PGP-UK Genomics Report for uk5E5B15

1 Summary

This is the genome report was produced using collaborative research tools, including SNPedia and GetEvidence. This section shows an overview of all the small variants which were found in the genome for this individual, when compared with a reference genome. These variants are summarised in Table 1 and the pie-charts in Figures 2, 3 and 4.

This report was generated automatically and is not clinically approved. It is provided for personal and research purposes only.

This document contains hyperlinks, shown in grey, that will take you to external websites where you can find more detailed explanations. Some of the technical terms are also explained in more detail in the Ensembl Glossary. We would welcome your feedback about this report, for example, if you would like more information about anything or if any of the links have become inactive. You can contact us on: pgp-uk@ucl.ac.uk.

This summary shows an overview of all the variants which were found in the genome for this individual. The "variants remaining after filtering" refers to any differences in the DNA identified when compared to the reference genome. Of these, the majority will have already been found in some other sequenced individual and put on a database (existing variants) while others have not yet been annotated (novel variants).

"Overlapped genes" refers to the number of times where a variant was found in a region of the genome containing a gene. The diagram in Figure 1 is a simplification of the usual gene structure. "Exon" refers to the part of the gene which goes on to form a protein, and variants in this part of the gene are more likely to cause changes in the shape of the protein. Upstream, downstream, intronic and intergenic variants are more likely to alter the regulation of that gene but will not change the protein itself.

A transcript for a protein-coding gene can include the exons, introns and other gene features that are transcribed and important for gene function but might not be translated into the final protein. Not all transcripts are for protein-coding genes, with many containing non-coding RNAs that can be overlapping other genes, in introns or in intergenic regions.

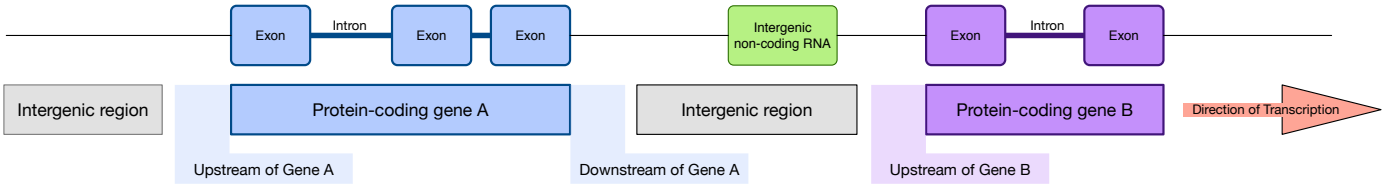

Figure 1: Diagram of gene structure indicating locations of potential variants

Feature	Count
Lines of input read	4946648
Variants filtered out	0
Novel / existing variants	492747 (10.0) / 4440447 (90.0)
Overlapped genes	56701
Overlapped transcripts	67480
Overlapped regulatory features	166164

Table 1: Variant calling summary

There are several different types of genomic variants. The most common change is when one single building block of the DNA (called a nucleotide) is changed, called a single nucleotide variants (SNV). Other variant types include insertions, where the DNA in the individual is longer than the reference sequence due to the insertion of one or more nucleotides; and deletions, where a few nucleotides are missing compared to the reference sequence.

Some of these changes will have no effect on the protein, while some changes may alter the protein function to varying degrees. The PolyPhen analysis software attempts to quantify the effect each mutation will have on the protein function. This ranges from "benign" where no change to the protein function is expected, to "probably damaging" where it is predicted that the mutation will affect protein function. It is nevertheless important to note that what is "damaging" for the protein is not necessarily damaging for the individual.

