

# PGP-UK Genomics Report for ukF6D075

## 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	4976113
Variants filtered out	0
Novel / existing variants	499920 (10.1) / 4463821 (89.9)
Overlapped genes	56739
Overlapped transcripts	67535
Overlapped regulatory features	166866

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.

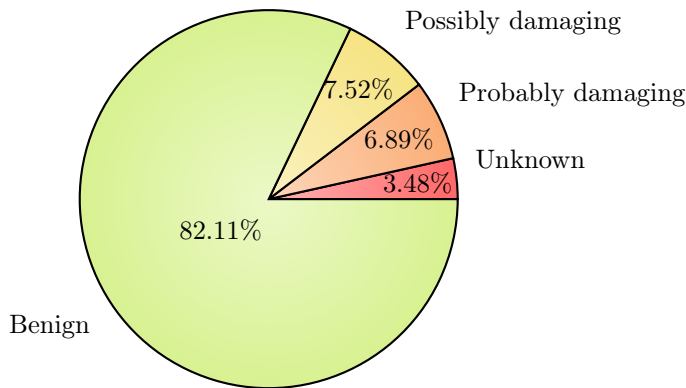


Figure 2: PolyPhen Summary

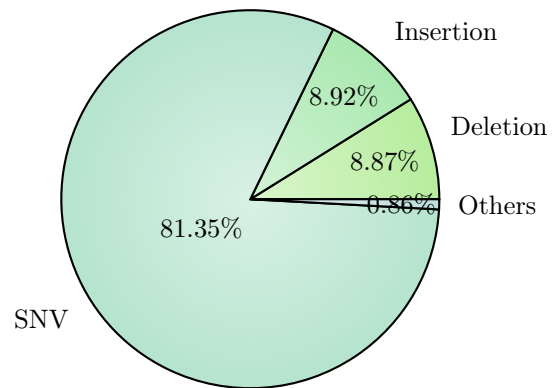


Figure 3: Variant Class

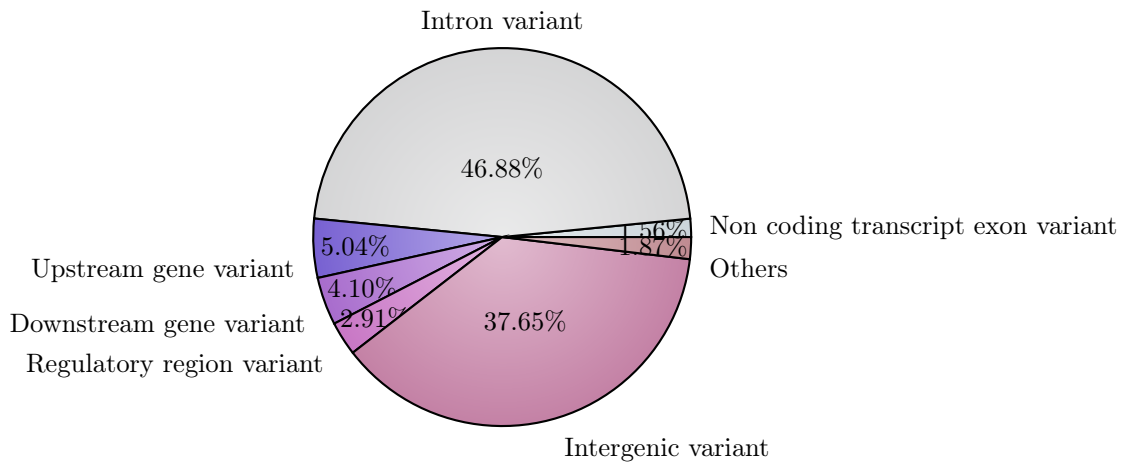


Figure 4: Consequence type

## 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).

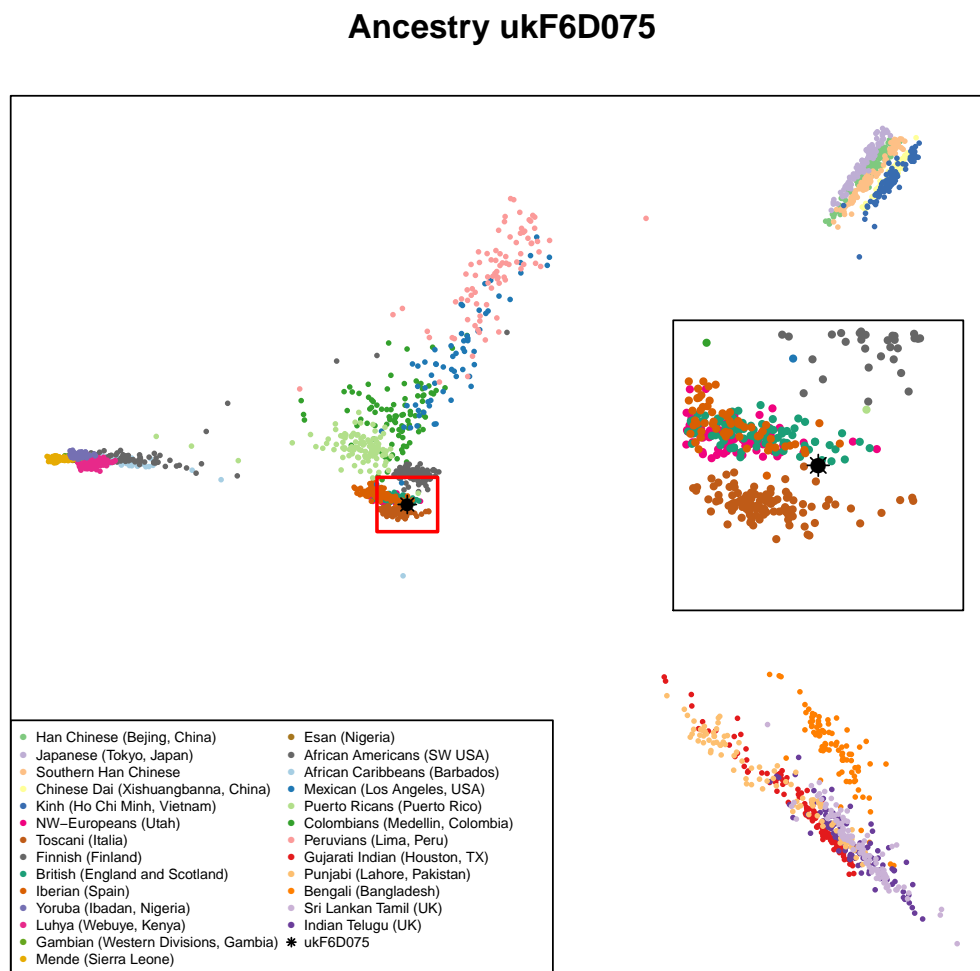


Figure 5: Ancestry Principal Component Analysis

### 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	rs1012053	(A;C)	0.625x reduced risk of Bipolar Disorder.		<a href="#">Link</a>	
2	rs10468017	(T;T)	Associated with higher HDL cholesterol		<a href="#">Link</a>	
2	rs10503669	(A;C)	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	rs12678919	(A;G)	Associated with higher HDL cholesterol		<a href="#">Link</a>	
2	rs1799884	(G;G)	Mothers have typical Birth-Weight babies. Sligh...			
2	rs1864163	(G;G)	Associated with higher HDL cholesterol		<a href="#">Link</a>	
