

# PGP-UK Genomics Report for ukB4A6DF

## 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	5034217
Variants filtered out	0
Novel / existing variants	516183 (10.3) / 4506638 (89.7)
Overlapped genes	56664
Overlapped transcripts	67490
Overlapped regulatory features	168005

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.

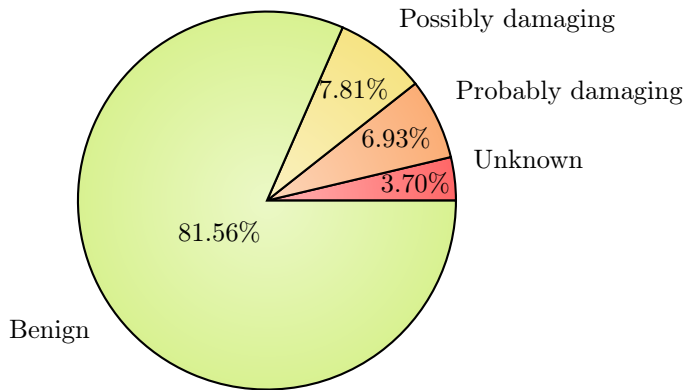


Figure 2: PolyPhen Summary

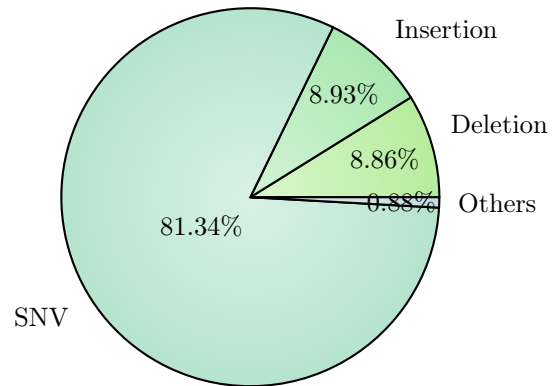


Figure 3: Variant Class

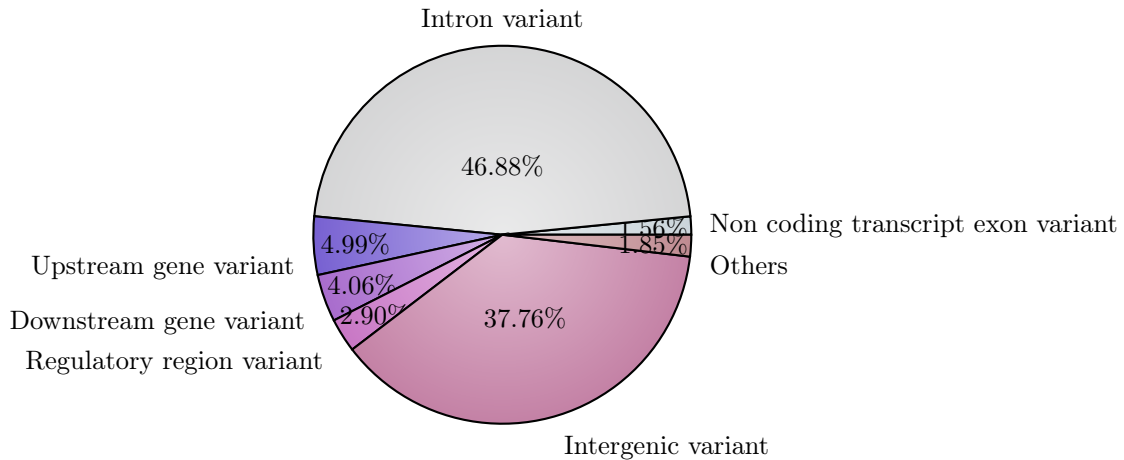


Figure 4: Consequence type

## 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).

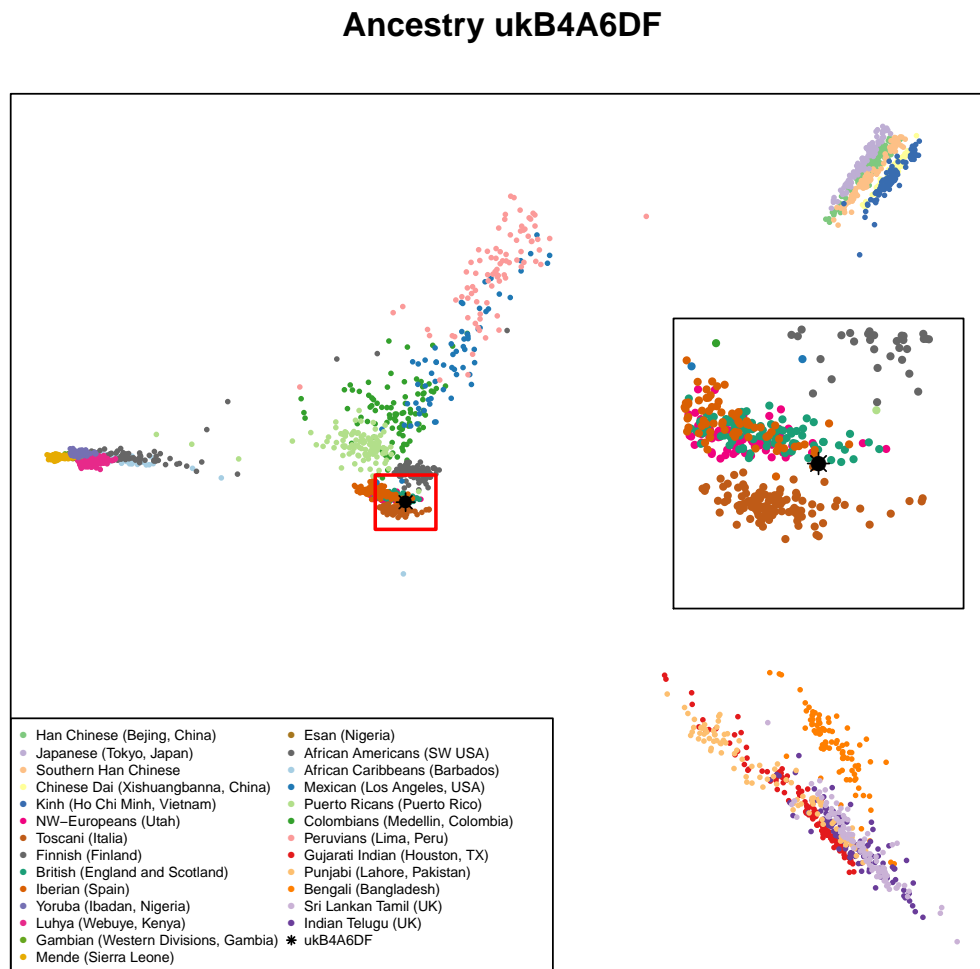


Figure 5: Ancestry Principal Component Analysis

### 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
2.5	rs2943634	(A;A)	Lower risk of ischemic stroke		<a href="#">Link</a>	
2.1	rs2511989	(A;G)	0.63x decreased age-related macular degeneratio...		<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	rs12979860	(C;C)	~80% of such hepatitis C patients respond to tr...		<a href="#">Link</a>	<a href="#">Link</a>
2	rs1800972	(G;G)	Reduced risk for Crohn's disease; reduced risk ...	<a href="#">Link</a>		
2	rs1864163	(A;G)	Associated with higher HDL cholesterol		<a href="#">Link</a>	
2	rs2241423	(A;G)	0.79 decreased risk for obesity			
