PGP-UK Genomics Report for uk2FAAF0

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.

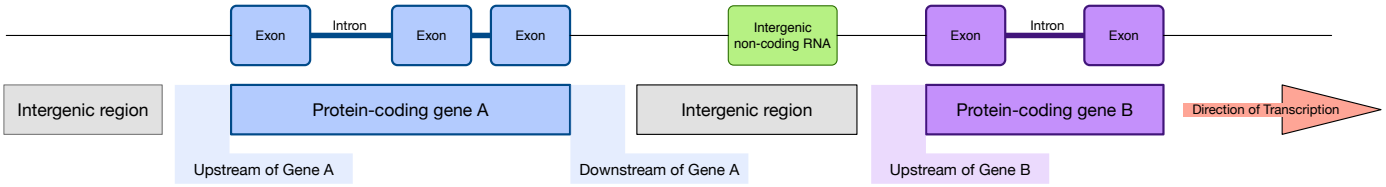

Figure 1: Diagram of gene structure indicating locations of potential variants

Feature	Count
Lines of input read	4690676
Variants filtered out	3723619
Novel / existing variants	0 (0.0) / 967057 (100.0)
Overlapped genes	51586
Overlapped transcripts	58951
Overlapped regulatory features	46721

Table 1: Variant calling summary

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

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

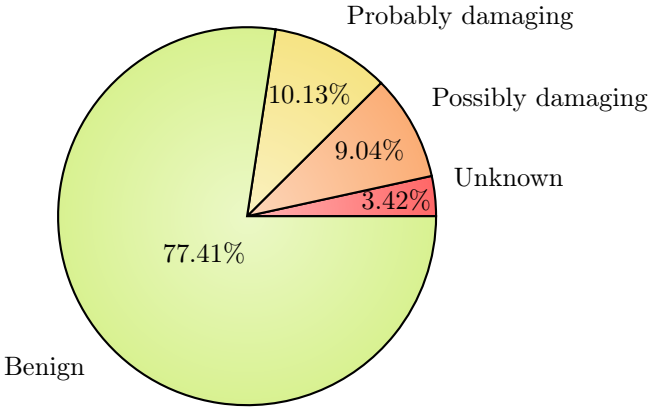

Figure 2: PolyPhen Summary

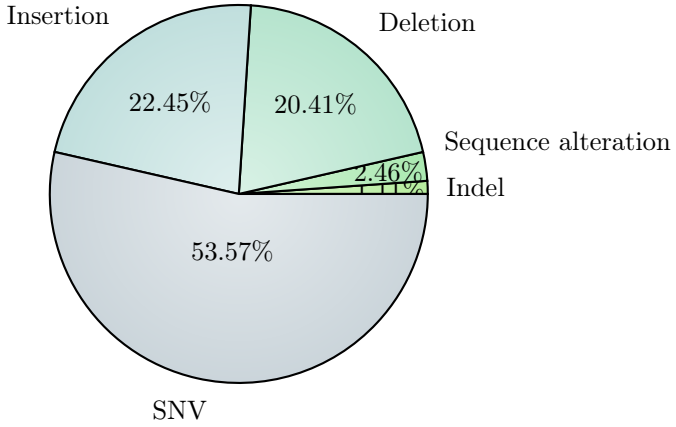

Figure 3: Variant Class

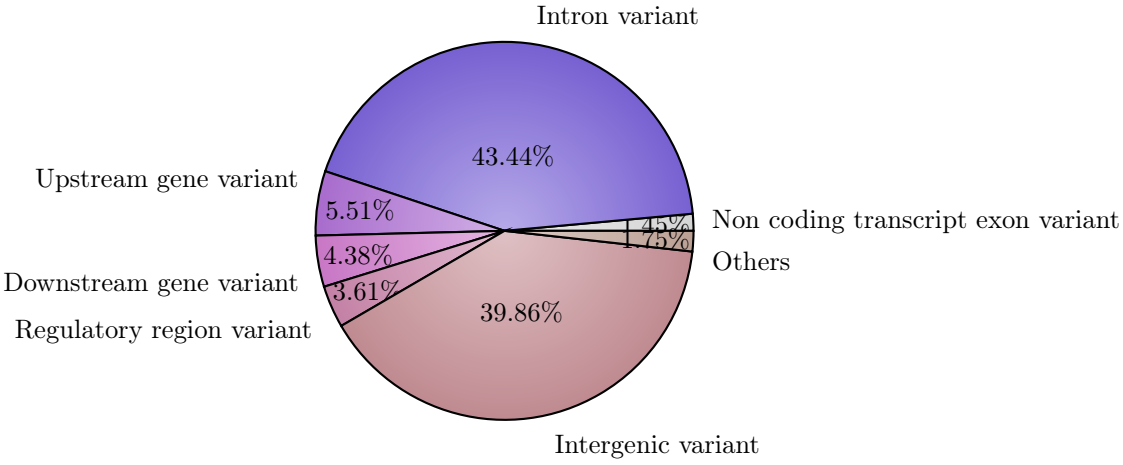

Figure 4: Consequence type

2 Ancestry

This plot shows the distribution of the genomes of different populations. Data from several studies which used whole genome sequencing was used to see the relationships between the genomes of the populations. It shows how closely related certain populations are genetically: Groups which cluster closely are more genetically similar than groups