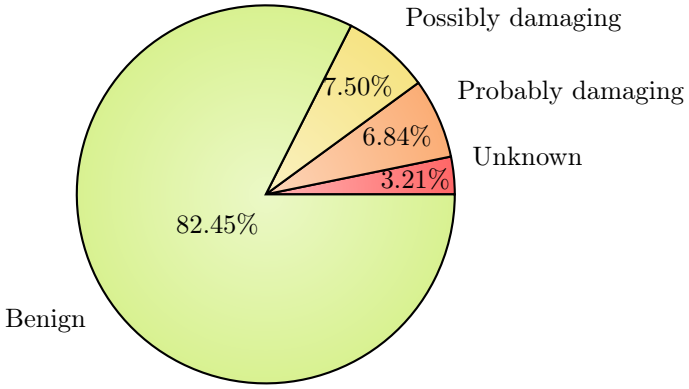

Figure 2: PolyPhen Summary

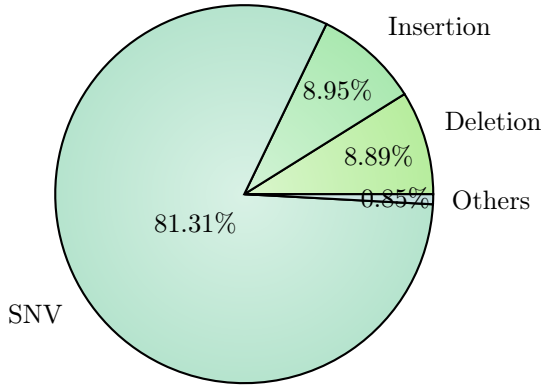

Figure 3: Variant Class

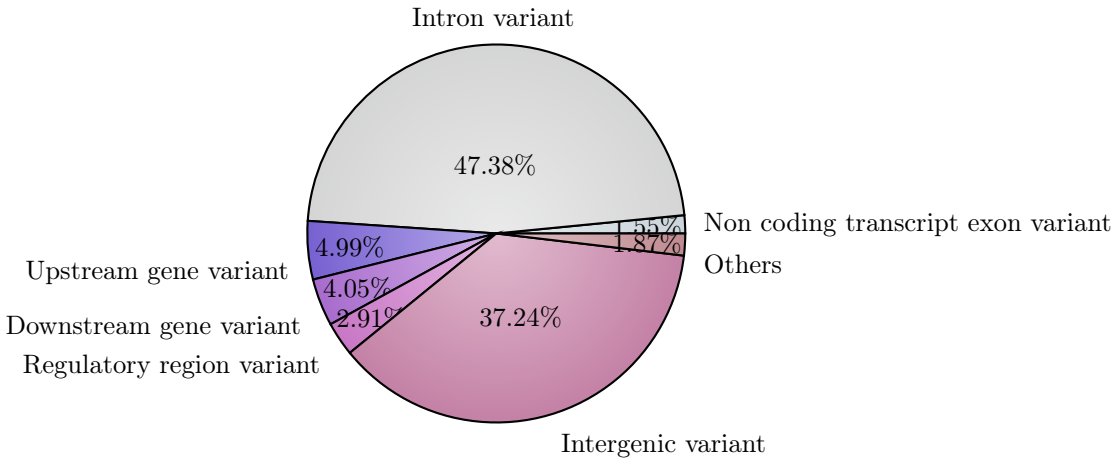

Figure 4: Consequence type

2 Ancestry

This plot shows the distribution of the genomes of different populations. Data from several studies which used whole genome sequencing was used to see the relationships between the genomes of the populations. It shows how closely related certain populations are genetically: Groups which cluster closely are more genetically similar than groups which are further apart. The black star symbol shows where this PGP-UK participant sits in relation to other populations, indicating their ancestry and their most closely related populations according to genetic sequence.

Based on the populations defined in the 1000 genomes project (1kGP), the ancestry composition for this individual is inferred to be 100.0 percent European [British in England and Scotland].

Please note that this analysis is limited by the populations available in the 1kGP data. If there are European subpopulations reported, and the ancestry of the participant does not correspond to any of the 1kGP populations, the closest 1kGP sampled subpopulation will be shown (even though it might be different from the participant's actual ancestry).

