

Genomics Report for PGP-UK2/uk2E2AAE

1 Summary

This is the genome report for participant PGP-UK2/uk2E2AAE . It was produced using collaborative research tools, including SNPedia and GetEvidence. This summary shows an overview of all the variants which were found in the genome for this individual. They have been compared with a reference genome.

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. "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. The diagram in Figure 1 is a simplification of the usual gene structure.

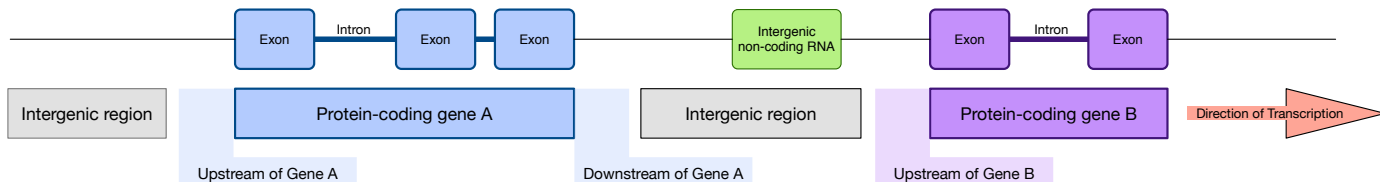


Figure 1: Diagram of gene structure indicating locations of potential variants

Feature	Count
Lines of input read	4154008
Variants remaining after filtering	4133129
Novel / existing variants	115090 (2.8%) / 4018039 (97.2%)
Overlapped genes	54665
Overlapped transcripts	64378
Overlapped regulatory features	212022

Table 1: Variant calling summary

There are several different types of genomic variants. The most common are single nucleotide variants (SNV) that correspond to the change of a single nucleotide in the DNA. 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.