which are further apart. The black star symbol shows where this PGP-UK participant sits in relation to other populations, indicating their ancestry and their most closely related populations according to genetic sequence.

Please note that this analysis is limited by the populations available in the 1000 genomes project (1kGP) data. If there are European subpopulations reported, and the ancestry of the participant does not correspond to any of the 1kGP populations, the closest 1kGP sampled subpopulation will be shown (even though it might be different from the participant's actual ancestry).

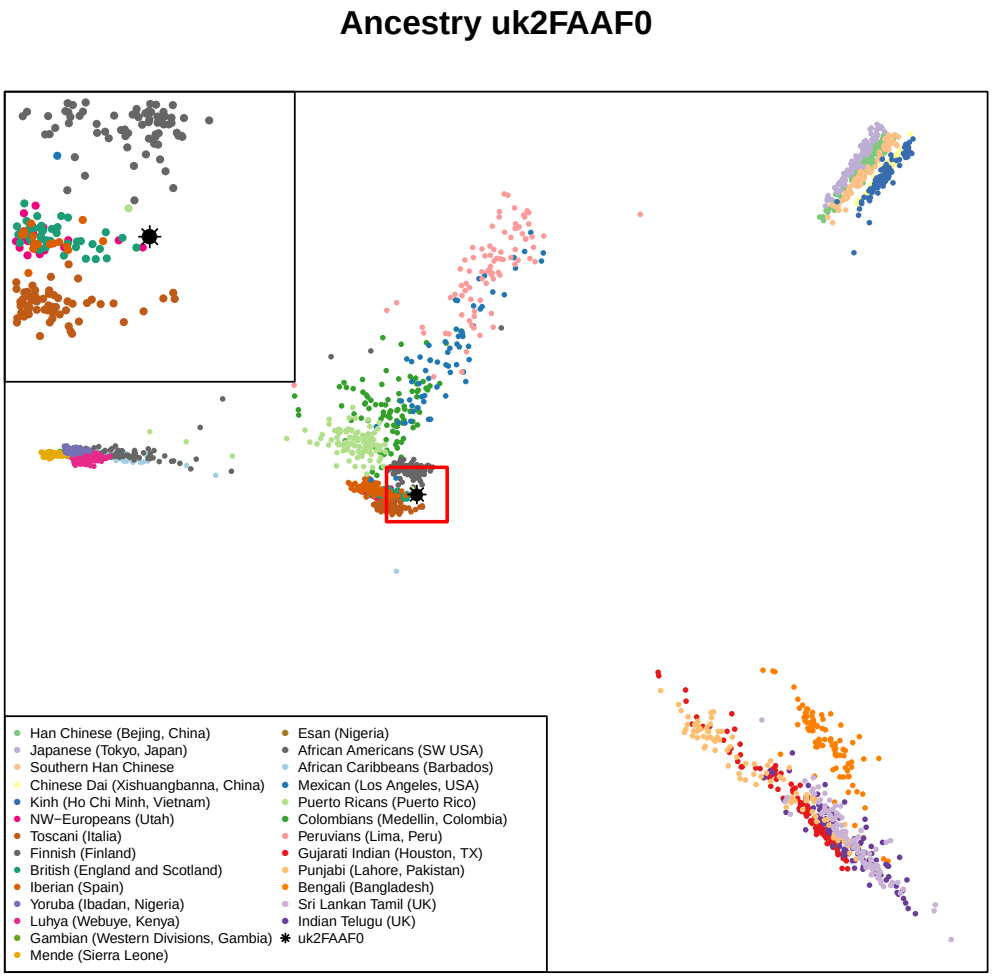

Figure 5: Ancestry Principal Component Analysis

3 Traits (based on SNPedia information)

Existing research has associated many variants with phenotypic traits, some of which can be perceived as beneficial while others appear to have a harmful effect. Some traits are complex and can be affected by several variants. It is likely that some of these would confer a higher risk while others a lower risk of trait manifestation. These can not be combined linearly to produce an actual risk of disease.

