Genomics Report for PGP-UK6/uk0C72FF

1 Summary

This is the genome report for participant PGP-UK6/uk0C72FF . 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.

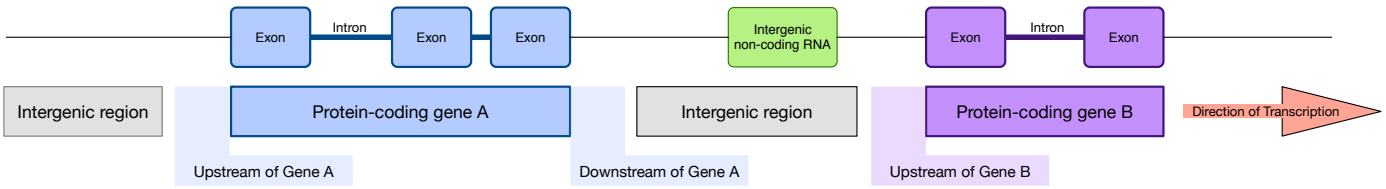

Figure 1: Diagram of gene structure indicating locations of potential variants

Feature	Count
Lines of input read	4138711
Variants remaining after filtering	4111625
Novel / existing variants	108299 (2.6%) / 4003326 (97.4%)
Overlapped genes	54592
Overlapped transcripts	64300
Overlapped regulatory features	211668

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.

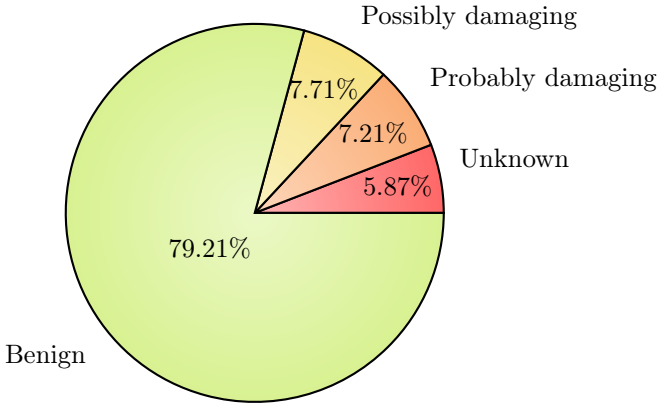

Figure 2: PolyPhen Summary

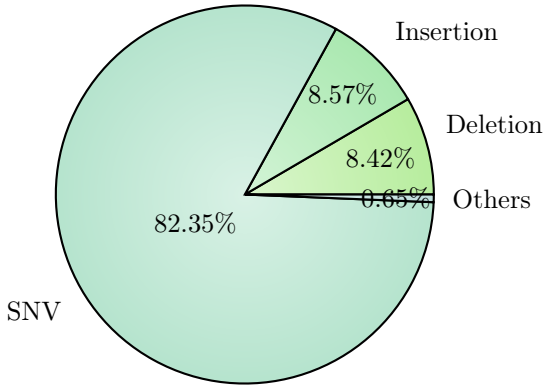

Figure 3: Variant Class

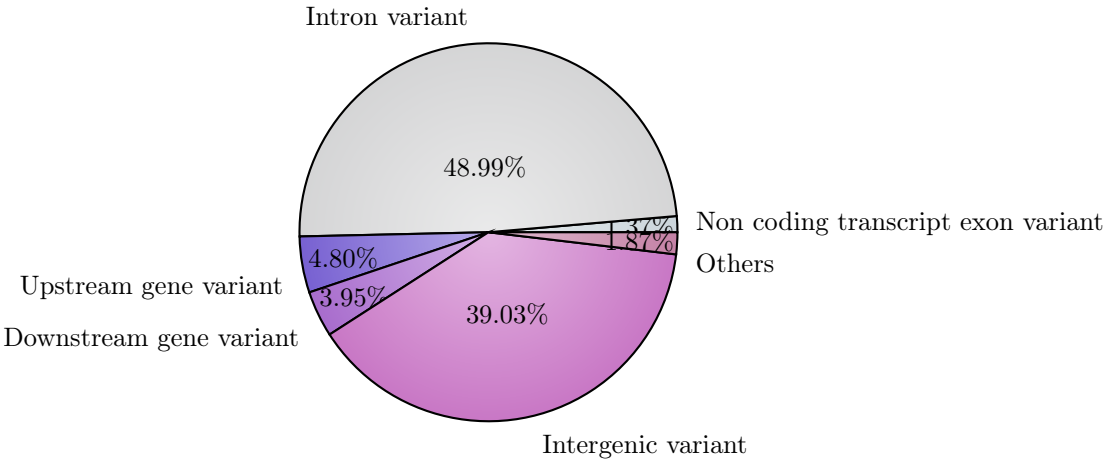

Figure 4: Consequence type

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.

