

# PGP-UK Genomics Report for ukAC4950

## 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	4971960
Variants filtered out	0
Novel / existing variants	491719 (9.9) / 4468685 (90.1)
Overlapped genes	56812
Overlapped transcripts	67555
Overlapped regulatory features	166800

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.

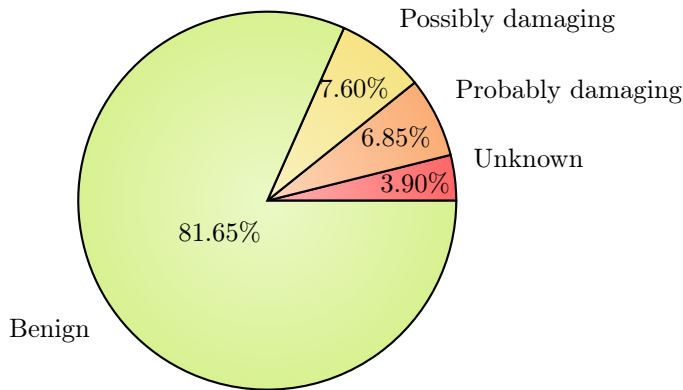


Figure 2: PolyPhen Summary

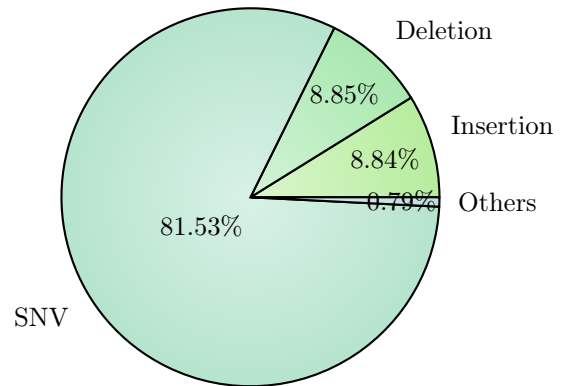


Figure 3: Variant Class

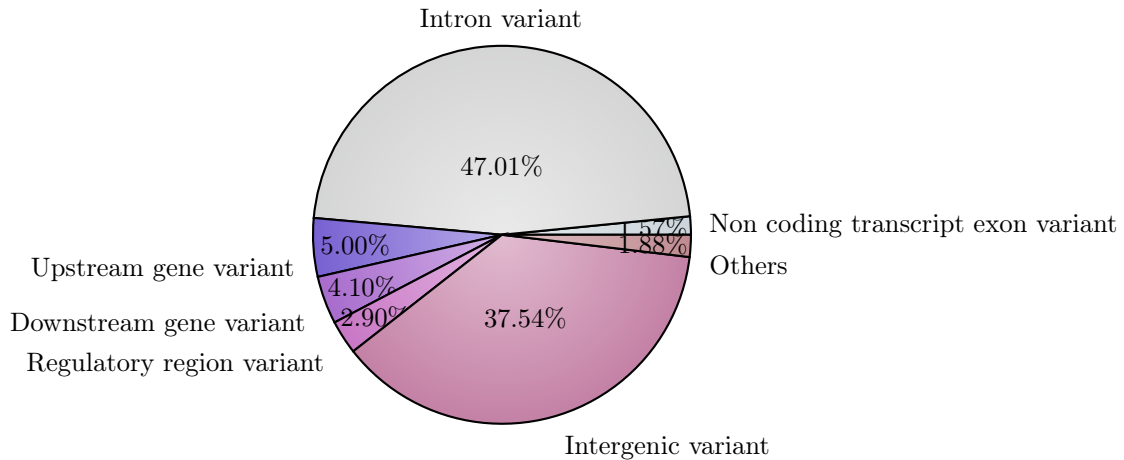


Figure 4: Consequence type

## 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).

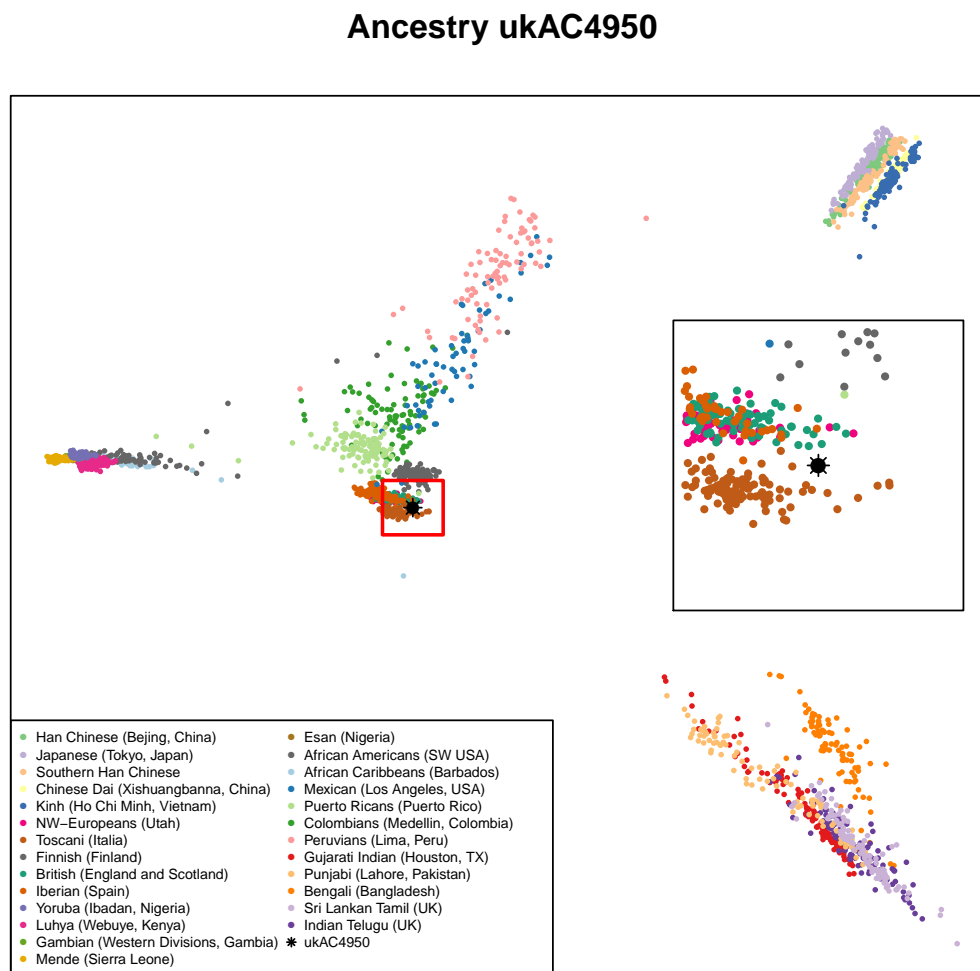


Figure 5: Ancestry Principal Component Analysis

### 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
3	rs7294919	(C;C)	Enhanced hippocampal volume			
2.5	rs3782179	(C;C)	9x lower odds of testicular cancer			
2.1	rs1136410	(C;C)	0.80x reduced risk for glioblastoma	Link	Link	
2.1	rs2511989	(A;G)	0.63x decreased age-related macular degeneratio...		Link	
2.1	rs9332739	(C;G)	0.47x decreased risk for AMD	Link	Link	Link
2.1	rs995030	(A;A)	Reduced risk of testicular cancer		Link	
2	rs1012053	(A;C)	0.625x reduced risk of Bipolar Disorder.		Link	
2	rs11045585	(A;A)	24% chance (lower than average) of docetaxel-in...		Link	
