

# PGP-UK Genomics Report for uk09B2EE

## 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.

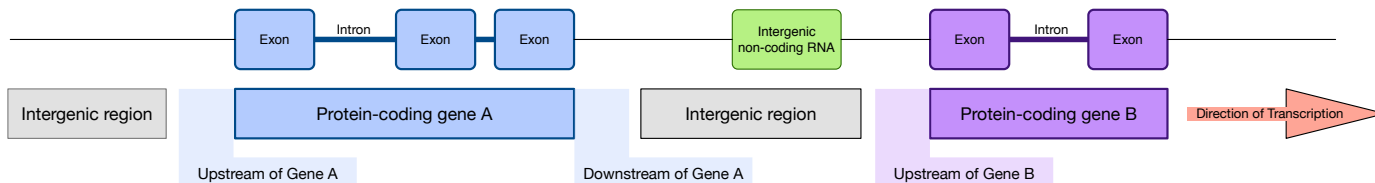


Figure 1: Diagram of gene structure indicating locations of potential variants

Feature	Count
Lines of input read	4931956
Variants filtered out	0
Novel / existing variants	466477 (9.5) / 4454901 (90.5)
Overlapped genes	56642
Overlapped transcripts	67409
Overlapped regulatory features	166517

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.

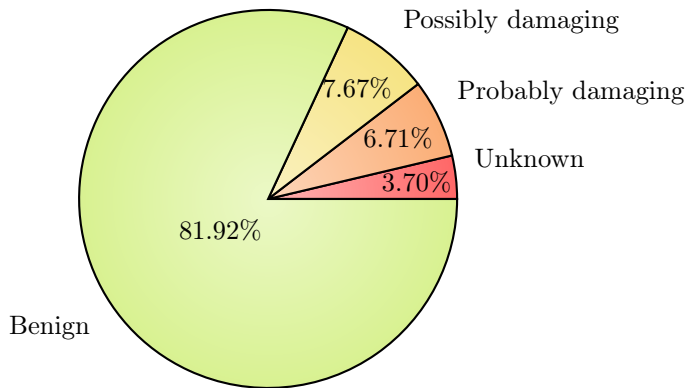


Figure 2: PolyPhen Summary

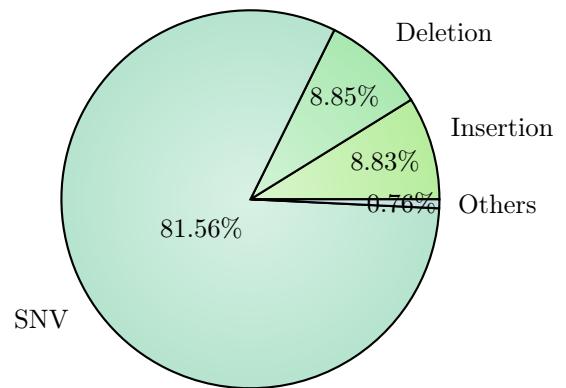


Figure 3: Variant Class

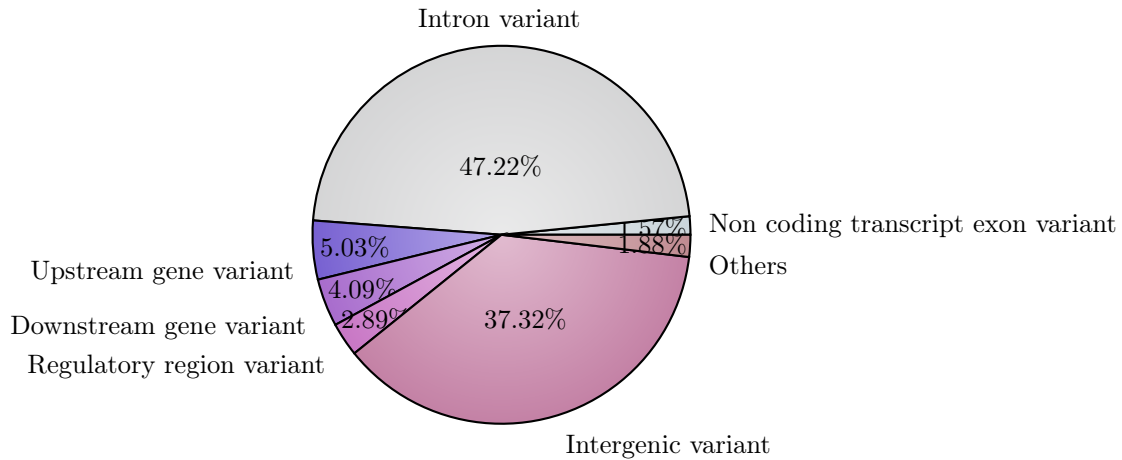


Figure 4: Consequence type

## 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).

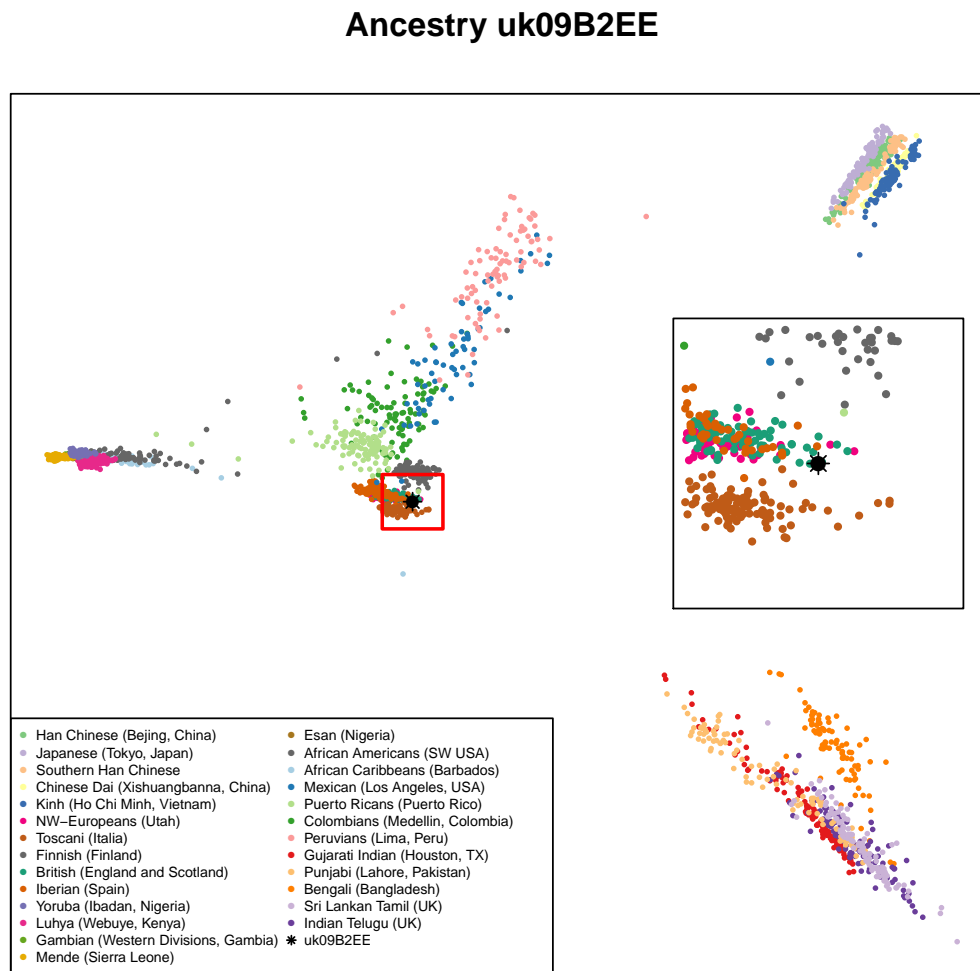


Figure 5: Ancestry Principal Component Analysis

### 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
3.5	rs11591147	(G;T)	2-3 fold lower risk of heart disease	<a href="#">Link</a>	<a href="#">Link</a>	<a href="#">Link</a>
2.5	rs3782179	(C;C)	9x lower odds of testicular cancer			
2.4	rs9272346	(G;G)	0.08x risk type-1 diabetes		<a href="#">Link</a>	
2.2	rs2511989	(A;A)	0.44x decreased age-related macular degeneratio...		<a href="#">Link</a>	
2.1	rs547154	(A;C)	0.47x decreased risk for AMD			<a href="#">Link</a>