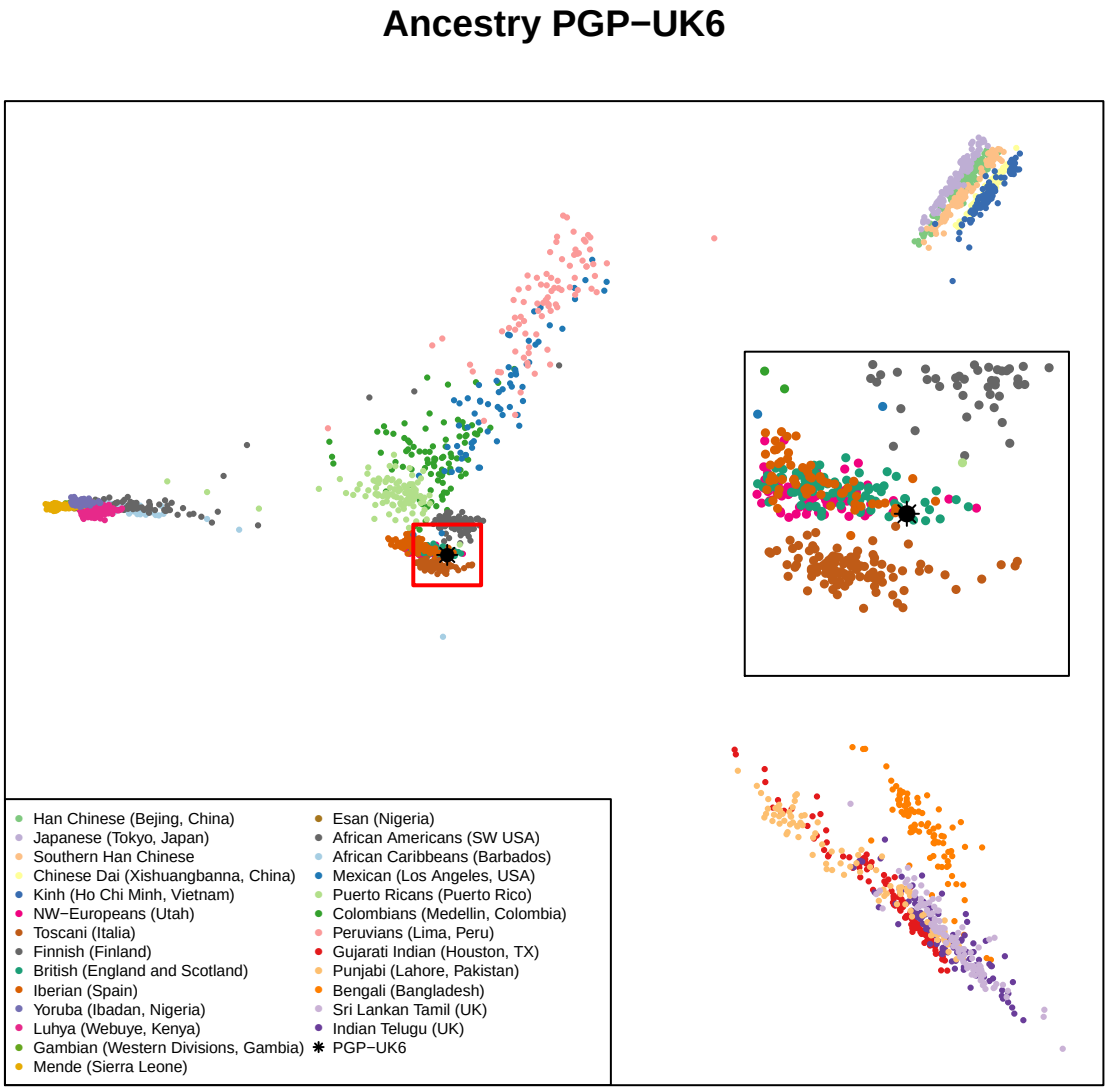

Figure 5: Ancestry Principal Component Analysis

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
2.1	rs2511989	(A;G)	0.63x decreased age-related macular degeneratio...		Link	
2.1	rs3775291	(A;G)	0.71x decreased risk for dry age related macula...	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	rs10088218	(A;A)	0.5x decreased risk for ovarian cancer			
2	rs10468017	(T;T)	Associated with higher HDL cholesterol		Link	
2	rs10503669	(A;C)	Associated with higher HDL cholesterol		Link	
2	rs11045585	(A;A)	24% chance (lower than average) of docetaxel-in...		Link	
2	rs12678919	(A;G)	Associated with higher HDL cholesterol		Link	
2	rs12979860	(C;C)	~80% of such hepatitis C patients respond to tr...		Link	
2	rs1544410	(G;G)	Decreased risk of low bone mineral density diso...		Link	
2	rs17070145	(C;T)	Increased memory performance			Link
2	rs1799884	(G;G)	Mothers have typical Birth-Weight babies. Sligh...			
2	rs1864163	(G;G)	Associated with higher HDL cholesterol		Link	
2	rs2542052	(C;C)	Better odds of living to 100			
2	rs3738579	(C;T)	0.5x decreased risk for cervical cancer: HNSCC:...			
2	rs3764261	(G;T)	Associated with higher HDL cholesterol		Link	Link
2	rs3782179	(C;T)	3x lower odds of testicular cancer risk for men...			
2	rs3819331	(T;T)	Lower risk of autism	Link		
2	rs3914132	(C;T)	Lower otosclerosis risk		Link	
2	rs4143094	(G;G)	No increased risk of colorectal cancer correlat...			
2	rs4149268	(A;G)	Associated with higher HDL cholesterol		Link	
2	rs505922	(T;T)	Blood type O		Link	
2	rs763110	(C;T)	~0.80x reduced cancer risk			Link
2	rs7776725	(T;T)	Stronger bones		Link	
2	rs801114	(T;T)	0.78x decreased Basal Cell Carcinoma risk.		Link	
2	rs9642880	(G;G)	Slightly lower risk of Bladder Cancer.		Link	
1.5	rs1026732	(A;G)	0.70x risk for restless legs		Link	

