

PGP-UK Genomics Report for uk0D2BB4

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	4929539
Variants filtered out	0
Novel / existing variants	473666 (9.6) / 4444107 (90.4)
Overlapped genes	56631
Overlapped transcripts	67393
Overlapped regulatory features	165808

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.

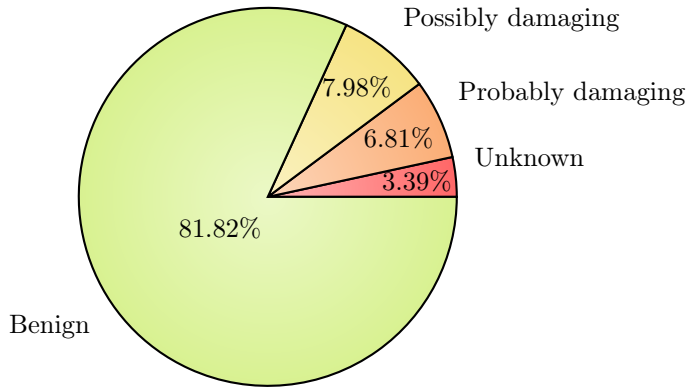


Figure 2: PolyPhen Summary

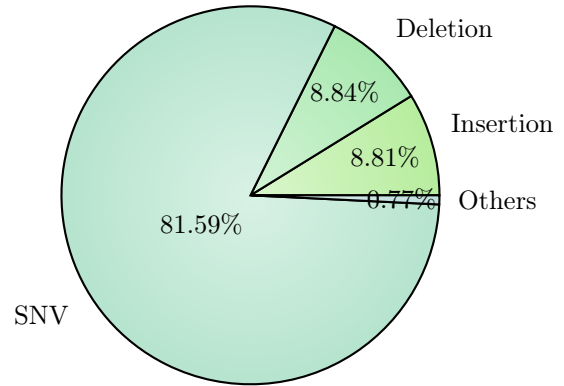


Figure 3: Variant Class

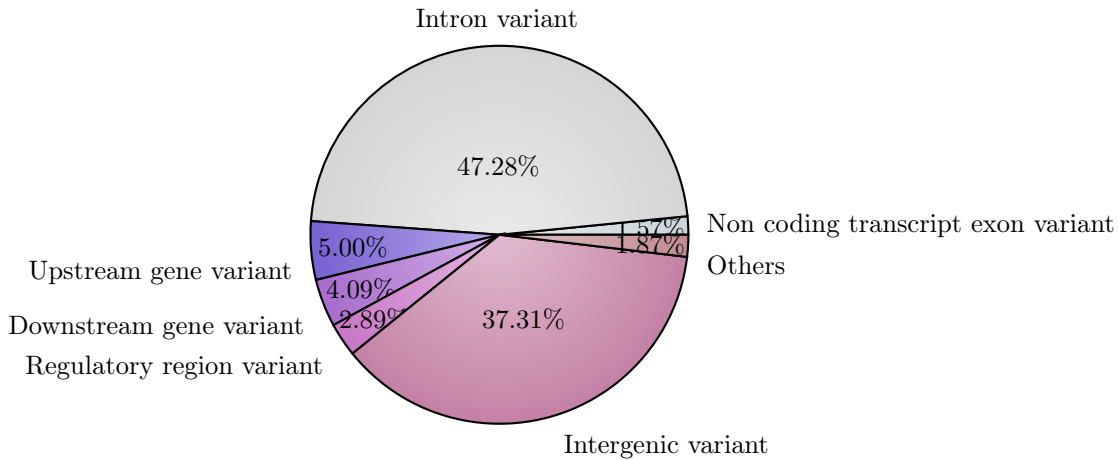


Figure 4: Consequence type

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).