2	rs2060793	(A;A)	Lower serum levels of vitamin D			
2	rs2241423	(A;G)	0.79 decreased risk for obesity			
2	rs3736309	(A;G)	0.44x decreased risk for chronic obstructive pu...			
2	rs3750817	(C;T)	0.78x reduced risk for breast cancer			
2	rs3782179	(C;T)	3x lower odds of testicular cancer risk for men...			
2	rs3819331	(T;T)	Lower risk of autism	<a href="#">Link</a>		
2	rs3914132	(C;T)	Lower otosclerosis risk		<a href="#">Link</a>	
2	rs4073582	(A;A)	Lower risk for gout	<a href="#">Link</a>		
2	rs4307059	(C;C)	Reduced Autism risk		<a href="#">Link</a>	
2	rs6505162	(A;C)	0.58x decreased risk for esophageal cancer	<a href="#">Link</a>		
2	rs6855911	(A;G)	0.62x decreased risk for gout		<a href="#">Link</a>	
2	rs763110	(C;T)	~0.80x reduced cancer risk			<a href="#">Link</a>
2	rs7776725	(T;T)	Stronger bones		<a href="#">Link</a>	
2	rs800292	(T;T)	5% decreased risk of macular degeneration	<a href="#">Link</a>	<a href="#">Link</a>	<a href="#">Link</a>
2	rs801114	(T;T)	0.78x decreased Basal Cell Carcinoma risk.		<a href="#">Link</a>	
2	rs925391	(C;T)	Lower odds of going bald			
2	rs9642880	(G;G)	Slightly lower risk of Bladder Cancer.		<a href="#">Link</a>	

