

# PGP-UK Genomics Report for ukFCACC7

## 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.

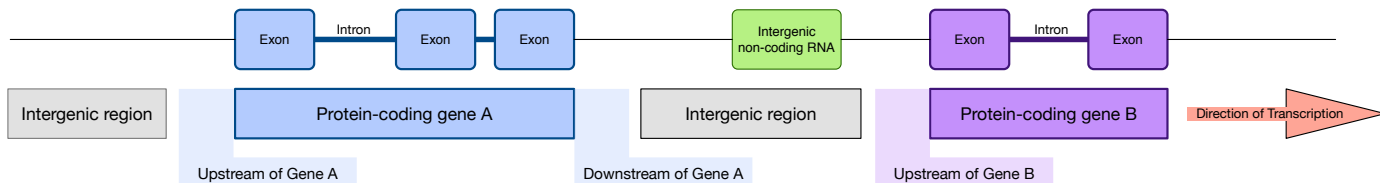


Figure 1: Diagram of gene structure indicating locations of potential variants

Feature	Count
Lines of input read	4951551
Variants filtered out	0
Novel / existing variants	497136 (10.1) / 4440848 (89.9)
Overlapped genes	56743
Overlapped transcripts	67565
Overlapped regulatory features	166952

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.

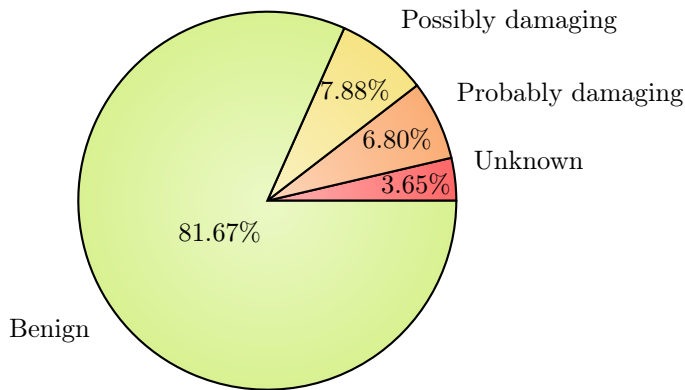


Figure 2: PolyPhen Summary

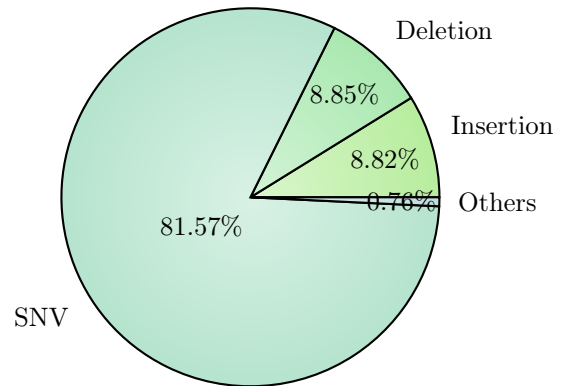


Figure 3: Variant Class

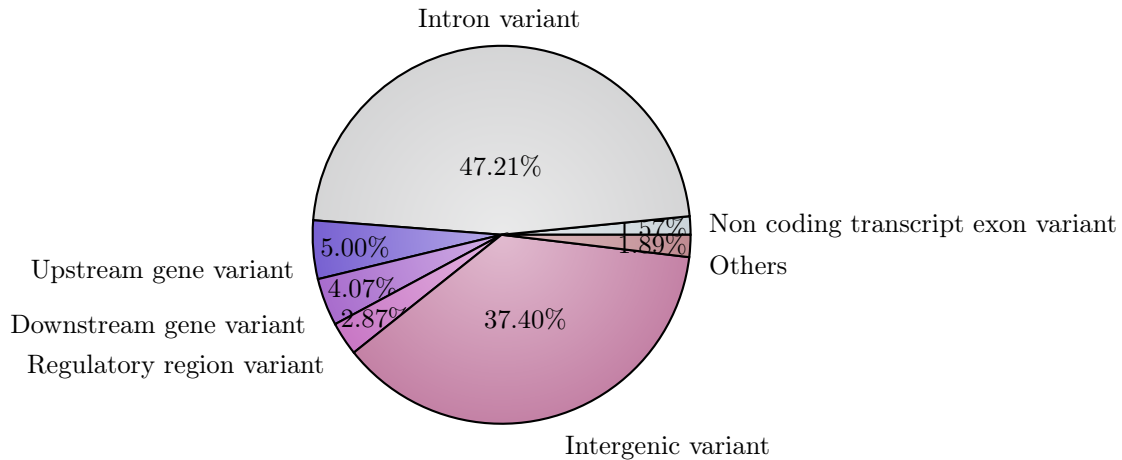


Figure 4: Consequence type

## 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 17.6 percent South Asian, 82.4 percent European.

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

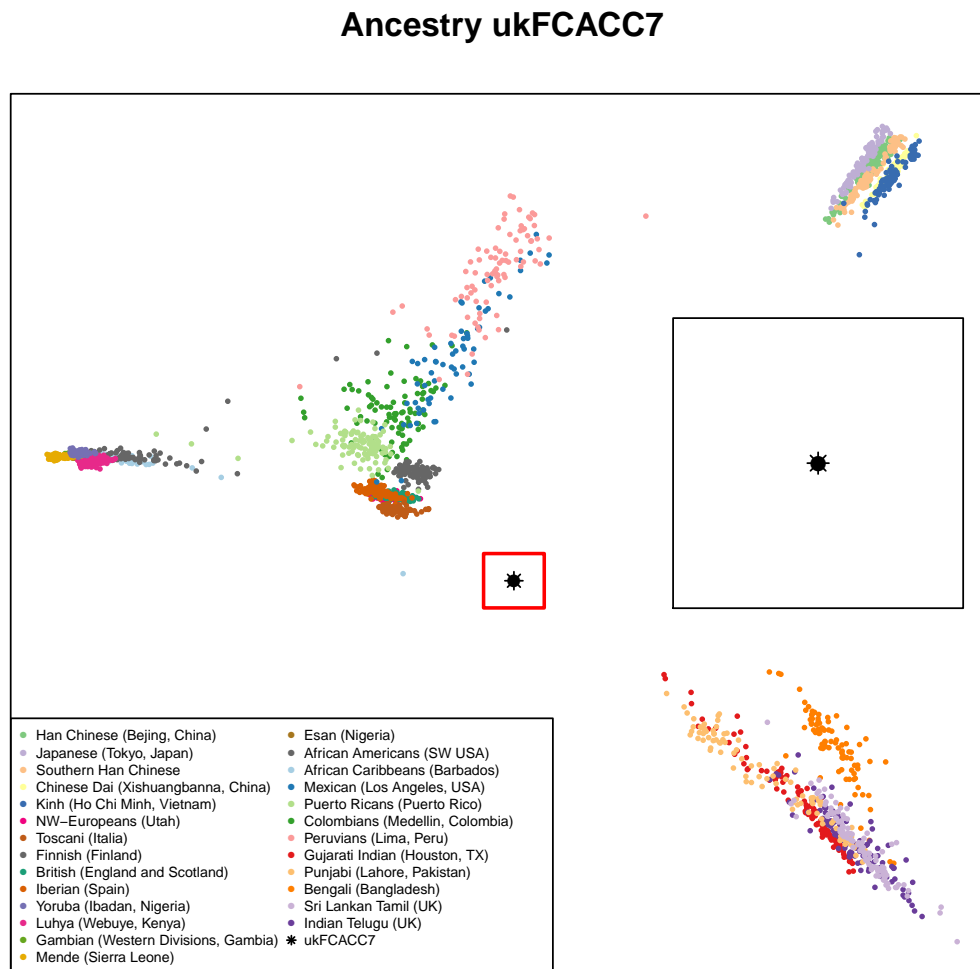


Figure 5: Ancestry Principal Component Analysis

### 3 Traits (based on SNPedia information)

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

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

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

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

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

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