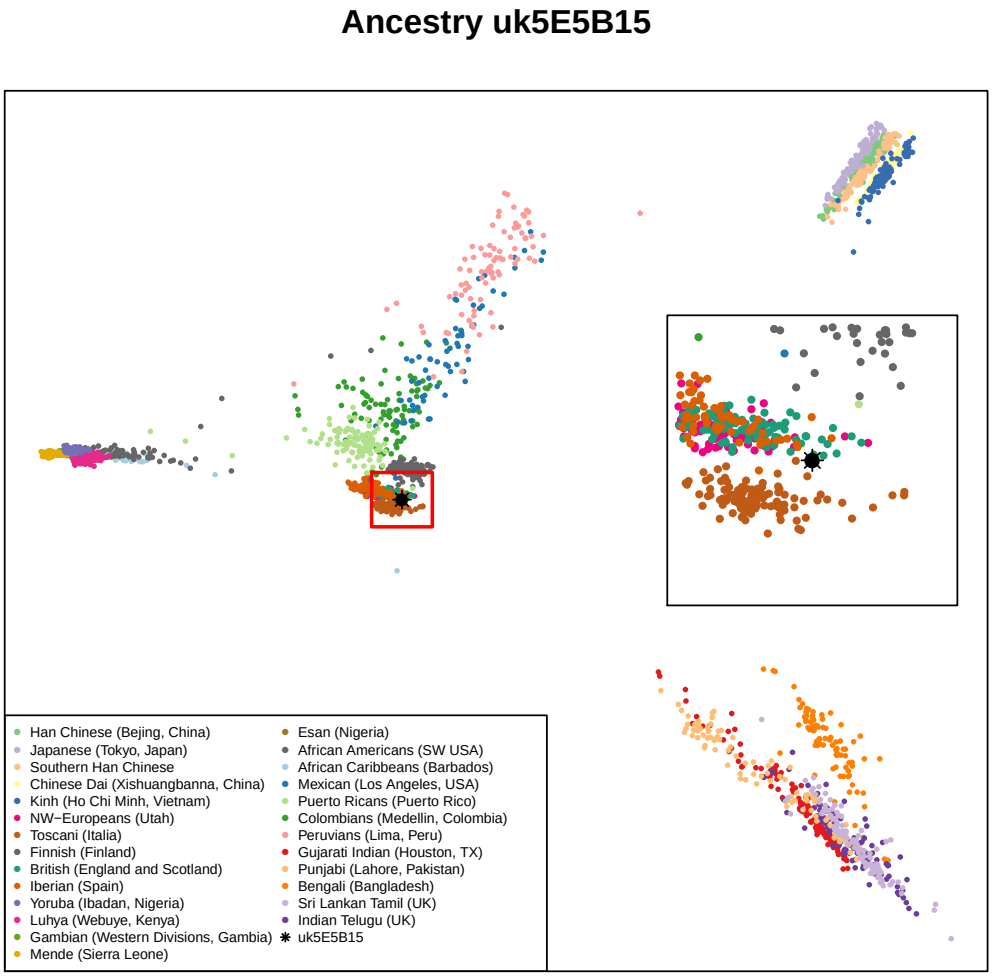

Figure 5: Ancestry Principal Component Analysis

3 Traits (based on SNPedia information)

Existing research has associated many variants with phenotypic traits, some of which can be perceived as beneficial while others appear to have a harmful effect. Some traits are complex and can be affected by several variants. It is likely that some of these would confer a higher risk while others a lower risk of trait manifestation. These can not be combined linearly to produce an actual risk of disease.

It is important to note that in most cases genomic data is probabilistic, not deterministic- i.e. having a genetic predisposition for a disease is not a diagnosis; rather, it shows an increased likelihood of developing that disease. Also, one person can have both potentially beneficial and harmful variants in the same gene, or associated with the same disease.