2	rs12193446	(G;G)	Lower risk of nearsightedness			
2	rs12979860	(C;C)	~80% of such hepatitis C patients respond to tr...		Link	Link
2	rs1799884	(G;G)	Mothers have typical Birth-Weight babies. Sligh...			
2	rs2241423	(A;G)	0.79 decreased risk for obesity			
2	rs261332	(A;G)	Associated with higher HDL cholesterol			
2	rs3750817	(C;T)	0.78x reduced risk for breast cancer			
2	rs3819331	(T;T)	Lower risk of autism	Link		
2	rs3914132	(C;T)	Lower otosclerosis risk		Link	
2	rs4585	(G;G)	Slightly higher (1.35x) odds of good metformin ...			
2	rs6505162	(A;C)	0.58x decreased risk for esophageal cancer	Link		
2	rs6855911	(A;G)	0.62x decreased risk for gout		Link	
2	rs763110	(C;T)	~0.80x reduced cancer risk			Link
2	rs8070723	(A;G)	0.18x reduced risk of developing progressive su...			
1.9	rs1015362	(A;A)	Probably tans instead of freckles and sunburns....		Link	
1.8	rs1746048	(C;T)	0.94 decreased risk for coronary heart disease		Link	
1.8	rs1800588	(C;T)	Higher HDL-C levels	Link	Link	
1.8	rs187238	(C;G)	Hypertension not a risk factor for sudden cardi...			
1.8	rs266729	(C;G)	0.73x decreased risk for colorectal cancer		Link	
1.8	rs3814113	(C;T)	0.8x decreased risk for ovarian cancer		Link	