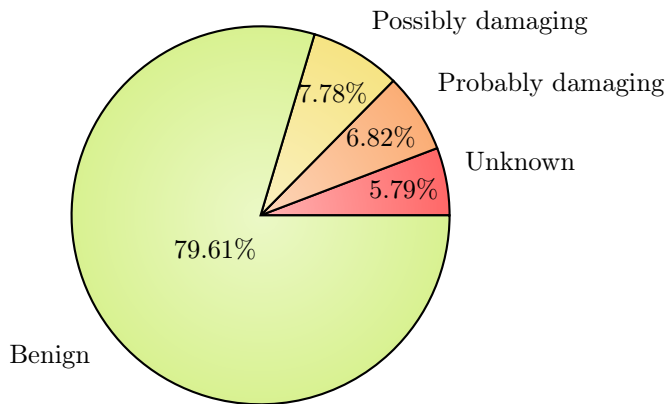


Figure 2: PolyPhen Summary

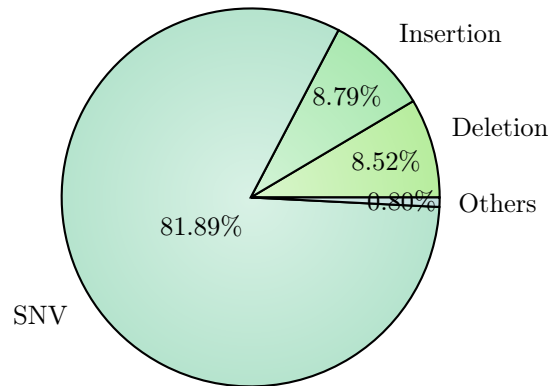


Figure 3: Variant Class

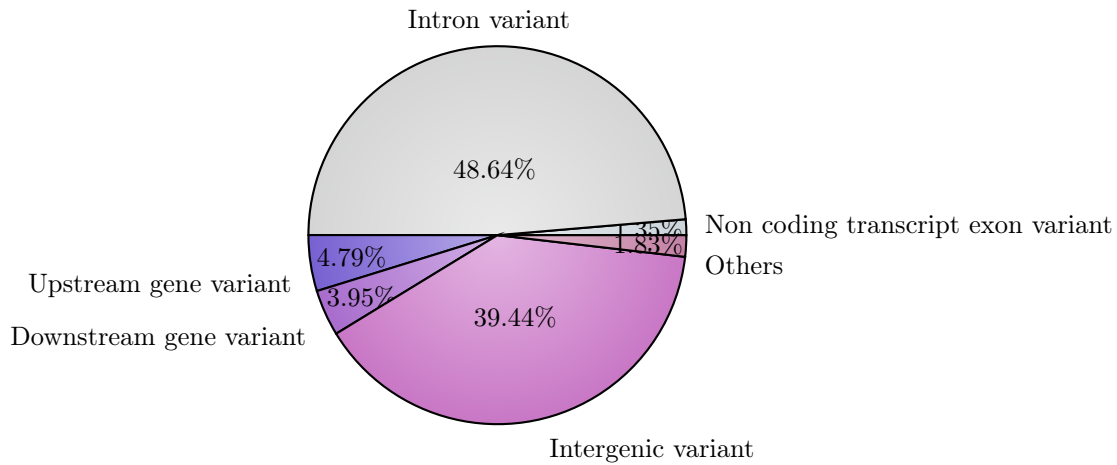


Figure 4: Consequence type

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.

