Genomics Report for PGP-UK5/uk33D02F

1 Summary

This is the genome report for participant PGP-UK5/uk33D02F . 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.

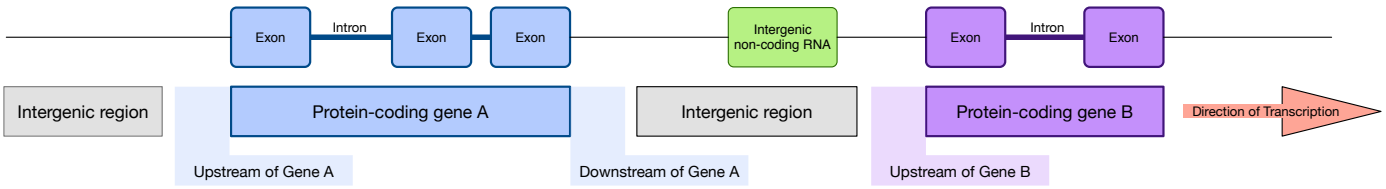

Figure 1: Diagram of gene structure indicating locations of potential variants

Feature	Count
Lines of input read	4214134
Variants remaining after filtering	4187518
Novel / existing variants	119829 (2.9%) / 4067689 (97.1%)
Overlapped genes	54728
Overlapped transcripts	64562
Overlapped regulatory features	214210

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.

