PGP-UK Genomics Report for uk915C7E

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.

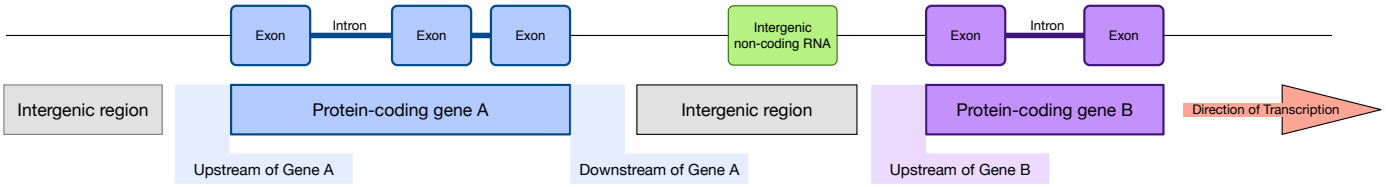

Figure 1: Diagram of gene structure indicating locations of potential variants

Feature	Count
Lines of input read	4898537
Variants filtered out	3298219
Novel / existing variants	0 (0.0) / 1600318 (100.0)
Overlapped genes	55857
Overlapped transcripts	64840
Overlapped regulatory features	147342

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.

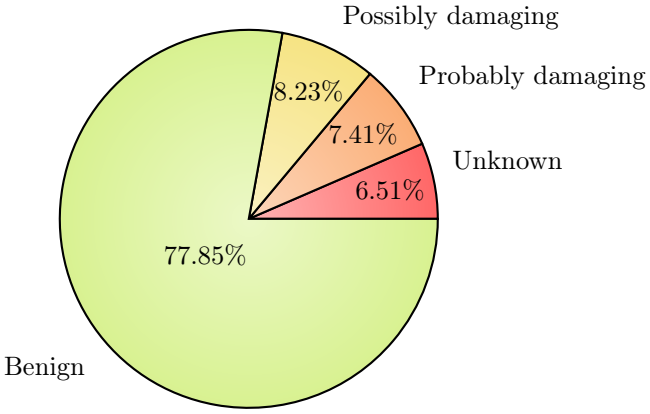

Figure 2: PolyPhen Summary

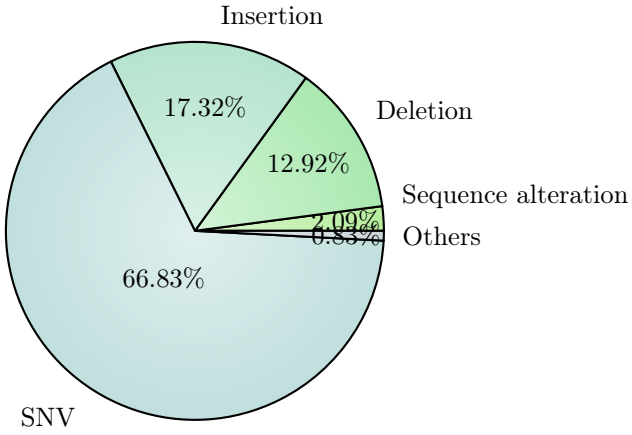

Figure 3: Variant Class

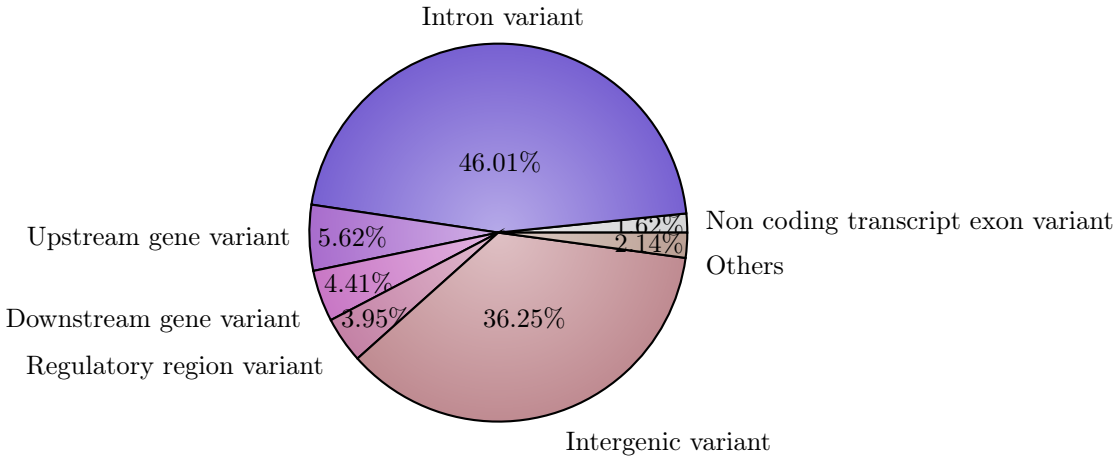

Figure 4: Consequence type

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).