Some variants can also affect certain populations more, or will only affect a particular gender. For example, a variant for higher risk of endometriosis in the sequence of a male will not directly affect that person, but can be passed on to descendants.

While many traits are the result of a unique variant, many are the combination of several variants throughout the genome. In SNPedia, these are called genosets. These can integrate some of the information already present in the single variant tables, or be the combination of variants that have no phenotypic effect on their own, but contribute to a trait when together.

The variants in the following tables are sorted by magnitude. This is an subjective measure defined in SNPedia to highlight the perceived importance of the genotype described. At the moment this scale goes from 0 to 10. You can read more about it by visiting their explanatory webpage.

As our knowledge grows, the interpretation of the effect of certain variants might change. Clicking on the links in the genome report tables will take you to websites containing more information about each variant.

3.1 Possibly Beneficial Traits

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
3	rs7294919	(C;T)	Moderately enhanced hippocampal volume			
2.5	rs3764261	(T;T)	Associated with higher HDL cholesterol. HDL		Link	Link
2.4	rs9272346	(G;G)	0.08x risk type-1 diabetes		Link	
2.1	rs3775291	(A;G)	0.71x decreased risk for dry age related macula...	Link	Link	Link
2.1	rs547154	(A;C)	0.47x decreased risk for AMD			Link
2	rs1012053	(A;C)	0.625x reduced risk of Bipolar Disorder.		Link	
2	rs10468017	(C;T)	Associated with higher HDL cholesterol		Link	
2	rs11045585	(A;A)	24% chance (lower than average) of docetaxel-in...		Link	
2	rs1136410	(C;T)	0.80x reduced risk for glioblastoma	Link	Link	
2	rs1160312	(G;G)	Reduced risk of Baldness.		Link	
2	rs1544410	(G;G)	Decreased risk of low bone mineral density diso...		Link	
2	rs1799884	(G;G)	Mothers have typical Birth-Weight babies. Sligh...			
2	rs1864163	(G;G)	Associated with higher HDL cholesterol		Link	
2	rs2073963	(T;T)	Reduced risk of baldness			
2	rs2542052	(C;C)	Better odds of living to 100			
2	rs25487	(A;A)	0.7x lower risk for skin cancer	Link	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	rs3819331	(T;T)	Lower risk of autism	Link		
2	rs4149268	(G;G)	Associated with higher HDL cholesterol		Link	
2	rs6505162	(A;C)	0.58x decreased risk for esophageal cancer	Link		
2	rs6855911	(G;G)	Rare: but 0.62x decreased risk for gout		Link	
2	rs801114	(T;T)	0.78x decreased Basal Cell Carcinoma risk.		Link	
2	rs9642880	(G;G)	Slightly lower risk of Bladder Cancer.		Link	
2.0	rs3790844	(C;C)	Reduced risk (0.59x) of pancreatic cancer			
1.8	rs1128535	(A;G)	0.77x risk for Crohn's disease			
1.8	rs187238	(C;G)	Hypertension not a risk factor for sudden cardi...			

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
1.8	rs4714156	(C;C)	<0.61x risk for restless legs			
1.8	rs7101429	(A;G)	0.70x reduced risk for Alzheimer's risk			
1.6	rs1061170	(T;T)	Lower risk for AMD: generally longer live than ...	Link	Link	Link
1.6	rs3775948	(C;C)	Slightly lower risk for gout			
1.5	rs11136000	(C;T)	0.84x decreased risk for Alzheimer's disease		Link	
1.5	rs1165205	(A;A)	0.85x decreased gout risk		Link	
1.5	rs4149274	(C;C)	Associated with higher HDL (good) cholesterol.			
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	
1.2	rs4320932	(A;G)	0.87x decreased risk for ovarian cancer			
1.1	rs11172113	(C;T)	0.9x lower risk for migraines			
1.1	rs2293347	(G;G)	Among NSCLC patients: better Gefitinib response...	Link		Link
1.1	rs7568369	(G;T)	0.90x reduced risk of obesity			
1	rs182549	(C;T)	Can digest milk.			Link
1	rs2952768	(C;T)	Slightly less drug dependence: decreased effect...			Link
1	rs7850258	(A;G)	Typical odds of developing primary hypothyroidi...			
1	rs800292	(C;T)	1% decreased risk of macular degeneration	Link	Link	Link
0.5	rs36094464	(A;T)	Most likely benign: though reported years ago t...	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	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	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	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	rs5065	(A;A)	1.12x risk on diuretic; if hypertensive: better...	Link	Link	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	rs9394492	(C;C)	<0.76x risk for restless legs			