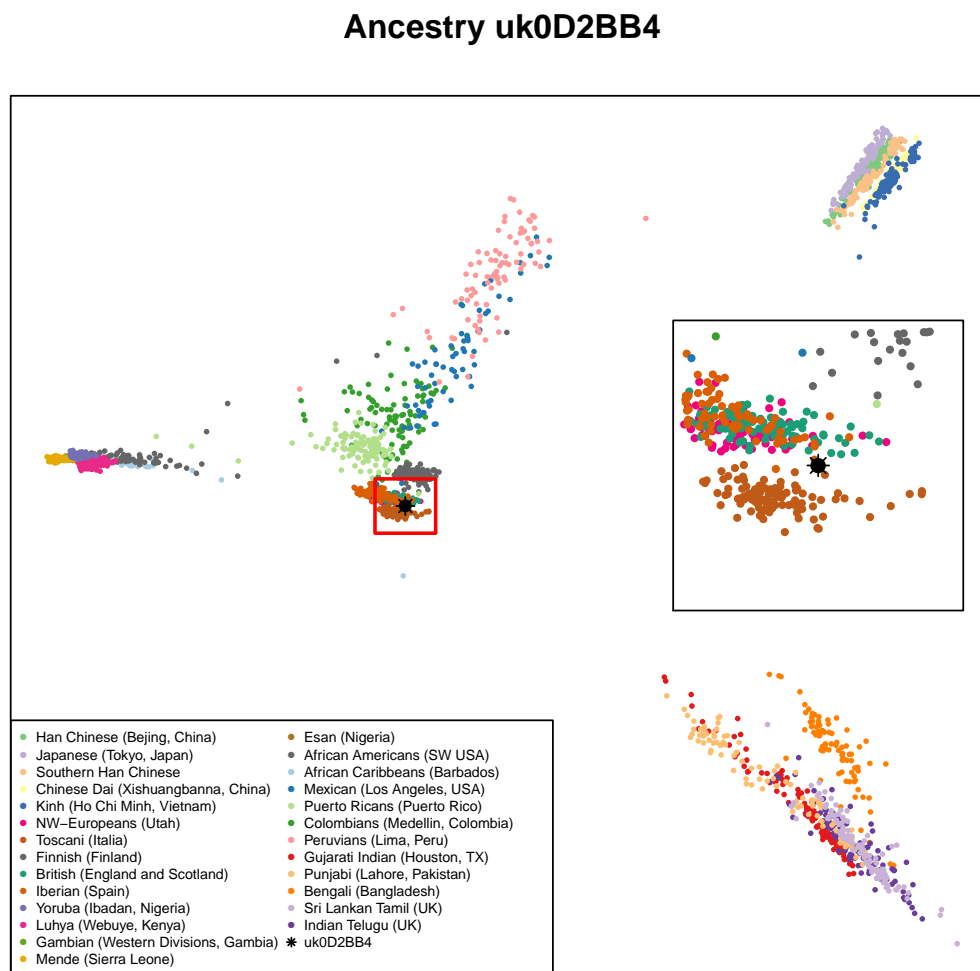


Figure 5: Ancestry Principal Component Analysis

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
2.4	rs2802288	(A;A)	Longer lifespan			
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	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	rs2073963	(T;T)	Reduced risk of baldness			
2	rs2241423	(A;G)	0.79 decreased risk for obesity			
2	rs2542052	(C;C)	Better odds of living to 100			
2	rs261332	(A;A)	Associated with higher HDL cholesterol			
2	rs3218536	(A;G)	Lower risk for breast: ovarian cancer	Link	Link	
2	rs3736309	(G;G)	0.44x decreased risk for chronic obstructive pu...			
2	rs3750817	(C;T)	0.78x reduced risk for breast cancer			
2	rs3819331	(T;T)	Lower risk of autism	Link		
2	rs4073582	(A;A)	Lower risk for gout	Link		
2	rs6505162	(A;C)	0.58x decreased risk for esophageal cancer	Link		
2	rs6807362	(G;G)	Decreased autism risk	Link	Link	
2	rs6855911	(A;G)	0.62x decreased risk for gout		Link	
2	rs7216389	(C;C)	0.69x lower risk of Childhood Asthma.		Link	
2	rs763110	(C;T)	~0.80x reduced cancer risk			Link
2	rs7776725	(T;T)	Stronger bones		Link	
2	rs8070723	(A;G)	0.18x reduced risk of developing progressive su...			
2	rs9642880	(G;G)	Slightly lower risk of Bladder Cancer.		Link	
1.8	rs1128535	(A;G)	0.77x risk for Crohn's disease			
1.8	rs1800588	(T;T)	Higher HDL-C levels	Link	Link	
1.8	rs187238	(C;G)	Hypertension not a risk factor for sudden cardi...			