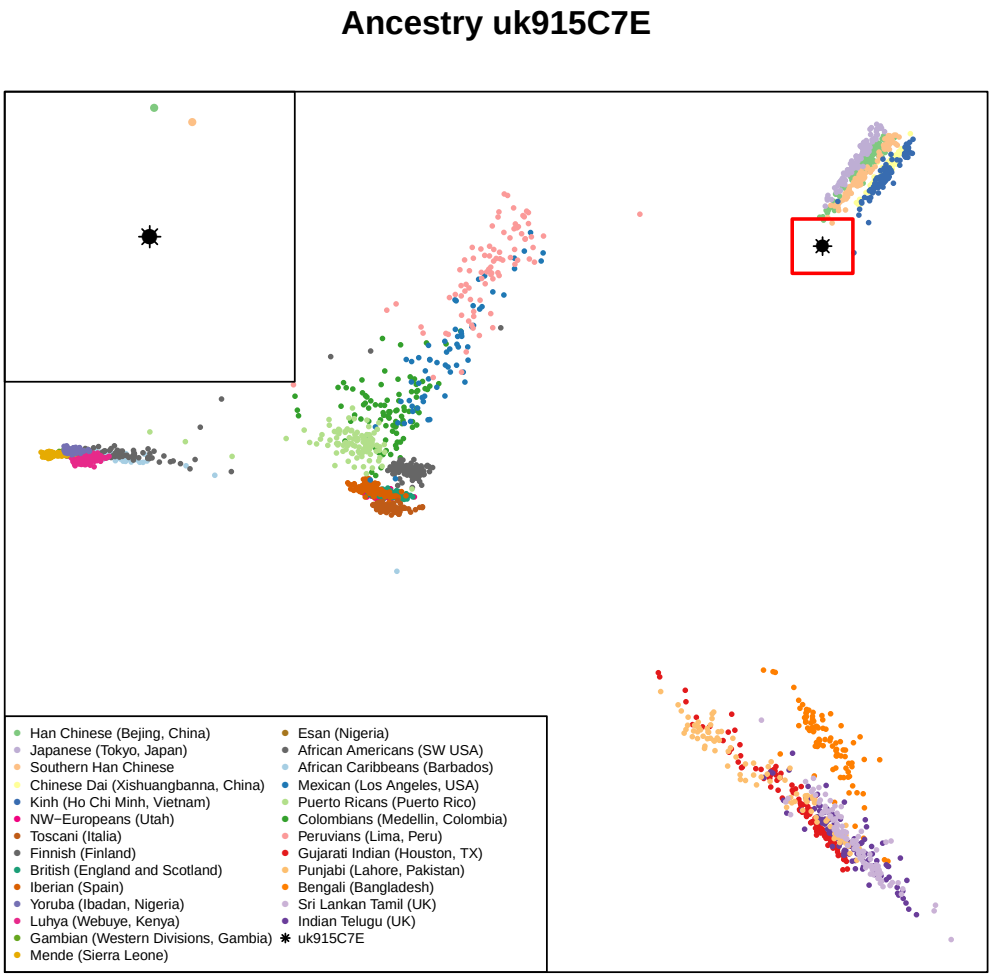

Figure 5: Ancestry Principal Component Analysis

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	rs7294919	(C;T)	Moderately enhanced hippocampal volume	Link		
2.1	rs2511989	(A;G)	0.63x decreased age-related macular degeneratio...	Link	Link	
2	rs10936599	(C;C)	Longer telomeres: longer life?	Link		Link
2	rs1128535	(G;G)	Reduced risk (0.77x) for Crohn's disease	Link		
2	rs1136410	(C;T)	0.80x reduced risk for glioblastoma	Link	Link	
2	rs1229984	(A;A)	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	rs1544410	(G;G)	Decreased risk of low bone mineral density diso...	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	rs2241423	(A;G)	0.79 decreased risk for obesity	Link		
2	rs2241766	(G;T)	Slightly lower risk of breast cancer	Link		
2	rs2243250	(T;T)	0.33x decreased risk for myocardial infarction ...	Link		
2	rs2542052	(C;C)	Better odds of living to 100	Link		
2	rs261332	(A;G)	Associated with higher HDL cholesterol	Link		
2	rs3178250	(C;C)	Lower otosclerosis risk	Link		
2	rs3782179	(C;T)	3x lower odds of testicular cancer risk for men...	Link		
2	rs4149268	(A;G)	Associated with higher HDL cholesterol	Link	Link	
2	rs4307059	(C;C)	Reduced Autism risk	Link	Link	
2	rs763110	(C;T)	~0.80x reduced cancer risk	Link		Link
2	rs9525638	(C;C)	Stronger bones	Link		
2.0	rs3790844	(C;C)	Reduced risk (0.59x) of pancreatic cancer	Link		
1.8	rs1800588	(C;T)	Higher HDL-C levels	Link	Link	
1.8	rs6897932	(C;T)	0.91x decreased risk for multiple sclerosis	Link	Link	Link
1.5	rs1026732	(A;G)	0.70x risk for restless legs	Link	Link	
1.5	rs1050631	(C;C)	Mean Survival Time of 32 months for esophageal ...	Link		

Mag.	Identifier	Genotype	Summary	GnomAD	GetEvidence	ClinVar
1.5	rs1063192	(C;T)	0.71x reduced risk of myocardial infarction	Link		
1.5	rs11635424	(A;G)	0.70x risk for restless legs	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	rs3851179	(A;G)	0.85x decreased risk for Alzheimer's disease	Link	Link	
1.5	rs4149274	(C;T)	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	rs464049	(C;C)	Decreased risk of schizophrenia in limited stud...	Link		
1.5	rs4939883	(C;C)	Associated with higher HDL cholesterol	Link	Link	
1.5	rs9939609	(T;T)	Lower risk of obesity and Type-2 diabetes	Link	Link	
1.4	rs10513789	(G;T)	0.8x decreased risk of Parkinson's disease	Link		
1.4	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		
1.3	rs10166942	(C;C)	0.7x lower risk for migraines	Link		
1.2	rs11246226	(A;C)	Decreased risk of schizophrenia in limited stud...	Link	Link	
1.1	rs11172113	(C;T)	0.9x lower risk for migraines	Link		
1.1	rs7568369	(G;T)	0.90x reduced risk of obesity	Link		
1	rs10784502	(C;T)	Slightly higher intracranial volume	Link		
1	rs2351299	(G;T)	Possible reduced risk of Autism	Link		
1	rs2494732	(T;T)	Lower odds of psychosis	Link	Link	
1	rs2952768	(C;T)	Slightly less drug dependence: decreased effect...	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