3.2 Possibly Harmful Traits

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
3	rs10897346	(C;C)	If depressed: 2.6x more likely to not respond t...			
3	rs13266634	(C;C)	Increased risk for type-2 diabetes	Link	Link	Link
3	rs2306402	(C;C)	1.18x increased risk for late-onset Alzheimer's...			
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
2.9	rs1061235	(A;T)	26% risk of bad reaction to anti-epileptic carb...			Link
2.7	rs10830963	(C;G)	Increased type-2 diabetes risk; higher gestatio...		Link	
2.6	rs110419	(A;A)	2.6x increased risk for neuroblastoma			
2.6	rs8034191	(C;C)	1.80x lung cancer risk; decreased response to a...		Link	
2.5	rs10490924	(G;T)	2.7x risk for age related macular degeneration	Link	Link	Link
2.5	rs11190870	(T;T)	Possibly even more increased risk of scoliosis			
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	rs2254958	(C;C)	1.61x increased risk for Alzheimer's			
2.5	rs2943634	(C;C)	Slightly higher risk of ischemic stroke		Link	
2.5	rs339331	(T;T)	Prostate cancer risk			
2.3	rs1859962	(G;G)	1.28x increased risk for prostate cancer		Link	
2.3	rs37973	(G;G)	Among asthmatics: 2.3x more likely to show less...			Link
2.2	rs2004640	(G;T)	1.4x increased risk for SLE		Link	Link
2.2	rs2231137	(G;G)	~1.5-3x increased risk for ischemic stroke	Link	Link	Link
2.2	rs944289	(T;T)	1.69x increased thyroid cancer risk		Link	
2.1	rs10427255	(C;C)	Highest odds of photic sneeze reflex			
2.1	rs1050152	(T;T)	2.1x increased risk of Crohn's disease	Link	Link	Link
2.1	rs1360780	(T;T)	1.3x increased risk for depression		Link	
2.1	rs17070145	(C;C)	Reduced memory abilities			Link
2.1	rs4149056	(C;T)	Reduced breakdown of some drugs; 5x increased m...	Link	Link	Link
2.1	rs4363657	(C;T)	4.5x increased myopathy risk for statin users		Link	
2.1	rs646776	(A;A)	1.2x risk of coronary artery disease		Link	
2	rs10086908	(C;T)	1.7x increased risk for prostate cancer			
2	rs10096097	(G;G)	Increased Anorexia Nervosa risk			
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	rs10513789	(G;T)	Increased risk of Parkinson's disease			
2	rs1051730	(C;T)	1.3x increased risk of lung cancer	Link	Link	Link
2	rs10811661	(C;T)	1.2x increased risk for type-2 diabetes		Link	
2	rs10984447	(A;G)	1.17x increased risk for multiple sclerosis		Link	
2	rs11983225	(T;T)	7x less likely to respond to certain antidepres...		Link	
2	rs12037606	(A;A)	1.52x risk of developing Crohn's disease			
2	rs1219648	(A;G)	1.20x risk for breast cancer		Link	
2	rs12696304	(G;G)	Prone to aging faster: at least in European pop...			
2	rs13254738	(C;C)	1.18x prostate cancer risk		Link	
2	rs1333048	(A;C)	1.3x increased coronary artery disease risk			
2	rs1537415	(G;G)	2x increased risk for periodontitis		Link	
2	rs16942	(A;G)	Very slightly increased breast cancer risk	Link	Link	Link
2	rs17001266	(-;C)	1.58x increased risk for schizophrenia in males...			
2	rs17228212	(C;T)	1.26x increased risk for heart disease		Link	
2	rs1734791	(A;A)	1.4x increased risk for lupus			
2	rs17576	(A;G)	Higher risk for MI and lung cancer: and COPD in...	Link	Link	
2	rs1799732	(-;C)	1.3x increased adenoma recurrence risk		Link	
2	rs1975197	(T;T)	>1.3x increased risk of developing restless leg...		Link	
2	rs1994090	(G;G)	Increased risk of developing Parkinson's Diseas...		Link	
2	rs2143340	(C;T)	Increased risk of dyslexia and poor reading per...			