#### 3.1 Possibly Beneficial Traits

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
3	rs7294919	(C;T)	Moderately enhanced hippocampal volume			
2.1	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	rs11045585	(A;A)	24% chance (lower than average) of docetaxel-in...		<a href="#">Link</a>	
2	rs1128535	(G;G)	Reduced risk (0.77x) for Crohn's disease			
2	rs1160312	(G;G)	Reduced risk of Baldness.		<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	rs17070145	(C;T)	Increased memory performance			<a href="#">Link</a>
2	rs2073963	(T;T)	Reduced risk of baldness			
2	rs2243250	(C;T)	0.6x decreased risk for myocardial infarction i...			
2	rs2292813	(C;T)	Decreased risk of autism			
2	rs2542052	(C;C)	Better odds of living to 100			
2	rs2707466	(A;A)	Stronger bones	<a href="#">Link</a>	<a href="#">Link</a>	
2	rs3218536	(A;G)	Lower risk for breast: ovarian cancer	<a href="#">Link</a>	<a href="#">Link</a>	
2	rs3736309	(A;G)	0.44x decreased risk for chronic obstructive pu...			
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	rs4307059	(C;C)	Reduced Autism risk		<a href="#">Link</a>	
2	rs4585	(G;G)	Slightly higher (1.35x) odds of good metformin ...			
2	rs6807362	(G;G)	Decreased autism risk	<a href="#">Link</a>	<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	rs8070723	(A;G)	0.18x reduced risk of developing progressive su...			