2	rs2243250	(C;T)	0.6x decreased risk for myocardial infarction i...			
2	rs2542052	(C;C)	Better odds of living to 100			
2	rs3736309	(G;G)	0.44x decreased risk for chronic obstructive pu...			
2	rs3738579	(C;T)	0.5x decreased risk for cervical cancer: HNSCC:...			
2	rs3750817	(C;T)	0.78x reduced risk for breast cancer			
2	rs3764261	(G;T)	Associated with higher HDL cholesterol		<a href="#">Link</a>	<a href="#">Link</a>
2	rs3819331	(T;T)	Lower risk of autism	<a href="#">Link</a>		
2	rs3914132	(C;T)	Lower otosclerosis risk		<a href="#">Link</a>	
2	rs4149268	(G;G)	Associated with higher HDL cholesterol		<a href="#">Link</a>	
2	rs763110	(T;T)	~0.80x reduced cancer risk			<a href="#">Link</a>
2	rs7776725	(T;T)	Stronger bones		<a href="#">Link</a>	
2	rs8070723	(A;G)	0.18x reduced risk of developing progressive su...			
2	rs9272346	(A;G)	0.3x risk type-1 diabetes		<a href="#">Link</a>	
1.8	rs1128535	(A;G)	0.77x risk for Crohn's disease			
1.8	rs1746048	(C;T)	0.94 decreased risk for coronary heart disease		<a href="#">Link</a>	
1.8	rs187238	(C;G)	Hypertension not a risk factor for sudden cardi...			
1.8	rs9402571	(G;G)	0.85x decreased risk for type-2 diabetes			
1.5	rs1026732	(A;G)	0.70x risk for restless legs		<a href="#">Link</a>	
1.5	rs11635424	(A;G)	0.70x risk for restless legs		<a href="#">Link</a>	