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
1.8	rs187238	(C;G)	Hypertension not a risk factor for sudden cardi...			
1.8	rs3814113	(C;T)	0.8x decreased risk for ovarian cancer		Link	
1.8	rs4714156	(C;C)	<0.61x risk for restless legs			
1.6	rs1061170	(T;T)	Lower risk for AMD: generally longer live than ...	Link	Link	Link
1.5	rs1026732	(A;G)	0.70x risk for restless legs		Link	
1.5	rs1063192	(C;C)	0.71x reduced risk of myocardial infarction			
1.5	rs11136000	(C;T)	0.84x decreased risk for Alzheimer's disease		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	rs3851179	(A;A)	0.85x decreased risk for Alzheimer's disease		Link	
1.5	rs4149274	(C;T)	Associated with higher HDL (good) cholesterol			
1.5	rs4489954	(G;T)	0.69x risk risk of developing restless legs syn...		Link	
1.5	rs4939883	(C;C)	Associated with higher HDL cholesterol		Link	
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	rs9402571	(G;T)	Slightly decreased risk for type-2 diabetes			
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			
1.2	rs4686484	(G;G)	Slightly decreased risk for celiac disease			
1.1	rs2293347	(G;G)	Among NSCLC patients: better Gefitinib response...	Link		Link
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	rs2494732	(T;T)	Lower odds of psychosis	Link	Link	
1	rs2546890	(G;G)	Lower risk of multiple sclerosis			
1	rs2952768	(C;C)	Less drug dependence: decreased effectiveness o...			Link
1	rs7850258	(A;G)	Typical odds of developing primary hypothyroidi...			
0.5	rs36094464	(A;T)	Most likely benign: though reported years ago t...	Link	Link	Link
0	rs10427255	(T;T)	Lowest odds of photic sneeze reflex			
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	rs1799945	(C;C)	Not a H63D hemochromatosis carrier.	Link	Link	Link
0	rs1800562	(G;G)	Not a C282Y hemochromatosis carrier.	Link	Link	Link
0	rs28933385	(G;G)	Prion protein Codon 200 (E) - Non pathogenic va...			Link
0	rs312481	(C;C)	Better response to certain calcium channel bloc...			
0	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	rs9394492	(C;C)	<0.76x risk for restless legs			

### 3.2 Possibly Harmful Traits

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
4	rs118020901	(A;C)	Corneal dystrophy: fuchs endothelial: 6	Link		Link
3	rs10897346	(C;C)	If depressed: 2.6x more likely to not respond t...			
3	rs2306402	(C;C)	1.18x increased risk for late-onset Alzheimer's...			
3	rs3738579	(T;T)	1.5x - 2x increased risk for cervical cancer: H...			
3	rs6920220	(A;G)	1.2x risk Rheumatoid Arthritis		Link	
2.5	rs1121980	(C;T)	1.67x risk for obesity		Link	
2.5	rs13266634	(C;T)	Increased risk for type-2 diabetes	Link	Link	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	rs2073963	(G;G)	Increased risk of baldness			
2.5	rs2943634	(C;C)	Slightly higher risk of ischemic stroke		Link	
2.5	rs5888	(C;T)	3x higher risk for age-related macular degenera...	Link		
2.5	rs613872	(G;T)	~5 fold higher risk for Fuchs' dystrophy: a cor...			
2.5	rs8034191	(C;T)	1.27x lung cancer risk		Link	
2.5	rs891512	(A;G)	Higher blood pressure than G;G	Link		
2.4	rs7966230	(G;G)	Slightly lower levels of plasma VWF			
2.3	rs1859962	(G;G)	1.28x increased risk for prostate cancer		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	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	rs5186	(A;C)	~1.4x increased risk of hypertension	Link	Link	Link
2.1	rs5751876	(T;T)	Significantly higher anxiety levels after moder...	Link		
2.1	rs646776	(A;A)	1.2x risk of coronary artery disease		Link	
2	rs10248420	(A;A)	7x less likely to respond to certain antidepres...		Link	
2	rs1041981	(A;A)	Higher myocardial infarction risk	Link	Link	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	rs10871777	(A;G)	Adults likely to be 0.22 BMI units higher			
2	rs10937823	(C;T)	Some association with bipolar disorder			
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;G)	1.6x increased risk of Male Pattern Baldness.		Link	
2	rs11983225	(T;T)	7x less likely to respond to certain antidepres...		Link	
2	rs12567232	(A;G)	Increased risk for Crohn's Disease		Link	
2	rs1265181	(C;G)	Increased risk for psoriasis		Link	
2	rs12696304	(C;G)	Prone to aging faster: at least in European pop...			
2	rs13254738	(A;C)	1.18x prostate cancer risk		Link	
2	rs1360780	(C;T)	1.3x increased risk for depression		Link	
2	rs1585215	(A;G)	2x increased risk for Hodgkin lymphoma			
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	rs17782313	(C;T)	Adults likely to be 0.22 BMI units higher		Link	Link
2	rs1800629	(A;G)	Complex; generally higher risk for certain dise...	Link	Link	Link
2	rs1800896	(A;G)	1.6x increased prostate cancer risk			
2	rs2201841	(C;T)	1.5x increased risk for Crohn's disease; 2x inc...		Link	