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
2	rs2156921	(A;G)	1.29x increased risk for depression			
2	rs2201841	(T;T)	2.4x increased risk for Graves' disease		Link	
2	rs2230201	(G;G)	>1.4x risk of lupus	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	rs2383206	(A;G)	1.4x increased risk for heart disease			
2	rs2383207	(A;G)	Increased risk for heart disease			
2	rs241448	(C;T)	1.51x increased risk for Alzheimer's	Link		Link
2	rs2420946	(C;T)	1.20x risk for breast cancer			
2	rs2736100	(T;T)	Higher risk of Interstitial lung disease: and t...		Link	
2	rs3212227	(A;C)	Significantly increased risk of developing cerv...			
2	rs326	(A;A)	Lower HDL cholesterol		Link	Link
2	rs351855	(C;T)	1.2x increased risk for prostate cancer	Link	Link	Link
2	rs358806	(C;C)	1.78x increased risk of developing Type-2 diabe...		Link	
2	rs3793784	(C;G)	1.5x risk for ARMD		Link	Link
2	rs4027132	(A;A)	1.51x increased risk of developing bipolar diso...			
2	rs4148739	(A;A)	7x less likely to respond to certain antidepres...		Link	
2	rs4402960	(G;T)	1.2x increased risk for type-2 diabetes: ~1x ri...		Link	Link
2	rs4420638	(A;G)	~3x increased Alzheimer's risk; 1.4x increased ...		Link	Link
2	rs4792311	(A;A)	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	rs4961	(G;T)	1.8x increased risk for high blood pressure	Link	Link	Link
2	rs4994	(C;C)	2x higher risk in certain women for cardiac eve...	Link	Link	Link
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	rs6896702	(T;T)	Increased risk of developing Parkinson's Diseas...			
2	rs6897932	(C;C)	1.08x increased risk for multiple sclerosis	Link	Link	Link
2	rs6997709	(G;T)	1.2x higher risk for hypertension			
2	rs7190458	(A;G)	Slightly higher pancreatic cancer risk	Link		
2	rs7250872	(T;T)	Increased risk of developing bipolar disorder	Link	Link	
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	rs7961152	(A;A)	1.5x higher risk for hypertension			
2	rs854560	(A;T)	Higher risk for heart disease: diabetic retinop...	Link	Link	Link
2	rs9525638	(T;T)	Weaker bones			
2	rs9543325	(C;C)	1.37x Slightly higher pancreatic cancer risk			
2	rs965513	(A;G)	1.77x increased thyroid cancer risk		Link	
2.0	rs1434536	(A;A)	1.94x increased breast cancer risk			
1.8	rs2278206	(T;T)	1.16x increased risk for asthma	Link	Link	
1.8	rs4474514	(A;G)	3x increased testicular cancer risk for men		Link	
1.8	rs6700125	(C;T)	1.2x increased risk for ALS			
1.8	rs733618	(A;G)	1.87x risk for myasthenia gravis			
1.7	rs2024513	(A;A)	1.7x higher risk for schizophrenia (among Han C...			
1.7	rs8055236	(G;T)	1.9x risk for heart disease		Link	
1.6	rs3764880	(A;A)	1.2 - 1.8x increased tuberculosis risk	Link	Link	
1.6	rs4959039	(G;G)	1.6x higher risk for multiple sclerosis			
1.5	rs10492519	(A;G)	Slightly increased risk of developing prostate ...			
1.5	rs10757272	(C;T)	1.30x increased risk for Coronary artery diseas...			
1.5	rs10859871	(A;C)	Slight (~1.2x) increase in endometriosis risk			

