

PGP-UK Genomics Report for uk3CAFB3

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.

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	5003112
Variants filtered out	0
Novel / existing variants	490013 (9.8) / 4502031 (90.2)
Overlapped genes	56730
Overlapped transcripts	67621
Overlapped regulatory features	167800

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.

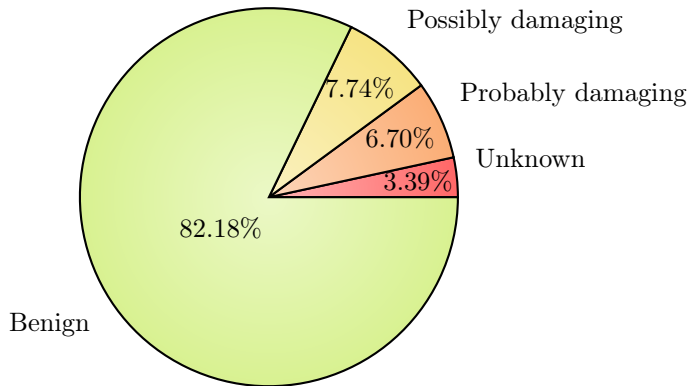


Figure 2: PolyPhen Summary

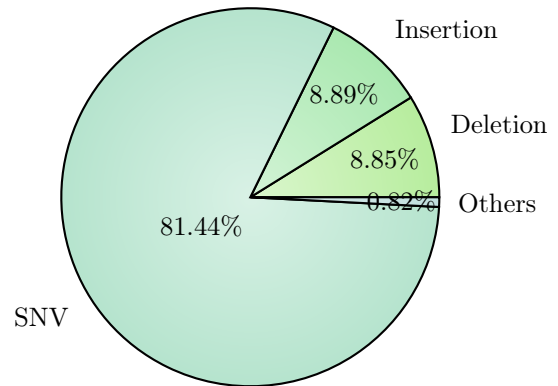


Figure 3: Variant Class

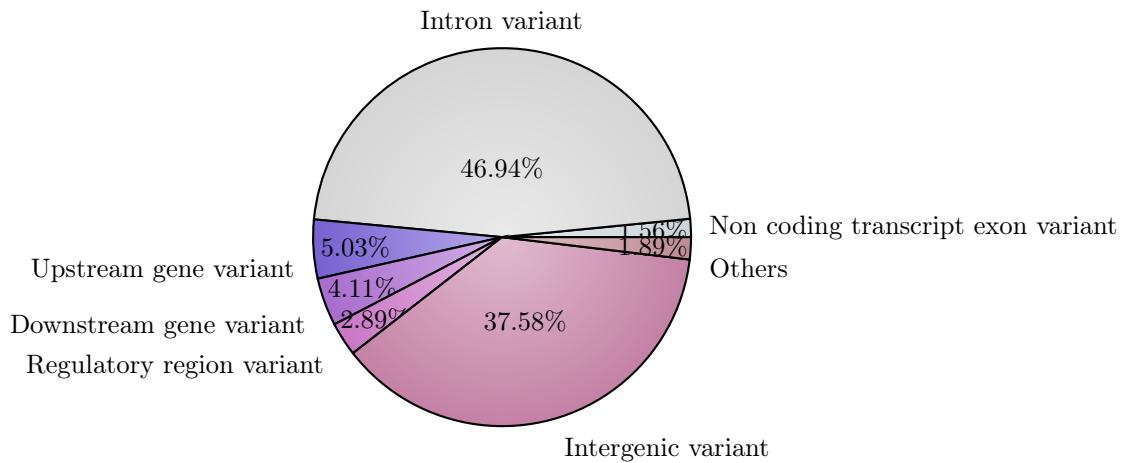


Figure 4: Consequence type

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).

