Genomics Report for PGP-UK10/uk481F67

1 Summary

This is the genome report for participant PGP-UK10/uk481F67. 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 <u>personal and research purposes</u> 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.

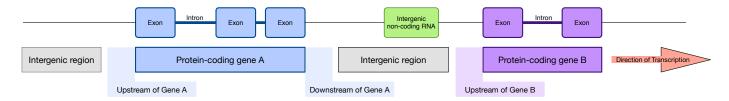


Figure 1: Diagram of gene structure indicating locations of potential variants

Feature	Count
Lines of input read	4932231
Variants remaining after filtering	4900960
Novel / existing variants	$165833 \; (3.4\%) \; / \; 4735127 \; (96.6\%)$
Overlapped genes	55493
Overlapped transcripts	65993
Overlapped regulatory features	242649

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.

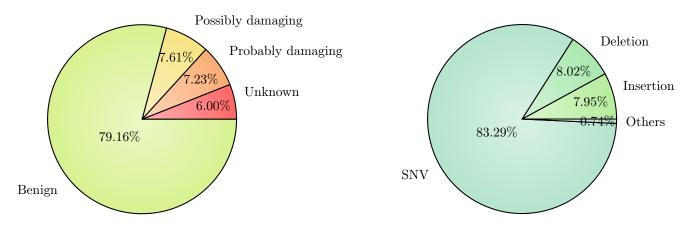


Figure 2: PolyPhen Summary

Figure 3: Variant Class

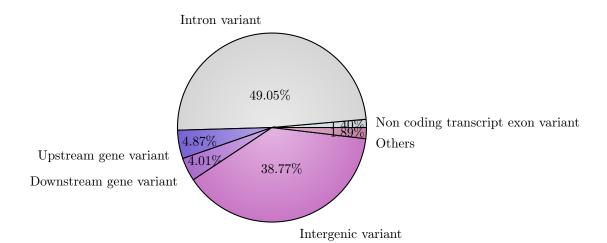


Figure 4: Consequence type

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.

Ancestry PGP-UK10

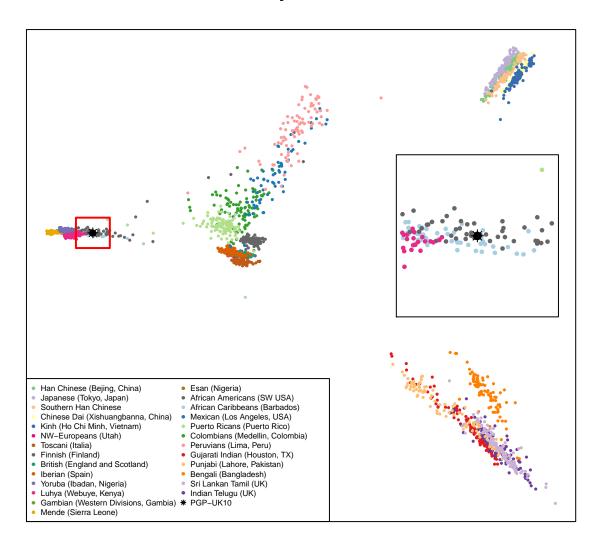


Figure 5: Ancestry Principal Component Analysis

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
3	rs925391	(T;T)	Unlikely to go bald			
2.5	rs3782179	(C;C)	9x lower odds of testicular cancer			
2.4	rs2802288	(A;A)	Longer lifespan			
2.1	rs2511989	(A;G)	0.63x decreased age-related macular degeneratio		Link	
2.1	rs6505162	(A;A)	0.43x decreased risk for esophageal cancer	Link		
2.1	rs995030	(A;A)	Reduced risk of testicular cancer		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	rs1160312	(G;G)	Reduced risk of Baldness.		Link	
2	rs11635424	(A;A)	<0.70x risk for restless legs		Link	
2	rs12593813	(A;A)	<0.71x risk for restless legs		Link	
2	rs13706	(A;A)	Decreased risk for certain cancers	Link	Link	Link
2	rs1544410	(G;G)	Decreased risk of low bone mineral density diso		Link	
2	rs16890979	(T;T)	Lower systolic blood pressure: serum uric acid	Link	Link	
2	rs1799884	(G;G)	Mothers have typical Birth-Weight babies. Sligh			
2	rs1864163	(A;G)	Associated with higher HDL cholesterol		Link	
2	rs2235015	(G;T)	Somewhat more likely to respond to certain anti	Link	Link	
2	rs2243250	(T;T)	0.33x decreased risk for myocardial infarction			
2	rs2707466	(A;A)	Stronger bones	Link	Link	
2	rs2764264	(C;C)	Greater odds of living to 95			
2	rs2908004	(T;T)	Stronger bones	Link	Link	
2	rs3178250	(C;C)	Lower otosclerosis risk			
2	rs3764261	(G;T)	Associated with higher HDL cholesterol		Link	Link
2	rs3819331	(T;T)	Lower risk of autism	Link		
2	rs3914132	(C;T)	Lower otosclerosis risk		Link	
2	rs4149268	(A;G)	Associated with higher HDL cholesterol		Link	
2	rs505922	(T;T)	Blood type O		Link	
2	rs6855911	(G;G)	Rare: but 0.62x decreased risk for gout		Link	