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
1.5	rs1063192	(C;C)	0.71x reduced risk of myocardial infarction			
1.5	rs11635424	(A;G)	0.70x risk for restless legs		Link	
1.5	rs1165205	(A;A)	0.85x decreased gout risk		Link	
1.5	rs12593813	(A;G)	0.71x risk for restless legs		Link	
1.5	rs16991615	(A;G)	Slight increase (11 months) in avg age at menop...	Link	Link	
1.5	rs3784709	(C;T)	0.71x risk of developing restless legs syndrome...		Link	
1.5	rs3790844	(C;T)	Slightly reduced risk (0.77x) for pancreatic ca...			
1.5	rs3851179	(A;G)	0.85x decreased risk for Alzheimer's disease		Link	
1.5	rs4149274	(C;T)	Associated with higher HDL (good) cholesterol			
1.5	rs4489954	(G;T)	0.69x risk risk of developing restless legs syn...		Link	
1.5	rs4939883	(C;T)	Associated with higher HDL cholesterol		Link	
1.5	rs5968255	(C;C)	Slower AIDS progression (8 years)			
1.5	rs610932	(A;A)	A allele associated with reduced risk of Alzhei...			
1.5	rs729302	(A;C)	0.89x decreased risk of developing rheumatoid a...			
1.2	rs11246226	(A;C)	Decreased risk of schizophrenia in limited stud...		Link	
1.1	rs13333226	(A;G)	Slightly lower risk for hypertension			Link
1	rs11601907	(C;T)	Variant allele is designated benign in ClinVar	Link		Link
1	rs182549	(C;T)	Can digest milk.			Link
1	rs2351299	(G;T)	Possible reduced risk of Autism			
1	rs2546890	(G;G)	Lower risk of multiple sclerosis			
1	rs2952768	(C;T)	Slightly less drug dependence: decreased effect...			
1	rs4939827	(C;T)	0.86x decreased risk for colorectal cancer		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	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	rs16947	(A;A)	Homozygous for CYP2D6 variants (non-CYP2D6*1)	Link	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	rs1799782	(C;C)	Lower risk for skin cancer	Link	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	rs2306402	(C;C)	1.18x increased risk for LOAD			
0	rs242941	(G;G)	Better response to inhaled corticosteroid in pa...		Link	
0	rs28933385	(G;G)	Prion protein Codon 200 (E) - Non pathogenic va...			Link
0	rs312481	(C;C)	Better response to certain calcium channel bloc...			
0	rs403016	(C;C)	2x risk for lupus		Link	
0	rs5746059	(A;A)	Slightly higher fat mass			
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
3	rs13266634	(C;C)	Increased risk for type-2 diabetes	Link	Link	Link
3	rs2066844	(C;T)	3x higher risk for Crohn's disease	Link	Link	Link
3	rs2981582	(C;T)	1.3x higher risk of ER+ breast cancer		Link	
3	rs4244285	(A;G)	Poorer metabolizer of several popular medicines...	Link	Link	Link
3	rs6920220	(A;G)	1.2x risk Rheumatoid Arthritis		Link	
2.7	rs10830963	(C;G)	Increased type-2 diabetes risk; higher gestatio...		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: l...	Link	Link	Link
