PGP-UK Genomics Report for ukBA4FF7

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.

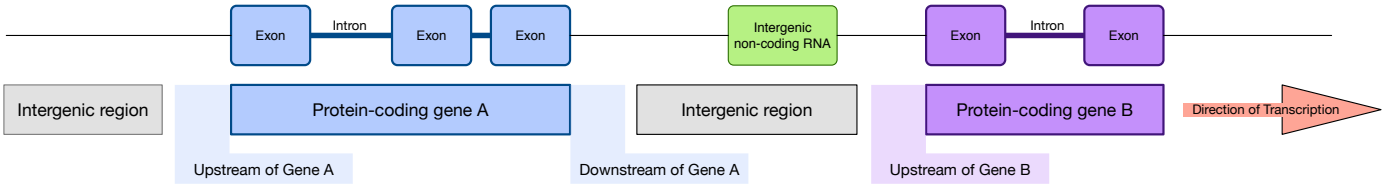

Figure 1: Diagram of gene structure indicating locations of potential variants

Feature	Count
Lines of input read	5037497
Variants filtered out	0
Novel / existing variants	507606 (10.1) / 4517929 (89.9)
Overlapped genes	56787
Overlapped transcripts	67653
Overlapped regulatory features	167887

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.

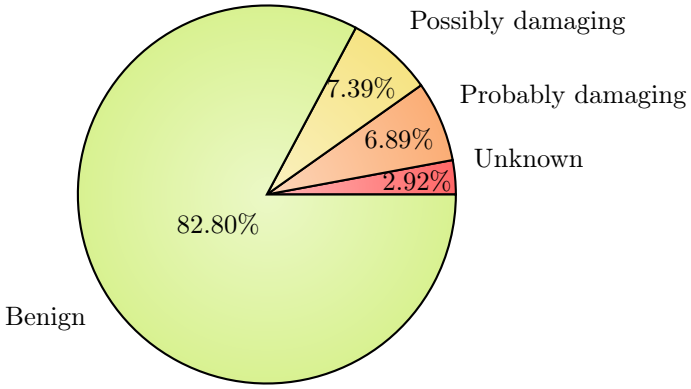

Figure 2: PolyPhen Summary

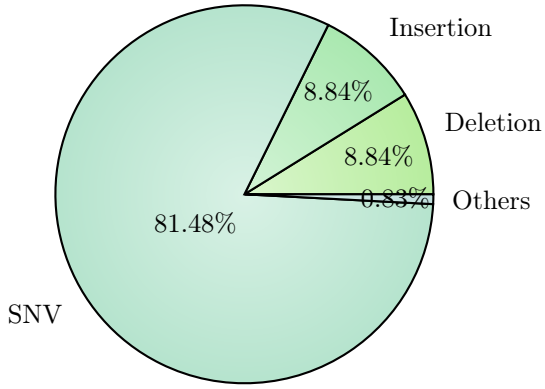

Figure 3: Variant Class

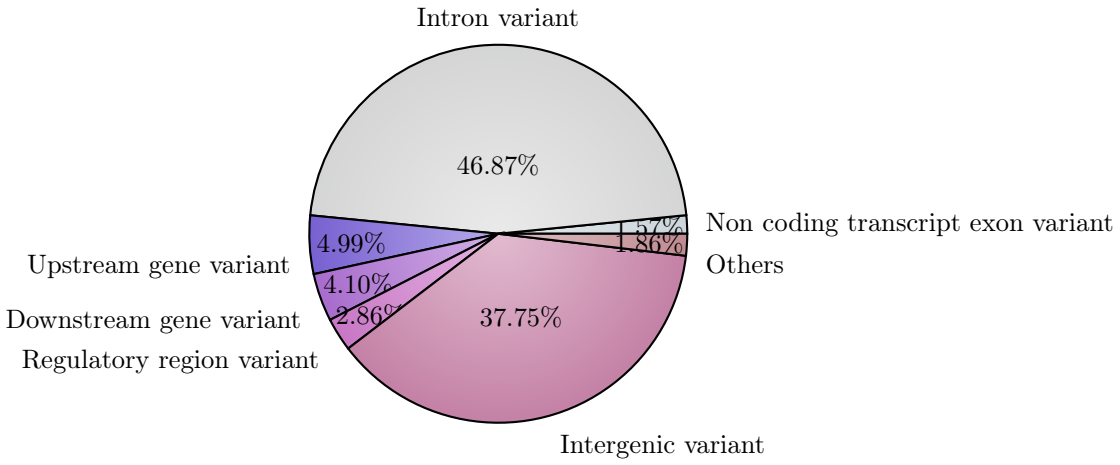

Figure 4: Consequence type

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).