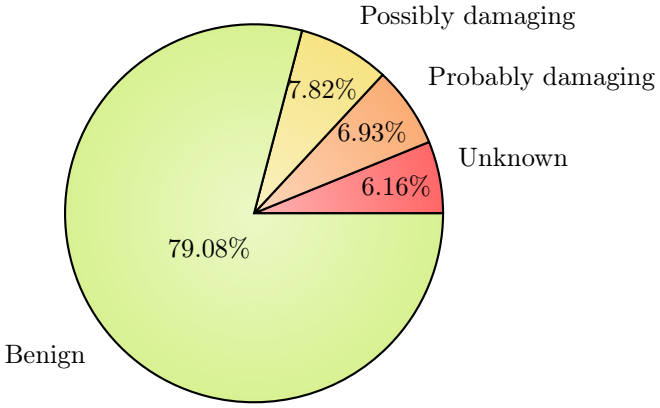

Figure 2: PolyPhen Summary

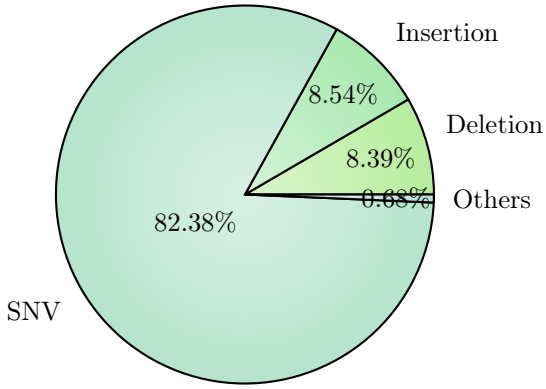

Figure 3: Variant Class

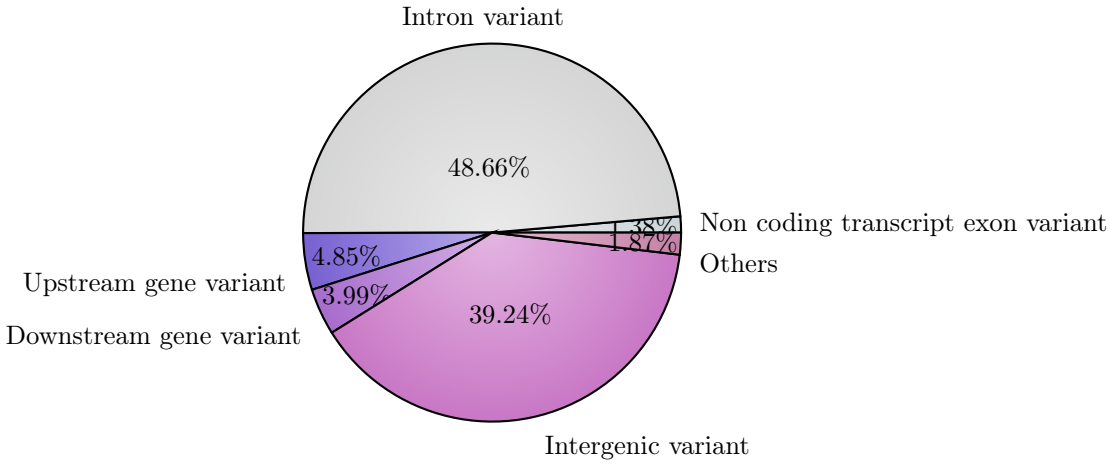

Figure 4: Consequence type

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.