2.5	rs1799971	(A;G)	Stronger cravings for alcohol. if alcoholic: na...	Link	Link	Link
2.5	rs2073963	(G;G)	Increased risk of baldness			
2.5	rs2254958	(C;C)	1.61x increased risk for Alzheimer's			
2.5	rs2943634	(C;C)	Higher risk of ischemic stroke		Link	
2.5	rs339331	(T;T)	Prostate cancer risk			
2.5	rs3738919	(C;C)	1.94x risk of developing rheumatoid arthritis			
2.5	rs5219	(T;T)	2.5x increased risk for type-2 diabetes	Link	Link	Link
2.5	rs5888	(C;T)	3x higher risk for age-related macular degenera...	Link		
2.5	rs664143	(C;T)	Higher risk for number of cancers			
2.5	rs7574865	(G;T)	1.3x risk of rheumatoid arthritis; 1.55x risk o...		Link	Link
2.5	rs8034191	(C;T)	1.27x lung cancer risk		Link	
2.5	rs9934438	(A;A)	Coumadin resistance		Link	Link
2.4	rs1143679	(A;G)	1.78x increased risk for SLE	Link	Link	
2.2	rs1052133	(G;G)	2x increased bladder cancer risk; 4.5x increase...	Link	Link	
2.2	rs2231137	(G;G)	~1.5-3x increased risk for ischemic stroke	Link	Link	Link
2.1	rs10811661	(T;T)	1.2x increased risk for type-2 diabetes		Link	
2.1	rs1329428	(G;G)	2x increased risk for macular degeneration			
2.1	rs1695	(G;G)	3.5x asthma risk in certain populations	Link	Link	
2.1	rs17563	(C;C)	Risk for otosclerosis	Link	Link	Link
2.1	rs4402960	(T;T)	1.2x increased risk for type-2 diabetes		Link	Link
2.1	rs5186	(A;C)	~1.4x increased risk of hypertension	Link	Link	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	rs1024611	(C;T)	Increased risk of exercise induced ischemia			Link
2	rs10248420	(A;A)	7x less likely to respond to certain antidepres...		Link	
2	rs1045642	(C;T)	Slower metaboliser for some drugs	Link	Link	
2	rs1050152	(C;T)	2.1x increased risk of Crohn's disease	Link	Link	Link
2	rs1050631	(C;T)	Mean Survival Time of 25 months for esophageal ...	Link		
2	rs1051730	(C;T)	1.3x increased risk of lung cancer	Link	Link	Link
2	rs10984447	(A;A)	>1.17x increased risk for multiple sclerosis		Link	
2	rs11171739	(C;C)	1.75x risk of developing Type-1 diabetes		Link	
2	rs11190870	(C;T)	Possibly increased risk of scoliosis			
2	rs1160312	(A;G)	1.6x increased risk of Male Pattern Baldness.		Link	
2	rs11983225	(T;T)	7x less likely to respond to certain antidepres...		Link	
2	rs1219648	(A;G)	1.20x risk for breast cancer		Link	
2	rs12567232	(A;A)	Increased risk for Crohn's Disease		Link	
2	rs1585215	(A;G)	2x increased risk for Hodgkin lymphoma			
2	rs1691053	(A;G)	Increased risk of developing prostate cancer			
2	rs16944	(G;G)	Increased risk of mental disorders		Link	
2	rs17228212	(C;C)	>1.26x increased risk for heart disease		Link	
2	rs1734791	(A;A)	1.4x increased risk for lupus			
2	rs1800896	(A;A)	1.8x increased prostate cancer risk			
2	rs187238	(G;G)	Hypertension increases risk 3.75x for sudden ca...			