1.8	rs187238	(C;C)	Hypertension not a risk factor for sudden cardi...			

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
1.6	rs1061170	(T;T)	Lower risk for AMD: generally longer live than ...	Link	Link	Link
1.6	rs10801935	(C;C)	0.3x decreased risk of breast cancer			
1.6	rs3025786	(C;T)	Slightly decreased Alzheimer's disease risk amo...	Link		
1.5	rs1063192	(C;T)	0.71x reduced risk of myocardial infarction			
1.5	rs11136000	(C;T)	0.84x decreased risk for Alzheimer's disease		Link	
1.5	rs11212617	(C;C)	Somewhat increased likelihood of treatment succ...			Link
1.5	rs3851179	(A;G)	0.85x decreased risk for Alzheimer's disease		Link	
1.5	rs464049	(C;C)	Decreased risk of schizophrenia in limited stud...			
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	rs2294008	(C;C)	Lower risk of gastric and bladder cancer	Link	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	rs4686484	(G;G)	Slightly decreased risk for celiac disease			
1.2	rs6048	(G;G)	Slightly lower risk (10-20%) of deep vein throm...	Link	Link	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	rs2293347	(G;G)	Among NSCLC patients: better Gefitinib response...	Link		Link
1.1	rs7568369	(G;T)	0.90x reduced risk of obesity			
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	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	rs74315403	(G;G)	PrP codon 178 (D) - non pathogenic variant			Link