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
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	rs3851179	(A;G)	0.85x decreased risk for Alzheimer's disease		Link	
1.5	rs4149274	(C;C)	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;C)	Associated with higher HDL cholesterol		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	rs6700125	(C;C)	0.7x decreased risk for ALS			
1.2	rs11246226	(A;C)	Decreased risk of schizophrenia in limited stud...		Link	
1.2	rs6048	(A;G)	Slightly lower risk (10-20%) of deep vein throm...	Link	Link	Link
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	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
1.0	rs6583817	(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
0.1	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	rs16947	(A;A)	Homozygous for CYP2D6 variants (non-CYP2D6*1)	Link	Link	Link
0	rs16990018	(A;A)	PrP Codon 171 Asn - Non-pathogenic variant	Link		Link
0	rs17244841	(A;A)	More responsive to statin treatment		Link	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	rs41303129	(C;T)	Likely to be a benign variant according to Clin...	Link		Link
0	rs5065	(A;A)	1.12x risk on diuretic; if hypertensive: better...	Link	Link	Link
0	rs74315403	(G;G)	PrP codon 178 (D) - non pathogenic variant			Link
0	rs7495174	(A;A)	Blue/gray eyes more likely		Link	

### 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	
3.8	rs5186	(C;C)	7.3x increased risk of hypertension	Link	Link	Link
3	rs10897346	(C;C)	If depressed: 2.6x more likely to not respond t...			
3	rs1983132	(C;T)	2 - 3x higher prostate cancer risk if routinely...			
3	rs2306402	(C;T)	1.18x increased risk for late-onset Alzheimer's...			
3	rs2981582	(C;T)	1.3x higher risk of ER+ breast cancer		Link	
3	rs55705857	(A;G)	6x increased risk of glioma of IDH1/IDH2 subtyp...			
3	rs6920220	(A;G)	1.2x risk Rheumatoid Arthritis		Link	
3	rs7754840	(C;G)	1.3x increased risk for type-2 diabetes		Link	
2.7	rs10830963	(C;G)	Increased type-2 diabetes risk; higher gestatio...		Link	
2.5	rs10490924	(G;T)	2.7x risk for age related macular degeneration	Link	Link	Link
2.5	rs1057910	(A;C)	CYP2C9*3 carrier; average 40% reduction in warf...	Link	Link	Link
2.5	rs1121980	(C;T)	1.67x risk for obesity		Link	
2.5	rs1421085	(C;T)	~1.3x increased obesity risk		Link	Link
2.5	rs16969968	(A;G)	Slightly higher risk for nicotine dependence: l...	Link	Link	Link
2.5	rs17782313	(C;C)	Adults likely to be 0.44 BMI units higher		Link	Link
2.5	rs3780374	(A;G)	Substantially increased odds of developing V617...			
2.5	rs5888	(C;T)	3x higher risk for age-related macular degenera...	Link		
2.5	rs8034191	(C;T)	1.27x lung cancer risk		Link	
2.4	rs1143679	(A;G)	1.78x increased risk for SLE	Link	Link	
2.4	rs7966230	(G;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	rs10811661	(T;T)	1.2x increased risk for type-2 diabetes		Link	
2.1	rs10871777	(G;G)	Adults likely to be 0.44 BMI units higher			
2.1	rs17070145	(C;C)	Reduced memory abilities			Link
2.1	rs17077540	(A;G)	1.6x major depressive disorder risk			
2.1	rs2231142	(A;C)	1.74x increased gout risk; gefinitib takers 4x ...	Link	Link	Link
2.1	rs2254958	(C;T)	1.24x increased risk for Alzheimer's			
2.1	rs2270641	(G;G)	3.7x higher risk for schizophrenia	Link	Link	
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	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	rs646776	(A;A)	1.2x risk of coronary artery disease		Link	
2.1	rs795484	(A;G)	Increased morphine dose requirement and postope...			
2	rs10086908	(C;T)	1.7x increased risk for prostate cancer			
2	rs10248420	(A;A)	7x less likely to respond to certain antidepres...		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	rs10984447	(A;A)	>1.17x increased risk for multiple sclerosis		Link	
2	rs11171739	(C;C)	1.75x risk of developing Type-1 diabetes		Link	
2	rs11190870	(C;T)	Possibly increased risk of scoliosis			
2	rs1160312	(A;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	rs13254738	(C;C)	1.18x prostate cancer risk		Link	
2	rs1360780	(C;T)	1.3x increased risk for depression		Link	
2	rs1361600	(G;G)	~2x increased risk for adult-onset asthma in Ja...			
2	rs144848	(G;G)	Very slightly increased breast cancer risk	Link	Link	Link