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
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	rs11212617	(C;C)	Somewhat increased likelihood of treatment succ...			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;G)	0.85x decreased risk for Alzheimer's disease		Link	
1.5	rs6427528	(A;G)	For rheumatoid arthritis patients: better respo...			
1.5	rs9939609	(T;T)	Lower risk of obesity and Type-2 diabetes		Link	
1.4	rs1165205	(A;T)	0.85x decreased gout risk		Link	
1.4	rs9402571	(G;T)	Slightly decreased risk for type-2 diabetes			
1.25	rs10088218	(A;G)	0.76x decreased risk for ovarian cancer			
1.2	rs11246226	(A;C)	Decreased risk of schizophrenia in limited stud...		Link	
1.2	rs4320932	(A;G)	0.87x decreased risk for ovarian cancer			
1.2	rs9306160	(C;T)	0.75x (reduced) risk for metastasis in LN-/ER+ ...	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
1.1	rs7568369	(T;T)	0.90x reduced risk of obesity			
1	rs182549	(C;T)	Can digest milk.			Link
1	rs2351299	(G;T)	Possible reduced risk of Autism			
1	rs7850258	(A;A)	Slightly lower odds of developing primary hypot...			
0.5	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 alle...	Link		
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	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	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	rs74315403	(G;G)	PrP codon 178 (D) - non pathogenic variant			Link
0	rs7495174	(A;A)	Blue/gray eyes more likely		Link	
0	rs7997012	(A;A)	~18% more likely to respond to citalopram		Link	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	rs13266634	(C;C)	Increased risk for type-2 diabetes	Link	Link	Link
3	rs1801282	(C;G)	Unconfirmed higher risk of cardiovascular disea...	Link	Link	Link
3	rs1983132	(C;T)	2 - 3x higher prostate cancer risk if routinely...			
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	rs4151667	(A;T)	Age related macular degeneration	Link	Link	Link
3	rs6920220	(A;G)	1.2x risk Rheumatoid Arthritis		Link	
2.7	rs10830963	(C;G)	Increased type-2 diabetes risk; higher gestatio...		Link	
2.5	rs10484554	(C;T)	2.8x increased risk for psoriasis		Link	
2.5	rs16847548	(C;C)	2.6x increased risk for sudden cardiac death in...			
2.5	rs16969968	(A;G)	Slightly higher risk for nicotine dependence: l...	Link	Link	Link
2.5	rs2004640	(T;T)	1.4x increased risk for SLE		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	rs664143	(T;T)	Higher risk for number of cancers			
2.5	rs8034191	(C;T)	1.27x lung cancer risk		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	
2.3	rs37973	(G;G)	Among asthmatics: 2.3x more likely to show less...			Link
2.3	rs6025	(A;G)	Prone to thrombosis	Link	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	rs1360780	(T;T)	1.3x increased risk for depression		Link	
2.1	rs1585215	(G;G)	3.5x increased risk for Hodgkin lymphoma			
2.1	rs17070145	(C;C)	Reduced memory abilities			Link
2.1	rs17563	(C;C)	Risk for otosclerosis	Link	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	rs10086908	(C;T)	1.7x increased risk for prostate cancer			
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	rs10757272	(T;T)	1.54x increased risk for Coronary artery diseas...			
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	rs11983225	(T;T)	7x less likely to respond to certain antidepres...		Link	
2	rs1219648	(A;G)	1.20x risk for breast cancer		Link	
2	rs12469063	(G;G)	Increased risk of developing restless legs synd...			
2	rs12567232	(A;G)	Increased risk for Crohn's Disease		Link	
2	rs1265181	(C;G)	Increased risk for psoriasis		Link	
2	rs13254738	(A;C)	1.18x prostate cancer risk		Link	
2	rs17228212	(C;T)	1.26x increased risk for heart disease		Link	
2	rs1734791	(A;A)	1.4x increased risk for lupus			
2	rs17696736	(A;G)	1.34x risk of type-1 diabetes		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			