## 3.2 Possibly Harmful Traits

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
3.1	rs1421085	(C;C)	~1.7x increased obesity risk		Link	Link
3	rs10897346	(C;C)	If depressed: 2.6x more likely to not respond t...			
3	rs1121980	(T;T)	2.76x risk for obesity		Link	
3	rs1801282	(C;G)	Unconfirmed higher risk of cardiovascular disea...	Link	Link	Link
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	rs3738579	(T;T)	1.5x - 2x increased risk for cervical cancer: H...			
3	rs7754840	(C;G)	1.3x increased risk for type-2 diabetes		Link	
2.6	rs4958847	(A;A)	2.6x increased risk for Crohn's disease			
2.5	rs10490924	(G;T)	2.7x risk for age related macular degeneration	Link	Link	Link
2.5	rs12803066	(A;G)	Increased risk of myopia			
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	rs2254958	(C;C)	1.61x increased risk for Alzheimer's			
2.5	rs2943634	(C;C)	Slightly higher risk of ischemic stroke		Link	
2.5	rs339331	(T;T)	Prostate cancer risk			
2.5	rs3738919	(C;C)	1.94x risk of developing rheumatoid arthritis			
2.5	rs5888	(C;T)	3x higher risk for age-related macular degenera...	Link		
2.5	rs6441286	(G;G)	3.08x chance of developing primary biliary cirr...		Link	
2.5	rs664143	(T;T)	Higher risk for number of cancers			
2.5	rs7574865	(G;T)	1.3x risk of rheumatoid arthritis; 1.55x risk o...		Link	Link
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	rs2274223	(G;G)	1.9x increased risk for stomach and esophageal ...	Link	Link	Link
2.4	rs7966230	(G;G)	Slightly lower levels of plasma VWF			
2.2	rs2004640	(G;T)	1.4x increased risk for SLE		Link	Link
2.2	rs2231137	(G;G)	~1.5-3x increased risk for ischemic stroke	Link	Link	Link
2.2	rs2305089	(T;T)	Higher risk for chordoma	Link	Link	
2.1	rs10811661	(T;T)	1.2x increased risk for type-2 diabetes		Link	
2.1	rs1360780	(T;T)	1.3x increased risk for depression		Link	
2.1	rs2187668	(A;G)	Somewhat increased autoimmune disorder (lupus: ...			
2.1	rs2494732	(C;C)	Greater odds of cannabis-associated psychosis	Link	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	rs944289	(C;T)	1.3x increased thyroid cancer risk		Link	
2	rs1018381	(T;T)	Impaired cognitive ability			
2	rs1024611	(C;T)	Increased risk of exercise induced ischemia			Link
2	rs10248420	(A;A)	7x less likely to respond to certain antidepres...		Link	
2	rs10513789	(G;T)	Increased risk of Parkinson's disease			
2	rs1051730	(C;T)	1.3x increased risk of lung cancer	Link	Link	Link
2	rs10984447	(A;A)	>1.17x increased risk for multiple sclerosis		Link	
2	rs11190870	(C;T)	Possibly increased risk of scoliosis			
2	rs11983225	(T;T)	7x less likely to respond to certain antidepres...		Link	
2	rs12037606	(A;A)	1.52x risk of developing Crohn's disease			
2	rs1219648	(A;G)	1.20x risk for breast cancer		Link	
2	rs12696304	(C;G)	Prone to aging faster: at least in European pop...			
2	rs1333048	(A;C)	1.3x increased coronary artery disease risk			
2	rs1537415	(G;G)	2x increased risk for periodontitis		Link	
2	rs16942	(A;G)	Very slightly increased breast cancer risk	Link	Link	Link
2	rs17001266	(-;C)	1.58x increased risk for schizophrenia in males...			
2	rs17228212	(C;C)	>1.26x increased risk for heart disease		Link	
2	rs1734791	(A;A)	1.4x increased risk for lupus			
2	rs17576	(G;G)	Higher risk for lung cancer: and COPD in smoker...	Link	Link	