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
1.5	rs10883365	(A;G)	1.2x increased risk for developing Crohn's dise...		Link	
1.5	rs11171739	(C;T)	1.34x risk of developing Type-1 diabetes		Link	
1.5	rs1169300	(A;G)	~1.5x increased lung cancer risk			
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	rs13149290	(C;T)	Slightly increased risk of developing prostate ...			
1.5	rs1375144	(C;C)	1.59x 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	rs16944	(A;G)	Minorly increased risk of mental illness and os...		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	rs2177369	(C;C)	1.5x increased risk for Alzheimer's disease			
1.5	rs2240340	(A;G)	Slightly increased (1.5x) risk for RA	Link		
1.5	rs2241880	(C;T)	1.4x increased risk for Crohn's disease in Cauc...	Link	Link	Link
1.5	rs2272127	(C;C)	Associated with herpes and schizophrenia			
1.5	rs2280714	(A;A)	1.4x increased risk of SLE			
1.5	rs2286812	(C;T)	~2x higher risk for Fuchs' dystrophy: a corneal...			
1.5	rs2464196	(C;T)	~1.5x increased lung cancer risk	Link	Link	Link
1.5	rs27388	(A;G)	Slightly increased risk of developing schizophr...			
1.5	rs2881766	(T;T)	Slightly increased risk for pregnancy-induced h...			
1.5	rs3087243	(G;G)	Increased risk for autoimmune diseases		Link	
1.5	rs309375	(T;T)	Larger mosquito bites			
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;T)	~1.2x increased risk for several types of cance...		Link	
1.5	rs4464148	(C;T)	1.10x increased risk for colorectal cancer			
1.5	rs4585	(T;T)	Slightly poorer (0.75x) response to metformin i...			
1.5	rs464049	(C;T)	Increased risk of schizophrenia in limited stud...			
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	rs5746059	(A;A)	Slightly higher fat mass			
1.5	rs619203	(C;G)	Increases susceptibility to Myocardial Infarcti...	Link	Link	
1.5	rs642961	(A;G)	1.68x increased risk of cleft lip		Link	
1.5	rs6601764	(C;T)	1.16x increased risk of developing Crohn's dise...		Link	
1.5	rs7341475	(G;G)	1.58x increased schizophrenia risk for women		Link	
1.5	rs763035	(C;T)	1.2x increased risk for rosacea			
1.5	rs7774434	(C;T)	Slightly increased risk of developing primary b...			
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	rs9652490	(A;G)	Slightly increased risk of developing Parkinson...		Link	
1.4	rs10865331	(A;A)	1.4x higher risk for ankylosing spondylitis			
1.4	rs1801157	(A;G)	1.4x higher risk for breast cancer			
1.4	rs2046210	(C;T)	1.4x increased breast cancer risk		Link	Link
1.4	rs2665390	(C;C)	1.4x increased risk for ovarian cancer			
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	
1.34	rs17465637	(C;C)	1.34x higher risk for myocardial infarction	Link	Link	
1.3	rs1260326	(C;T)	Slightly higher risk for gout	Link	Link	Link
1.3	rs16847548	(C;T)	1.3x increased risk for sudden cardiac death in...			
1.3	rs1746048	(C;C)	1.03 increased risk for coronary heart disease		Link	
1.3	rs2059693	(C;T)	1.3x increased risk for testicular cancer			
1.3	rs34330	(C;T)	1.3x higher risk for endometrial cancer (in Chi...			
1.3	rs4295627	(G;T)	1.36x higher risk for glioma development		Link	