It is important to note that in most cases genomic data is probabilistic, not deterministic- i.e. having a genetic predisposition for a disease is not a diagnosis; rather, it shows an increased likelihood of developing that disease. Also, one person can have both potentially beneficial and harmful variants in the same gene, or associated with the same disease.

Some variants can also affect certain populations more, or will only affect a particular gender. For example, a variant for higher risk of endometriosis in the sequence of a male will not directly affect that person, but can be passed on to descendants.

While many traits are the result of a unique variant, many are the combination of several variants throughout the genome. In SNPedia, these are called genosets. These can integrate some of the information already present in the single variant tables, or be the combination of variants that have no phenotypic effect on their own, but contribute to a trait when together.

The variants in the following tables are sorted by magnitude. This is an subjective measure defined in SNPedia to highlight the perceived importance of the genotype described. At the moment this scale goes from 0 to 10. You can read more about it by visiting their explanatory webpage.

As our knowledge grows, the interpretation of the effect of certain variants might change. Clicking on the links in the genome report tables will take you to websites containing more information about each variant.

3.1 Possibly Beneficial Traits

Mag.	Identifier	Genotype	Summary	GnomAD	GetEvidence	ClinVar
2.5	rs11649743	(A;A)	Lower prostate cancer risk?	Link	Link	
2.1	rs2511989	(A;G)	0.63x decreased age-related macular degeneratio...	Link	Link	
2	rs10503669	(A;C)	Associated with higher HDL cholesterol	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	rs12678919	(A;G)	Associated with higher HDL cholesterol	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	rs1799884	(G;G)	Mothers have typical Birth-Weight babies. Sligh...	Link		
2	rs1864163	(G;G)	Associated with higher HDL cholesterol	Link	Link	
2	rs2056202	(T;T)	Rare decreased risk of autism	Link		
2	rs2292813	(C;T)	Decreased risk of autism	Link		
2	rs2542052	(C;C)	Better odds of living to 100	Link		
2	rs2707466	(A;A)	Stronger bones	Link	Link	
2	rs2908004	(T;T)	Stronger bones	Link	Link	
2	rs3738579	(C;T)	0.5x decreased risk for cervical cancer: HNSCC:...	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	rs6855911	(A;G)	0.62x decreased risk for gout	Link	Link	
2	rs7105934	(A;G)	0.69 times lower odds of developing renal cell ...	Link		
2	rs7776725	(T;T)	Stronger bones	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	rs7101429	(A;G)	0.70x reduced risk for Alzheimer's risk	Link		

Mag.	Identifier	Genotype	Summary	GnomAD	GetEvidence	ClinVar
1.6	rs3025786	(C;T)	Slightly decreased Alzheimer's disease risk amo...	Link		
1.5	rs1050631	(C;C)	Mean Survival Time of 32 months for esophageal ...	Link		
1.5	rs11212617	(A;C)	Somewhat increased likelihood of treatment succ...	Link		Link
1.5	rs2229169	(C;C)	1.5x decreased risk of heart attack and stroke ...	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	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	rs729302	(A;C)	0.89x decreased risk of developing rheumatoid a...	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		
1.3	rs9306160	(T;T)	0.75x (reduced) risk for metastasis in LN-/ER+ ...	Link	Link	
1.2	rs11172113	(C;C)	0.8x lower risk for migraines	Link		
1.2	rs4867568	(T;T)	Decreased risk for knee osteoporosis	Link		
1.2	rs6048	(A;G)	Slightly lower risk (10-20%) of deep vein throm...	Link	Link	Link
1.1	rs2293347	(G;G)	Among NSCLC patients: better Gefitinib response...	Link		Link
1.1	rs7611694	(C;C)	Lower prostate cancer risk?	Link		
1	rs10784502	(C;T)	Slightly higher intracranial volume	Link		
1	rs182549	(C;T)	Can digest milk.	Link		Link
1	rs1991517	(C;G)	May have higher bone density and lower TSH	Link	Link	Link
1	rs2351299	(G;T)	Possible reduced risk of Autism	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		
1.0	rs11246226	(C;C)	Decreased risk of schizophrenia in limited stud...	Link	Link	
1.0	rs2283123	(C;T)	Decreased risk of schizophrenia in limited stud...	Link		
1.0	rs6583817	(C;T)	~0.80x (lower) risk for late onset Alzheimer's ...	Link		
0.1	rs1726866	(C;C)	Can taste bitter	Link	Link	Link