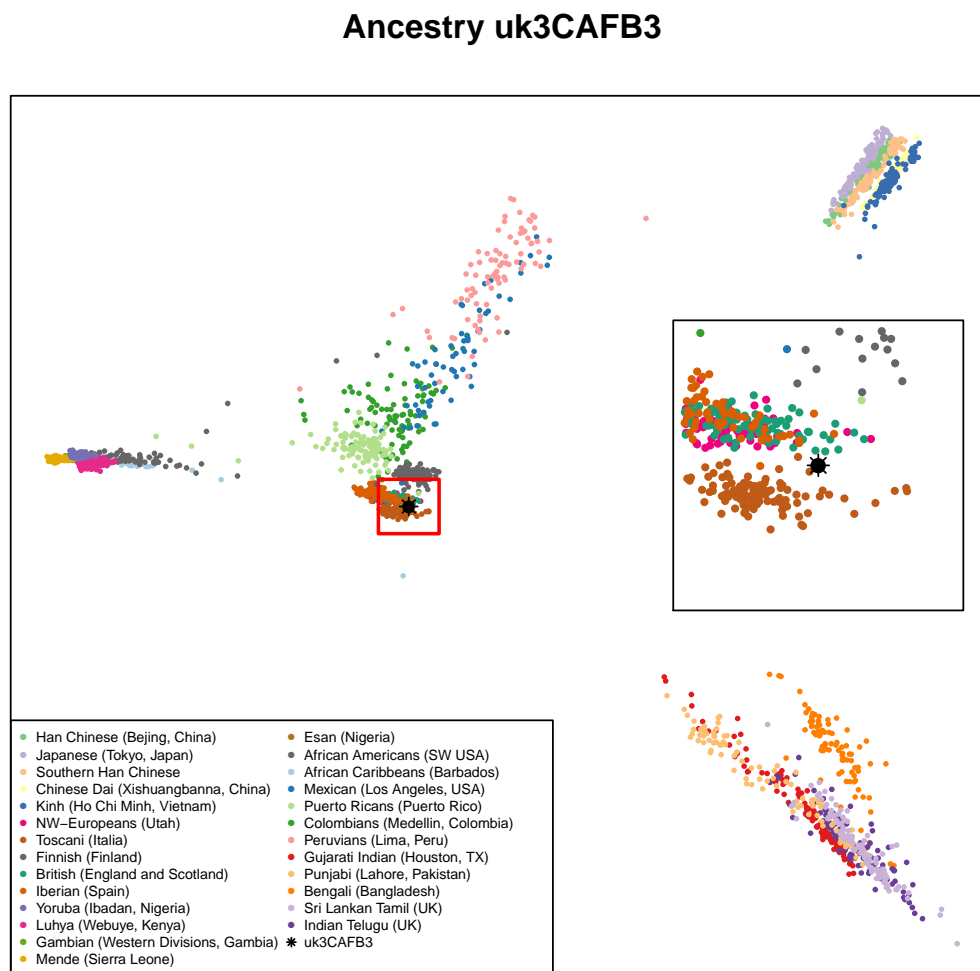


Figure 5: Ancestry Principal Component Analysis

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...		Link	
2.1	rs3775291	(A;G)	0.71x decreased risk for dry age related macula...	Link	Link	Link
2	rs10468017	(C;T)	Associated with higher HDL cholesterol		Link	
2	rs11045585	(A;A)	24% chance (lower than average) of docetaxel-in...		Link	
2	rs1160312	(G;G)	Reduced risk of Baldness.		Link	
2	rs12979860	(C;C)	~80% of such hepatitis C patients respond to tr...		Link	Link
2	rs17070145	(C;T)	Increased memory performance			Link
2	rs1799884	(G;G)	Mothers have typical Birth-Weight babies. Sligh...			
2	rs1864163	(A;G)	Associated with higher HDL cholesterol		Link	
2	rs2073963	(T;T)	Reduced risk of baldness			
2	rs2241423	(A;G)	0.79 decreased risk for obesity			
2	rs2243250	(C;T)	0.6x decreased risk for myocardial infarction i...			
2	rs2542052	(C;C)	Better odds of living to 100			
2	rs3738579	(C;T)	0.5x decreased risk for cervical cancer: HNSCC:...			
2	rs3819331	(T;T)	Lower risk of autism	Link		
2	rs4149268	(G;G)	Associated with higher HDL cholesterol		Link	
2	rs6505162	(A;C)	0.58x decreased risk for esophageal cancer	Link		
2	rs6807362	(G;G)	Decreased autism risk	Link	Link	
2	rs6855911	(A;G)	0.62x decreased risk for gout		Link	
2	rs763110	(C;T)	~0.80x reduced cancer risk			Link
2	rs7776725	(T;T)	Stronger bones		Link	
2	rs801114	(T;T)	0.78x decreased Basal Cell Carcinoma risk.		Link	
2	rs8070723	(A;G)	0.18x reduced risk of developing progressive su...			
2	rs925391	(C;T)	Lower odds of going bald			
2	rs9272346	(A;G)	0.3x risk type-1 diabetes		Link	
1.8	rs1128535	(A;G)	0.77x risk for Crohn's disease			
1.8	rs3814113	(C;T)	0.8x decreased risk for ovarian cancer		Link	