3.5	rs10490924	(T;T)	8.2x risk for age related macular degeneration	Link	Link	Link
3.2	rs2981582	(T;T)	1.7x higher risk of ER+ breast cancer	Link	Link	
3	rs1799999	(T;T)	Insulin resistance	Link	Link	Link
3	rs1983132	(C;T)	2 - 3x higher prostate cancer risk if routinely...	Link		
3	rs3738579	(T;T)	1.5x - 2x increased risk for cervical cancer: H...	Link		
3	rs4244285	(A;G)	Poorer metabolizer of several popular medicines...	Link	Link	Link
3	rs7754840	(C;C)	1.3x increased risk for type-2 diabetes	Link	Link	
2.7	rs10830963	(C;G)	Increased type-2 diabetes risk; higher gestatio...	Link	Link	
2.5	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	rs12803066	(A;G)	Increased risk of myopia	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	rs2073963	(G;G)	Increased risk of baldness	Link		
2.5	rs2943634	(C;C)	Slightly higher risk of ischemic stroke	Link	Link	
2.5	rs3738919	(C;C)	1.94x risk of developing rheumatoid arthritis	Link		
2.5	rs4495487	(C;T)	Increased odds (2 fold?) of developing V617F-as...	Link		
2.5	rs5888	(C;T)	3x higher risk for age-related macular degenera...	Link		
2.5	rs6441286	(G;G)	3.08x chance of developing primary biliary cirr...	Link	Link	
2.5	rs7574865	(G;T)	1.3x risk of rheumatoid arthritis; 1.55x risk o...	Link	Link	Link
2.5	rs9934438	(A;A)	Coumadin resistance	Link	Link	Link
2.3	rs3798220	(C;T)	2-3x higher risk for cardiovascular events: whi...	Link	Link	
2.3	rs7966230	(C;G)	Slightly lower levels of plasma VWF	Link		
2.2	rs7913069	(C;T)	1.47x risk for uterine fibroids	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	rs1585215	(G;G)	3.5x increased risk for Hodgkin lymphoma	Link		
2.1	rs2231142	(A;C)	1.74x increased gout risk; gefinitib takers 4x ...	Link	Link	Link
2.1	rs2306402	(C;C)	1.18x increased risk for late-onset Alzheimer's...	Link		
2.1	rs2420946	(T;T)	1.64x risk for breast cancer	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	rs4961	(T;T)	1.8x increased risk for high blood pressure	Link	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	rs1024611	(C;T)	Increased risk of exercise induced ischemia	Link		Link
2	rs1042838	(G;T)	1.28x risk for endometrial ovarian cancer; over...	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	rs10883365	(G;G)	1.62x increased risk for developing Crohn's dis...	Link	Link	
2	rs10937823	(C;T)	Some association with bipolar disorder	Link		
2	rs10984447	(A;G)	1.17x increased risk for multiple sclerosis	Link	Link	
2	rs11045585	(A;G)	63% chance (higher than average) of docetaxel-i...	Link	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	rs12567232	(A;A)	Increased risk for Crohn's Disease	Link	Link	
2	rs13254738	(C;C)	1.18x prostate cancer risk	Link	Link	
2	rs1360780	(C;T)	1.3x increased risk for depression	Link	Link	Link
2	rs16944	(G;G)	Slightly increased (~2x or less) risk for certa...	Link	Link	