3.2 Possibly Harmful Traits

Mag.	Identifier	Genotype	Summary	GnomAD	GetEvidence	ClinVar
3.2	rs2981582	(T;T)	1.7x higher risk of ER+ breast cancer	Link	Link	
3	rs1801282	(C;G)	Unconfirmed higher risk of cardiovascular disea...	Link	Link	Link
3	rs7754840	(C;G)	1.3x increased risk for type-2 diabetes	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	rs1421085	(C;T)	~1.3x increased obesity risk	Link	Link	Link
2.5	rs16969968	(A;G)	Slightly higher risk for nicotine dependence: l...	Link	Link	Link
2.5	rs187238	(G;G)	Hypertension increases risk 3.75x for sudden ca...	Link		
2.5	rs339331	(T;T)	Prostate cancer risk	Link		
2.5	rs3780374	(A;G)	Substantially increased odds of developing V617...	Link		
2.5	rs4495487	(C;T)	Increased odds (2 fold?) of developing V617F-as...	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	rs8034191	(C;T)	1.27x lung cancer risk	Link	Link	
2.5	rs891512	(A;G)	Higher blood pressure than G;G	Link		Link
2.4	rs7966230	(G;G)	Slightly lower levels of plasma VWF	Link		
2.2	rs2004640	(G;T)	1.4x increased risk for SLE	Link	Link	
2.1	rs10427255	(C;C)	Highest odds of photic sneeze reflex	Link		
2.1	rs10811661	(T;T)	1.2x increased risk for type-2 diabetes	Link	Link	
2.1	rs1219648	(G;G)	1.64x risk for breast cancer	Link	Link	
2.1	rs1329428	(G;G)	2x increased risk for macular degeneration	Link		
2.1	rs2294008	(T;T)	Increased risk of gastric and bladder cancer	Link	Link	
2.1	rs2306402	(C;C)	1.18x increased risk for late-onset Alzheimer's...	Link		
2.1	rs2420946	(T;T)	1.64x risk for breast cancer	Link		
2.1	rs4430796	(A;A)	1.38x increased risk for prostate cancer	Link	Link	
2.1	rs629242	(T;T)	Somewhat higher risk for prostate cancer	Link		
2.1	rs646776	(A;A)	1.2x risk of coronary artery disease	Link	Link	
2.1	rs944289	(C;T)	1.3x increased thyroid cancer risk	Link	Link	
2	rs1024611	(C;T)	Increased risk of exercise induced ischemia	Link		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	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	rs11170164	(A;A)	1.35x risk of basal cell carcinoma	Link	Link	Link
2	rs11229030	(C;C)	Higher odds of Crohn's disease	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	rs12431733	(T;T)	Increased risk of developing Parkinson's Diseas...	Link	Link	
2	rs12696304	(C;G)	Prone to aging faster: at least in European pop...	Link		
2	rs13254738	(A;C)	1.18x prostate cancer risk	Link	Link	
2	rs1333048	(A;C)	1.3x increased coronary artery disease risk	Link		
2	rs16944	(A;A)	Increased risk (~3x) for osteoarthritis	Link	Link	
2	rs1734791	(A;T)	1.4x increased risk for lupus	Link		
2	rs17435	(A;T)	1.4x increased risk for lupus	Link		
2	rs17487223	(C;T)	Higher lung cancer risk?	Link		
2	rs17576	(A;G)	Higher risk for MI and lung cancer: and COPD in...	Link	Link	Link
2	rs1799732	(-;C)	1.3x increased adenoma recurrence risk	Link	Link	
2	rs1800896	(A;G)	1.6x increased prostate cancer risk	Link		
2	rs2073963	(G;T)	Increased risk of baldness	Link		
2	rs2143340	(C;T)	Increased risk of dyslexia and poor reading per...	Link		

