

# PGP-UK Genomics Report for uk2ABD6B

## 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](mailto: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	4950605
Variants filtered out	0
Novel / existing variants	515102 (10.4) / 4423055 (89.6)
Overlapped genes	56718
Overlapped transcripts	67571
Overlapped regulatory features	166341

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.

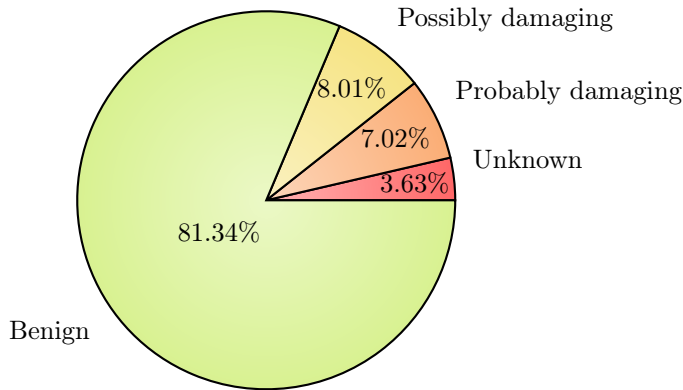


Figure 2: PolyPhen Summary

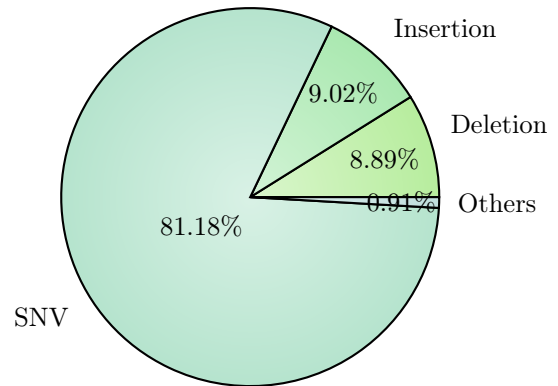


Figure 3: Variant Class

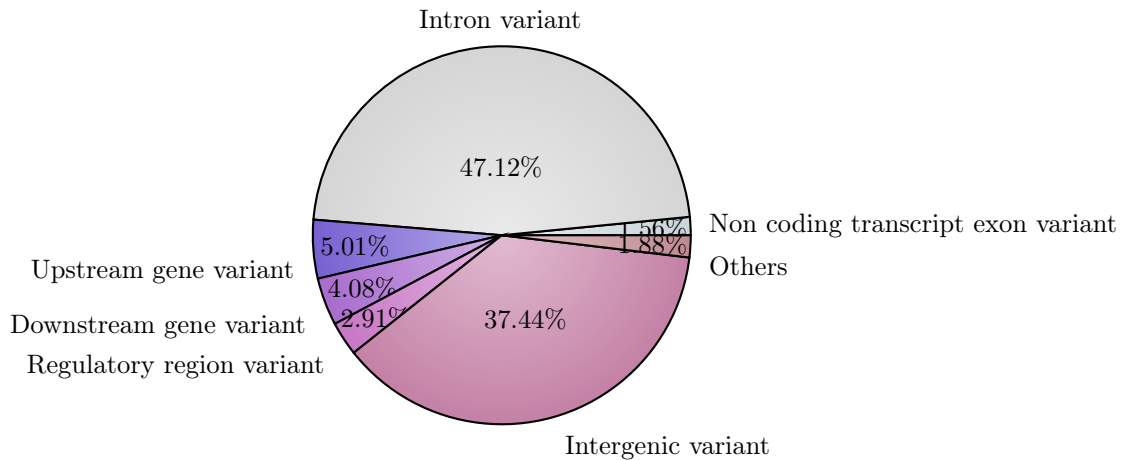


Figure 4: Consequence type

## 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).

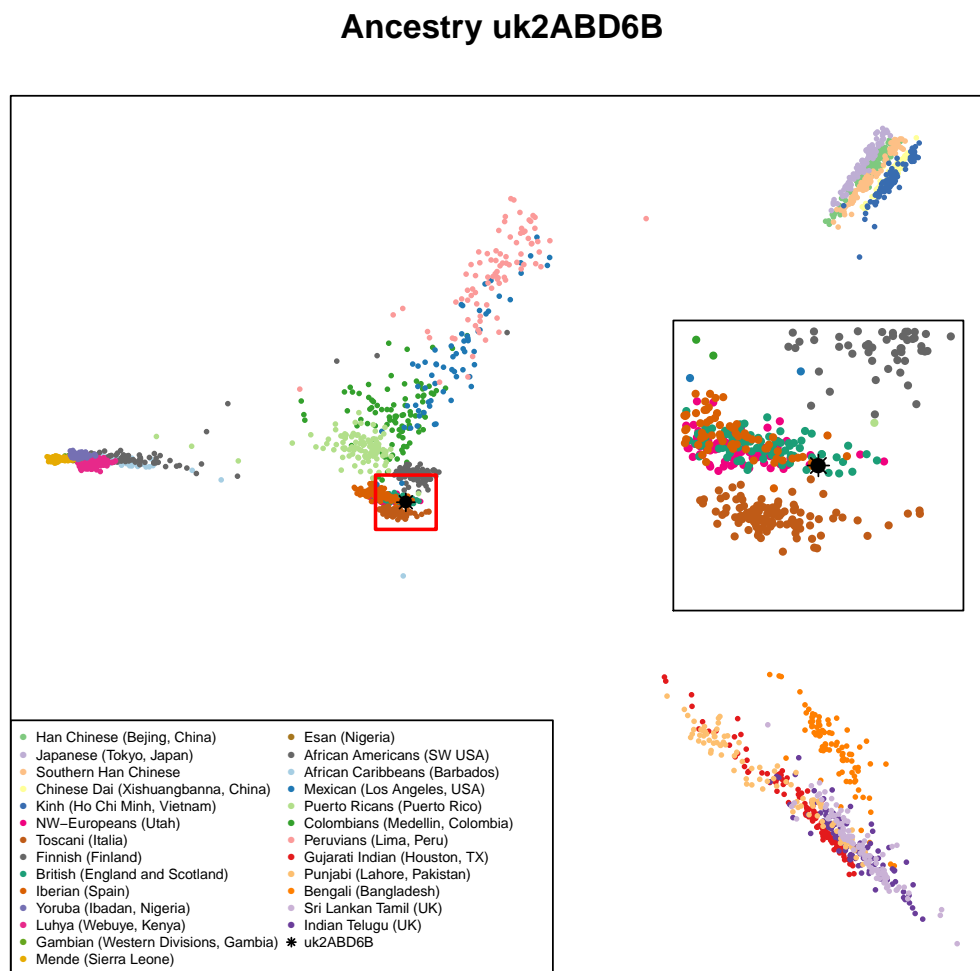


Figure 5: Ancestry Principal Component Analysis

### 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
2.1	rs2511989	(A;G)	0.63x decreased age-related macular degeneratio...		<a href="#">Link</a>	
2.1	rs3775291	(A;G)	0.71x decreased risk for dry age related macula...	<a href="#">Link</a>	<a href="#">Link</a>	<a href="#">Link</a>
2	rs10468017	(C;T)	Associated with higher HDL cholesterol		<a href="#">Link</a>	
2	rs10504861	(A;G)	Reduced risk of migraine without aura			
2	rs11045585	(A;A)	24% chance (lower than average) of docetaxel-in...		<a href="#">Link</a>	
2	rs1136410	(C;T)	0.80x reduced risk for glioblastoma	<a href="#">Link</a>	<a href="#">Link</a>	
2	rs12979860	(C;C)	~80% of such hepatitis C patients respond to tr...		<a href="#">Link</a>	<a href="#">Link</a>
2	rs1544410	(G;G)	Decreased risk of low bone mineral density diso...		<a href="#">Link</a>	
