585	(G;G)	Slightly higher (1.35x) odds of good metformin ...	Link		Link
2	rs6505162	(A;C)	0.58x decreased risk for esophageal cancer	Link		
2	rs6855911	(A;G)	0.62x decreased risk for gout	Link	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	rs8070723	(A;G)	0.18x reduced risk of developing progressive su...	Link		

Mag.	Identifier	Genotype	Summary	GnomAD	GetEvidence	ClinVar
1.8	rs1128535	(A;G)	0.77x risk for Crohn's disease	Link		
1.8	rs1746048	(C;T)	0.94 decreased risk for coronary heart disease	Link	Link	
1.8	rs1800588	(C;T)	Higher HDL-C levels	Link	Link	
1.8	rs4714156	(C;C)	<0.61x risk for restless legs	Link		
1.8	rs6897932	(C;T)	0.91x decreased risk for multiple sclerosis	Link	Link	Link
1.8	rs854560	(T;T)	0.5x lower risk of ovarian cancer	Link	Link	Link
1.6	rs1061170	(T;T)	Lower risk for AMD: generally longer live than ...	Link	Link	Link
1.5	rs1050631	(C;C)	Mean Survival Time of 32 months for esophageal ...	Link		
1.5	rs1063192	(C;C)	0.71x reduced risk of myocardial infarction	Link		
1.5	rs11136000	(T;T)	0.84x decreased risk for Alzheimer's disease	Link	Link	
1.5	rs11212617	(C;C)	Somewhat increased likelihood of treatment succ...	Link		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;T)	Associated with higher HDL cholesterol	Link	Link	
1.5	rs610932	(A;A)	A allele associated with reduced risk of Alzhei...	Link		
1.4	rs1165205	(A;T)	0.85x decreased gout risk	Link	Link	
1.4	rs6495446	(C;T)	0.8x reduced risk for chronic kidney disease	Link		
1.4	rs9402571	(G;T)	Slightly decreased risk for type-2 diabetes	Link		
1.3	rs9306160	(T;T)	0.75x (reduced) risk for metastasis in LN-/ER+ ...	Link	Link	
1.2	rs11246226	(A;C)	Decreased risk of schizophrenia in limited stud...	Link	Link	
1.2	rs4686484	(G;G)	Slightly decreased risk for celiac disease	Link		
1.2	rs4867568	(T;T)	Decreased risk for knee osteoporosis	Link		
1.1	rs10166942	(C;T)	0.85x 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
1.1	rs7568369	(G;T)	0.90x reduced risk of obesity	Link		
1.1	rs7611694	(C;C)	Lower prostate cancer risk?	Link		
1	rs11601907	(C;T)	Variant allele is designated benign in ClinVar	Link		Link
1	rs182549	(T;T)	Can digest milk.	Link		Link
1	rs2546890	(G;G)	Lower risk of multiple sclerosis	Link		
1	rs4752566	(G;T)	Associated with thicker hair in Asians	Link		
1	rs4939827	(C;T)	0.86x decreased risk for colorectal cancer	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
4	rs118020901	(A;C)	Corneal dystrophy: fuchs endothelial: 6	Link		Link
3.2	rs2981582	(T;T)	1.7x higher risk of ER+ breast cancer	Link	Link	
3.1	rs1421085	(C;C)	~1.7x increased obesity risk	Link	Link	Link
3	rs1121980	(T;T)	Moderate increase (2.76x) in risk for obesity	Link	Link	
3	rs7754840	(C;G)	1.3x increased risk for type-2 diabetes	Link	Link	
2.5	rs10490924	(G;T)	2.7x risk for age related macular degeneration	Link	Link	Link
2.5	rs1057910	(A;C)	CYP2C9*3 carrier; average 40% reduction in warf...	Link	Link	Link
2.5	rs11190870	(T;T)	Possibly even more increased risk of scoliosis	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	rs2241880	(C;C)	2x-3x increased risk for Crohn's disease in Cau...	Link	Link	Link
2.5	rs2943634	(C;C)	Slightly higher risk of ischemic stroke	Link	Link	
2.5	rs324420	(A;A)	Significantly increased risk for substance use ...	Link	Link	Link
2.5	rs4143094	(G;T)	Slightly (17%) higher risk of colorectal cancer...	Link		
2.5	rs5888	(C;T)	3x higher risk for age-related macular degenera...	Link		
2.5	rs664143	(T;T)	Higher risk for number of cancers	Link		
2.5	rs795484	(A;A)	Even more increased morphine dose requirement a...	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	
2.1	rs17077540	(A;G)	1.6x major depressive disorder risk	Link		
2.1	rs17563	(C;C)	Risk for otosclerosis	Link	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	rs4444903	(G;G)	3.5x risk of hep-cancer in cirrhosis patients; ...	Link		Link
2.1	rs5186	(A;C)	~1.4x increased risk of hypertension	Link	Link	Link
2.1	rs6457617	(T;T)	5.2x risk of rheumatoid arthritis	Link	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	rs1041981	(A;A)	Higher myocardial infarction risk	Link	Link	Link
2	rs1045642	(C;T)	Slower metaboliser for some drugs	Link	Link	Link
2	rs1050152	(C;T)	2.1x increased risk of Crohn's disease	Link	Link	Link
2	rs10811661	(C;T)	1.2x increased risk for type-2 diabetes	Link	Link	
2	rs10889677	(A;C)	1.5x increased risk for certain autoimmune dise...	Link	Link	
2	rs10984447	(A;G)	1.17x increased risk for multiple sclerosis	Link	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	rs1169300	(A;A)	~2x increased lung cancer risk	Link		
2	rs1219648	(A;G)	1.20x risk for breast cancer	Link	Link	
2	rs12469063	(G;G)	Increased risk of developing restless legs synd...	Link		
2	rs12567232	(A;G)	Increased risk for Crohn's Disease	Link	Link	
2	rs12696304	(C;G)	Prone to aging faster: at least in European pop...	Link		
2	rs1361600	(G;G)	~2x increased risk for adult-onset asthma in Ja...	Link		
2	rs1585215	(A;G)	2x increased risk for Hodgkin lymphoma	Link		
2	rs16944	(G;G)	Slightly increased (~2x or less) risk for certa...	Link	Link	
2	rs17228212	(C;C)	>1.26x increased risk for heart disease	Link	Link	
2	rs17435	(T;T)	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	rs2073963	(G;T)	Increased risk of baldness	Link		
2	rs2201841	(C;T)	1.5x increased risk for Crohn's disease; 2x inc...	Link	Link	
2	rs2230199	(C;G)	1.6x+ risk of ARMD	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