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
1.8	rs7101429	(A;G)	0.70x reduced risk for Alzheimer's risk			
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	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	rs4149274	(C;C)	Associated with higher HDL (good) cholesterol.			
1.5	rs4489954	(G;T)	0.69x risk risk of developing restless legs syn...		Link	
1.5	rs4939883	(C;C)	Associated with higher HDL cholesterol		Link	
1.5	rs6427528	(A;G)	For rheumatoid arthritis patients: better respo...			
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	rs6700125	(C;C)	0.7x decreased risk for ALS			
1.2	rs6048	(G;G)	Slightly lower risk (10-20%) of deep vein throm...	Link	Link	Link
1.1	rs11172113	(C;T)	0.9x lower risk for migraines			
1.1	rs4988235	(T;T)	Can digest milk			Link
1.1	rs7568369	(G;T)	0.90x reduced risk of obesity			
1	rs182549	(T;T)	Can digest milk.			Link
1	rs2494732	(T;T)	Lower odds of psychosis	Link	Link	
1	rs2546890	(G;G)	Lower risk of multiple sclerosis			
1	rs2952768	(C;T)	Slightly less drug dependence: decreased effect...			Link
1	rs800292	(C;T)	1% decreased risk of macular degeneration	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	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	rs5065	(A;A)	1.12x risk on diuretic; if hypertensive: better...	Link	Link	Link
0	rs7305115	(A;A)	Individuals showed a significantly lower risk o...	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
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
3.8	rs5186	(C;C)	7.3x increased risk of hypertension	Link	Link	Link
3.5	rs875858	(C;T)	Docetaxel sensitive?			
3.2	rs2981582	(T;T)	1.7x higher risk of ER+ breast cancer		Link	
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	rs16969968	(A;A)	Higher risk for nicotine dependence: lower risk...	Link	Link	Link
3	rs1983132	(C;T)	2 - 3x higher prostate cancer risk if routinely...			
3	rs2306402	(C;T)	1.18x increased risk for late-onset Alzheimer's...			
3	rs4244285	(A;G)	Poorer metabolizer of several popular medicines...	Link	Link	Link
3	rs7754840	(C;G)	1.3x increased risk for type-2 diabetes		Link	
2.7	rs10830963	(C;G)	Increased type-2 diabetes risk; higher gestatio...		Link	
2.6	rs8034191	(C;C)	1.80x lung cancer risk; decreased response to a...		Link	
2.5	rs10484554	(C;T)	2.8x increased risk for psoriasis		Link	
2.5	rs1051730	(T;T)	1.8x increased risk of lung cancer; reduced res...	Link	Link	Link
2.5	rs11190870	(T;T)	Possibly even more increased risk of scoliosis			
2.5	rs13266634	(C;T)	Increased risk for type-2 diabetes	Link	Link	Link
2.5	rs17696736	(G;G)	1.94x risk of type-1 diabetes		Link	
2.5	rs187238	(G;G)	Hypertension increases risk 3.75x for sudden ca...			
2.5	rs2943634	(C;C)	Slightly higher risk of ischemic stroke		Link	
2.5	rs339331	(T;T)	Prostate cancer risk			
2.5	rs7574865	(G;T)	1.3x risk of rheumatoid arthritis; 1.55x risk o...		Link	Link
2.5	rs891512	(A;G)	Higher blood pressure than G;G	Link		
2.4	rs1143679	(A;G)	1.78x increased risk for SLE	Link	Link	
2.3	rs37973	(G;G)	Among asthmatics: 2.3x more likely to show less...			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
2.2	rs4656461	(G;G)	2.2x increased risk for open angle glaucoma			
2.2	rs944289	(T;T)	1.69x increased thyroid cancer risk		Link	
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	rs1219648	(G;G)	1.64x risk for breast cancer		Link	
2.1	rs2254958	(C;T)	1.24x increased risk for Alzheimer's			
2.1	rs2420946	(T;T)	1.64x risk for breast cancer			
2.1	rs4444903	(G;G)	3.5x risk of hep-cancer in cirrhosis patients; ...			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	rs10306114	(A;G)	Higher risk of bleeding during coronary angiogr...			Link
2	rs1042838	(G;T)	1.28x risk for endometrial ovarian cancer; over...	Link	Link	
2	rs10759932	(C;C)	Increased risk for chorionic plate inflammation...			
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	rs11983225	(T;T)	7x less likely to respond to certain antidepres...		Link	
2	rs12431733	(T;T)	Increased risk of developing Parkinson's Diseas...		Link	
2	rs12567232	(A;G)	Increased risk for Crohn's Disease		Link	
2	rs1265181	(C;G)	Increased risk for psoriasis		Link	
2	rs1333048	(A;C)	1.3x increased coronary artery disease risk			
2	rs1585215	(A;G)	2x increased risk for Hodgkin lymphoma			
2	rs16942	(A;G)	Very slightly increased breast cancer risk	Link	Link	Link
2	rs17228212	(C;C)	>1.26x increased risk for heart disease		Link	
2	rs1734791	(A;A)	1.4x increased risk for lupus			
2	rs17782313	(C;T)	Adults likely to be 0.22 BMI units higher		Link	Link
2	rs1800896	(A;G)	1.6x increased prostate cancer risk			