2	rs1799884	(G;G)	Mothers have typical Birth-Weight babies. Sligh...			
2	rs1864163	(A;G)	Associated with higher HDL cholesterol		<a href="#">Link</a>	
2	rs2235015	(G;T)	Somewhat more likely to respond to certain anti...	<a href="#">Link</a>	<a href="#">Link</a>	
2	rs2241423	(A;G)	0.79 decreased risk for obesity			
2	rs3736309	(A;G)	0.44x decreased risk for chronic obstructive pu...			
2	rs3738579	(C;T)	0.5x decreased risk for cervical cancer: HNSCC:...			
2	rs3764261	(G;T)	Associated with higher HDL cholesterol		<a href="#">Link</a>	<a href="#">Link</a>
2	rs4073582	(A;A)	Lower risk for gout	<a href="#">Link</a>		
2	rs6505162	(A;C)	0.58x decreased risk for esophageal cancer	<a href="#">Link</a>		
2	rs763110	(C;T)	~0.80x reduced cancer risk			<a href="#">Link</a>
2	rs801114	(T;T)	0.78x decreased Basal Cell Carcinoma risk.		<a href="#">Link</a>	
2	rs9272346	(A;G)	0.3x risk type-1 diabetes		<a href="#">Link</a>	
2	rs9525638	(C;C)	Stronger bones			
1.8	rs1128535	(A;G)	0.77x risk for Crohn's disease			
1.8	rs3814113	(C;T)	0.8x decreased risk for ovarian cancer		<a href="#">Link</a>	
1.8	rs4714156	(C;C)	<0.61x risk for restless legs			
1.5	rs1026732	(A;G)	0.70x risk for restless legs		<a href="#">Link</a>	
1.5	rs1063192	(C;T)	0.71x reduced risk of myocardial infarction			
1.5	rs11212617	(A;C)	Somewhat increased likelihood of treatment succ...			<a href="#">Link</a>

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
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	rs309375	(G;G)	Smaller mosquito bites			