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
2	rs763110	(T;T)	~0.80x reduced cancer risk			Link
2	rs7776725	(T;T)	Stronger bones		Link	
1.6	rs10801935	(C;C)	0.3x decreased risk of breast cancer			
1.5	rs1026732	(A;G)	0.70x risk for restless legs		Link	
1.5	rs1050631	(C;C)	Mean Survival Time of 32 months for esophageal	Link		
1.5	rs11136000	(C;T)	0.84x decreased risk for Alzheimer's disease		Link	
1.5	rs2007153	(A;A)	Decreased risk of schizophrenia in limited stud			
1.5	rs28362263	(A;G)	Modest reduction in LDL-C and coronary risk	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	rs6427528	(A;G)	For rheumatoid arthritis patients: better respo			
1.5	rs729302	(A;C)	0.89x decreased risk of developing rheumatoid a			
1.5	rs9939609	(T;T)	Lower risk of obesity and Type-2 diabetes		Link	
1.3	rs10166942	(C;C)	0.7x lower risk for migraines			
1.2	rs11246226	(A;C)	Decreased risk of schizophrenia in limited stud		Link	
1.1	rs11172113	(C;T)	0.9x lower risk for migraines			
1.1	rs13333226	(A;G)	Slightly lower risk for hypertension	T	T . 1	Link
1.1	rs16891982	(C;C)	Generally non-European: but if European: 7x mor	Link	Link	Link
1.1	rs2293347	(G;G)	Among NSCLC patients: better Gefitinib response	Link		
1.1	rs7568369	(G;T)	0.90x reduced risk of obesity		T • 1	
1	rs10248420	(G;G)	7x more likely to respond to certain antidepres		Link	
1	rs11983225	(C;T)	7x more likely to respond to certain antidepres		Link	T . 1
1	rs182549	(C;T)	Can digest milk.	T · 1	T · 1	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	rs2546890	(G;G)	Lower risk of multiple sclerosis		Timl.	
1	rs4148739	(A;G)	7x more likely to respond to certain antidepres 0.86x decreased risk for colorectal cancer		Link Link	
1 1	rs4939827 rs800292	(C;T)	1% decreased risk of macular degeneration	Link	Link	Link
1.0	rs2283123	(C;T)	Decreased risk of schizophrenia in limited stud	LIIIK	LIIIK	LIIIK
1.0	rs6583817	(C;T) (C;T)	~0.80x (lower) risk for late onset Alzheimer's			
0.5	rs36094464	(A;T)	Most likely benign: though reported years ago t	Link	Link	Link
0.3	rs891512	(G;G)	Lower blood pressure than those with an A allel	Link	LIIIK	LIIIK
0.1	rs1047781	(A;A)	ABH blood group "Secretor" status if Japanese	Link	Link	Link
0	rs1056836	(G;G)	0.3x decreased risk for prostate cancer	Link	Link	Link
0	rs10897346	(C;C)	If depressed: 2.6x more likely to not respond t	TIIII	1311111	Lillix
0	rs12252	(T;T)	More resistant to influenza	Link		Link
0	rs16990018	(A;A)	PrP Codon 171 Asn - Non-pathogenic variant	Link		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	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	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	rs185919705	(C;T)	Corneal dystrophy: fuchs endothelial: 8	Link		Link
4	rs7158782	(G;G)	4x higher risk of adverse side-effect if taking			
3	rs13266634	(C;C)	Increased risk for type-2 diabetes	Link	Link	Link
3	rs3738579	(T;T)	1.5x - 2x increased risk for cervical cancer: H			
3	rs4244285	(A;G)	Poorer metabolizer of several popular medicines	Link	Link	Link
2.9	rs16901979	(A;A)	1.5x increased risk for prostate cancer		Link	
2.6	rs13361189	(C;C)	2.6x increased risk for Crohn's disease		Link	
2.6	rs4958847	(A;A)	2.6x increased risk for Crohn's disease			
2.5	rs1121980	(C;T)	1.67x risk for obesity		Link	
2.5	rs12803066	(A;G)	Increased risk of myopia			
2.5	rs17595731	(C;G)	~5 fold higher risk for Fuchs' dystrophy: a cor			
2.5	rs2241880	(C;C)	2x-3x increased risk for Crohn's disease in Cau	Link	Link	Link
2.5	rs339331	(T;T)	Prostate cancer risk			
2.5	rs3780374	(A;G)	Substantially increased odds of developing V617			
2.5	rs4143094	(G;T)	Slightly (17%) higher risk of colorectal cancer			
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			
2.3	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
2.1	rs10090154	(T;T)	1.4x increased risk for prostate cancer			
2.1	rs10411210	(T;T)	1.15x increased risk of colorectal cancer		Link	
2.1	rs1219648	(G;G)	1.64x risk for breast cancer		Link	
2.1	rs1329428	(G;G)	2x increased risk for macular degeneration			
2.1	rs17070145	(C;C)	Reduced memory abilities			Link
2.1	rs2254958	(C;T)	1.24x increased risk for Alzheimer's			
2.1	rs2294008	(T;T)	Increased risk of gastric and bladder cancer	Link	Link	
2.1	rs2383207	(G;G)	Increased risk for heart disease			
2.1	rs2420946	(T;T)	1.64x risk for breast cancer			
2.1	rs380390	(C;C)	Increased risk for ARMD		Link	
2.1	rs4402960	(T;T)	1.2x increased risk for type-2 diabetes		Link	Link
2.1	rs4430796	(A;A)	1.38x increased risk for prostate cancer		Link	
2.1	rs4444903	(G;G)	3.5x risk of hep-cancer in cirrhosis patients;			
2.1	rs7837688	(G;G)	1.7x increased risk for prostate cancer			
2.1	rs9272346	(A;G)	5.5x risk type-1 diabetes		Link	
2	rs10086908	(C;T)	1.7x increased risk for prostate cancer			
2	rs1024611	(C;T)	Increased risk of exercise induced ischemia			Link
2	rs10811661	(C;T)	1.2x increased risk for type-2 diabetes		Link	
2	rs10889677	(C;C)	1x increased risk for certain autoimmune diseas		Link	
2	rs10984447	(A;A)	>1.17x increased risk for multiple sclerosis		Link	
2	rs11229030	(C;C)	Higher odds of Crohn's disease		T . 1	
2	rs12431733	(T;T)	Increased risk of developing Parkinson's Diseas		Link	
2	rs13254738	(A;C)	1.18x prostate cancer risk		Link	
2	rs1333048	(A;C)	1.3x increased coronary artery disease risk			
2	rs1375144	(C;C)	1.59x increased risk of developing bipolar diso	T · 1	T 1	т. 1
2	rs16942	(A;G)	Very slightly increased breast cancer risk	Link	Link	Link
2	rs17228212	(C;T)	1.26x increased risk for heart disease		Link	
2	rs17435	(T;T)	1.4x increased risk for lupus		T : 1	
2	rs1799732	(-;C)	1.3x increased adenoma recurrence risk		Link	
2	rs2073963	(G;T)	Increased risk of baldness			
2	rs2143340	(C;T)	Increased risk of dyslexia and poor reading per			
2	rs2156921	(A;G)	1.29x increased risk for depression		T 1 1	
2	rs2201841	(T;T)	2.4x increased risk for Graves' disease	T . 1	Link	
2	rs2230201	(G;G)	>1.4x risk of lupus	Link		