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
2	rs2156921	(A;G)	1.29x increased risk for depression			
2	rs2201841	(C;C)	1.5x increased risk for Crohn's disease		Link	
2	rs2235015	(G;G)	Somewhat less likely to respond to certain anti...	Link	Link	
2	rs2235040	(G;G)	7x less likely to respond to certain antidepres...	Link	Link	
2	rs2235067	(G;G)	7x less likely to respond to certain antidepres...			
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	rs2352028	(T;T)	Increased risk of lung cancer in non-smokers an...		Link	
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	rs2707466	(G;G)	Weaker bones	Link	Link	
2	rs27388	(A;A)	Increased risk of developing schizophrenia			
2	rs2908004	(C;C)	Weaker bones	Link	Link	
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	rs3746444	(C;T)	~1.2x increased risk for cancer	Link		
2	rs3775948	(G;G)	Slightly higher risk for gout			
2	rs4148739	(A;A)	7x less likely to respond to certain antidepres...		Link	
2	rs4420638	(A;G)	~3x increased Alzheimer's risk; 1.4x increased ...		Link	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	rs4961	(G;T)	1.8x increased risk for high blood pressure	Link	Link	Link
2	rs520354	(A;G)	Increased risk in men for biliary conditions			
2	rs6457617	(C;T)	2.3x risk of rheumatoid arthritis		Link	
2	rs6498169	(A;A)	>1.14x risk of multiple sclerosis		Link	
2	rs6601764	(C;C)	1.52x increased risk of developing Crohn's dise...		Link	
2	rs663048	(G;T)	Significantly increased risk of developing lung...	Link	Link	
2	rs669	(G;G)	3.8x or higher increased risk for Alzheimers	Link	Link	Link
2	rs6897932	(C;T)	1.3x increased risk for multiple sclerosis	Link	Link	Link
2	rs6997709	(G;T)	1.2x 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	rs7639618	(C;T)	1.45x increased osteoarthritis risk	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	(T;T)	Increased risk of bladder cancer and 2x risk of...			
2	rs9525638	(T;T)	Weaker bones			
2	rs9652490	(A;A)	~2x increased risk for Parkinson's disease: and...		Link	
2.0	rs4911414	(G;T)	2-4x higher risk of sun sensitivity if part of ...		Link	
1.9	rs7923837	(A;G)	1.6x risk for T2D			
1.8	rs37973	(A;G)	Among asthmatics: 1.5x more likely to show less...			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	rs11523871	(A;C)	1.6x increased breast cancer risk for women ove...	Link	Link	
1.6	rs1537415	(C;G)	1.6x increased risk for periodontitis		Link	
1.6	rs2981745	(C;T)	1.6x increased risk for breast cancer in female...			
1.5	rs10492519	(A;G)	Slightly increased risk of developing prostate ...			
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	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	rs13181	(G;T)	1.12x increased risk for cutaneous melanoma	Link	Link	Link