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
1.25	rs748404	(T;T)	Slightly increased risk (1.25) for lung cancer...		Link	
1.2	rs12050604	(A;A)	Slightly increased risk for lung cancer			
1.2	rs143383	(C;T)	1.1x increased risk for osteoarthritis		Link	Link
1.2	rs1800693	(A;G)	Slight (1.2x) increase in risk for multiple scl...	Link	Link	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	rs2814707	(A;G)	1.2x increased risk for ALS		Link	
1.2	rs3849942	(A;G)	1.2x increased risk for ALS		Link	
1.2	rs4324715	(C;C)	>1.5x increased testicular cancer risk for men			
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	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	rs3802842	(A;C)	1.17x increased risk of colorectal cancer		Link	
1.1	rs11037909	(C;T)	1.27x type II diabetes risk	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	rs2295190	(G;T)	Slightly increased risk for ovarian cancer in w...	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	
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		Link
1.1	rs3818361	(C;T)	1.15x increased risk for late-onset Alzheimer's...			
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	rs925391	(C;C)	More likely to go bald; common			
1.07	rs2291834	(C;C)	Very slightly higher risk for myocardial infarc...			
1	rs10504861	(G;G)	Major allele: normal risk of migraine			
1	rs10761659	(A;G)	1.2x risk of Crohn's disease		Link	
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	rs6932590	(C;T)	1.1x increased risk for schizophrenia		Link	
0.1	rs601338	(A;G)	Susceptible to Norovirus infections	Link	Link	Link
0	rs1004819	(C;C)	1.5x risk of Crohn's disease		Link	
0	rs1061646	(C;C)	1.16x increased risk for breast cancer	Link		Link
0	rs1128503	(T;T)	Likely to require more methadone during heroin ...	Link	Link	Link
0	rs4293393	(T;T)	1.25x Increased Risk of CKD for T allele in ...			
0	rs440446	(G;G)	Increased risk in men for biliary conditions	Link		
0	rs6277	(C;C)	1.6x higher schizophrenia risk	Link	Link	Link
0	rs7787082	(G;G)	7x less likely to respond to certain antidepres...		Link	

3.3 Genosets (Multi-variant Phenotypes)

Magnitude	Identifier	Summary
2.5	gs155	CYP3A5 non-expressor
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	gs101	Probably able to digest milk
2	gs156	NAT2 Rapid metabolizer.
2	gs181	CYP2D6*2
2	gs188	One copy of APOE4 is possible: but not certain
2	gs249	Parkinson's Disease Risk
1.5	gs220	HLA-B*1502?
1.2	gs184	Able to taste bitterness.
1	gs163	CYP2D6*2A
0	gs158	CYP1A2 normal metabolizer

4 Raw Data

The raw data used to create this report has been assigned the identifier ERS1176636 in the European Nucleotide Archive (ENA) hosted at the European Bioinformatics Institute (EBI).

These data will not be accessible unless the report is approved. This will happen by default one month after the report is issued, or if the report is approved for immediate release within the one month period. Participants can also withdraw from the study at any time in which case the report and the data will not be released and will be deleted.

If the data has already been released, it can be accessed at: http://www.ebi.ac.uk/ena/data/view/ERS1176636

5 Report Metadata

Resource	Version	Website
Genome	GRCh38	Link
BWA	0.7.12	Link
SAMtools	1.3	Link
GATK	3.4-46	Link
PLINK	v1.90b3.35	Link
VEP	88	Link
SNPedia	30-Jul-2017	Link
ExAC	v0.3.1	Link
GetEvidence	16-Dec-2016	Link
ClinVar	16-Dec-2016	Link

Table 5: Analysis Pipeline Versions

Report generated on August 2, 2017.