2.1	rs995030	(A;A)	Reduced risk of testicular cancer		<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	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	rs1799884	(G;G)	Mothers have typical Birth-Weight babies. Sligh...			
2	rs1864163	(A;G)	Associated with higher HDL cholesterol		<a href="#">Link</a>	
2	rs2060793	(A;A)	Lower serum levels of vitamin D			
2	rs2073963	(T;T)	Reduced risk of baldness			
2	rs3736309	(A;G)	0.44x decreased risk for chronic obstructive pu...			
2	rs3738579	(C;T)	0.5x decreased risk for cervical cancer: HNSCC:...			
2	rs3819331	(T;T)	Lower risk of autism	<a href="#">Link</a>		
2	rs4149268	(A;G)	Associated with higher HDL cholesterol		<a href="#">Link</a>	
2	rs4585	(G;G)	Slightly higher (1.35x) odds of good metformin ...			
2	rs6505162	(A;C)	0.58x decreased risk for esophageal cancer	<a href="#">Link</a>		
2	rs6511720	(G;T)	Slightly lower odds of developing CHD.		<a href="#">Link</a>	<a href="#">Link</a>
2	rs6807362	(G;G)	Decreased autism risk	<a href="#">Link</a>	<a href="#">Link</a>	
2	rs7216389	(C;C)	0.69x lower risk of Childhood Asthma.		<a href="#">Link</a>	
2	rs763110	(C;T)	~0.80x reduced cancer risk			<a href="#">Link</a>
2	rs801114	(T;T)	0.78x decreased Basal Cell Carcinoma risk.		<a href="#">Link</a>	
1.9	rs1015362	(A;A)	Probably tans instead of freckles and sunburns....		<a href="#">Link</a>	
1.8	rs1128535	(A;G)	0.77x risk for Crohn's disease			