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
2	rs1801160	(A;G)	Possible 5-fluorouracil toxicity	Link	Link	Link
2	rs2073963	(G;T)	Increased risk of baldness			
2	rs2143340	(C;T)	Increased risk of dyslexia and poor reading per...			
2	rs2201841	(C;T)	1.5x increased risk for Crohn's disease; 2x inc...		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	rs2305795	(A;G)	1.28x higher risk of narcolepsy compared to (G;...			Link
2	rs241448	(C;T)	1.51x increased risk for Alzheimer's	Link		Link
2	rs2420946	(C;T)	1.20x risk for breast cancer			
2	rs25487	(G;G)	2x higher risk for skin cancer; possibly other ...	Link	Link	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	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	rs3745516	(A;A)	Increased risk of developing primary biliary ci...			
2	rs3775948	(G;G)	Slightly higher risk for gout			
2	rs3793784	(C;G)	1.5x risk for ARMD		Link	Link
2	rs3802842	(C;C)	>1.17x increased risk of colorectal cancer		Link	
2	rs4027132	(A;A)	1.51x increased risk of developing bipolar diso...			
2	rs4148739	(A;A)	7x less likely to respond to certain antidepres...		Link	
2	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	(C;T)	Higher risk for endometrial 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	rs4961	(G;T)	1.8x increased risk for high blood pressure	Link	Link	Link
2	rs4968451	(A;C)	1.61x increased risk for meningioma			
2	rs520354	(A;A)	Increased risk in men for biliary conditions			
2	rs587776825	(-,C)	Associated with MODY3; maturity onset of diabet...	Link		Link
2	rs638405	(G;G)	2x increased ALZ risk in ApoE4 carriers	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	rs6997709	(G;G)	1.5x higher risk for hypertension			
2	rs699	(C;T)	Increased risk of hypertension	Link	Link	Link
2	rs744373	(C;C)	1.17x risk of Alzheimer's			
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;G)	1.3x risk for Crohn's disease		Link	
2	rs7961152	(A;C)	1.2x higher risk for hypertension			
2	rs800292	(C;C)	5% higher risk of Age related macular degenerat...	Link	Link	Link
2	rs828907	(T;T)	Increased risk of bladder cancer and 2x risk of...			
2	rs854560	(A;T)	Higher risk for heart disease: diabetic retinop...	Link	Link	Link
2	rs9652490	(A;A)	~2x increased risk for Parkinson's disease: and...		Link	
2	rs965513	(A;A)	3.1x 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