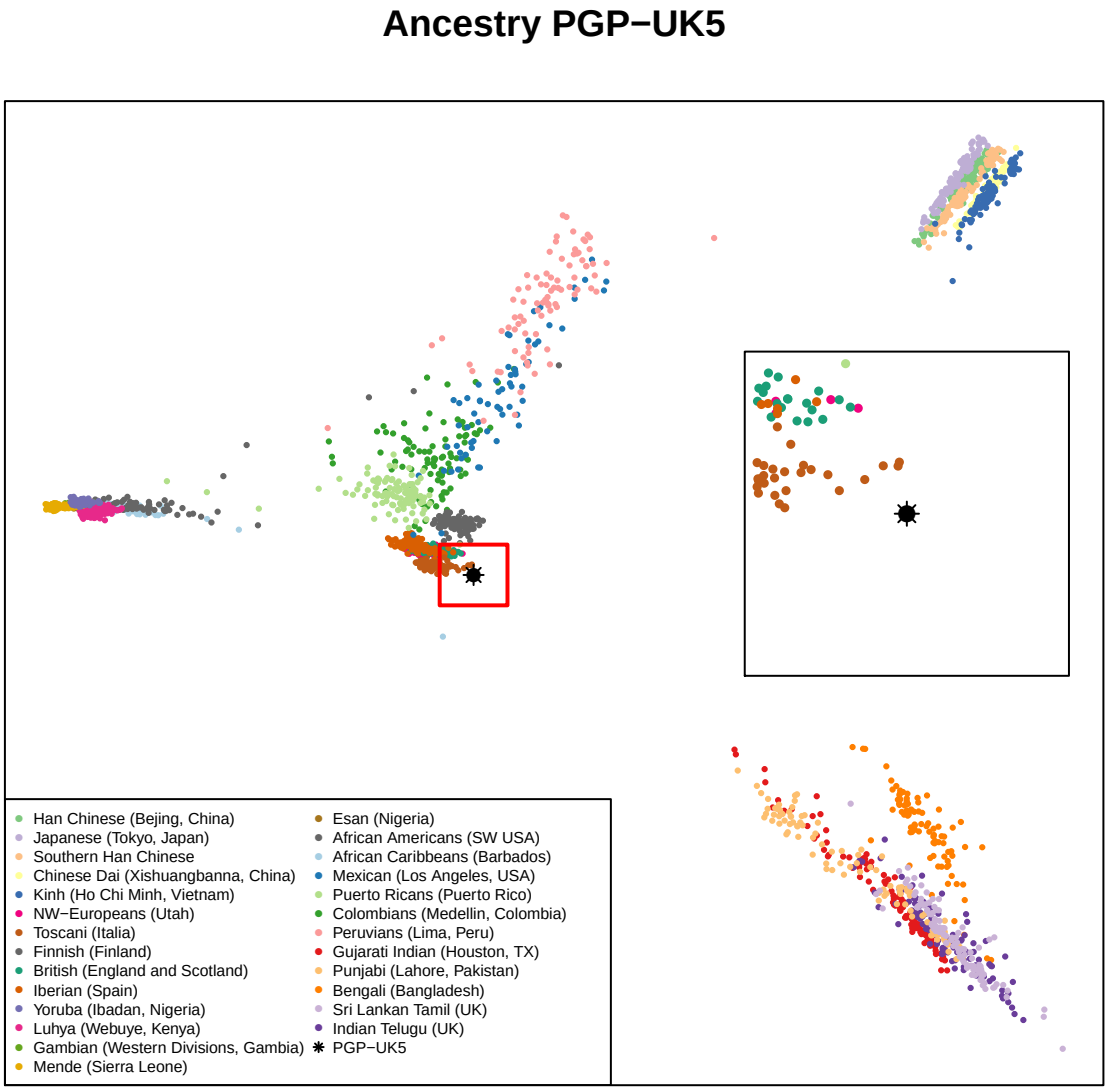

Figure 5: Ancestry Principal Component Analysis

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
2.8	rs6046	(T;T)	Better blood pressure: lower risk of myocardial...	Link	Link	Link
2.5	rs2943634	(A;A)	Lower risk of ischemic stroke		Link	
2	rs1012053	(A;C)	0.625x reduced risk of Bipolar Disorder.		Link	
2	rs10468017	(T;T)	Associated with higher HDL cholesterol		Link	
2	rs11045585	(A;A)	24% chance (lower than average) of docetaxel-in...		Link	
2	rs17070145	(C;T)	Increased memory performance			Link
2	rs1799884	(G;G)	Mothers have typical Birth-Weight babies. Sligh...			
2	rs1864163	(A;G)	Associated with higher HDL cholesterol		Link	
2	rs2073963	(T;T)	Reduced risk of baldness			
2	rs2235015	(G;T)	Somewhat more likely to respond to certain anti...	Link	Link	
2	rs261332	(A;G)	Associated with higher HDL cholesterol			
2	rs3738579	(C;T)	0.5x decreased risk for cervical cancer: HNSCC:...			
2	rs4143094	(G;G)	No increased risk of colorectal cancer correlat...			
2	rs4149268	(A;G)	Associated with higher HDL cholesterol		Link	
2	rs6505162	(A;C)	0.58x decreased risk for esophageal cancer	Link		
2	rs7216389	(C;C)	0.69x lower risk of Childhood Asthma.		Link	
2	rs801114	(T;T)	0.78x decreased Basal Cell Carcinoma risk.		Link	
2	rs8070723	(A;G)	0.18x reduced risk of developing progressive su...			
2	rs925391	(C;T)	Lower odds of going bald			
2	rs9642880	(G;G)	Slightly lower risk of Bladder Cancer.		Link	
1.6	rs10801935	(C;C)	0.3x decreased risk of breast cancer			
1.5	rs11136000	(C;T)	0.84x decreased risk for Alzheimer's disease		Link	
1.5	rs309375	(G;G)	Smaller mosquito bites			
1.5	rs3790844	(C;T)	Slightly reduced risk (0.77x) for pancreatic ca...			
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	
1.5	rs5888	(C;C)	Higher HDL cholesterol but lower risk for age-r...	Link		