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
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	rs25487	(G;G)	2x higher risk for skin cancer; possibly other ...	Link	Link	Link
2	rs2707466	(G;G)	Weaker bones	Link	Link	
2	rs27388	(A;A)	Increased risk of developing schizophrenia			
2	rs2908004	(C;C)	Weaker bones	Link	Link	
2	rs3212227	(A;C)	Significantly increased risk of developing cerv...			
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	rs3738919	(A;C)	1.94x risk of developing rheumatoid arthritis			
2	rs3793784	(C;G)	1.5x risk for ARMED		Link	Link
2	rs4148739	(A;A)	7x less likely to respond to certain antidepres...		Link	
2	rs4420638	(A;G)	~3x increased Alzheimer's risk; 1.4x increased ...		Link	Link
2	rs4444903	(A;G)	3.5x risk of hep-cancer in cirrhosis patients; ...			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	rs5174	(A;G)	1.3x increased risk for heart disease	Link	Link	Link
2	rs520354	(A;A)	Increased risk in men for biliary conditions			
2	rs629242	(C;T)	Somewhat higher risk for prostate cancer			
2	rs6435862	(G;G)	2.8x higher risk of aggressive neuroblastoma		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	rs662799	(A;G)	1.4x higher early heart attack risk; less weigh...		Link	
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	rs699	(C;T)	Increased risk of hypertension	Link	Link	Link
2	rs744373	(C;T)	1.17x risk of Alzheimer's			
2	rs7807268	(C;C)	1.4x risk for Crohn's disease		Link	
2	rs828907	(G;T)	Slightly increased risk of bladder cancer and 2...			
2	rs854560	(A;T)	Higher risk for heart disease: diabetic retinop...	Link	Link	Link
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	rs9954153	(G;T)	~2.5x higher risk for Fuchs' dystrophy: a corne...			
2.0	rs1044396	(C;C)	Increased risk of Nicotine dependence among mal...	Link	Link	Link
2.0	rs2156921	(G;G)	1.29x increased risk for depression			
2.0	rs4911414	(T;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	rs37973	(A;G)	Among asthmatics: 1.5x more likely to show less...			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			
1.7	rs2024513	(A;A)	1.7x higher risk for schizophrenia (among Han C...			
1.6	rs11523871	(A;C)	1.6x increased breast cancer risk for women ove...	Link	Link	
1.6	rs2736100	(G;G)	1.6x higher risk for glioma development		Link	
1.6	rs2981745	(C;T)	1.6x increased risk for breast cancer in female...			
1.6	rs3764880	(A;A)	1.2 - 1.8x increased tuberculosis risk	Link	Link	
1.6	rs3775948	(C;G)	Slightly higher risk for gout			
1.5	rs10492519	(A;G)	Slightly increased risk of developing prostate ...			
1.5	rs10883365	(A;G)	1.2x increased risk for developing Crohn's dise...		Link	
1.5	rs10980705	(C;T)	2.3x increased risk for knee osteoarthritis			
1.5	rs1154155	(G;T)	1.94x increased risk for narcolepsy		Link	
1.5	rs12037606	(A;G)	1.22x risk of developing Crohn's disease			