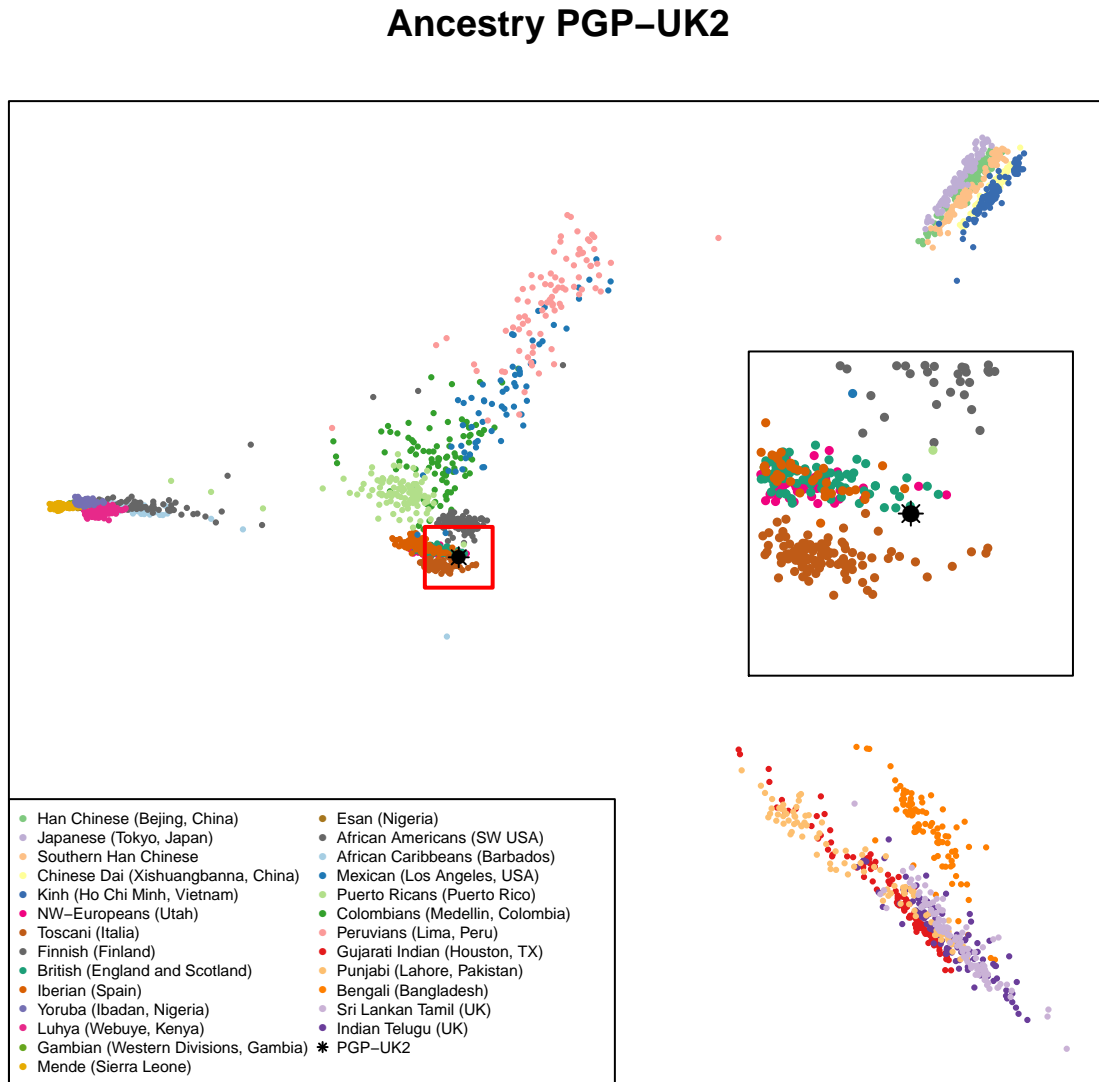


Figure 5: Ancestry Principal Component Analysis

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.

- Possibly Beneficial Traits

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
2.2	rs2511989	(A;A)	0.44x decreased age-related macular degeneratio...		Link	
2.2	rs3816873	(C;C)	Reduced risk of type-2 diabetes	Link	Link	Link
2.1	rs6505162	(A;A)	0.43x decreased risk for esophageal cancer	Link		
2.1	rs738409	(G;G)	Most common genotype; slightly less damage from...	Link	Link	
2	rs1012053	(A;C)	0.625x reduced risk of Bipolar Disorder.		Link	
2	rs10468017	(C;T)	Associated with higher HDL cholesterol		Link	
2	rs10504861	(A;G)	Reduced risk of migraine without aura			
2	rs11045585	(A;A)	24% chance (lower than average) of docetaxel-in...		Link	
2	rs1544410	(G;G)	Decreased risk of low bone mineral density diso...		Link	
2	rs1864163	(G;G)	Associated with higher HDL cholesterol		Link	
2	rs2235015	(G;T)	Somewhat more likely to respond to certain anti...	Link	Link	
2	rs2243250	(C;T)	0.6x decreased risk for myocardial infarction i...			
2	rs2542052	(C;C)	Better odds of living to 100			
2	rs261332	(A;G)	Associated with higher HDL cholesterol			
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:...			
2	rs3782179	(C;T)	3x lower odds of testicular cancer risk for men...			
2	rs4143094	(G;G)	No increased risk of colorectal cancer correlat...			
2	rs4149268	(A;G)	Associated with higher HDL cholesterol		Link	
2	rs4585	(G;G)	Slightly higher (1.35x) odds of good metformin ...			
2	rs763110	(C;T)	~0.80x reduced cancer risk			Link
2	rs801114	(T;T)	0.78x decreased Basal Cell Carcinoma risk.		Link	
2	rs925391	(C;T)	Lower odds of going bald			
1.5	rs11136000	(C;T)	0.84x decreased risk for Alzheimer's disease		Link	
1.5	rs4149274	(C;T)	Associated with higher HDL (good) cholesterol			
1.5	rs464049	(C;C)	Decreased risk of schizophrenia in limited stud...			
1.5	rs4939883	(C;C)	Associated with higher HDL cholesterol		Link	