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
1.5	rs729302	(A;C)	0.89x decreased risk of developing rheumatoid a...			
1.4	rs4320932	(G;G)	0.74x decreased risk for ovarian cancer			
1.2	rs6048	(A;G)	Slightly lower risk (10-20%) of deep vein throm...	Link	Link	Link
1.1	rs11172113	(C;T)	0.9x lower risk for migraines			
1.1	rs2293347	(G;G)	Among NSCLC patients: better Gefitinib response...	Link		
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	rs2494732	(T;T)	Lower odds of psychosis	Link	Link	
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;C)	0.73x decreased risk for colorectal cancer		Link	
1	rs7850258	(A;G)	Typical odds of developing primary hypothyroidi...			
1.0	rs11246226	(C;C)	Decreased risk of schizophrenia in limited stud...		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		
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	rs12252	(T;T)	More resistant to influenza	Link		Link
0	rs16990018	(A;A)	PrP Codon 171 Asn - Non-pathogenic variant	Link		Link
0	rs17244841	(A;A)	More responsive to statin treatment		Link	
0	rs1799945	(C;C)	Not a H63D hemochromatosis carrier.	Link	Link	Link
0	rs1800562	(G;G)	Not a C282Y hemochromatosis carrier.	Link	Link	Link
0	rs2240203	(A;A)	Blue eye color more likely			
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	rs9394492	(C;C)	<0.76x risk for restless legs			
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	rs1333049	(C;C)	1.9x increased risk for CAD		Link	
3.1	rs1421085	(C;C)	~1.7x increased obesity risk		Link	Link
3	rs1121980	(T;T)	2.76x risk for obesity		Link	
2.5	rs13266634	(C;T)	Increased risk for type-2 diabetes	Link	Link	Link
2.5	rs1800255	(A;A)	Increased risk for pelvic organ prolapse	Link	Link	Link
2.5	rs339331	(T;T)	Prostate cancer risk			
2.5	rs6441286	(G;G)	3.08x chance of developing primary biliary cirr...		Link	
2.5	rs7574865	(G;T)	1.3x risk of rheumatoid arthritis; 1.55x risk o...		Link	Link
2.4	rs7966230	(G;G)	Slightly lower levels of plasma VWF			
2.3	rs1859962	(G;G)	1.28x increased risk for prostate cancer		Link	
2.2	rs2231137	(G;G)	~1.5-3x increased risk for ischemic stroke	Link	Link	Link
2.2	rs2305089	(T;T)	Higher risk for chordoma	Link	Link	
2.1	rs10811661	(T;T)	1.2x increased risk for type-2 diabetes		Link	
2.1	rs1695	(G;G)	3.5x asthma risk in certain populations	Link	Link	
2.1	rs17077540	(A;G)	1.6x major depressive disorder risk			
2.1	rs2254958	(C;T)	1.24x increased risk for Alzheimer's			
2.1	rs2270641	(G;G)	3.7x higher risk for schizophrenia	Link	Link	
2.1	rs2383207	(G;G)	Increased risk for heart disease			
2.1	rs5186	(A;C)	~1.4x increased risk of hypertension	Link	Link	Link
2.1	rs5751876	(T;T)	Significantly higher anxiety levels after moder...	Link		
2.1	rs6457617	(T;T)	5.2x risk of rheumatoid arthritis		Link	
2.1	rs7837688	(G;G)	1.7x increased risk for prostate cancer			
2.1	rs795484	(A;G)	Increased morphine dose requirement and postope...			
2.1	rs9272346	(A;G)	5.5x risk type-1 diabetes		Link	
2.1	rs944289	(C;T)	1.3x increased thyroid cancer risk		Link	
2	rs10086908	(C;T)	1.7x increased risk for prostate cancer			
2	rs10306114	(A;G)	Higher risk of bleeding during coronary angiogr...			
2	rs1050631	(C;T)	Mean Survival Time of 25 months for esophageal ...	Link		
2	rs10513789	(G;T)	Increased risk of Parkinson's disease			
2	rs1064395	(A;G)	Having any copies of A at this SNP heightens yo...			
2	rs10757272	(T;T)	1.54x increased risk for Coronary artery diseas...			
2	rs11190870	(C;T)	Possibly increased risk of scoliosis			
2	rs1136287	(C;T)	1.5x increased risk of wet ARMD	Link	Link	
2	rs1160312	(A;G)	1.6x increased risk of Male Pattern Baldness.		Link	
2	rs12567232	(A;A)	Increased risk for Crohn's Disease		Link	
2	rs12770228	(A;A)	2x increased risk for meningioma			
2	rs13254738	(A;C)	1.18x prostate cancer risk		Link	
2	rs1360780	(C;T)	1.3x increased risk for depression		Link	
2	rs1585215	(A;G)	2x increased risk for Hodgkin lymphoma			
2	rs16942	(A;G)	Very slightly increased breast cancer risk	Link	Link	Link
2	rs16944	(G;G)	Increased risk of mental disorders		Link	
2	rs17228212	(C;T)	1.26x increased risk for heart disease		Link	
2	rs17435	(T;T)	1.4x increased risk for lupus			
2	rs1805007	(C;T)	Increased response to anesthetics	Link		Link
2	rs187238	(G;G)	Hypertension increases risk 3.75x for sudden ca...			
2	rs2156921	(A;G)	1.29x increased risk for depression			
2	rs2201841	(C;C)	1.5x increased risk for Crohn's disease		Link	
2	rs2230199	(C;G)	1.6x+ risk of ARMD	Link	Link	Link
2	rs2274223	(A;G)	1.5x increased risk for stomach and esophageal ...	Link	Link	
2	rs2305480	(T;T)	If 4 years old or younger: ~3x increased asthma...	Link	Link	
2	rs2305795	(A;G)	1.28x higher risk of narcolepsy compared to (G;...			Link
2	rs25487	(G;G)	2x higher risk for skin cancer; possibly other ...	Link	Link	
2	rs27388	(A;A)	Increased risk of developing schizophrenia			