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
1.5	rs12210050	(C;T)	Slightly higher risk for basal cell carcinoma		Link	
1.5	rs12431733	(C;T)	Slightly increased risk of developing Parkinson...		Link	
1.5	rs13149290	(C;C)	Slightly increased risk of developing prostate ...			
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	rs1801274	(C;T)	Complex; generally greater risk for cancer prog...	Link	Link	Link
1.5	rs1994090	(G;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;G)	1.4x increased risk of SLE			
1.5	rs2736990	(C;T)	Slightly increased risk of developing Parkinson...		Link	
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	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	rs393152	(A;A)	Increased risk of both PD and AD	Link	Link	
1.5	rs401681	(C;C)	~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	rs4626664	(A;G)	1.44x increased risk of developing restless leg...		Link	
1.5	rs464049	(C;T)	Increased risk of schizophrenia in limited stud...			
1.5	rs4656461	(A;G)	1.5x increased risk for open angle glaucoma			
1.5	rs4785763	(A;C)	1.5x higher risk for melanoma		Link	
1.5	rs4845618	(G;T)	1.7x increased melanoma risk			
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	rs619203	(C;G)	Increases susceptibility to Myocardial Infarcti...	Link	Link	
1.5	rs6498169	(A;G)	1.14x risk of multiple sclerosis		Link	
1.5	rs6896702	(C;T)	Slightly increased risk of developing Parkinson...			
1.5	rs6908425	(C;T)	1.63x increased risk of developing Crohn's dise...		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	rs7454108	(C;T)	Single HLA-DQ8 haplotype			
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	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	rs9561778	(G;T)	~2x increased risk of adverse drug reactions fr...		Link	
1.4	rs1126497	(T;T)	1.4x increased risk for breast cancer	Link	Link	Link
1.4	rs12770228	(A;G)	1.4x increased risk for meningioma			
1.4	rs2046210	(C;T)	1.4x increased breast cancer risk		Link	Link
1.4	rs2230201	(A;G)	1.4x risk of lupus	Link		
1.4	rs4977756	(G;G)	1.93x higher risk for glioma development		Link	
1.4	rs6010620	(G;G)	1.4x higher risk for glioma development; but th...		Link	
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	rs1434536	(A;G)	1.29x increased breast cancer risk			
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	rs356219	(A;G)	1.3x increased risk for Parkinson's disease			



Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
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			
1.2	rs2252586	(A;G)	1.2x higher risk for glioma development			
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	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	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	rs9960767	(A;C)	1.2x increased risk for schizophrenia		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	
1.1	rs11110912	(C;C)	1.3x high blood pressure risk			
1.1	rs1344706	(G;T)	1.1x increased risk for schizophrenia		Link	
1.1	rs1800450	(A;G)	Carrier of mannose binding deficiency but of lo...	Link	Link	Link
1.1	rs249954	(C;T)	Potentially increased risk of Breast Cancer			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	rs688034	(C;T)	1.1x risk higher risk for coronary artery disea...		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	rs889312	(A;C)	Very slightly higher risk for breast cancer		Link	
1.09	rs12050604	(A;C)	Very slightly increased risk for lung cancer			
1.05	rs2291834	(C;T)	Very slightly higher risk for myocardial infarc...			
1	rs10761659	(A;G)	1.2x risk of Crohn's disease		Link	
1	rs1143674	(A;G)	1.3x increased autism risk	Link		
1	rs2282679	(A;C)	Somewhat lower vitamin D levels			
1	rs3194051	(A;G)	1.12x risk of type-1 diabetes	Link	Link	Link
1	rs6932590	(C;T)	1.1x increased risk for schizophrenia		Link	
0	rs4293393	(T;T)	1.25x Increased Risk of CKD for T allele in ...			
0	rs4712653	(T;T)	2x increased risk for neuroblastoma			
0	rs6314	(C;C)	Higher risk for RA	Link	Link	
0	rs6684865	(A;A)	1.5x risk of rheumatoid arthritis			
0	rs7787082	(G;G)	7x less likely to respond to certain antidepres...		Link	

### 3.3 Genosets (Multi-variant Phenotypes)

Magnitude	Identifier	Summary
2.9	gs192	MTHFR polymorphisms affecting homocysteine
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	gs173	CYP2D6*10
2	gs187	HLA-B*5801 homozygosity is possible. too common...
2	gs188	One copy of APOE4 is possible: but not certain
1.5	gs186	HLA-B*5801 heterozygosity is possible: unfortun...
1.5	gs220	HLA-B*1502?
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 ERS1176611 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/ERS1176611>

## 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.