Mag.	Identifier	Genotype	Summary	GnomAD	GetEvidence	ClinVar
2	rs17435	(T;T)	1.4x increased risk for lupus	Link		
2	rs17576	(G;G)	Higher risk for lung cancer: and COPD in smoker...	Link	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	
2	rs1800896	(A;A)	1.8x increased prostate cancer risk	Link		
2	rs2201841	(C;C)	1.5x increased risk for Crohn's disease	Link	Link	
2	rs2383207	(A;G)	Increased risk for heart disease	Link		
2	rs241448	(C;T)	1.51x increased risk for Alzheimer's	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	rs3025039	(C;T)	2.6x increased risk for ARMD in a Taiwanese pop...	Link		
2	rs3212227	(A;C)	Significantly increased risk of developing cerv...	Link		Link
2	rs326	(A;A)	Lower HDL cholesterol	Link	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	rs3745516	(A;A)	Increased risk of developing primary biliary ci...	Link		
2	rs3775948	(G;G)	Slightly higher risk for gout	Link		
2	rs3825776	(G;G)	>1.3x increased risk for ALS	Link	Link	
2	rs4027132	(A;A)	1.51x increased risk of developing bipolar diso...	Link		
2	rs4129148	(C;G)	3x risk of schizophrenia.	Link	Link	
2	rs4242382	(A;G)	1.7x increased risk for prostate cancer	Link	Link	
2	rs4402960	(G;T)	1.2x increased risk for type-2 diabetes: ~1x ri...	Link	Link	Link
2	rs4538475	(G;G)	Increased risk of developing Parkinson's Diseas...	Link	Link	
2	rs4626664	(A;A)	>1.44x increased risk of developing restless le...	Link	Link	
2	rs493258	(A;G)	1.15x risk of Age Related Macular Degeneration	Link		
2	rs4968451	(A;C)	1.61x increased risk for meningioma	Link		
2	rs6498169	(A;A)	>1.14x risk of multiple sclerosis	Link	Link	
2	rs6601764	(C;C)	1.52x increased risk of developing Crohn's dise...	Link	Link	
2	rs6603272	(G;G)	>2.74x increased risk of developing schizophren...	Link		
2	rs663048	(G;T)	Significantly increased risk of developing lung...	Link	Link	
2	rs6908425	(C;C)	1.95x increased risk of developing Crohn's dise...	Link	Link	
2	rs6997709	(G;G)	1.5x higher risk for hypertension	Link		
2	rs699	(C;C)	Increased risk of hypertension	Link	Link	Link
2	rs7216389	(T;T)	1.5x increased risk for Childhood Asthma.	Link	Link	
2	rs738409	(C;G)	Increased liver fat: odds of alcoholic liver di...	Link	Link	Link
2	rs7442295	(A;A)	~4x higher risk for hyperuracemia	Link	Link	Link
2	rs744373	(C;C)	1.17x risk of Alzheimer's	Link		
2	rs7639618	(C;T)	1.45x increased osteoarthritis risk	Link		
2	rs7807268	(C;C)	1.4x risk for Crohn's disease	Link	Link	
2	rs854560	(A;A)	Higher risk for heart disease: diabetic retinop...	Link	Link	Link
2	rs965513	(A;G)	1.77x increased thyroid cancer risk	Link	Link	
2.0	rs4911414	(G;T)	2-4x higher risk of sun sensitivity if part of ...	Link	Link	
2.0	rs9642880	(T;T)	1.5x increased bladder cancer risk	Link	Link	
1.8	rs1136287	(C;T)	1.5x increased risk of wet ARMD in a Taiwanese ...	Link	Link	Link
1.8	rs4474514	(A;G)	3x increased testicular cancer risk for men	Link	Link	
1.8	rs4807015	(C;C)	>1.74x risk of type 2 diabetes	Link		
1.8	rs733618	(A;G)	1.87x risk for myasthenia gravis	Link		
1.7	rs10181656	(C;G)	1.7x increased SLE risk	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	rs2046210	(T;T)	1.6x increased breast cancer risk in certain wo...	Link	Link	Link