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
2	rs2274223	(A;G)	1.5x increased risk for stomach and esophageal	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	rs2383206	(A;G)	1.4x increased risk for heart disease			
2	rs25487	(A;G)	2x higher risk for skin cancer; possibly other	Link	Link	
2	rs2736990	(C;C)	Increased risk of developing Parkinson's Diseas		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	rs3790565	(C;C)	Increased risk of developing primary biliary ci			
2	rs4242382	(A;A)	1.7x increased risk for prostate cancer		Link	
2	rs449647	(A;A)	Lower levels of ApoE			
2	rs4538475	(G;G)	Increased risk of developing Parkinson's Diseas		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	rs4825476	(G;G)	1.9x higher risk of suicidal thoughts when taki		Link	
2	rs493258	(A;G)	1.15x risk of Age Related Macular Degeneration			
2	rs4968451	(A;C)	1.61x increased risk for meningioma			
2	rs520354	(A;G)	Increased risk in men for biliary conditions			
2	rs6441286	(G;T)	1.54x chance of developing primary biliary cirr		Link	
2	rs6457617	(C;T)	2.3x risk of rheumatoid arthritis		Link	
2	rs6498169	(A;A)	>1.14x risk of multiple sclerosis		Link	
2	rs6532197	(G;G)	Increased risk of developing Parkinson's Diseas		Link	
2	rs663048	(G;T)	Significantly increased risk of developing lung	Link	Link	
2	rs6897932	(C;C)	1.5x increased risk for multiple sclerosis	Link	Link	Link
2	rs6908425	(C;C)	1.95x increased risk of developing Crohn's dise	LIIIK	Link	LIIIK
2	rs6997709	(G;G)	1.5x higher risk for hypertension		Lilik	
2	rs699	(C;C)	Increased risk of hypertension	Link	Link	Link
2	rs7190458	(A;G)	Slightly higher pancreatic cancer risk	Link	Lilik	LIIIK
2	rs738409	(C;G)	Increased liver fat: odds of alcoholic liver di	Link	Link	
2	rs744373	(C;T)	1.17x risk of Alzheimer's	LIIIK	Lilik	
2	rs763361	(C,T) $(T;T)$	Increased risk for multiple autoimmune diseases	Link	Link	
2	rs7774434	(C;C)	Increased risk of developing primary biliary ci	LIIIK	LIIIK	
2	rs7794745	(T;T)	Slightly increased risk for autism		Link	Link
2	rs7807268	(C;C)	1.4x risk for Crohn's disease		Link	LIIIK
$\frac{2}{2}$	rs7923837	(G;G)	3.2x risk for T2D		LIIIK	
2	rs7961152	(A;C)	1.2x higher risk for hypertension			
2	rs9303277	(T;T)	1.46x Increased risk of developing primary bili			
$\frac{2}{2}$	rs9543325	(C;C)	Slightly higher pancreatic cancer risk			
$\frac{2}{2.0}$	rs9642880	(C,C)	1.5x increased bladder cancer risk		Link	
1.7	rs1447295	(A;A)	1.5x increased bladder cancer risk 1.7x increased risk of prostate cancer		Link	
1.6	rs1447295 rs11523871	(A;A) (A;C)	1.6x increased breast cancer risk for women ove	Link	Link	
1.6	rs1537415	(C;G)	1.6x increased breast cancer risk for women ove 1.6x increased risk for periodontitis	LIIIK	Link	
1.6	rs2059693	(C;G) (T;T)	1.6x increased risk for testicular cancer		THIK	
1.6	rs2981745	(C;T)	1.6x increased risk for testicular cancer 1.6x increased risk for breast cancer in female			
1.6	rs356219	(G;G)	1.6x increased risk for Parkinson's disease			
1.6	rs3775948	(C;G)	Slightly higher risk for gout			
1.5	rs10260404	(C;G) (C;T)	1.20x risk of developing ALS		Link	
1.5	rs10757272	(C;T)	1.30x increased risk for Coronary artery diseas		THIK	
1.5	rs10757272		Slight (~1.2x) increase in endometriosis risk			
		(A;C)	- · · /		Link	
1.5	rs10883365	(A;G)	1.2x increased risk for developing Crohn's dise		Link	
1.5 1.5	rs11171739 rs12037606	(C;T)	1.34x risk of developing Type-1 diabetes 1.22x risk of developing Crohn's disease		LIIIK	
1.5	rs13149290	(A;G)	Slightly increased risk of developing prostate			
		(C;C)			Link	
1.5	rs1360517	(A;G)	Higher susceptibility for AIDS		Link	