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
2	rs3129934	(C;T)	Increased risk of Multiple Sclerosis.		Link	
2	rs3184504	(C;T)	Increased risk for celiac disease	Link	Link	
2	rs3212227	(A;C)	Significantly increased risk of developing cerv...			
2	rs358806	(C;C)	1.78x increased risk of developing Type-2 diabe...		Link	
2	rs3738919	(A;C)	1.94x risk of developing rheumatoid arthritis			
2	rs3775948	(G;G)	Slightly higher risk for gout			
2	rs4444903	(A;G)	3.5x risk of hep-cancer in cirrhosis patients; ...			
2	rs4633	(T;T)	Higher risk for endometrial cancer	Link	Link	
2	rs520354	(A;G)	Increased risk in men for biliary conditions			
2	rs6498169	(A;A)	>1.14x risk of multiple sclerosis		Link	
2	rs6807362	(C;C)	Increased autism risk	Link	Link	
2	rs6896702	(T;T)	Increased risk of developing Parkinson's Diseas...			
2	rs6908425	(C;C)	1.95x increased risk of developing Crohn's dise...		Link	
2	rs6997709	(G;T)	1.2x higher risk for hypertension			
2	rs7442295	(A;A)	~4x higher risk for hyperuracemia		Link	
2	rs744373	(C;T)	1.17x risk of Alzheimer's			
2	rs7794745	(A;T)	Slightly increased risk for autism		Link	Link
2	rs7807268	(C;G)	1.3x risk for Crohn's disease		Link	
2	rs7923837	(G;G)	3.2x risk for T2D			
2	rs800292	(C;C)	5% higher risk of Age related macular degenerat...	Link	Link	Link
2	rs9303277	(T;T)	1.46x Increased risk of developing primary bili...			
2	rs9525638	(T;T)	Weaker bones			
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
1.6	rs11523871	(A;C)	1.6x increased breast cancer risk for women ove...	Link	Link	
1.6	rs2736100	(G;G)	1.6x higher risk for glioma development		Link	
1.6	rs33980500	(C;T)	1.6x increase in risk for psoriatic arthritis	Link	Link	Link
1.5	rs10883365	(A;G)	1.2x increased risk for developing Crohn's dise...		Link	
1.5	rs10889677	(A;A)	1.5x increased risk for certain autoimmune dise...		Link	
1.5	rs1154155	(G;T)	1.94x increased risk for narcolepsy		Link	
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	
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	rs1375144	(C;T)	1.32x increased risk of developing bipolar diso...			
1.5	rs140701	(A;G)	Increased risk for anxiety disorders			
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	
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			
1.5	rs1975197	(C;T)	1.3x increased risk of developing restless legs...		Link	
1.5	rs1994090	(G;T)	Slightly increased risk of developing Parkinson...		Link	
1.5	rs199533	(C;T)	Slightly increased risk of developing Parkinson...	Link		
1.5	rs2280714	(A;G)	1.4x increased risk of SLE			
1.5	rs28694718	(A;G)	2x higher risk for schizophrenia			
1.5	rs3087243	(A;G)	Increased risk for auto-immune diseases		Link	
1.5	rs3764880	(A;G)	Possible 1.2 - 1.8x increased tuberculosis susc...	Link	Link	
1.5	rs3790565	(C;T)	Slightly increased risk of developing primary b...			
1.5	rs3814570	(C;T)	1.3x increased risk for Crohn's disease with il...			
1.5	rs401681	(C;T)	~1.2x increased risk for several types of cance...		Link	
1.5	rs4027132	(A;G)	1.39x increased risk of developing bipolar diso...			