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
1.5	rs6427528	(A;G)	For rheumatoid arthritis patients: better respo...			
1.4	rs2294008	(C;C)	Lower risk of cancer	Link	Link	
1.2	rs11246226	(A;C)	Decreased risk of schizophrenia in limited stud...		Link	
1.2	rs4686484	(G;G)	Slightly decreased risk for celiac disease			
1.2	rs6048	(G;G)	Slightly lower risk (10-20%) of deep vein throm...	Link	Link	Link
1.1	rs10166942	(C;T)	0.85x lower risk for migraines			
1.1	rs11172113	(C;T)	0.9x lower risk for migraines			
1.1	rs2293347	(G;G)	Among NSCLC patients: better Gefitinib response...	Link		
1.1	rs7568369	(G;T)	0.90x reduced risk of obesity			
1	rs10248420	(A;G)	7x more likely to respond to certain antidepres...		Link	
1	rs11983225	(C;T)	7x more likely to respond to certain antidepres...		Link	
1	rs2235040	(A;G)	7x more likely to respond to certain antidepres...	Link	Link	
1	rs2235067	(A;G)	7x more likely to respond to certain antidepres...			
1	rs2952768	(C;T)	Slightly less drug dependence: decreased effect...			
1	rs4148739	(A;G)	7x more likely to respond to certain antidepres...		Link	
1	rs4939827	(C;T)	0.86x decreased risk for colorectal cancer		Link	
1	rs7850258	(A;G)	Typical odds of developing primary hypothyroidi...			
0.1	rs891512	(G;G)	Lower blood pressure than those with an A allel...	Link		
0	rs1047781	(A;A)	ABH blood group "Secretor" status if Japanese	Link	Link	Link
0	rs10897346	(C;C)	If depressed: 2.6x more likely to not respond t...			
0	rs1126742	(T;T)	Higher hypertension risk	Link	Link	
0	rs12252	(T;T)	More resistant to influenza	Link		Link
0	rs12593929	(A;A)	Blue eye color more likely			
0	rs16990018	(A;A)	PrP Codon 171 Asn - Non-pathogenic variant	Link		Link
0	rs17244841	(A;A)	More responsive to statin treatment		Link	
0	rs1799782	(C;C)	Lower risk for skin cancer	Link	Link	
0	rs1799945	(C;C)	Not a H63D hemochromatosis carrier.	Link	Link	Link
0	rs2240203	(A;A)	Blue eye color more likely			
0	rs2306402	(C;C)	1.18x increased risk for LOAD			
0	rs28933385	(G;G)	Prion protein Codon 200 (E) - Non pathogenic va...			Link
0	rs403016	(C;C)	2x risk for lupus		Link	
0	rs6259	(G;G)	Best inverse correlation between tea-drinking: ...	Link	Link	
0	rs74315403	(G;G)	PrP codon 178 (D) - non pathogenic variant			Link
0	rs7495174	(A;A)	Blue/gray eyes more likely		Link	
0	rs8028689	(T;T)	Blue eye color if part of blue eye color haplot...			
0	rs9951307	(A;G)	0.10 decreased risk for brain edema after a str...			