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
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	rs1800896	(A;G)	1.6x increased prostate cancer risk			
2	rs2073963	(G;T)	Increased risk of baldness			
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	(C;T)	3.5x increase in risk of asthma for Han Chinese...	Link	Link	
2	rs2420946	(C;T)	1.20x risk for breast cancer			
2	rs25487	(G;G)	2x higher risk for skin cancer; possibly other ...	Link	Link	Link
2	rs2707466	(G;G)	Weaker bones	Link	Link	
2	rs2908004	(C;C)	Weaker bones	Link	Link	
2	rs3129934	(C;T)	Increased risk of Multiple Sclerosis.		Link	
2	rs326	(A;A)	Lower HDL cholesterol		Link	Link
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	rs3746444	(C;T)	~1.2x increased risk for cancer	Link		
2	rs3775948	(G;G)	Slightly higher risk for gout			
2	rs3790565	(C;C)	Increased risk of developing primary biliary ci...			
2	rs3793784	(C;G)	1.5x risk for ARMD		Link	Link
2	rs4148739	(A;A)	7x less likely to respond to certain antidepres...		Link	
2	rs4444903	(A;G)	3.5x risk of hep-cancer in cirrhosis patients; ...			Link
2	rs4626664	(A;A)	>1.44x increased risk of developing restless le...		Link	
2	rs4633	(C;T)	Higher risk for endometrial cancer	Link	Link	Link
2	rs4792311	(A;G)	Increased risk of prostate cancer	Link	Link	Link
2	rs493258	(G;G)	1.15x risk of Age Related Macular Degeneration			
2	rs5174	(A;A)	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	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	rs6908425	(C;C)	1.95x increased risk of developing Crohn's dise...		Link	
2	rs6997709	(G;G)	1.5x higher risk for hypertension			
2	rs7442295	(A;A)	~4x higher risk for hyperuracemia		Link	
2	rs744373	(C;C)	1.17x risk of Alzheimer's			
2	rs7639618	(C;T)	1.45x increased osteoarthritis risk	Link		
2	rs7794745	(A;T)	Slightly increased risk for autism		Link	Link
2	rs7807268	(C;C)	1.4x risk for Crohn's disease		Link	
2	rs7923837	(G;G)	3.2x risk for T2D			
2	rs7961152	(A;C)	1.2x higher risk for hypertension			
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	rs9525638	(T;T)	Weaker bones			
2	rs9652490	(A;A)	~2x increased risk for Parkinson's disease: and...		Link	
2	rs965513	(A;G)	1.77x increased thyroid cancer risk		Link	
2.0	rs1434536	(A;A)	1.94x increased breast cancer risk			