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
1.5	rs4506565	(A;T)	1.4x increased risk for type-2 diabetes		Link	
1.5	rs4538475	(A;G)	Slightly increased risk of developing Parkinson...		Link	
1.5	rs4585	(T;T)	Slightly poorer (0.75x) response to metformin i...			
1.5	rs4785763	(A;A)	2x 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	rs4977574	(G;G)	Most studies find a somewhat elevated (~1.5x) r...		Link	
1.5	rs6435862	(G;T)	1.7x higher risk of aggressive neuroblastoma		Link	
1.5	rs7341475	(G;G)	1.58x increased schizophrenia risk for women		Link	
1.5	rs7536563	(A;G)	1.12x risk of multiple sclerosis		Link	
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	rs966221	(C;C)	1.5x increased stroke risk certain populations			
1.5	rs995030	(G;G)	Non-protective against testicular cancer		Link	
1.4	rs1126497	(T;T)	1.4x increased risk for breast cancer	Link	Link	Link
1.4	rs1545843	(A;A)	1.4x increased risk for depression (for those u...			
1.4	rs1800693	(G;G)	Slight (1.4x) increase in risk for multiple scl...	Link	Link	Link
1.4	rs1801157	(A;G)	1.4x higher risk for breast cancer			
1.4	rs2230201	(A;G)	1.4x risk of lupus	Link		
1.4	rs2252586	(A;A)	1.4x higher risk for glioma development			
1.4	rs3131296	(G;G)	1.4x increased risk for schizophrenia		Link	
1.4	rs4959039	(A;G)	1.4x higher risk for multiple sclerosis			
1.4	rs8050136	(A;A)	1.4x increased risk for T2D in some populations...		Link	
1.34	rs17465637	(C;C)	1.34x higher risk for myocardial infarction	Link	Link	
1.3	rs1047031	(A;A)	1.3x increased risk for periodontitis	Link		
1.3	rs1047286	(C;T)	1.3x increased risk for age-related macular deg...	Link	Link	Link
1.3	rs10947262	(C;C)	1.3x increased risk for osteoarthritis			
1.3	rs1260326	(C;T)	Slightly higher risk for gout	Link	Link	Link
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...			
1.25	rs13387042	(A;A)	1.24x increased risk for breast cancer		Link	
1.25	rs748404	(T;T)	Slightly increased risk (1.25) for lung cancer...		Link	
1.2	rs10865331	(A;G)	1.2x higher risk for ankylosing spondylitis			
1.2	rs11037909	(T;T)	1.47x type II diabetes risk	Link		
1.2	rs1344706	(T;T)	1.2x increased risk for schizophrenia		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	rs2076295	(G;T)	One copy of the risk allele (G): slightly incre...			
1.2	rs2651899	(G;G)	1.2x higher risk for migraines			
1.2	rs2814707	(A;G)	1.2x increased risk for ALS		Link	
1.2	rs3740878	(A;A)	1.46x type II diabetes risk; common	Link		
1.2	rs3849942	(A;G)	1.2x increased risk for ALS		Link	
1.2	rs393152	(A;G)	Slight increased risk of both PD and AD	Link	Link	
1.2	rs4795067	(A;G)	Slight increase in risk for psoriatic arthritis...			
1.2	rs498872	(C;T)	1.2x higher risk for glioma development		Link	
1.2	rs6010620	(A;G)	1.2x higher risk for glioma development: 1.17x ...		Link	
1.17	rs3802842	(A;C)	1.17x increased risk of colorectal cancer		Link	
1.1	rs11110912	(C;C)	1.3x high blood pressure risk			
1.1	rs11650494	(A;G)	Slightly higher prostate cancer risk			
1.1	rs1799966	(A;G)	Very slightly increased breast cancer risk	Link	Link	Link
1.1	rs1800450	(A;G)	Mannose binding deficiency but of low clinical ...	Link	Link	Link
1.1	rs34516635	(G;G)	Less longevity for Ashkenazi Jewish women.	Link		Link
1.1	rs4324715	(C;T)	1.5x increased testicular cancer risk for men			
1.1	rs6495446	(C;T)	1.2x increased risk for chronic kidney disease			
1.1	rs688034	(C;T)	1.1x risk higher risk for coronary artery disea...		Link	