1.6	rs356219	(G;G)	1.6x increased risk for Parkinson's disease	Link		
1.6	rs4712653	(C;C)	Slightly (~1.6x) increased risk for neuroblasto...	Link		
1.5	rs10757272	(C;T)	1.30x increased risk for Coronary artery diseas...	Link		
1.5	rs10889677	(A;A)	1.5x increased risk for certain autoimmune 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	rs1169300	(A;G)	~1.5x increased lung cancer risk	Link		
1.5	rs12037606	(A;G)	1.22x risk of developing Crohn's disease	Link		
1.5	rs1223271	(A;G)	Slightly increased risk of developing Parkinson...	Link	Link	
1.5	rs12498742	(A;A)	1.25 increased risk for gout	Link		
1.5	rs13149290	(C;T)	Slightly increased risk of developing prostate ...	Link		
1.5	rs1375144	(C;C)	1.59x increased risk of developing bipolar diso...	Link		
1.5	rs140701	(A;A)	Increased risk for anxiety disorders	Link		
1.5	rs17115100	(G;T)	Slightly increased risk of developing Parkinson...	Link	Link	
1.5	rs1801020	(T;T)	1.31x increased risk of heart disease	Link		Link
1.5	rs1801274	(C;T)	Complex; generally greater risk for cancer prog...	Link	Link	Link
1.5	rs2007153	(G;G)	Increased risk of schizophrenia in limited stud...	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	rs2240340	(A;G)	Slightly increased (1.5x) risk for RA	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	rs2305089	(T;T)	Higher risk for chordoma reported in one study;...	Link	Link	
1.5	rs2464196	(C;T)	~1.5x increased lung cancer risk	Link	Link	Link
1.5	rs2697962	(A;G)	Slightly increased risk of developing Parkinson...	Link		
1.5	rs28694718	(A;G)	2x higher risk for schizophrenia	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	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	rs393152	(A;A)	Increased risk of both PD and AD	Link	Link	
1.5	rs401681	(C;T)	~1.2x increased risk for several types of cance...	Link	Link	
1.5	rs4585	(T;T)	Slightly poorer (0.75x) response to metformin i...	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	rs6532197	(A;G)	Slightly increased risk of developing Parkinson...	Link	Link	
1.5	rs6896702	(C;T)	Slightly increased risk of developing Parkinson...	Link		
1.5	rs699473	(C;C)	~1.5x increased brain tumor risk	Link		
1.5	rs7341475	(G;G)	1.58x increased schizophrenia risk for women	Link	Link	
1.5	rs7536563	(A;G)	1.12x risk of multiple sclerosis	Link	Link	
1.5	rs7774434	(C;T)	Slightly increased risk of developing primary b...	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	rs9561778	(T;T)	~2x increased risk of adverse drug reactions fr...	Link	Link	
1.5	rs9652490	(A;G)	Slightly increased risk of developing Parkinson...	Link	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	rs1447295	(A;C)	1.4x increased risk of prostate cancer	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	rs4795067	(G;G)	Slight increase in risk for psoriatic arthritis...	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	rs13361189	(C;T)	1.3x increased risk for Crohn's disease	Link	Link	
1.3	rs1746048	(C;C)	1.03 increased risk for coronary heart disease	Link	Link	
1.3	rs2295490	(A;G)	1.32x increased risk of early-onset type-2 diab...	Link	Link	