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
1.6	rs3775948	(C;C)	Slightly lower risk for gout			
1.5	rs1026732	(A;G)	0.70x risk for restless legs		Link	
1.5	rs11212617	(A;C)	Somewhat increased likelihood of treatment succ...			Link
1.5	rs11465804	(G;T)	0.68x lower risk for spondylitis	Link	Link	
1.5	rs11635424	(A;G)	0.70x risk for restless legs		Link	
1.5	rs12593813	(A;G)	0.71x risk for restless legs		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	rs4489954	(G;T)	0.69x risk risk of developing restless legs syn...		Link	
1.5	rs4939883	(C;C)	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...			
1.4	rs1165205	(A;T)	0.85x decreased gout risk		Link	
1.25	rs10088218	(A;G)	0.76x decreased risk for ovarian cancer			
1.2	rs11246226	(A;C)	Decreased risk of schizophrenia in limited stud...		Link	
1.2	rs9306160	(C;T)	0.75x (reduced) risk for metastasis in LN-/ER+ ...	Link	Link	
1.1	rs10166942	(C;T)	0.85x lower risk for migraines			
1.1	rs11172113	(C;T)	0.9x lower risk for migraines			
1.1	rs4988235	(T;T)	Can digest milk			Link
1	rs182549	(T;T)	Can digest milk.			Link
1	rs2351299	(G;T)	Possible reduced risk of Autism			
1	rs7850258	(A;A)	Slightly lower odds of developing primary hypot...			
0	rs1047781	(A;A)	ABH blood group "Secretor" status if Japanese	Link	Link	Link
0	rs1126809	(A;G)	Slight increase in skin cancer risk	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	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	rs312481	(C;C)	Better response to certain calcium channel bloc...			
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	rs7305115	(A;A)	Individuals showed a significantly lower risk o...	Link	Link	
0	rs74315403	(G;G)	PrP codon 178 (D) - non pathogenic variant			Link
0	rs9951307	(A;G)	0.10 decreased risk for brain edema after a str...			