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
1.5	rs13376333	(C;T)	1.5x higher risk of atrial fibrillation		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	rs2076295	(G;G)	Slightly increased risk for pulmonary fibrosis ...			
1.5	rs2241880	(C;T)	1.4x increased risk for Crohn's disease in Cauc...	Link	Link	Link
1.5	rs2280714	(A;A)	1.4x increased risk of SLE			
1.5	rs28694718	(A;G)	2x higher risk for schizophrenia			
1.5	rs2881766	(T;T)	Slightly increased risk for pregnancy-induced h...			
1.5	rs3087243	(A;G)	Increased risk for auto-immune diseases		Link	
1.5	rs3825776	(A;G)	1.3x increased risk for ALS		Link	
1.5	rs393152	(A;A)	Increased risk of both PD and AD	Link	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	rs4506565	(A;T)	1.4x increased risk for type-2 diabetes		Link	
1.5	rs464049	(T;T)	Increased risk of schizophrenia in limited stud...			
1.5	rs486907	(A;G)	1.5x increased prostate cancer risk	Link	Link	Link
1.5	rs6435862	(G;T)	1.7x higher risk of aggressive neuroblastoma		Link	
1.5	rs6896702	(C;T)	Slightly increased risk of developing Parkinson...			
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	rs7850258	(G;G)	Slightly higher odds of developing primary hypo...			
1.5	rs807701	(C;T)	Slightly increased dyslexia risk			
1.5	rs9303277	(C;T)	1.46x Slightly increased risk of developing pri...			
1.5	rs966221	(C;C)	1.5x increased stroke risk certain populations			
1.4	rs1545843	(A;A)	1.4x increased risk for depression (for those u...			
1.4	rs2046210	(C;T)	1.4x increased breast cancer risk		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	rs4977756	(G;G)	1.4x higher risk for glioma development		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...			
1.3	rs2736100	(G;T)	1.3x higher risk for glioma development		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	rs11842874	(A;G)	+17% increased risk for osteoarthritis			
1.2	rs143383	(C;T)	1.1x increased risk for osteoarthritis		Link	Link
1.2	rs2056116	(A;G)	1.18x risk for breast cancer			
1.2	rs2252586	(A;G)	1.2x higher risk for glioma development			
1.2	rs2665390	(C;T)	1.2x increased risk for ovarian cancer			
1.2	rs3740878	(A;A)	1.46x type II diabetes risk; common	Link		
1.2	rs419788	(A;G)	2.0x risk for lupus	Link		
1.2	rs4686484	(A;A)	Slightly increased risk for celiac disease			
1.2	rs6010620	(A;G)	1.2x higher risk for glioma development: 1.17x ...		Link	
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	
1.1	rs11110912	(C;C)	1.3x high blood pressure risk			
1.1	rs13387042	(A;G)	1.12x increased risk for breast cancer		Link	
1.1	rs1800450	(A;G)	Mannose binding deficiency but of low clinical ...	Link	Link	Link
1.1	rs2651899	(A;G)	1.1x higher risk for migraines			
1.1	rs2653349	(G;G)	2-6x increased risk for cluster headaches	Link	Link	

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
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	rs6897876	(C;T)	Slight increase in testicular cancer risk for m...			
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
1.1	rs925391	(C;C)	More likely to go bald; common			
1.05	rs2291834	(C;T)	Very slightly higher risk for myocardial infarc...			
1	rs10504861	(G;G)	Major allele: normal risk of migraine			
1	rs1143674	(A;A)	1.3x increased autism risk	Link		
1	rs12718541	(A;A)	Nicotine dependence			
1	rs3194051	(A;G)	1.12x risk of type-1 diabetes	Link	Link	Link
1	rs6932590	(T;T)	1.1x increased risk for schizophrenia		Link	
1	rs987525	(A;C)	2.5x increased risk for cleft lip		Link	
0	rs10761659	(A;A)	1.5x risk of Crohn's disease		Link	
0	rs1333040	(C;T)	1.24x increased myocardial infarction risk: 1.2...		Link	
0	rs3761418	(A;A)	1.3x increased risk for depression			
0	rs440446	(G;G)	Increased risk in men for biliary conditions	Link		
0	rs6314	(C;C)	Higher risk for RA	Link	Link	
0	rs7787082	(G;G)	7x less likely to respond to certain antidepres...		Link	
0	rs9298506	(A;A)	Higher Risk Aneurysm		Link	

- Genosets (Multi-variant Phenotypes)

Magnitude	Identifier	Summary
4	gs144	Male
3.1	gs122	7x risk of baldness
3.1	gs191	Problem metabolizing NSAIDs
3	gs114	Haplogroup R1b1b2a1a1
3	gs127	Intermediate warfarin metabolizer
3	gs241	Lighter green: brown or hazel eye color
2.5	gs155	CYP3A5 non-expressor
2.5	gs161	CYP2C9 Intermediate Metabolizers
2.5	gs259	Homozygous for eye color haplotype #3
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 ...
2.3	gs255	Homozygous eye color haplotype #1
2	gs101	Probably able to digest milk
2	gs159	CYP1A2 fast metabolizer
2	gs213	Haplogroup R (Y-DNA)
2	gs246	APOE3/APOE3
1.5	gs247	Parkinson's Disease Risk
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).