1.5	rs3784709	(C;T)	0.71x risk of developing restless legs syndrome...		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	rs464049	(C;C)	Decreased risk of schizophrenia in limited stud...			
1.5	rs4939883	(C;T)	Associated with higher HDL cholesterol		Link	
1.5	rs5888	(C;C)	Higher HDL cholesterol but lower risk for age-r...	Link		
1.5	rs729302	(A;C)	0.89x decreased risk of developing rheumatoid a...			
1.4	rs1165205	(A;T)	0.85x decreased gout risk		Link	
1.4	rs6495446	(C;T)	0.8x reduced risk for chronic kidney disease			
1.4	rs6700125	(C;C)	0.7x decreased risk for ALS			
1.1	rs2293347	(G;G)	Among NSCLC patients: better Gefitinib response...	Link		Link
1.1	rs4988235	(T;T)	Can digest milk			Link
1.1	rs7568369	(T;T)	0.90x reduced risk of obesity			
1	rs10248420	(A;G)	7x more likely to respond to certain antidepres...		Link	
1	rs11983225	(C;T)	7x more likely to respond to certain antidepres...		Link	
1	rs182549	(T;T)	Can digest milk.			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	rs2952768	(C;T)	Slightly less drug dependence: decreased effect...			Link
1	rs4148739	(A;G)	7x more likely to respond to certain antidepres...		Link	
1.0	rs11246226	(C;C)	Decreased risk of schizophrenia in limited stud...		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	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	rs7014851	(C;T)	Very likely to be a benign allele	Link	Link	Link
0	rs74315403	(G;G)	PrP codon 178 (D) - non pathogenic variant			Link
0	rs9394492	(C;C)	<0.76x risk for restless legs			