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
1.8	rs4714156	(C;C)	<0.61x risk for restless legs			
1.8	rs6897932	(C;T)	0.91x decreased risk for multiple sclerosis	Link	Link	Link
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	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	rs4149274	(C;T)	Associated with higher HDL (good) cholesterol			
1.5	rs4939883	(C;C)	Associated with higher HDL cholesterol		Link	
1.5	rs5888	(C;C)	Higher HDL cholesterol but lower risk for age-r...	Link		
1.5	rs610932	(A;A)	A allele associated with reduced risk of Alzhei...			
1.5	rs729302	(A;C)	0.89x decreased risk of developing rheumatoid a...			
1.5	rs9939609	(T;T)	Lower risk of obesity and Type-2 diabetes		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			
1.3	rs9306160	(T;T)	0.75x (reduced) risk for metastasis in LN-/ER+ ...	Link	Link	
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			
1.1	rs11172113	(C;T)	0.9x lower risk for migraines			
1.1	rs13333226	(A;G)	Slightly lower risk for hypertension			Link
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
1.0	rs6583817	(C;T)	~0.80x (lower) risk for late onset Alzheimer's ...			
0	rs1047781	(A;A)	ABH blood group "Secretor" status if Japanese	Link	Link	Link
0	rs12252	(T;T)	More resistant to influenza	Link		Link
0	rs16990018	(A;A)	PrP Codon 171 Asn - Non-pathogenic variant	Link		Link
0	rs17244841	(A;A)	More responsive to statin treatment		Link	Link
0	rs1799782	(C;C)	Lower risk for skin cancer	Link	Link	
0	rs1799945	(C;C)	Not a H63D 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	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	rs9394492	(C;C)	<0.76x risk for restless legs			
0	rs9951307	(A;G)	0.10 decreased risk for brain edema after a str...			