3.2 Possibly Harmful Traits

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
4	rs1333049	(C;C)	1.9x increased risk for coronary artery disease...		Link	
3.1	rs10830963	(G;G)	Increased type-2 diabetes risk; higher gestatio...		Link	
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	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
3	rs6920220	(A;G)	1.2x risk Rheumatoid Arthritis		Link	
3	rs7754840	(C;C)	1.3x increased risk for type-2 diabetes		Link	
2.8	rs3780374	(A;A)	Substantially increased odds of developing V617...			
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	rs2241880	(C;C)	2x-3x increased risk for Crohn's disease in Cau...	Link	Link	Link
2.5	rs3738919	(C;C)	1.94x risk of developing rheumatoid arthritis			
2.5	rs613872	(G;T)	~5 fold higher risk for Fuchs' dystrophy: a cor...			
2.5	rs663048	(T;T)	3x increased risk of developing lung cancer	Link	Link	
2.5	rs664143	(C;T)	Higher risk for number of cancers			
2.5	rs8034191	(C;T)	1.27x lung cancer risk		Link	
2.5	rs891512	(A;G)	Higher blood pressure than G;G	Link		
2.3	rs7966230	(C;G)	Slightly lower levels of plasma VWF			
2.2	rs2231137	(G;G)	~1.5-3x increased risk for ischemic stroke	Link	Link	Link
2.1	rs10811661	(T;T)	1.2x increased risk for type-2 diabetes		Link	
2.1	rs2383207	(G;G)	Increased risk for heart disease			
2.1	rs4402960	(T;T)	1.2x increased risk for type-2 diabetes: 1.5x r...		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; ...			Link
2.1	rs4968451	(C;C)	2.33x increased risk for meningioma			
2.1	rs6457617	(T;T)	5.2x risk of rheumatoid arthritis		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	rs10455872	(A;G)	1:51x increased Coronary Heart disease risk			Link
2	rs1045642	(C;T)	Slower metaboliser for some drugs	Link	Link	Link
2	rs1051730	(C;T)	1.3x increased risk of lung cancer	Link	Link	Link
2	rs10757272	(T;T)	1.54x increased risk for Coronary artery diseas...			
2	rs10871777	(A;G)	Adults likely to be 0.22 BMI units higher			
2	rs10883365	(G;G)	1.62x increased risk for developing Crohn's dis...		Link	
2	rs10984447	(A;G)	1.17x increased risk for multiple sclerosis		Link	
2	rs11190870	(C;T)	Possibly increased risk of scoliosis			
2	rs1160312	(A;A)	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	rs12431733	(T;T)	Increased risk of developing Parkinson's Diseas...		Link	
2	rs1265181	(C;G)	Increased risk for psoriasis		Link	
2	rs1360780	(C;T)	1.3x increased risk for depression		Link	
2	rs1585215	(A;G)	2x increased risk for Hodgkin lymphoma			
2	rs16942	(G;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	

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
2	rs1734791	(A;A)	1.4x increased risk for lupus			
2	rs17696736	(A;G)	1.34x risk of type-1 diabetes		Link	
2	rs17782313	(C;T)	Adults likely to be 0.22 BMI units higher		Link	Link
2	rs1800896	(A;G)	1.6x increased prostate cancer risk			
2	rs1867277	(A;A)	2x increased risk for thyroid cancer			
2	rs2056116	(G;G)	1.41x risk for breast cancer			
2	rs2143340	(C;T)	Increased risk of dyslexia and poor reading per...			
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	rs2274223	(A;G)	1.5x increased risk for stomach and esophageal ...	Link	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	rs241448	(C;T)	1.51x increased risk for Alzheimer's	Link		Link
2	rs2420946	(C;T)	1.20x risk for breast cancer			
2	rs25487	(A;G)	2x higher risk for skin cancer; possibly other ...	Link	Link	Link
2	rs2707466	(G;G)	Weaker bones	Link	Link	
2	rs2736100	(T;T)	Higher risk of Interstitial lung disease: and t...		Link	
2	rs2908004	(C;C)	Weaker bones	Link	Link	
2	rs3025039	(C;T)	2.6x increased risk for ARMD in a Taiwanese pop...			
2	rs326	(A;A)	Lower HDL cholesterol		Link	Link
2	rs3746444	(C;T)	~1.2x increased risk for cancer	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	rs4633	(C;T)	Higher risk for endometrial cancer	Link	Link	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	rs5174	(A;G)	1.3x increased risk for heart disease	Link	Link	Link
2	rs520354	(A;G)	Increased risk in men for biliary conditions			
2	rs5759167	(T;T)	Higher prostate cancer risk		Link	
2	rs629242	(C;T)	Somewhat higher risk for prostate cancer			
2	rs6441286	(G;T)	1.54x chance of developing primary biliary cirr...		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	rs6908425	(C;C)	1.95x increased risk of developing Crohn's dise...		Link	
2	rs6997709	(G;T)	1.2x higher risk for hypertension			
2	rs7190458	(A;G)	Slightly higher pancreatic cancer risk	Link		
2	rs744373	(C;T)	1.17x risk of Alzheimer's			
2	rs7923837	(G;G)	3.2x risk for T2D			
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	rs854560	(A;T)	Higher risk for heart disease: diabetic retinop...	Link	Link	Link
2	rs9303277	(T;T)	1.46x Increased risk of developing primary bili...			
2	rs9543325	(C;C)	1.37x Slightly higher pancreatic cancer risk			
2	rs965513	(A;A)	3.1x increased thyroid cancer risk		Link	
2.0	rs1434536	(A;A)	1.94x increased breast cancer risk			
2.0	rs2156921	(G;G)	1.29x increased risk for depression			
1.8	rs1136287	(C;T)	1.5x increased risk of wet ARMD in a Taiwanese ...	Link	Link	
1.8	rs37973	(A;G)	Among asthmatics: 1.5x more likely to show less...			Link
1.8	rs6700125	(C;T)	1.2x increased risk for ALS			
1.6	rs33980500	(C;T)	1.6x increase in risk for psoriatic arthritis	Link	Link	Link