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
1.1	rs6897876	(C;T)	Slight increase in testicular cancer risk for m...			
1.1	rs7171755	(A;A)	Very slight descrease in cortical thickness and...			
1.1	rs7412	(C;C)	More likely to gain weight if taking olanzapine...	Link	Link	Link
1.1	rs889312	(A;C)	Very slightly higher risk for breast cancer		Link	
1.07	rs2291834	(C;C)	Very slightly higher risk for myocardial infarc...			
1	rs10504861	(G;G)	Major allele: normal risk of migraine			
1	rs1143674	(A;G)	1.3x increased autism risk	Link		
1	rs12752888	(C;C)	Faster progression of mild cognitive impairment...			
1	rs1804197	(A;C)	Increased risk of familial colorectal cancer an...			Link
1	rs2273697	(A;G)	Adverse reaction more likely to carbamazepine i...	Link	Link	
1	rs2546890	(A;A)	Higher risk of multiple sclerosis			
1	rs3194051	(A;A)	>1.1x risk of type-1 diabetes	Link	Link	Link
1	rs6932590	(C;T)	1.1x increased risk for schizophrenia		Link	
1	rs761100	(G;G)	Higher risk for dyslexia			
0.1	rs601338	(A;G)	Susceptible to Norovirus infections	Link	Link	Link
0	rs10761659	(A;A)	1.5x risk of Crohn's disease		Link	
0	rs2296336	(C;C)	2.9x risk of type-1 diabetes			
0	rs4293393	(T;T)	1.25x Increased Risk of CKD for T allele in ...			
0	rs4714156	(C;C)	<0.61x risk for restless legs			
0	rs4795400	(T;T)	If 4 years old or younger: ~2.5x increased asth...		Link	
0	rs6314	(C;C)	Higher risk for RA	Link	Link	

- Genosets (Multi-variant Phenotypes)

Magnitude	Identifier	Summary
4	gs145	Female
3.1	gs191	Problem metabolizing NSAIDs
3	gs127	Intermediate warfarin metabolizer
2.5	gs100	Lactose intolerance risk
2.5	gs155	CYP3A5 non-expressor
2.5	gs157	More stimulated by coffee
2.5	gs161	CYP2C9 Intermediate Metabolizers
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	You will lose 2.5x as much weight on a low fat ...
2.2	gs280	Light hair color for europeans
2	gs129	Unable to classify your ABO blood type
2	gs246	APOE3/APOE3
2	gs249	Parkinson's Disease Risk
1.5	gs1001	Mitochondrial Haplogroup H
1.2	gs184	Able to taste bitterness.
1	gs182	CYP2D6*39
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).