Mag.	Identifier	Genotype	Summary	GnomAD	GetEvidence	ClinVar
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	(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	rs2305480	(C;T)	3.5x increase in risk of asthma for Han Chinese...	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	rs2464196	(T;T)	~2x increased lung cancer risk	Link	Link	Link
2	rs25487	(A;G)	2x higher risk for skin cancer; possibly other ...	Link	Link	Link
2	rs2736990	(C;C)	Slightly increased risk of developing Parkinson...	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	rs3746444	(C;T)	~1.2x increased risk for cancer	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	rs4444903	(A;G)	3.5x risk of hep-cancer in cirrhosis patients; ...	Link		Link
2	rs4464148	(C;C)	1.35x increased risk for colorectal cancer	Link		
2	rs4633	(T;T)	Higher risk for endometrial cancer	Link	Link	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	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	rs6601764	(C;C)	1.52x increased risk of developing Crohn's dise...	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	rs6922269	(A;A)	1.6x risk of coronary artery disease	Link	Link	
2	rs6997709	(G;G)	1.5x higher risk for hypertension	Link		
2	rs699	(C;T)	Increased risk of hypertension	Link	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;G)	1.3x risk for Crohn's disease	Link	Link	
2	rs7961152	(A;C)	1.2x higher risk for hypertension	Link		
2	rs800292	(C;C)	5% higher risk of Age related macular degenerat...	Link	Link	Link
2	rs854560	(A;A)	Higher risk for heart disease: diabetic retinop...	Link	Link	Link
2	rs9525638	(T;T)	Weaker bones	Link		
2	rs965513	(A;G)	1.77x increased thyroid cancer risk	Link	Link	
2.0	rs1044396	(C;C)	Increased risk of Nicotine dependence among mal...	Link	Link	Link
2.0	rs2305795	(A;A)	1.64x higher risk of narcolepsy compared to (G;...	Link		Link
1.9	rs7923837	(A;G)	1.6x risk for T2D	Link		
1.8	rs2278206	(T;T)	1.16x increased risk for asthma	Link	Link	
1.7	rs1047286	(T;T)	1.7x increased risk for age-related macular deg...	Link	Link	Link
1.7	rs2024513	(A;A)	1.7x higher risk for schizophrenia (among Han C...	Link		
1.7	rs8055236	(G;T)	1.9x risk for heart disease	Link	Link	
1.6	rs11523871	(C;C)	>1.6x increased breast cancer risk for women ov...	Link	Link	
1.6	rs2736100	(G;G)	1.6x higher risk for glioma development	Link	Link	Link
1.6	rs2981745	(T;T)	>1.6x increased risk for breast cancer in femal...	Link		
1.6	rs356219	(G;G)	1.6x increased risk for Parkinson's disease	Link		
1.6	rs3775948	(C;G)	Slightly higher risk for gout	Link		
1.6	rs4712653	(C;C)	Slightly (~1.6x) increased risk for neuroblasto...	Link		
1.5	rs10260404	(C;T)	1.20x risk of developing ALS	Link	Link	
1.5	rs10464059	(A;G)	Slightly increased risk of developing Parkinson...	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		