Mag.	Identifier	Genotype	Summary	GnomAD	GetEvidence	ClinVar
1.3	rs2736100	(G;T)	1.3x higher risk for glioma development: 2.1x r...	Link	Link	Link
1.3	rs34330	(C;T)	1.3x higher risk for endometrial cancer (in Chi...	Link		Link
1.3	rs4295627	(G;T)	1.36x higher risk for glioma development	Link	Link	
1.3	rs4958847	(A;G)	1.3x increased risk for Crohn's disease	Link		
1.3	rs7234029	(A;G)	Slightly increased (1.36x) risk for Crohn's dis...	Link		
1.25	rs748404	(T;T)	Slightly increased risk (1.25) for lung cancer...	Link	Link	
1.2	rs10210302	(C;T)	1.2x increased risk for Crohn's disease	Link	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	rs2072590	(G;T)	1.2x increased risk for ovarian cancer	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	rs419788	(A;G)	2.0x risk for lupus	Link		
1.2	rs4977756	(A;G)	1.39x higher risk for glioma development	Link	Link	
1.2	rs7514229	(G;G)	Associated with early-onset autoimmune thyroid ...	Link		
1.2	rs851715	(A;A)	Risk of nonsense-word repetition problems if sp...	Link		
1.17	rs17465637	(A;C)	1.17x higher risk for myocardial infarction	Link	Link	
1.17	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	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	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	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	rs6707530	(G;G)	In colorectal cancer: may allow cancer cells to...	Link		
1.1	rs6897876	(C;T)	Slight increase in testicular cancer risk for m...	Link		
1.1	rs7171755	(A;G)	Very slight decrease 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;T)	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	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	rs1804197	(A;C)	Increased risk of familial colorectal cancer an...	Link		Link
1	rs2273697	(A;G)	Adverse reaction more likely to carbamazepine i...	Link	Link	Link
1	rs2282679	(A;C)	Somewhat lower vitamin D levels	Link		
1	rs2435357	(A;A)	Slightly higher (2x?) risk for Hirschsprung dis...	Link		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	rs5326	(A;G)	Possible psychiatric risks	Link		
1	rs6932590	(T;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		
0.5	rs1566734	(G;T)	Somatic mutation: cancer associated	Link	Link	Link
0.1	rs3095870	(G;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	gs296	Lower heart attack risk than average
2.5	gs100	Lactose intolerance risk
2.5	gs155	CYP3A5 non-expressor
2.5	gs242	Increased risk of individuals with prostate can...
2.5	gs256	Carrier for a type of blue eyes
2.5	gs277	Increased risk of Atrial Fibrillation in one of...
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	gs159	CYP1A2 fast metabolizer
2	gs173	CYP2D6*10
2	gs221	Autoimmune disorder risk in Europeans
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	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
1.2	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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