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
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	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
1.7	rs2024513	(A;A)	1.7x higher risk for schizophrenia (among Han C...			
1.6	rs1537415	(C;G)	1.6x increased risk for periodontitis		Link	
1.6	rs2059693	(T;T)	1.6x increased risk for testicular cancer			
1.6	rs3764880	(A;A)	1.2 - 1.8x increased tuberculosis risk	Link	Link	
1.5	rs12037606	(A;G)	1.22x risk of developing Crohn's disease			
1.5	rs12498742	(A;A)	1.25 increased risk for gout			
1.5	rs13149290	(C;C)	Slightly increased risk of developing prostate ...			
1.5	rs13181	(G;T)	1.12x increased risk for cutaneous melanoma	Link	Link	Link
1.5	rs1375144	(C;C)	1.59x increased risk of developing bipolar diso...			
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	rs17115100	(G;T)	Slightly increased risk of developing Parkinson...	Link	Link	
1.5	rs17221417	(C;G)	1.3x higher risk for Crohn's disease		Link	
1.5	rs1799950	(A;G)	Very slightly increased breast cancer risk	Link	Link	Link
1.5	rs1867277	(A;G)	1.5x increased risk for thyroid cancer			
1.5	rs1975197	(C;T)	1.3x increased risk of developing restless legs...		Link	
1.5	rs1994090	(G;T)	Slightly increased risk of developing Parkinson...		Link	
1.5	rs199533	(C;T)	Slightly increased risk of developing Parkinson...	Link		
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	rs27388	(A;G)	Slightly increased risk of developing schizophr...			
1.5	rs2881766	(G;T)	Slightly increased risk for pregnancy-induced h...			
1.5	rs3087243	(A;G)	Increased risk for auto-immune diseases		Link	
1.5	rs3212227	(A;A)	1.43x increased risk of developing psoriasis an...			
1.5	rs3814570	(C;T)	1.3x increased risk for Crohn's disease with il...			
1.5	rs3825776	(A;G)	1.3x increased risk for ALS		Link	
1.5	rs401681	(C;T)	~1.2x increased risk for several types of cance...		Link	
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	rs4785763	(A;C)	1.5x higher risk for melanoma		Link	
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	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	rs699473	(C;T)	~1.5x increased brain tumor risk			
1.5	rs7536563	(A;G)	1.12x risk of multiple sclerosis		Link	
1.5	rs807701	(C;T)	Slightly increased dyslexia risk			
1.5	rs872071	(G;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	rs9642880	(G;T)	1.2x increased bladder cancer risk		Link	
1.5	rs966221	(C;C)	1.5x increased stroke risk certain populations			
1.5	rs995030	(G;G)	Non-protective against testicular cancer		Link	
1.4	rs12770228	(A;G)	1.4x increased risk for meningioma			
1.4	rs1801157	(A;A)	1.4x higher risk for breast cancer			
1.4	rs2046210	(C;T)	1.4x increased breast cancer risk		Link	Link
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	rs4959039	(A;G)	1.4x higher risk for multiple sclerosis			
1.34	rs17465637	(C;C)	1.34x higher risk for myocardial infarction	Link	Link	



Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
1.3	rs1042713	(A;G)	1.3x increased risk that pediatric inhaler use ...	Link	Link	Link
1.3	rs10947262	(C;C)	1.3x increased risk for osteoarthritis			
1.3	rs2736100	(G;T)	1.3x higher risk for glioma development: 2.1x r...		Link	
1.3	rs4295627	(G;T)	1.36x higher risk for glioma development		Link	
1.3	rs501120	(A;G)	1.3x increased risk for heart disease		Link	
1.25	rs748404	(T;T)	Slightly increased risk (1.25) for lung cancer...		Link	
1.2	rs11037909	(T;T)	1.47x type II diabetes risk	Link		
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	rs2056116	(A;G)	1.18x risk for breast cancer			
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	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	rs419788	(A;G)	2.0x risk for lupus	Link		
1.2	rs4686484	(A;A)	Slightly increased risk for celiac disease			
1.2	rs6010620	(A;G)	1.2x higher risk for glioma development: 1.17x ...		Link	
1.2	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;G)	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	rs2295190	(G;T)	Slightly increased risk for ovarian cancer in w...	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	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			
1.07	rs2291834	(C;C)	Very slightly higher risk for myocardial infarc...			
1	rs1143674	(A;G)	1.3x increased autism risk	Link		
1	rs2546890	(A;A)	Higher risk of multiple sclerosis			
1	rs3194051	(A;A)	>1.1x risk of type-1 diabetes	Link	Link	Link
1	rs3735684	(C;T)	Associated with increased colorectal cancer ris...	Link	Link	
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	rs10761659	(A;A)	1.5x risk of Crohn's disease		Link	
0	rs2296336	(C;C)	2.9x risk of type-1 diabetes			
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	rs6314	(C;C)	Higher risk for RA	Link	Link	
0	rs7787082	(G;G)	7x less likely to respond to certain antidepres...		Link	

### 3.3 Genosets (Multi-variant Phenotypes)

Magnitude	Identifier	Summary
3.1	gs191	Problem metabolizing NSAIDs
3	gs241	Lighter green: brown or hazel eye color
2.5	gs155	CYP3A5 non-expressor
2.5	gs161	CYP2C9 Intermediate Metabolizers
2.5	gs281	Part of the 88% of the population claimed not t...
2	gs101	Probably able to digest milk
2	gs156	NAT2 Rapid metabolizer.
2	gs239	Reduced conversion of beta-carotene to retinol
2	gs246	APOE3/APOE3
1.5	gs185	The beta blocker metoprolol is effective with 1...
1.5	gs247	Parkinson's Disease Risk
1.2	gs184	Able to taste bitterness.
0	gs158	CYP1A2 normal metabolizer

## 4 Raw Data

The raw data used to create this report has been assigned the identifier ERS1176589 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/ERS1176589>

## 5 Report Metadata

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

Table 5: Analysis Pipeline Versions

Report generated on August 2, 2017.