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
2	rs2156921	(A;G)	1.29x increased risk for depression			
2	rs2201841	(C;T)	1.5x increased risk for Crohn's disease; 2x inc...		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	rs2383206	(A;G)	1.4x increased risk for heart disease			
2	rs2383207	(A;G)	Increased risk for heart disease			
2	rs241448	(C;T)	1.51x increased risk for Alzheimer's	Link		Link
2	rs25487	(A;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	rs3738919	(A;C)	1.94x risk of developing rheumatoid arthritis			
2	rs3775948	(G;G)	Slightly higher risk for gout			
2	rs3793784	(C;G)	1.5x risk for ARMD		Link	Link
2	rs3842787	(C;T)	Higher risk of bleeding during coronary angiogr...	Link	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	rs4402960	(G;T)	1.2x increased risk for type-2 diabetes: ~1x ri...		Link	Link
2	rs4464148	(C;C)	1.35x increased risk for colorectal cancer			
2	rs4633	(C;T)	Higher risk for endometrial cancer	Link	Link	Link
2	rs4792311	(A;G)	Increased risk of prostate cancer	Link	Link	Link
2	rs493258	(A;G)	1.15x risk of Age Related Macular Degeneration			
2	rs4968451	(A;C)	1.61x increased risk for meningioma			
2	rs520354	(A;G)	Increased risk in men for biliary conditions			
2	rs629242	(C;T)	Somewhat higher risk for prostate cancer			
2	rs6457617	(C;T)	2.3x risk of rheumatoid arthritis		Link	
2	rs6498169	(A;A)	>1.14x risk of multiple sclerosis		Link	
2	rs6601764	(C;C)	1.52x increased risk of developing Crohn's dise...		Link	
2	rs6897932	(C;C)	1.08x increased risk for multiple sclerosis	Link	Link	Link
2	rs6908425	(C;C)	1.95x increased risk of developing Crohn's dise...		Link	
2	rs6922269	(A;A)	1.6x risk of coronary artery disease		Link	
2	rs6997709	(G;T)	1.2x higher risk for hypertension			
2	rs699	(C;C)	Increased risk of hypertension	Link	Link	Link
2	rs7442295	(A;A)	~4x higher risk for hyperuracemia		Link	
2	rs744373	(C;T)	1.17x risk of Alzheimer's			
2	rs7536563	(A;A)	>1.12x risk of multiple sclerosis		Link	
2	rs7807268	(C;G)	1.3x risk for Crohn's disease		Link	
2	rs7923837	(G;G)	3.2x risk for T2D			
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	rs9525638	(T;T)	Weaker bones			
2	rs9543325	(C;C)	1.37x Slightly higher pancreatic cancer risk			
2	rs9652490	(A;A)	~2x increased risk for Parkinson's disease: and...		Link	
2.0	rs2305795	(A;A)	1.64x higher risk of narcolepsy compared to (G;...			Link
1.8	rs2278206	(T;T)	1.16x increased risk for asthma	Link	Link	
1.7	rs8055236	(G;T)	1.9x risk for heart disease		Link	
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...			
1.6	rs3764880	(A;A)	1.2 - 1.8x increased tuberculosis risk	Link	Link	
1.5	rs10464059	(A;G)	Slightly increased risk of developing Parkinson...			
1.5	rs10757272	(C;T)	1.30x increased risk for Coronary artery diseas...			