Mag.	Identifier	Genotype	Summary	GnomAD	GetEvidence	ClinVar
2	rs2306402	(C;T)	1.18x increased risk for late-onset Alzheimer's...	Link		
2	rs2420946	(C;T)	1.20x risk for breast cancer	Link		
2	rs2464196	(T;T)	~2x increased lung cancer risk	Link	Link	Link
2	rs25487	(G;G)	2x higher risk for skin cancer; possibly other ...	Link	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	rs3738919	(A;C)	1.94x risk of developing rheumatoid arthritis	Link		
2	rs4129148	(C;G)	3x risk of schizophrenia.	Link	Link	
2	rs4825476	(G;G)	1.9x higher risk of suicidal thoughts when taki...	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;G)	Increased risk in men for biliary conditions	Link		
2	rs5759167	(T;T)	Higher prostate cancer risk	Link	Link	
2	rs6232	(A;G)	Higher risk of obesity and insulin sensitivity	Link	Link	Link
2	rs6441286	(G;T)	1.54x chance of developing primary biliary cirr...	Link	Link	
2	rs6498169	(A;A)	>1.14x risk of multiple sclerosis	Link	Link	
2	rs6603272	(G;T)	2.74x increased risk of developing schizophre...	Link		
2	rs669	(G;G)	3.8x or higher increased risk for Alzheimers	Link	Link	Link
2	rs6997709	(G;G)	1.5x higher risk for hypertension	Link		
2	rs744373	(C;T)	1.17x risk of Alzheimer's	Link		
2	rs7536563	(A;A)	>1.12x risk of multiple sclerosis	Link	Link	
2	rs7639618	(C;T)	1.45x increased osteoarthritis risk	Link		
2	rs7807268	(C;G)	1.3x risk for Crohn's disease	Link	Link	
2	rs7961152	(A;C)	1.2x higher risk for hypertension	Link		
2	rs9954153	(G;T)	~2.5x higher risk for Fuchs' dystrophy: a corne...	Link		
2.0	rs1044396	(C;C)	Increased risk of Nicotine dependence among mal...	Link	Link	Link
2.0	rs1434536	(A;A)	1.94x increased breast cancer risk	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	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	
1.8	rs4807015	(C;C)	>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	rs1537415	(C;G)	1.6x increased risk for periodontitis	Link	Link	
1.6	rs2736100	(G;G)	1.6x higher risk for glioma development	Link	Link	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	
1.6	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	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	rs11171739	(C;T)	1.34x risk of developing Type-1 diabetes	Link	Link	
1.5	rs12210050	(C;T)	Slightly higher risk for basal cell carcinoma	Link	Link	
1.5	rs13149290	(C;C)	Slightly increased risk of developing prostate ...	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	rs1801274	(C;T)	Complex; generally greater risk for cancer prog...	Link	Link	Link
1.5	rs199533	(C;T)	Slightly increased risk of developing Parkinson...	Link		
1.5	rs2076295	(G;G)	Slightly increased risk for pulmonary fibrosis ...	Link		
1.5	rs2177369	(C;C)	1.5x increased risk for Alzheimer's disease	Link		
1.5	rs2272127	(C;C)	Associated with herpes and schizophrenia	Link		
1.5	rs2305089	(T;T)	Higher risk for chordoma reported in one study;...	Link	Link	