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
1.5	rs140701	(A;G)	Increased risk for anxiety disorders			
1.5	rs16944	(A;G)	Minorly increased risk of mental illness and os		Link	
1.5	rs1801020	(C;T)	1.31x increased risk of heart disease	Link		Link
1.5	rs2280714	(A;G)	1.4x increased risk of SLE			
1.5	rs28694718	(A;G)	2x higher risk for schizophrenia			
1.5	rs309375	(T;T)	Larger mosquito bites			
1.5	rs356220	(T;T)	Increased risk of Parkinson's Disease			
1.5	rs3745516	(A;G)	Slightly increased risk of developing primary b			
1.5	rs3825776	(A;G)	1.3x increased risk for ALS		Link	
1.5	rs464049	(T;T)	Increased risk of schizophrenia in limited stud			
1.5	rs4656461	(A;G)	1.5x increased risk for open angle glaucoma			
1.5	rs4785763	(A;C)	1.5x higher risk for melanoma		Link	
1.5	rs4979462	(C;T)	Slightly increased risk of developing primary b			
1.5	rs699473	(C;T)	~1.5x increased brain tumor risk			
1.5	rs7850258	(G;G)	Slightly higher odds of developing primary hypo			
1.5	rs966221	(C;C)	1.5x increased stroke risk certain populations			
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	rs2046210	(C;T)	1.4x increased breast cancer risk		Link	
1.4	rs3131296	(G;G)	1.4x increased risk for schizophrenia		Link	
1.4	rs6010620	(G;G)	1.4x higher risk for glioma development; but th		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	rs2736100	(G;T)	1.3x higher risk for glioma development		Link	
1.3	rs34330	(C;T)	1.3x higher risk for endometrial cancer (in Chi			
1.3	rs501120	(A;G)	1.3x increased risk for heart disease		Link	
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	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	rs3740878	(A;A)	1.46x type II diabetes risk; common	Link		
1.2	rs4496877	(T;T)	For type-1 diabetics: 1.6x increased nephropath			
1.2	rs4795067	(A;G)	Slight increase in risk for psoriatic arthritis			
1.2	rs4977756	(A;G)	1.2x higher risk for glioma development		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	rs11650354	(C;T)	Possible risk for allergic asthma	Link		
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	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	rs4324715	(C;T)	1.5x increased testicular cancer risk for men			
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	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	
1.09	rs12050604	(A;C)	Very slightly increased risk for lung cancer			
1.05	rs2291834	(C;T)	Very slightly higher risk for myocardial infarc			
1	rs10761659	(A;G)	1.2x risk of Crohn's disease		Link	