• Possibly Harmful Traits

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
4	i5012680	(A;G)	Congenital Disorder of Glycosylation Type 1a ca...			
4	rs28936415	(A;G)	Carrier of Congenital Disorder of Glycosylation...	Link		Link
3	rs13266634	(C;C)	Increased risk for type-2 diabetes	Link	Link	Link
3	rs1801282	(C;G)	Watch out for high fat in diet	Link	Link	Link
3	rs2981582	(C;T)	1.3x higher risk of ER+ breast cancer		Link	
3	rs3892097	(A;A)	CYP2D6 poor metabolizer; many associations rela...	Link	Link	Link
3	rs3903239	(C;C)	Higher frequency of atrial fibrillation			
3	rs55705857	(A;G)	6x increased risk of glioma of IDH1/IDH2 subtyp...			
3	rs6920220	(A;G)	1.2x risk Rheumatoid Arthritis		Link	
3	rs7754840	(C;C)	1.3x increased risk for type-2 diabetes		Link	
2.5	rs10484554	(C;T)	2.8x increased risk for psoriasis		Link	
2.5	rs10490924	(G;T)	2.7x risk for age related macular degeneration	Link	Link	Link
2.5	rs1121980	(C;T)	1.67x risk for obesity		Link	
2.5	rs1421085	(C;T)	~1.3x increased obesity risk		Link	Link
2.5	rs16969968	(A;G)	Slightly higher risk for nicotine dependence: 1...	Link	Link	Link
2.5	rs2943634	(C;C)	Higher risk of ischemic stroke		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	
2.5	rs664143	(T;T)	Higher risk for number of cancers			
2.5	rs8034191	(C;T)	1.27x lung cancer risk		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			
2.2	rs2231137	(G;G)	~1.5-3x increased risk for ischemic stroke	Link	Link	Link
2.2	rs944289	(T;T)	2.6x increased thyroid cancer risk		Link	
2.1	rs10811661	(T;T)	1.2x increased risk for type-2 diabetes		Link	
2.1	rs11887534	(C;G)	2x increased risk for gallstones	Link	Link	Link
2.1	rs1329428	(G;G)	2x increased risk for macular degeneration			
2.1	rs1360780	(T;T)	1.3x increased risk for depression		Link	
2.1	rs17070145	(C;C)	Reduced memory abilities			Link
2.1	rs2254958	(C;T)	1.24x increased risk for Alzheimer's			
2.1	rs241448	(C;C)	2.14x increased risk for Alzheimer's	Link		Link
2.1	rs380390	(C;C)	Increased risk for ARMD		Link	
2.1	rs4149056	(C;T)	Reduced breakdown of some drugs; 5x increased m...	Link	Link	
2.1	rs4363657	(C;T)	4.5x increased myopathy risk for statin users		Link	
2.1	rs6457617	(T;T)	5.2x risk of rheumatoid arthritis		Link	
2.1	rs795484	(A;G)	Increased morphine dose requirement and postope...			
2.1	rs9272346	(A;G)	5.5x risk type-1 diabetes		Link	
2	rs10090154	(C;T)	1.4x increased risk for prostate cancer			
2	rs1024611	(C;T)	Increased risk of exercise induced ischemia			Link
2	rs1045642	(C;T)	Slower metaboliser for some drugs	Link	Link	
2	rs1050631	(C;T)	Mean Survival Time of 25 months for esophageal ...	Link		
2	rs10513789	(G;T)	Increased risk of Parkinson's disease			
2	rs1051730	(C;T)	1.3x increased risk of lung cancer	Link	Link	Link
2	rs10871777	(A;G)	Adults likely to be 0.22 BMI units higher			
2	rs10889677	(A;C)	1.5x increased risk for certain autoimmune dise...		Link	
2	rs10984447	(A;A)	>1.17x increased risk for multiple sclerosis		Link	
2	rs1136287	(C;T)	1.5x increased risk of wet ARMD	Link	Link	
2	rs1160312	(A;A)	1.6x increased risk of Male Pattern Baldness.		Link	
2	rs1219648	(A;G)	1.20x risk for breast cancer		Link	
2	rs12567232	(A;G)	Increased risk for Crohn's Disease		Link	
2	rs1265181	(C;G)	Increased risk for psoriasis		Link	
2	rs13254738	(A;C)	1.18x prostate cancer risk		Link	
2	rs1333048	(A;C)	1.3x increased coronary artery disease risk			