Mag.	Identifier	Genotype	Summary	GnomAD	GetEvidence	ClinVar
1.5	rs2881766	(G;T)	Slightly increased risk for pregnancy-induced h...	Link		
1.5	rs3087243	(G;G)	Increased risk for autoimmune diseases	Link	Link	Link
1.5	rs3825776	(A;G)	1.3x increased risk for ALS	Link	Link	
1.5	rs401681	(C;C)	~1.2x increased risk for several types of cance...	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	(C;T)	Increased risk of schizophrenia in limited stud...	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	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	rs6435862	(G;T)	1.7x higher risk of aggressive neuroblastoma	Link	Link	
1.5	rs6710341	(A;G)	Slightly increased risk of developing restless ...	Link		
1.5	rs6896702	(C;T)	Slightly increased risk of developing Parkinson...	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	rs7850258	(G;G)	Slightly higher odds of developing primary hypo...	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	rs9642880	(G;T)	1.2x increased bladder cancer risk	Link	Link	
1.5	rs9652490	(A;G)	Slightly increased risk of developing Parkinson...	Link	Link	
1.5	rs995030	(G;G)	Non-protective against testicular cancer	Link	Link	
1.4	rs1545843	(A;A)	1.4x increased risk for depression (for those u...	Link		
1.4	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	rs2230201	(A;G)	1.4x risk of lupus	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	rs4795067	(G;G)	Slight increase in risk for psoriatic arthritis...	Link		
1.4	rs4959039	(A;G)	1.4x higher risk for multiple sclerosis	Link		
1.4	rs4977756	(G;G)	1.93x higher risk for glioma development	Link	Link	
1.4	rs8050136	(A;A)	1.4x increased risk for T2D in some populations...	Link	Link	
1.3	rs1047031	(A;A)	1.3x increased risk for periodontitis	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	rs1260326	(C;T)	Slightly higher risk for gout	Link	Link	Link
1.3	rs1375144	(C;T)	1.32x increased risk of developing bipolar diso...	Link		
1.3	rs2024513	(A;G)	1.3x higher risk for schizophrenia (among Han C...	Link		
1.3	rs4712653	(C;T)	Very slightly (~1.3x) increased risk for neurob...	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	rs11037909	(T;T)	1.47x type II diabetes risk	Link		
1.2	rs11842874	(A;G)	+17% increased risk for osteoarthritis	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		
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	rs3176336	(T;T)	Slightly higher (1.25x) higher risk for breast ...	Link		
1.2	rs3740878	(A;A)	1.46x type II diabetes risk; common	Link		Link
1.2	rs393152	(A;G)	Slight increased risk of both PD and AD	Link	Link	
1.2	rs4324715	(C;C)	>1.5x increased testicular cancer risk for men	Link		
1.2	rs4496877	(T;T)	For type-1 diabetics: 1.6x increased nephropath...	Link		

Mag.	Identifier	Genotype	Summary	GnomAD	GetEvidence	ClinVar
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	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	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	rs249954	(C;T)	Potentially increased risk of Breast Cancer	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	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		
1.1	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;T)	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	rs17300539	(G;G)	Increased risk of insulin resistance	Link		
1	rs2282679	(A;C)	Somewhat lower vitamin D levels	Link		
1	rs3194051	(A;G)	1.12x risk of type-1 diabetes	Link	Link	Link
1	rs6166	(G;G)	Females slightly more likely to be sterile	Link	Link	Link
1	rs6932590	(T;T)	1.1x increased risk for schizophrenia	Link	Link	
1	rs7453920	(G;G)	Slight increase in risk for chronic hepatitis B...	Link		
0.1	rs2070744	(C;C)	Increased prostate cancer risk	Link	Link	Link
0.1	rs3095870	(G;G)	1.7x increased risk for SLE (lupus)	Link		
0.1	rs601338	(G;G)	Susceptible to Norovirus infections	Link	Link	Link

3.3 Genosets (Multi-variant Phenotypes)

Magnitude	Identifier	Summary
4	gs144	Male
3.1	gs191	Impaired NSAID drug metabolism
3.1	gs237	Blue eyes are more likely
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	gs161	CYP2C9 Intermediate Metabolizers
2.5	gs259	Homozygous for eye color haplotype #3
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
2.3	gs255	Homozygous eye color haplotype #1
2.1	gs223	One copy of GCH1 variant associated with lower ...
2	gs101	Probably able to digest milk
2	gs159	CYP1A2 fast metabolizer
2	gs173	CYP2D6*10
2	gs179	CYP2D6*41
2	gs211	Ethanol biodisposition
2	gs213	Haplogroup R (Y-DNA)
2	gs246	APOE E3/E3
2	gs313	Normal DPYD activity and thus 5-FU metabolism p...
1.7	gs233	Normal pain sensitivity; APS/APS: LPS/APS: and ...
1.6	gs236	Alzheimer's disease-related haplotype
1.5	gs185	The beta blocker metoprolol is effective: with ...
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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