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
1	rs1143674	(A;G)	1.3x increased autism risk	Link		
1	rs1804197	(A;C)	Increased risk of familial colorectal cancer an			Link
1	rs2228000	(T;T)	Statistically significant: but slight: increase	Link	Link	Link
1	rs3194051	(A;G)	1.12x risk of type-1 diabetes	Link	Link	Link
1	rs6932590	(C;T)	1.1x increased risk for schizophrenia		Link	
1	rs798766	(T;T)	Increased susceptibility urinary bladder cancer			
1	rs987525	(A;C)	2.5x increased risk for cleft lip		Link	
0.1	rs2070744	(C;C)	Increased prostate cancer risk		Link	Link
0.1	rs601338	(G;G)	Susceptible to Norovirus infections	Link	Link	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	rs6277	(C;C)	1.6x higher schizophrenia risk	Link	Link	Link

• Genosets (Multi-variant Phenotypes)

Magnitude	Identifier	Summary
4	gs144	Male
3	gs273	Lowest risk (13% of white women) of Atrial Fibr
2.5	gs157	More stimulated by coffee
2.5	gs281	Part of the 88% of the population claimed not t
2.2	gs280	Light hair color for europeans
2	gs101	Probably able to digest milk
2	gs136	Normal: but this haplotype in 8q24 increases ri
2	gs140	NAT2 slow metabolizer
2	gs154	NAT2 Slow metabolizer
2	gs173	CYP2D6*10
2	gs221	Autoimmune disorder risk in Europeans
2	gs246	APOE3/APOE3
1.5	gs247	Parkinson's Disease Risk
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).