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
1.5	rs10859871	(A;C)	Slight (~1.2x) increase in endometriosis risk			
1.5	rs10883365	(A;G)	1.2x increased risk for developing Crohn's dise...		Link	
1.5	rs1169300	(A;G)	~1.5x increased lung cancer risk			
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;C)	Slightly increased risk of developing prostate ...			
1.5	rs144848	(G;T)	Very slightly increased breast cancer risk	Link	Link	Link
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	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	rs199533	(C;T)	Slightly increased risk of developing Parkinson...	Link		
1.5	rs2076295	(G;G)	Slightly increased risk for pulmonary fibrosis ...			
1.5	rs2177369	(C;C)	1.5x increased risk for Alzheimer's disease			
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	rs2286812	(C;T)	~2x higher risk for Fuchs' dystrophy: a corneal...			
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	(G;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	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	rs3814570	(C;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	rs4585	(T;T)	Slightly poorer (0.75x) response to metformin i...			
1.5	rs464049	(T;T)	Increased risk of schizophrenia in limited stud...			
1.5	rs4785763	(A;A)	2x higher risk for melanoma		Link	
1.5	rs486907	(A;G)	1.5x increased prostate cancer risk	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	rs642961	(A;G)	1.68x increased risk of cleft lip		Link	
1.5	rs6435862	(G;T)	1.7x higher risk of aggressive neuroblastoma		Link	
1.5	rs6896702	(C;T)	Slightly increased risk of developing Parkinson...			
1.5	rs6974491	(A;A)	Higher risk of coeliac and/or inflammatory bowe...			
1.5	rs7341475	(G;G)	1.58x increased schizophrenia risk for women		Link	
1.5	rs763035	(C;T)	1.2x increased risk for rosacea			
1.5	rs7850258	(G;G)	Slightly higher odds of developing primary hypo...			
1.5	rs9642880	(G;T)	1.2x increased bladder cancer risk		Link	
1.5	rs995030	(G;G)	Non-protective against testicular cancer		Link	
1.4	rs1545843	(A;A)	1.4x increased risk for depression (for those u...			
1.4	rs1800693	(G;G)	Slight (1.4x) increase in risk for multiple scl...	Link	Link	Link
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	rs6010620	(G;G)	1.4x higher risk for glioma development; but th...		Link	
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	rs110419	(A;G)	1.3x increased risk for neuroblastoma			
1.3	rs1260326	(C;T)	Slightly higher risk for gout	Link	Link	Link
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	rs1746048	(C;C)	1.03 increased risk for coronary heart disease		Link	

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
1.3	rs2024513	(A;G)	1.3x higher risk for schizophrenia (among Han C...			
1.3	rs2295490	(A;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	rs4958847	(A;G)	1.3x increased risk for Crohn's disease			
1.2	rs10865331	(A;G)	1.2x higher risk for ankylosing spondylitis			
1.2	rs143383	(C;T)	1.1x increased risk for osteoarthritis		Link	Link
1.2	rs2072590	(G;T)	1.2x increased risk for ovarian cancer			
1.2	rs393152	(A;G)	Slight increased risk of both PD and AD	Link	Link	
1.2	rs419788	(A;G)	2.0x risk for lupus	Link		
1.2	rs449647	(A;T)	Possibly lower levels of ApoE			
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...			
1.17	rs17465637	(A;C)	1.17x higher risk for myocardial infarction	Link	Link	
1.15	rs748404	(C;T)	Very slightly increased risk (1.15) for lung ca...		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	rs11650494	(A;A)	Slightly higher prostate cancer 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	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	rs3740878	(A;G)	1.26x type II diabetes risk	Link		Link
1.1	rs688034	(C;T)	1.1x risk higher risk for coronary artery disea...		Link	
1.1	rs7412	(C;C)	More likely to gain weight if taking olanzapine...	Link	Link	Link
1.1	rs889312	(C;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	rs10504861	(G;G)	Major allele: normal risk of migraine			
1	rs1143674	(A;G)	1.3x increased autism risk	Link		
1	rs3194051	(A;G)	1.12x risk of type-1 diabetes	Link	Link	Link
1	rs5326	(A;G)	Possible psychiatric risks			
1	rs6932590	(T;T)	1.1x increased risk for schizophrenia		Link	
1.0	rs11246226	(A;A)	Increased risk of schizophrenia in limited stud...		Link	
0.1	rs601338	(A;G)	Susceptible to Norovirus infections	Link	Link	Link
0	rs10239794	(T;T)	>1.3x risk for ALS			
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	rs3813929	(C;C)	Possible weight gain if taking olanzapine		Link	Link
0	rs4293393	(T;T)	1.25x Increased Risk of CKD for T allele in ...			
0	rs6314	(C;C)	Higher risk for RA	Link	Link	
0	rs7787082	(G;G)	7x less likely to respond to certain antidepres...		Link	

3.3 Genosets (Multi-variant Phenotypes)

Magnitude	Identifier	Summary
3	gs241	Lighter green: brown or hazel eye color
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	gs246	APOE3/APOE3
1.5	gs185	The beta blocker metoprolol is effective with 1...
1.5	gs247	Parkinson's Disease Risk
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 ERS1176587 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/ERS1176587>

5 Report Metadata

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

Table 5: Analysis Pipeline Versions

Report generated on August 2, 2017.