### 3.2 Possibly Harmful Traits

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
3.5	rs199826652	(-;TCT)	Carrier for the most common cystic fibrosis mut...			
3.5	rs875858	(C;T)	Docetaxel sensitive?			
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	rs2066844	(C;T)	3x higher risk for Crohn's disease	Link	Link	Link
3	rs2306402	(C;C)	1.18x increased risk for late-onset Alzheimer's...			
3	rs7754840	(C;G)	1.3x increased risk for type-2 diabetes		Link	
2.5	rs10484554	(C;T)	2.8x increased risk for psoriasis		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	rs187238	(G;G)	Hypertension increases risk 3.75x for sudden ca...			
2.5	rs2943634	(C;C)	Slightly higher risk of ischemic stroke		Link	
2.5	rs5219	(T;T)	2.5x increased risk for type-2 diabetes	Link	Link	Link
2.5	rs664143	(C;T)	Higher risk for number of cancers			
2.5	rs9934438	(A;A)	Coumadin resistance		Link	Link
2.3	rs7966230	(C;G)	Slightly lower levels of plasma VWF			
2.2	rs1024611	(C;C)	Increased risk of exercise induced ischemia: In...			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
2.1	rs1050152	(T;T)	2.1x increased risk of Crohn's disease	Link	Link	Link
2.1	rs10811661	(T;T)	1.2x increased risk for type-2 diabetes		Link	
2.1	rs17070145	(C;C)	Reduced memory abilities			Link
2.1	rs17563	(C;C)	Risk for otosclerosis	Link	Link	Link
2.1	rs2187668	(A;G)	Somewhat increased autoimmune disorder (lupus: ...			
2.1	rs2254958	(C;T)	1.24x increased risk for Alzheimer's			
2.1	rs4402960	(T;T)	1.2x increased risk for type-2 diabetes: 1.5x r...		Link	Link
2.1	rs4444903	(G;G)	3.5x risk of hep-cancer in cirrhosis patients; ...			Link
2.1	rs5186	(A;C)	~1.4x increased risk of hypertension	Link	Link	Link
2.1	rs646776	(A;A)	1.2x risk of coronary artery disease		Link	
2.1	rs7837688	(G;T)	1.7x increased risk for prostate cancer			
2.1	rs944289	(C;T)	1.3x increased thyroid cancer risk		Link	
2	rs10090154	(C;T)	1.4x increased risk for prostate cancer			
2	rs1045642	(C;T)	Slower metaboliser for some drugs	Link	Link	Link
2	rs10513789	(G;T)	Increased risk of Parkinson's disease			
2	rs10883365	(G;G)	1.62x increased risk for developing Crohn's dis...		Link	
2	rs10984447	(A;A)	>1.17x increased risk for multiple sclerosis		Link	
2	rs11190870	(C;T)	Possibly increased risk of scoliosis			
2	rs1160312	(A;G)	1.6x increased risk of Male Pattern Baldness.		Link	
2	rs1265181	(C;G)	Increased risk for psoriasis		Link	
2	rs1333048	(A;C)	1.3x increased coronary artery disease risk			
2	rs1360780	(C;T)	1.3x increased risk for depression		Link	
2	rs1585215	(A;G)	2x increased risk for Hodgkin lymphoma			
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	rs1800629	(A;G)	Complex; generally higher risk for certain dise...	Link	Link	Link
2	rs1800896	(A;A)	1.8x increased prostate cancer risk			
2	rs2066843	(T;T)	4.09x higher risk for Crohn's disease	Link	Link	
2	rs2073963	(G;T)	Increased risk of baldness			
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		