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
2	rs1691053	(A;G)	Increased risk of developing prostate cancer			
2	rs16942	(A;G)	Very slightly increased breast cancer risk	Link	Link	Link
2	rs1734791	(A;T)	1.4x increased risk for lupus			
2	rs17435	(A;T)	1.4x increased risk for lupus			
2	rs17576	(G;G)	Higher risk for lung cancer: and COPD in smoker...	Link	Link	
2	rs17782313	(C;T)	Adults likely to be 0.22 BMI units higher		Link	
2	rs1800896	(A;G)	1.6x increased prostate cancer risk			
2	rs187238	(G;G)	Hypertension increases risk 3.75x for sudden ca...			
2	rs2073963	(G;T)	Increased risk of baldness			
2	rs2156921	(A;G)	1.29x increased risk for depression			
2	rs2201841	(C;T)	1.5x increased risk for Crohn's disease; 2x inc...		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
2	rs2383206	(A;G)	1.4x increased risk for heart disease			
2	rs2383207	(A;G)	Increased risk for heart disease			
2	rs2420946	(C;T)	1.20x risk for breast cancer			
2	rs25487	(G;G)	2x higher risk for skin cancer; possibly other ...	Link	Link	
2	rs326	(A;A)	Lower HDL cholesterol		Link	Link
2	rs351855	(C;T)	1.2x increased risk for prostate cancer	Link	Link	Link
2	rs3775948	(G;G)	Slightly higher risk for gout			
2	rs3793784	(C;G)	1.5x risk for ARMD		Link	Link
2	rs4242382	(A;G)	1.7x increased risk for prostate cancer		Link	
2	rs4633	(C;T)	Higher risk for endometrial cancer	Link	Link	
2	rs4792311	(A;G)	Increased risk of prostate cancer	Link	Link	Link
2	rs4968451	(A;C)	1.61x increased risk for meningioma			
2	rs6807362	(C;C)	Increased autism risk	Link	Link	
2	rs6896702	(T;T)	Increased risk of developing Parkinson's Diseas...			
2	rs6897932	(C;T)	1.3x increased risk for multiple sclerosis	Link	Link	Link
2	rs6908425	(C;C)	1.95x increased risk of developing Crohn's dise...		Link	
2	rs6997709	(G;G)	1.5x higher risk for hypertension			
2	rs699	(C;T)	Increased risk of hypertension	Link	Link	Link
2	rs7442295	(A;A)	~4x higher risk for hyperuracemia		Link	
2	rs763361	(T;T)	Increased risk for multiple autoimmune diseases...	Link	Link	
2	rs7774434	(C;C)	Increased risk of developing primary biliary ci...			
2	rs7776725	(C;C)	Weaker bones		Link	
2	rs7794745	(A;T)	Slightly increased risk for autism		Link	Link
2	rs7807268	(C;C)	1.4x risk for Crohn's disease		Link	
2	rs7961152	(A;C)	1.2x higher risk for hypertension			
2	rs800292	(C;C)	5% higher risk of Age related macular degenerat...	Link	Link	Link
2	rs828907	(G;T)	Slightly increased risk of bladder cancer and 2...			
2	rs9652490	(A;A)	~2x increased risk for Parkinson's disease: and...		Link	
2	rs965513	(A;G)	1.7x increased thyroid cancer risk		Link	
2.0	rs4911414	(G;T)	2-4x higher risk of sun sensitivity if part of ...		Link	
1.8	rs143383	(T;T)	1.3x increased risk for osteoarthritis		Link	Link
1.8	rs4474514	(A;G)	3x increased testicular cancer risk for men		Link	
1.7	rs8055236	(G;T)	1.9x risk for heart disease		Link	
1.6	rs1260326	(T;T)	Slightly higher risk for gout	Link	Link	Link
1.6	rs33980500	(C;T)	1.6x increase in risk for psoriatic arthritis	Link	Link	Link
1.5	rs10260404	(C;T)	1.20x risk of developing ALS		Link	
1.5	rs10757272	(C;T)	1.30x increased risk for Coronary artery diseas...			
1.5	rs10883365	(A;G)	1.2x increased risk for developing Crohn's dise...		Link	
1.5	rs1169300	(A;G)	~1.5x increased lung cancer risk			
1.5	rs12037606	(A;G)	1.22x risk of developing Crohn's disease			
1.5	rs1223271	(A;G)	Slightly increased risk of developing Parkinson...		Link	
1.5	rs12431733	(C;T)	Slightly increased risk of developing Parkinson...		Link	