### 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	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;C)	1.18x increased risk for late-onset Alzheimer's...			
3	rs2981582	(C;T)	1.3x higher risk of ER+ breast cancer		Link	
3	rs3803662	(T;T)	1.6x increased risk for breast cancer		Link	
3	rs4244285	(A;G)	Poorer metabolizer of several popular medicines...	Link	Link	Link
3	rs891512	(A;A)	Higher blood pressure than G;G	Link		
2.8	rs3780374	(A;A)	Substantially increased odds of developing V617...			
2.6	rs110419	(A;A)	2.6x increased risk for neuroblastoma			
2.5	rs13266634	(C;T)	Increased risk for type-2 diabetes	Link	Link	Link
2.5	rs187238	(G;G)	Hypertension increases risk 3.75x for sudden ca...			
2.5	rs2241880	(C;C)	2x-3x increased risk for Crohn's disease in Cau...	Link	Link	Link
2.5	rs3738919	(C;C)	1.94x risk of developing rheumatoid arthritis			
2.5	rs664143	(T;T)	Higher risk for number of cancers			
2.5	rs795484	(A;A)	Even more increased morphine dose requirement a...			
2.5	rs9934438	(A;A)	Coumadin resistance		Link	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	rs2231137	(G;G)	~1.5-3x increased risk for ischemic stroke	Link	Link	Link
2.2	rs944289	(T;T)	1.69x increased thyroid cancer risk		Link	
2.1	rs10427255	(C;C)	Highest odds of photic sneeze reflex			
2.1	rs10811661	(T;T)	1.2x increased risk for type-2 diabetes		Link	
2.1	rs11887534	(C;G)	2x increased risk for gallstones	Link	Link	Link
2.1	rs1329428	(G;G)	2x increased risk for macular degeneration			
2.1	rs17070145	(C;C)	Reduced memory abilities			Link
2.1	rs2254958	(C;T)	1.24x increased risk for Alzheimer's			
2.1	rs2383207	(G;G)	Increased risk for heart disease			
2.1	rs380390	(C;C)	Increased risk for ARMD		Link	
2.1	rs4149056	(C;T)	Reduced breakdown of some drugs; 5x increased m...	Link	Link	Link
2.1	rs4363657	(C;T)	4.5x increased myopathy risk for statin users		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	rs1042838	(G;T)	1.28x risk for endometrial ovarian cancer; over...	Link	Link	
2	rs1045642	(C;T)	Slower metaboliser for some drugs	Link	Link	Link
2	rs10492519	(G;G)	Increased risk of developing prostate cancer			
2	rs10757272	(T;T)	1.54x increased risk for Coronary artery diseas...			
2	rs10871777	(A;G)	Adults likely to be 0.22 BMI units higher			
2	rs10984447	(A;A)	>1.17x increased risk for multiple sclerosis		Link	
2	rs11190870	(C;T)	Possibly increased risk of scoliosis			
2	rs11229030	(C;C)	Higher odds of Crohn's disease			
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	rs12696304	(C;G)	Prone to aging faster: at least in European pop...			
2	rs13254738	(C;C)	1.18x prostate cancer risk		Link	
2	rs13376333	(T;T)	~2x higher risk of atrial fibrillation		Link	
2	rs1585215	(A;G)	2x increased risk for Hodgkin lymphoma			
2	rs16942	(G;G)	Very slightly increased breast cancer risk	Link	Link	Link
2	rs16944	(A;A)	Increased risk for osteoarthritis		Link	
2	rs17228212	(C;T)	1.26x increased risk for heart disease		Link	
2	rs1734791	(A;A)	1.4x increased risk for lupus			
2	rs17435	(A;T)	1.4x increased risk for lupus			

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
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	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	(T;T)	If 4 years old or younger: ~3x increased asthma...	Link	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	rs2736100	(T;T)	Higher risk of Interstitial lung disease: and t...		Link	
2	rs3129934	(C;T)	Increased risk of Multiple Sclerosis.		Link	
2	rs3775948	(G;G)	Slightly higher risk for gout			
2	rs4148739	(A;A)	7x less likely to respond to certain antidepres...		Link	
2	rs4792311	(A;G)	Increased risk of prostate cancer	Link	Link	Link
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	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	rs587776825	(-;C)	Associated with MODY3; maturity onset of diabet...	Link		Link
2	rs6441286	(G;T)	1.54x chance of developing primary biliary cirr...		Link	
2	rs6896702	(T;T)	Increased risk of developing Parkinson's Diseas...			
2	rs6997709	(G;G)	1.5x higher risk for hypertension			
2	rs7442295	(A;A)	~4x higher risk for hyperuracemia		Link	
2	rs744373	(C;T)	1.17x risk of Alzheimer's			
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	(G;T)	Slightly increased risk of bladder cancer and 2...			
2	rs854560	(A;A)	Higher risk for heart disease: diabetic retinop...	Link	Link	Link
2	rs9303277	(T;T)	1.46x Increased risk of developing primary bili...			
2.0	rs1044396	(C;C)	Increased risk of Nicotine dependence among mal...	Link	Link	Link
2.0	rs1434536	(A;A)	1.94x increased breast cancer risk			
2.0	rs2305795	(A;A)	1.64x higher risk of narcolepsy compared to (G;...			Link
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	rs2278206	(T;T)	1.16x increased risk for asthma	Link	Link	
1.8	rs37973	(A;G)	Among asthmatics: 1.5x more likely to show less...			Link
1.8	rs6700125	(C;T)	1.2x increased risk for ALS			
1.6	rs1537415	(C;G)	1.6x increased risk for periodontitis		Link	
1.6	rs2046210	(T;T)	1.6x increased breast cancer risk in certain wo...		Link	Link
1.6	rs3764880	(A;A)	1.2 - 1.8x increased tuberculosis risk	Link	Link	
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	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	rs12498742	(A;A)	1.25 increased risk for gout			
1.5	rs13149290	(C;C)	Slightly increased risk of developing prostate ...			
1.5	rs140701	(A;G)	Increased risk for anxiety disorders			
1.5	rs144848	(G;T)	Very slightly increased breast cancer risk	Link	Link	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	rs2240340	(A;G)	Slightly increased (1.5x) risk for RA	Link		
1.5	rs27388	(A;G)	Slightly increased risk of developing schizophr...			