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
2	rs2305480	(C;T)	3.5x increase in risk of asthma for Han Chinese...	Link	Link	
2	rs2383206	(A;G)	1.4x increased risk for heart disease			
2	rs2383207	(A;G)	Increased risk for heart disease			
2	rs25487	(G;G)	2x higher risk for skin cancer; possibly other ...	Link	Link	Link
2	rs2619522	(G;G)	Associated with lower attention capacity but al...			
2	rs2736100	(T;T)	Higher risk of Interstitial lung disease: and t...		Link	
2	rs2736990	(C;C)	Increased risk of developing Parkinson's Diseas...		Link	
2	rs27388	(A;A)	Increased risk of developing schizophrenia			
2	rs3025039	(C;T)	2.6x increased risk for ARMD in a Taiwanese pop...			
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	rs3775948	(G;G)	Slightly higher risk for gout			
2	rs3793784	(C;G)	1.5x risk for ARMD		Link	Link
2	rs4027132	(A;A)	1.51x increased risk of developing bipolar diso...			
2	rs4242382	(A;G)	1.7x increased risk for prostate cancer		Link	
2	rs4633	(T;T)	Higher risk for endometrial cancer	Link	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	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	rs638405	(G;G)	2x increased ALZ risk in ApoE4 carriers	Link		
2	rs6498169	(A;A)	>1.14x risk of multiple sclerosis		Link	
2	rs663048	(G;T)	Significantly increased risk of developing lung...	Link	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	rs699	(C;T)	Increased risk of hypertension	Link	Link	Link
2	rs7442295	(A;A)	~4x higher risk for hyperuracemia		Link	
2	rs744373	(C;T)	1.17x risk of Alzheimer's			
2	rs800292	(C;C)	5% higher risk of Age related macular degenerat...	Link	Link	Link
2	rs854560	(A;A)	Higher risk for heart disease: diabetic retinop...	Link	Link	Link
2.0	rs17221417	(G;G)	1.9x higher risk for Crohn's disease		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	rs143383	(T;T)	1.3x increased risk for osteoarthritis		Link	Link
1.8	rs2278206	(T;T)	1.16x increased risk for asthma	Link	Link	
1.8	rs37973	(A;G)	Among asthmatics: 1.5x more likely to show less...			Link
1.8	rs733618	(A;G)	1.87x risk for myasthenia gravis			
1.7	rs8055236	(G;T)	1.9x risk for heart disease		Link	
1.6	rs1537415	(C;G)	1.6x increased risk for periodontitis		Link	
1.6	rs3764880	(A;A)	1.2 - 1.8x increased tuberculosis risk	Link	Link	
1.6	rs763035	(T;T)	1.4x increased risk for rosacea			
1.5	rs10464059	(A;G)	Slightly increased risk of developing Parkinson...			
1.5	rs10757272	(C;T)	1.30x increased risk for Coronary artery diseas...			
1.5	rs10859871	(C;C)	Slight (~1.4x) increase in endometriosis risk			
1.5	rs10980705	(C;T)	2.3x increased risk for knee osteoarthritis			
1.5	rs12037606	(A;G)	1.22x risk of developing Crohn's disease			
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	rs13376333	(C;T)	1.5x higher risk of atrial fibrillation		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	rs1801274	(C;T)	Complex; generally greater risk for cancer prog...	Link	Link	Link
1.5	rs2240340	(A;A)	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	rs2697962	(A;G)	Slightly increased risk of developing Parkinson...			
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	rs358806	(A;C)	0.86x increased risk of developing Type-2 diabe...		Link	
1.5	rs3745516	(A;G)	Slightly increased risk of developing primary b...			
1.5	rs3814570	(T;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	rs4626664	(A;G)	1.44x increased risk of developing restless leg...		Link	
1.5	rs4785763	(A;C)	1.5x higher risk for melanoma		Link	
1.5	rs4845618	(G;T)	1.7x increased melanoma risk			
1.5	rs486907	(A;G)	1.5x increased prostate cancer risk	Link	Link	Link
1.5	rs5746059	(A;G)	Slightly higher fat mass			
1.5	rs6435862	(G;T)	1.7x higher risk of aggressive neuroblastoma		Link	
1.5	rs6974491	(A;A)	Higher risk of coeliac and/or inflammatory bowe...			
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	rs7850258	(G;G)	Slightly higher odds of developing primary hypo...			
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	rs9561778	(G;T)	~2x increased risk of adverse drug reactions fr...		Link	
1.5	rs9642880	(G;T)	1.2x increased bladder cancer risk		Link	
1.5	rs9652490	(A;G)	Slightly increased risk of developing Parkinson...		Link	
1.5	rs995030	(G;G)	Non-protective against testicular cancer		Link	
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	rs1447295	(A;C)	1.4x increased risk of prostate cancer		Link	
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	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	rs10947262	(C;C)	1.3x increased risk for osteoarthritis			
1.3	rs110419	(A;G)	1.3x increased risk for neuroblastoma			
1.3	rs13361189	(C;T)	1.3x increased risk for Crohn's disease		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	rs2024513	(A;G)	1.3x higher risk for schizophrenia (among Han C...			
1.3	rs2295490	(G;G)	1.32x increased risk of early-onset type-2 diab...	Link	Link	
1.3	rs4295627	(G;T)	1.36x higher risk for glioma development		Link	
1.3	rs4958847	(A;G)	1.3x increased risk for Crohn's disease			
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	rs1800693	(A;G)	Slight (1.2x) increase in risk for multiple scl...	Link	Link	Link
1.2	rs2056116	(A;G)	1.18x risk for breast cancer			



Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
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	rs2665390	(C;T)	1.2x increased risk for ovarian cancer			
1.2	rs3131296	(A;G)	1.2x increased risk for schizophrenia		Link	
1.2	rs3740878	(A;A)	1.46x type II diabetes risk; common	Link		Link
1.2	rs419788	(A;G)	2.0x risk for lupus	Link		
1.2	rs4977756	(A;G)	1.39x higher risk for glioma development		Link	
1.2	rs498872	(C;T)	1.2x higher risk for glioma development		Link	
1.2	rs6897876	(C;C)	Slight increase in testicular cancer risk for m...			
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	
1.1	rs11110912	(C;C)	1.3x high blood pressure risk			
1.1	rs1344706	(G;T)	1.1x increased risk for schizophrenia		Link	
1.1	rs249954	(C;T)	Potentially increased risk of Breast Cancer			Link
1.1	rs2651899	(A;G)	1.1x higher risk for migraines			
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	rs688034	(C;T)	1.1x risk higher risk for coronary artery disea...		Link	
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	
1.1	rs925391	(C;C)	More likely to go bald; common			
1.07	rs2291834	(C;C)	Very slightly higher risk for myocardial infarc...			
1	rs10761659	(A;G)	1.2x risk of Crohn's disease		Link	
1	rs1143674	(A;G)	1.3x increased autism risk	Link		
1	rs2273697	(A;G)	Adverse reaction more likely to carbamazepine i...	Link	Link	Link
1	rs2282679	(A;C)	Somewhat lower vitamin D levels			
1	rs2546890	(A;G)	Higher risk of multiple sclerosis			
1	rs3194051	(A;A)	>1.1x risk of type-1 diabetes	Link	Link	Link
1	rs761100	(G;G)	Higher risk for dyslexia			
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	rs1495965	(A;A)	1.2x higher risk for spondylitis			
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	rs4712653	(T;T)	2x increased risk for neuroblastoma			
0	rs6277	(C;C)	1.6x higher schizophrenia risk	Link	Link	Link
0	rs6314	(C;C)	Higher risk for RA	Link	Link	

### 3.3 Genosets (Multi-variant Phenotypes)

Magnitude	Identifier	Summary
6	gs216	2 copies of the <i>APO-Îµ4</i> allele
2.5	gs155	CYP3A5 non-expressor
2.5	gs281	Part of the 88% of the population claimed not t...
2	gs101	Probably able to digest milk
2	gs154	NAT2 Slow metabolizer
2	gs181	CYP2D6*2
2	gs249	Parkinson's Disease Risk
1.5	gs186	HLA-B*5801 heterozygosity is possible: unfortun...
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 ERS1176620 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/ERS1176620>

## 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 5, 2017.