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
1.5	rs12498742	(A;A)	1.25 increased risk for gout			
1.5	rs13149290	(C;C)	Slightly increased risk of developing prostate ...			
1.5	rs13376333	(C;T)	1.5x higher risk of atrial fibrillation		Link	
1.5	rs16944	(A;G)	Minorly increased risk of mental illness and os...		Link	
1.5	rs1801274	(T;T)	Complex; generally greater risk for cancer prog...	Link	Link	Link
1.5	rs2076295	(G;G)	Slightly increased risk for pulmonary fibrosis ...			
1.5	rs2280714	(A;A)	1.4x increased risk of SLE			
1.5	rs2464196	(C;T)	~1.5x increased lung cancer risk	Link	Link	Link
1.5	rs2736990	(C;T)	Slightly increased risk of developing Parkinson...		Link	
1.5	rs27388	(A;G)	Slightly increased risk of developing schizophr...			
1.5	rs2881766	(G;T)	Slightly increased risk for pregnancy-induced h...			
1.5	rs3087243	(A;G)	Increased risk for auto-immune diseases		Link	
1.5	rs3212227	(A;A)	1.43x increased risk of developing psoriasis an...			
1.5	rs358806	(A;C)	0.86x increased risk of developing Type-2 diabe...		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...			
1.5	rs393152	(A;A)	Increased risk of both PD and AD	Link	Link	
1.5	rs401681	(C;C)	~1.2x increased risk for several types of cance...		Link	
1.5	rs4027132	(A;G)	1.39x increased risk of developing bipolar diso...			
1.5	rs4464148	(C;T)	1.10x increased risk for colorectal cancer			
1.5	rs4538475	(A;G)	Slightly increased risk of developing Parkinson...		Link	
1.5	rs4785763	(A;C)	1.5x higher risk for melanoma		Link	
1.5	rs4845618	(G;T)	1.7x increased melanoma risk			
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	rs619203	(C;G)	Increases susceptibility to Myocardial Infarcti...	Link	Link	
1.5	rs6601764	(C;T)	1.16x increased risk of developing Crohn's dise...		Link	
1.5	rs7454108	(C;T)	Single HLA-DQ8 haplotype			
1.5	rs807701	(C;T)	Slightly increased dyslexia risk			
1.5	rs872071	(A;G)	~1.5x increased risk for chronic lymphocytic le...		Link	
1.5	rs9303277	(C;T)	1.46x Slightly increased risk of developing pri...			
1.5	rs9642880	(G;T)	1.2x increased bladder cancer risk		Link	
1.4	rs1126497	(C;T)	1.4x increased risk for breast cancer	Link	Link	Link
1.4	rs12770228	(A;G)	1.4x increased risk for meningioma			
1.4	rs1447295	(A;C)	1.4x increased risk of prostate cancer		Link	
1.4	rs1800693	(G;G)	Slight (1.4x) increase in risk for multiple scl...	Link	Link	Link
1.4	rs2230201	(A;G)	1.4x risk of lupus	Link		
1.4	rs3131296	(G;G)	1.4x increased risk for schizophrenia		Link	
1.4	rs4795067	(G;G)	Slight increase in risk for psoriatic arthritis...			
1.4	rs498872	(T;T)	1.4x higher risk for glioma development		Link	
1.4	rs6010620	(G;G)	1.4x higher risk for glioma development; but th...		Link	
1.3	rs1042713	(A;G)	1.3x increased risk that pediatric inhaler use ...	Link	Link	Link
1.3	rs10947262	(C;C)	1.3x increased risk for osteoarthritis			
1.3	rs110419	(A;G)	1.3x increased risk for neuroblastoma			
1.3	rs1434536	(A;G)	1.29x increased breast cancer risk			
1.3	rs16847548	(C;T)	1.3x increased risk for sudden cardiac death in...			
1.3	rs2059693	(C;T)	1.3x increased risk for testicular cancer			
1.3	rs2736100	(G;T)	1.3x higher risk for glioma development		Link	
1.3	rs356219	(A;G)	1.3x increased risk for Parkinson's disease			
1.3	rs4295627	(G;T)	1.3x higher risk for glioma development		Link	
1.25	rs13387042	(A;A)	1.24x increased risk for breast cancer		Link	
1.2	rs2056116	(A;G)	1.18x risk for breast cancer			
1.2	rs2072590	(G;T)	1.2x increased risk for ovarian cancer			
1.2	rs2252586	(A;G)	1.2x higher risk for glioma development			
1.2	rs6897876	(C;C)	Slight increase in testicular cancer risk for m...			