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
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	rs358806	(A;C)	0.86x increased risk of developing Type-2 diabe...		Link	
1.5	rs393152	(A;A)	Increased risk of both PD and AD	Link	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	rs4538475	(A;G)	Slightly increased risk of developing Parkinson...		Link	
1.5	rs464049	(T;T)	Increased risk of schizophrenia in limited stud...			
1.5	rs486907	(A;G)	1.5x increased prostate cancer risk	Link	Link	Link
1.5	rs5219	(C;T)	1.3x increased risk for type-2 diabetes	Link	Link	Link
1.5	rs6498169	(A;G)	1.14x risk of multiple sclerosis		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	rs7341475	(G;G)	1.58x increased schizophrenia risk for women		Link	
1.5	rs7536563	(A;G)	1.12x risk of multiple sclerosis		Link	
1.5	rs7850258	(G;G)	Slightly higher odds of developing primary hypo...			
1.5	rs872071	(G;G)	~1.5x increased risk for chronic lymphocytic le...		Link	
1.5	rs9561778	(G;T)	~2x increased risk of adverse drug reactions fr...		Link	
1.5	rs9642880	(G;T)	1.2x increased bladder cancer risk		Link	
1.5	rs9652490	(A;G)	Slightly increased risk of developing Parkinson...		Link	
1.4	rs1126497	(C;T)	1.4x increased risk for breast cancer	Link	Link	Link
1.4	rs3131296	(G;G)	1.4x increased risk for schizophrenia		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	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	rs1375144	(C;T)	1.32x increased risk of developing bipolar diso...			
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	rs34330	(C;T)	1.3x higher risk for endometrial cancer (in Chi...			
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	rs2295190	(T;T)	Slightly increased risk for ovarian cancer in w...	Link	Link	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	rs4496877	(T;T)	For type-1 diabetics: 1.6x increased nephropath...			
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	rs498872	(C;T)	1.2x higher risk for glioma development		Link	
1.1	rs11110912	(C;C)	1.3x high blood pressure risk			
1.1	rs13387042	(A;G)	1.12x increased risk for breast cancer		Link	
1.1	rs1344706	(G;T)	1.1x increased risk for schizophrenia		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	rs4324715	(C;T)	1.5x increased testicular cancer risk for men			
1.1	rs5030737	(C;T)	Carrier of mannose binding deficiency but of lo...	Link	Link	Link
1.1	rs688034	(C;T)	1.1x risk higher risk for coronary artery disea...		Link	
1.1	rs6897876	(C;T)	Slight increase in testicular cancer risk for m...			
1.1	rs7171755	(A;A)	Very slight decrease in cortical thickness and...			
1.1	rs7412	(C;C)	More likely to gain weight if taking olanzapine...	Link	Link	Link



Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
1.1	rs889312	(A;C)	Very slightly higher risk for breast cancer		Link	
1.1	rs925391	(C;C)	More likely to go bald; common			
1.09	rs12050604	(A;C)	Very slightly increased risk for lung cancer			
1.07	rs2291834	(C;C)	Very slightly higher risk for myocardial infarc...			
1	rs2546890	(A;A)	Higher risk of multiple sclerosis			
1	rs3194051	(A;G)	1.12x risk of type-1 diabetes	Link	Link	Link
1	rs6932590	(T;T)	1.1x increased risk for schizophrenia		Link	
0.1	rs2070744	(C;C)	Increased prostate cancer risk		Link	Link
0	rs1004819	(C;C)	1.5x risk of Crohn's disease		Link	
0	rs10761659	(A;A)	1.5x risk of Crohn's disease		Link	
0	rs1495965	(A;A)	1.2x higher risk for spondylitis			
0	rs3813929	(C;C)	Possible weight gain if taking olanzapine		Link	Link
0	rs4795400	(T;T)	If 4 years old or younger: ~2.5x increased asth...		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	gs134	Age related macular degeneration
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	gs179	CYP2D6*41
2	gs246	APOE3/APOE3
2	gs292	Possible 2x increased risk of Alzheimer's disea...
1.5	gs185	The beta blocker metoprolol is effective with 1...
1.5	gs247	Parkinson's Disease Risk
0	gs158	CYP1A2 normal metabolizer

## 4 Raw Data

The raw data used to create this report has been assigned the identifier ERS1176572 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/ERS1176572>

## 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.