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
2	rs17696736	(A;G)	1.34x risk of type-1 diabetes		Link	
2	rs1800896	(A;A)	1.8x increased prostate cancer risk			
2	rs1867277	(A;A)	2x increased risk for thyroid cancer			
2	rs2143340	(C;T)	Increased risk of dyslexia and poor reading per...			
2	rs2156921	(A;G)	1.29x increased risk for depression			
2	rs2201841	(T;T)	2.4x increased risk for Graves' disease		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	rs2305795	(A;G)	1.28x higher risk of narcolepsy compared to (G;...			Link
2	rs2383206	(A;G)	1.4x increased risk for heart disease			
2	rs2383207	(A;G)	Increased risk for heart disease			
2	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	rs2619522	(G;G)	Associated with lower attention capacity but al...			
2	rs27388	(A;A)	Increased risk of developing schizophrenia			
2	rs326	(A;A)	Lower HDL cholesterol		Link	Link
2	rs358806	(C;C)	1.78x increased risk of developing Type-2 diabe...		Link	
2	rs3775948	(G;G)	Slightly higher risk for gout			
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	rs4633	(C;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;G)	Increased risk in men for biliary conditions			
2	rs587776825	(-;C)	Associated with MODY3; maturity onset of diabet...	Link		Link
2	rs629242	(C;T)	Somewhat higher risk for prostate cancer			
2	rs638405	(G;G)	2x increased ALZ risk in ApoE4 carriers	Link		
2	rs6435862	(G;G)	2.8x higher risk of aggressive neuroblastoma		Link	
2	rs6457617	(C;T)	2.3x risk of rheumatoid arthritis		Link	
2	rs6498169	(A;A)	>1.14x risk of multiple sclerosis		Link	
2	rs6710341	(G;G)	Increased risk of developing restless legs synd...			
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	rs6922269	(A;A)	1.6x risk of coronary artery disease		Link	
2	rs6997709	(G;G)	1.5x higher risk for hypertension			
2	rs699	(C;T)	Increased risk of hypertension	Link	Link	Link
2	rs7216389	(T;T)	1.5x increased risk for Childhood Asthma.		Link	
2	rs7442295	(A;A)	~4x higher risk for hyperuracemia		Link	
2	rs7536563	(A;A)	>1.12x risk of multiple sclerosis		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	rs800292	(C;C)	5% higher risk of Age related macular degenerat...	Link	Link	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	rs9525638	(T;T)	Weaker bones			
2	rs9652490	(A;A)	~2x increased risk for Parkinson's disease: and...		Link	
2.0	rs4911414	(G;T)	2-4x higher risk of sun sensitivity if part of ...		Link	
2.0	rs9642880	(T;T)	1.5x increased bladder cancer risk		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			
1.6	rs11523871	(A;C)	1.6x increased breast cancer risk for women ove...	Link	Link	
1.6	rs2046210	(T;T)	1.6x increased breast cancer risk in certain wo...		Link	Link
1.6	rs2981745	(C;T)	1.6x increased risk for breast cancer in female...			

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
1.5	rs10260404	(C;T)	1.20x risk of developing ALS		Link	
1.5	rs10464059	(A;G)	Slightly increased risk of developing Parkinson...			
1.5	rs10859871	(A;C)	Slight (~1.2x) increase in endometriosis risk			
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	rs12431733	(C;T)	Slightly increased risk of developing Parkinson...		Link	
1.5	rs12469063	(A;G)	Slightly increased risk of developing restless ...			
1.5	rs12498742	(A;A)	1.25 increased risk for gout			
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	rs1360517	(A;G)	Higher susceptibility for AIDS		Link	
1.5	rs140701	(A;A)	Increased risk for anxiety disorders			
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	rs1801020	(C;T)	1.31x increased risk of heart disease	Link		Link
1.5	rs1975197	(C;T)	1.3x increased risk of developing restless legs...		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	rs2240340	(A;G)	Slightly increased (1.5x) risk for RA	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	(A;G)	Increased risk for auto-immune diseases		Link	
1.5	rs309375	(T;T)	Larger mosquito bites			
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	rs4027132	(A;G)	1.39x increased risk of developing bipolar diso...			
1.5	rs4506565	(A;T)	1.4x increased risk for type-2 diabetes		Link	
1.5	rs4785763	(A;C)	1.5x higher risk for melanoma		Link	
1.5	rs5746059	(A;A)	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	rs6908425	(C;T)	1.63x increased risk of developing Crohn's dise...		Link	
1.5	rs699473	(C;C)	~1.5x increased brain tumor risk			
1.5	rs7454108	(C;T)	Single HLA-DQ8 haplotype			
1.5	rs7774434	(C;T)	Slightly increased risk of developing primary b...			
1.5	rs7850258	(G;G)	Slightly higher odds of developing primary hypo...			
1.5	rs872071	(G;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	rs1545843	(A;A)	1.4x increased risk for depression (for those u...			
1.4	rs2230201	(A;G)	1.4x risk of lupus	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	rs3849942	(A;A)	1.4x increased risk for ALS		Link	
1.4	rs4959039	(A;G)	1.4x higher risk for multiple sclerosis			
1.4	rs8050136	(A;A)	1.4x increased risk for T2D in some populations...		Link	
1.3	rs10947262	(C;C)	1.3x increased risk for osteoarthritis			
1.3	rs1260326	(C;T)	Slightly higher risk for gout	Link	Link	Link



Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
1.3	rs1375144	(C;T)	1.32x increased risk of developing bipolar diso...			
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	rs2736100	(G;T)	1.3x higher risk for glioma development: 2.1x r...		Link	
1.3	rs356219	(A;G)	1.3x increased risk for Parkinson's disease			
1.3	rs501120	(A;G)	1.3x increased risk for heart disease		Link	
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	rs12050604	(A;A)	Slightly increased risk for lung cancer			
1.2	rs143383	(C;T)	1.1x increased risk for osteoarthritis		Link	Link
1.2	rs1800693	(A;G)	Slight (1.2x) increase in risk for multiple scl...	Link	Link	Link
1.2	rs2056116	(A;G)	1.18x risk for breast cancer			
1.2	rs2072590	(G;T)	1.2x increased risk for ovarian cancer			
1.2	rs2252586	(A;G)	1.2x higher risk for glioma development			
1.2	rs2814707	(A;A)	>1.2x increased risk for ALS		Link	
1.2	rs3176336	(T;T)	Slightly higher (1.25x) higher risk for breast ...			
1.2	rs393152	(A;G)	Slight increased risk of both PD and AD	Link	Link	
1.2	rs4795067	(A;G)	Slight increase in risk for psoriatic arthritis...			
1.2	rs4977756	(A;G)	1.39x higher risk for glioma development		Link	
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...			
1.17	rs17465637	(A;C)	1.17x higher risk for myocardial infarction	Link	Link	
1.1	rs11037909	(C;T)	1.27x type II diabetes risk	Link		
1.1	rs11110912	(C;G)	1.3x high blood pressure risk			
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	rs34516635	(G;G)	Less longevity for Ashkenazi Jewish women.	Link		Link
1.1	rs3740878	(A;G)	1.26x type II diabetes risk	Link		Link
1.1	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			
1.1	rs997669	(G;G)	Very slightly increased (1.18x) increased breas...			
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	rs2546890	(A;G)	Higher risk of multiple sclerosis			
1	rs3194051	(A;A)	>1.1x risk of type-1 diabetes	Link	Link	Link
1	rs5326	(A;G)	Possible psychiatric risks			
1	rs6166	(G;G)	Females slightly more likely to be sterile	Link	Link	Link
1	rs6932590	(T;T)	1.1x increased risk for schizophrenia		Link	
1	rs6974491	(A;G)	Higher risk of coeliac and/or inflammatory bowe...			
0.1	rs601338	(A;G)	Susceptible to Norovirus infections	Link	Link	Link
0	rs1004819	(C;C)	1.5x risk of Crohn's disease		Link	
0	rs1061646	(C;C)	1.16x increased risk for breast cancer	Link		Link
0	rs1128503	(T;T)	Likely to require more methadone during heroin ...	Link	Link	Link
0	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	rs6277	(C;C)	1.6x higher schizophrenia risk	Link	Link	Link
0	rs6314	(C;C)	Higher risk for RA	Link	Link	
0	rs6684865	(A;A)	1.5x risk of rheumatoid arthritis			

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
0	rs7652331	(T;T)	Somewhat higher risk for prostate cancer			
0	rs7787082	(G;G)	7x less likely to respond to certain antidepres...		Link	

### 3.3 Genosets (Multi-variant Phenotypes)

Magnitude	Identifier	Summary
2.5	gs100	Lactose intolerance risk
2.5	gs155	CYP3A5 non-expressor
2.5	gs281	Part of the 88% of the population claimed not t...
2.5	gs285	You will lose 2.5x as much weight on a low fat ...
2.4	gs297	Lower heart attack risk than average
2	gs104	Restless legs syndrome risk
2	gs154	NAT2 Slow metabolizer
2	gs188	One copy of APOE4 is possible: but not certain
2	gs249	Parkinson's Disease Risk
1.5	gs186	HLA-B*5801 heterozygosity is possible: unfortun...
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 ERS1176574 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/ERS1176574>

## 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.