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
1.2	rs8050136	(A;C)	1.2x increased risk for T2D in some populations...		Link	
1.2	rs9858542	(A;G)	1.1x risk Crohn's Disease	Link	Link	
1.17	rs17465637	(A;C)	1.17x higher risk for myocardial infarction	Link	Link	
1.15	rs748404	(C;T)	Very slightly increased risk (1.15) for lung ca...		Link	
1.1	rs11037909	(C;T)	1.27x type II diabetes risk	Link		
1.1	rs11110912	(C;G)	1.3x high blood pressure risk			
1.1	rs11650354	(C;T)	Possible risk for allergic asthma	Link		
1.1	rs11650494	(A;G)	Slightly higher prostate cancer risk			
1.1	rs1344706	(G;T)	1.1x increased risk for schizophrenia		Link	
1.1	rs1799966	(A;G)	Very slightly increased breast cancer risk	Link	Link	Link
1.1	rs34516635	(G;G)	Less longevity for Ashkenazi Jewish women.	Link		Link
1.1	rs3740878	(A;G)	1.26x type II diabetes risk	Link		
1.1	rs3818361	(C;T)	1.15x increased risk for late-onset Alzheimer's...			
1.1	rs4977574	(A;G)	Some studies - but not others - report a slight...		Link	
1.1	rs6495446	(C;T)	1.2x increased risk for chronic kidney disease			
1.1	rs7171755	(A;G)	Very slight decrease in cortical thickness and ...			
1.1	rs7412	(C;C)	More likely to gain weight if taking olanzapine...	Link	Link	Link
1.09	rs12050604	(A;C)	Very slightly increased risk for lung cancer			
1.05	rs2291834	(C;T)	Very slightly higher risk for myocardial infarc...			
1	rs1143674	(A;G)	1.3x increased autism risk	Link		
1	rs2282679	(A;C)	Somewhat lower vitamin D levels			
1	rs2546890	(A;G)	Higher risk of multiple sclerosis			
1	rs3194051	(A;A)	>1.1x risk of type-1 diabetes	Link	Link	Link
1	rs6932590	(T;T)	1.1x increased risk for schizophrenia		Link	
0.1	rs601338	(G;G)	Susceptible to Norovirus infections	Link	Link	Link
0	rs3761418	(A;A)	1.3x increased risk for depression			
0	rs4293393	(T;T)	1.25x Increased Risk of CKD for T allele in ...			
0	rs4712653	(T;T)	2x increased risk for neuroblastoma			
0	rs6277	(C;C)	1.6x higher schizophrenia risk	Link	Link	Link

- Genosets (Multi-variant Phenotypes)

Magnitude	Identifier	Summary
4	gs145	Female
3	gs273	Lowest risk (13% of white women) of Atrial Fibr...
2.5	gs100	Lactose intolerance risk
2.5	gs155	CYP3A5 non-expressor
2.5	gs157	More stimulated by coffee
2.5	gs256	Blue eyes
2.5	gs259	Homozygous for eye color haplotype #3
2.5	gs281	Part of the 88% of the population claimed not t...
2	gs129	Unable to classify your ABO blood type
2	gs156	NAT2 Rapid metabolizer.
2	gs239	Reduced conversion of beta-carotene to retinol
2	gs246	APOE3/APOE3
2	gs249	Parkinson's Disease Risk
2	gs292	Possible 2x increased risk of Alzheimer's disea...
1.5	gs1001	Mitochondrial Haplogroup H
1.5	gs139	NAT2 intermediate metabolizer
1.5	gs185	The beta blocker metoprolol is effective with 1...
0.1	gs233	Normal pain sensitivity

4 Report Metadata

Resource	Version	Website
Genome	GRCh37	Link
BWA	0.7.12	Link
SAMtools	1.2	Link
GATK	3.4-46	Link
PLINK	v1.90b3.35	Link
VEP	84	Link
SNPedia	8-Apr-2016	Link
ExAC	v0.3.1	Link
GetEvidence	8-Apr-2016	Link
ClinVar	4-Apr-2016	Link

Table 5: Analysis Pipeline Versions

Report generated on July 20, 2016 (using report generator version 16-174).