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
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	rs1136287	(C;T)	1.5x increased risk of wet ARMD in a Taiwanese ...	Link	Link	
1.8	rs6700125	(C;T)	1.2x increased risk for ALS			
1.7	rs1042713	(A;A)	1.7x increased risk that pediatric inhaler use ...	Link	Link	Link
1.6	rs1260326	(T;T)	Slightly higher risk for gout	Link	Link	Link
1.6	rs1537415	(C;G)	1.6x increased risk for periodontitis		Link	
1.6	rs2736100	(G;G)	1.6x higher risk for glioma development		Link	
1.6	rs33980500	(C;T)	1.6x increase in risk for psoriatic arthritis	Link	Link	Link
1.5	rs10260404	(C;T)	1.20x risk of developing ALS		Link	
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	rs11171739	(C;T)	1.34x risk of developing Type-1 diabetes		Link	
1.5	rs1169300	(A;G)	~1.5x increased lung cancer risk			
1.5	rs13149290	(C;T)	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	rs16944	(A;G)	Minorly increased risk of mental illness and os...		Link	
1.5	rs17221417	(C;G)	1.3x higher risk for Crohn's disease		Link	
1.5	rs1801020	(C;T)	1.31x increased risk of heart disease	Link		Link
1.5	rs1801274	(C;T)	Complex; generally greater risk for cancer prog...	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	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	rs2280714	(A;A)	1.4x increased risk of SLE			
1.5	rs2464196	(C;T)	~1.5x increased lung cancer risk	Link	Link	Link
1.5	rs2736990	(C;T)	Slightly increased risk of developing Parkinson...		Link	
1.5	rs2881766	(T;T)	Slightly increased risk for pregnancy-induced h...			
1.5	rs3087243	(G;G)	Increased risk for autoimmune diseases		Link	
1.5	rs309375	(T;T)	Larger mosquito bites			
1.5	rs3814570	(T;T)	1.3x increased risk for Crohn's disease with il...			
1.5	rs401681	(C;T)	~1.2x increased risk for several types of cance...		Link	
1.5	rs4464148	(C;T)	1.10x increased risk for colorectal cancer			
1.5	rs464049	(C;T)	Increased risk of schizophrenia in limited stud...			
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;G)	Slightly higher fat mass			
1.5	rs6710341	(A;G)	Slightly increased risk of developing restless ...			
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	rs7774434	(C;T)	Slightly increased risk of developing primary b...			
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	
1.4	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	rs3184504	(C;T)	Slightly increased risk for celiac disease	Link	Link	
1.4	rs4959039	(A;G)	1.4x higher risk for multiple sclerosis			
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			



Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
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	rs1434536	(A;G)	1.29x increased breast cancer risk			
1.3	rs2059693	(C;T)	1.3x increased risk for testicular cancer			
1.3	rs4958847	(A;G)	1.3x increased risk for Crohn'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	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	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	rs393152	(A;G)	Slight increased risk of both PD and AD	Link	Link	
1.2	rs4686484	(A;A)	Slightly increased risk for celiac disease			
1.2	rs4795067	(A;G)	Slight increase in risk for psoriatic arthritis...			
1.2	rs6897876	(C;C)	Slight increase in testicular cancer risk for m...			
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	rs11650354	(C;T)	Possible risk for allergic asthma	Link		
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	rs2651899	(A;G)	1.1x higher risk for migraines			
1.1	rs2653349	(G;G)	2-6x increased risk for cluster headaches	Link	Link	
1.1	rs34516635	(G;G)	Less longevity for Ashkenazi Jewish women.	Link		Link
1.1	rs3818361	(C;T)	1.15x increased risk for late-onset Alzheimer's...			
1.1	rs7171755	(A;G)	Very slight decrease in cortical thickness and ...			
1.1	rs7412	(C;T)	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	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	rs12752888	(C;C)	Faster progression of mild cognitive impairment...			
1	rs2546890	(A;G)	Higher risk of multiple sclerosis			
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	
1	rs6974491	(A;G)	Higher risk of coeliac and/or inflammatory bowe...			
1	rs761100	(G;G)	Higher risk for dyslexia			
0.1	rs601338	(A;G)	Susceptible to Norovirus infections	Link	Link	Link
0	rs1061646	(C;C)	1.16x increased risk for breast cancer	Link		Link
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	rs440446	(G;G)	Increased risk in men for biliary conditions	Link		
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	gs241	Lighter green: brown or hazel eye color
2.5	gs155	CYP3A5 non-expressor
2.5	gs189	Probably APOE E2/E4: but maybe E1/E3. E1 is the...
2.5	gs282	You are part of the 12% of the population who c...
2.5	gs285	You will lose 2.5x as much weight on a low fat ...
2	gs101	Probably able to digest milk
2	gs154	NAT2 Slow metabolizer
2	gs179	CYP2D6*41
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.
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 ERS1176583 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/ERS1176583>

## 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.