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
1.6	rs3764880	(A;A)	1.2 - 1.8x increased tuberculosis risk	Link	Link	
1.5	rs10464059	(A;G)	Slightly increased risk of developing Parkinson...			
1.5	rs10492519	(A;G)	Slightly increased risk of developing prostate ...			
1.5	rs10859871	(A;C)	Slight (~1.2x) increase in endometriosis risk			
1.5	rs10895068	(A;G)	2.5x increased odds of breast cancer among horm...			
1.5	rs10980705	(C;T)	2.3x increased risk for knee osteoarthritis			
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	rs12469063	(A;G)	Slightly increased risk of developing restless ...			
1.5	rs13149290	(C;T)	Slightly increased risk of developing prostate ...			
1.5	rs13181	(G;T)	1.12x increased risk for cutaneous melanoma	Link	Link	Link
1.5	rs13376333	(C;T)	1.5x higher risk of atrial fibrillation		Link	
1.5	rs140701	(A;G)	Increased risk for anxiety disorders			
1.5	rs1571801	(A;A)	>1.36x risk for prostate cancer			
1.5	rs16944	(A;G)	Minorly increased risk of mental illness and os...		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	rs199533	(C;T)	Slightly increased risk of developing Parkinson...	Link		
1.5	rs2007153	(G;G)	Increased risk of schizophrenia in limited stud...			
1.5	rs2076295	(G;G)	Slightly increased risk for pulmonary fibrosis ...			
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	rs2464196	(C;T)	~1.5x increased lung cancer risk	Link	Link	Link
1.5	rs2736990	(C;T)	Slightly increased risk of developing Parkinson...		Link	
1.5	rs27388	(A;G)	Slightly increased risk of developing schizophr...			
1.5	rs2881766	(T;T)	Slightly increased risk for pregnancy-induced h...			
1.5	rs3087243	(G;G)	Increased risk for autoimmune diseases		Link	
1.5	rs3212227	(A;A)	1.43x increased risk of developing psoriasis an...			
1.5	rs358806	(A;C)	0.86x increased risk of developing Type-2 diabe...		Link	
1.5	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	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	rs464049	(C;T)	Increased risk of schizophrenia in limited stud...			
1.5	rs5219	(C;T)	1.3x increased risk for type-2 diabetes	Link	Link	Link
1.5	rs5746059	(A;A)	Slightly higher fat mass			
1.5	rs6498169	(A;G)	1.14x risk of multiple sclerosis		Link	
1.5	rs6601764	(C;T)	1.16x increased risk of developing Crohn's dise...		Link	
1.5	rs6710341	(A;G)	Slightly increased risk of developing restless ...			
1.5	rs699473	(C;T)	~1.5x increased brain tumor risk			
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	rs763035	(C;T)	1.2x increased risk for rosacea			
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	rs995030	(G;G)	Non-protective against testicular cancer		Link	
1.4	rs10865331	(A;A)	1.4x higher risk for ankylosing spondylitis			
1.4	rs1126497	(C;T)	1.4x increased risk for breast cancer	Link	Link	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	rs3184504	(C;T)	Slightly increased risk for celiac disease	Link	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	rs1746048	(C;C)	1.03 increased risk for coronary heart disease		Link	
1.3	rs2059693	(C;T)	1.3x increased risk for testicular cancer			