Mag.	Identifier	Genotype	Summary	GnomAD	GetEvidence	ClinVar
1.5	rs10859871	(A;C)	Slight (~1.2x) increase in endometriosis risk	Link		
1.5	rs10883365	(A;G)	1.2x increased risk for developing Crohn's dise...	Link	Link	
1.5	rs12037606	(A;G)	1.22x risk of developing Crohn's disease	Link		
1.5	rs12469063	(A;G)	Slightly increased risk of developing restless ...	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	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	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	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	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	rs2280714	(A;A)	1.4x increased risk of SLE	Link		
1.5	rs2282679	(C;C)	Lower vitamin D levels	Link		
1.5	rs2286812	(C;T)	~2x higher risk for Fuchs' dystrophy: a corneal...	Link		
1.5	rs27388	(A;G)	Slightly increased risk of developing schizophr...	Link		
1.5	rs28694718	(A;G)	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	rs356220	(T;T)	Increased risk of Parkinson's Disease	Link		
1.5	rs3745516	(A;G)	Slightly increased risk of developing primary b...	Link		
1.5	rs3764880	(A;G)	Possible 1.2 - 1.8x increased tuberculosis susc...	Link	Link	
1.5	rs3814570	(C;T)	1.3x increased risk for Crohn's disease with il...	Link		
1.5	rs3825776	(A;G)	1.3x increased risk for ALS	Link	Link	
1.5	rs4506565	(A;T)	1.4x increased risk for type-2 diabetes	Link	Link	
1.5	rs4785763	(A;A)	2x 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	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	rs6498169	(A;G)	1.14x risk of multiple sclerosis	Link	Link	
1.5	rs699473	(C;T)	~1.5x increased brain tumor risk	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	rs1126497	(C;T)	1.4x increased risk for breast cancer	Link	Link	Link
1.4	rs1801157	(A;A)	1.4x higher risk for breast cancer	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	
1.34	rs17465637	(C;C)	1.34x higher risk for myocardial infarction	Link	Link	
1.3	rs110419	(A;G)	1.3x increased risk for neuroblastoma	Link		
1.3	rs1434536	(A;G)	1.29x increased breast cancer risk	Link		Link
1.3	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	
1.2	rs11037909	(T;T)	1.47x type II diabetes risk	Link		
1.2	rs11842874	(A;G)	+17% increased risk for osteoarthritis	Link		
1.2	rs1344706	(T;T)	1.2x increased risk for schizophrenia	Link	Link	
1.2	rs1800693	(A;G)	Slight (1.2x) increase in risk for multiple scl...	Link	Link	Link
1.2	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		

Mag.	Identifier	Genotype	Summary	GnomAD	GetEvidence	ClinVar
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	rs3850641	(A;G)	Increased risk of myocardial infarction in wome...	Link		
1.2	rs4324715	(C;C)	>1.5x increased testicular cancer risk for men	Link		
1.2	rs4795067	(A;G)	Slight increase in risk for psoriatic arthritis...	Link		
1.2	rs4977756	(A;G)	1.39x higher risk for glioma development	Link	Link	
1.2	rs7528684	(G;G)	1.2x risk of Rheumatoid Arthritis; various risk...	Link		
1.2	rs8050136	(A;C)	1.2x increased risk for T2D in some populations...	Link	Link	
1.1	rs10248420	(A;A)	Possibly less likely to remit on certain antide...	Link	Link	
1.1	rs11650494	(A;G)	Slightly higher prostate cancer risk	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	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	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	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	
1.1	rs997669	(G;G)	Very slightly increased (1.18x) increased breas...	Link		
1.09	rs12050604	(A;C)	Very slightly increased risk for lung cancer	Link		
1.07	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	rs10761659	(A;G)	1.2x risk of Crohn's disease	Link	Link	
1	rs1143674	(A;G)	1.3x increased autism risk	Link		
1	rs12718541	(A;A)	Nicotine dependence	Link		
1	rs1417066	(C;T)	Slightly increased risk of osteoarthritis	Link		
1	rs17300539	(G;G)	Increased risk of insulin resistance	Link		
1	rs2546890	(A;G)	Higher risk of multiple sclerosis	Link		
1	rs3194051	(A;G)	1.12x risk of type-1 diabetes	Link	Link	Link
1	rs3735684	(C;T)	Associated with increased colorectal cancer ris...	Link	Link	
1	rs6932590	(T;T)	1.1x increased risk for schizophrenia	Link	Link	
1	rs6974491	(A;G)	Higher risk of coeliac and/or inflammatory bowe...	Link		
1	rs987525	(A;C)	2.5x increased risk for cleft lip	Link	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	(G;G)	Susceptible to Norovirus infections	Link	Link	Link

3.3 Genosets (Multi-variant Phenotypes)

Magnitude	Identifier	Summary
4	gs145	Female
2.6	gs327	Somewhat increased risk of Alzheimer's disease
2.5	gs155	CYP3A5 non-expressor
2.5	gs157	More stimulated by coffee
2.5	gs242	Increased risk of individuals with prostate can...
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...
2.5	gs298	Increased surveillance for colorectal cancer re...
2.4	gs297	Lower heart attack risk than average
2	gs101	Probably able to digest milk
2	gs140	NAT2 slow metabolizer
2	gs154	NAT2 Slow metabolizer
2	gs194	Myocardial Infarction Risk
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	gs185	The beta blocker metoprolol is effective: with ...
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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