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
1.3	rs2295490	(A;G)	1.32x increased risk of early-onset type-2 diab...	Link	Link	
1.3	rs356219	(A;G)	1.3x increased risk for Parkinson's disease			
1.2	rs11037909	(T;T)	1.47x type II diabetes risk	Link		
1.2	rs11842874	(A;G)	+17% increased risk for osteoarthritis			
1.2	rs35677470	(A;G)	2x higher risk for scleroderma	Link	Link	
1.2	rs3740878	(A;A)	1.46x type II diabetes risk; common	Link		Link
1.2	rs393152	(A;G)	Slight increased risk of both PD and AD	Link	Link	
1.2	rs4324715	(C;C)	>1.5x increased testicular cancer risk for men			
1.2	rs449647	(A;T)	Possibly lower levels of ApoE			
1.2	rs4795067	(A;G)	Slight increase in risk for psoriatic arthritis...			
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	
1.17	rs3802842	(A;C)	1.17x increased risk of colorectal cancer		Link	
1.15	rs748404	(C;T)	Very slightly increased risk (1.15) for lung ca...		Link	
1.1	rs11110912	(C;C)	1.3x high blood pressure risk			
1.1	rs11650494	(A;G)	Slightly higher prostate cancer risk			
1.1	rs13387042	(A;G)	1.12x increased risk for breast cancer		Link	
1.1	rs1344706	(G;T)	1.1x increased risk for schizophrenia		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	rs2828520	(G;G)	1.35x major depressive disorder risk			
1.1	rs34516635	(G;G)	Less longevity for Ashkenazi Jewish women.	Link		Link
1.1	rs3818361	(C;T)	1.15x increased risk for late-onset Alzheimer's...			
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	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;A)	Higher risk of multiple sclerosis			
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	rs1004819	(C;C)	1.5x risk of Crohn's disease		Link	
0	rs10239794	(T;T)	>1.3x risk for ALS			
0	rs1495965	(A;A)	1.2x higher risk for spondylitis			
0	rs3761418	(A;A)	1.3x increased risk for depression			
0	rs3813929	(C;C)	Possible weight gain if taking olanzapine		Link	Link
0	rs4293393	(T;T)	1.25x Increased Risk of CKD for T allele in ...			
0	rs4795400	(T;T)	If 4 years old or younger: ~2.5x increased asth...		Link	
0	rs7787082	(G;G)	7x less likely to respond to certain antidepres...		Link	
0	rs855791	(T;T)	0.2 g/dL lower hemoglobin on average	Link	Link	Link

3.3 Genosets (Multi-variant Phenotypes)

Magnitude	Identifier	Summary
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 ...
2.4	gs297	Lower heart attack risk than average
2	gs101	Probably able to digest milk
2	gs104	Restless legs syndrome risk
2	gs154	NAT2 Slow metabolizer
2	gs173	CYP2D6*10
2	gs246	APOE3/APOE3
1.5	gs185	The beta blocker metoprolol is effective with 1...
1.5	gs186	HLA-B*5801 heterozygosity is possible: unfortun...
1.5	gs247	Parkinson's Disease Risk
1.2	gs184	Able to taste bitterness.
1	gs182	CYP2D6*39
0	gs158	CYP1A2 normal metabolizer

4 Raw Data

The raw data used to create this report has been assigned the identifier ERS1176606 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/ERS1176606>

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.