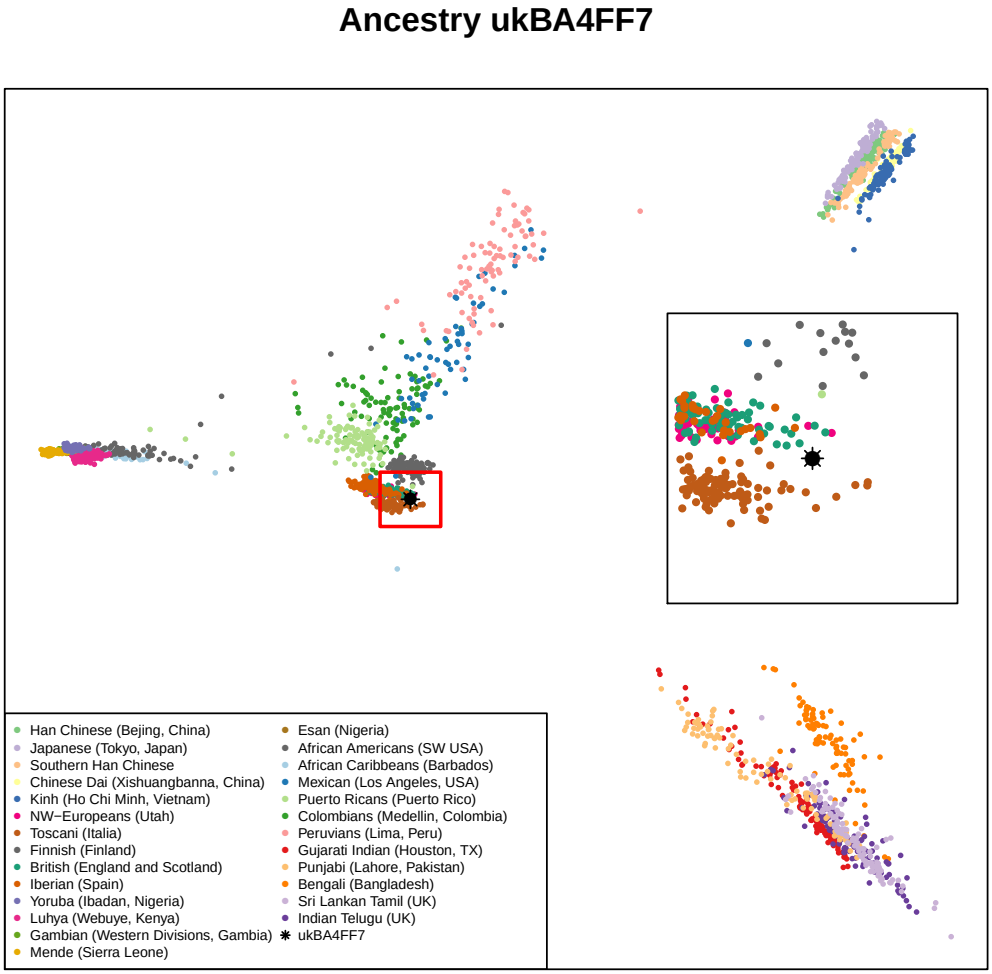

Figure 5: Ancestry Principal Component Analysis

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
2.5	rs2943634	(A;A)	Lower risk of ischemic stroke		Link	
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	rs1012053	(A;C)	0.625x reduced risk of Bipolar Disorder.		Link	
2	rs10468017	(T;T)	Associated with higher HDL cholesterol		Link	
2	rs10504861	(A;G)	Reduced risk of migraine without aura			
2	rs1136410	(C;T)	0.80x reduced risk for glioblastoma	Link	Link	
2	rs1160312	(G;G)	Reduced risk of Baldness.		Link	
2	rs1229984	(A;G)	0.56x decreased risk of oral/throat cancers	Link	Link	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	(G;G)	Associated with higher HDL cholesterol		Link	
2	rs2241423	(A;G)	0.79 decreased risk for obesity			
2	rs2292813	(C;T)	Decreased risk of autism			
2	rs2542052	(C;C)	Better odds of living to 100			
2	rs2707466	(A;A)	Stronger bones	Link	Link	
2	rs2908004	(T;T)	Stronger bones	Link	Link	
2	rs3738579	(C;T)	0.5x decreased risk for cervical cancer: HNSCC:...			
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	rs6505162	(A;C)	0.58x decreased risk for esophageal cancer	Link		
2	rs7216389	(C;C)	0.69x lower risk of Childhood Asthma.		Link	
2	rs801114	(T;T)	0.78x decreased Basal Cell Carcinoma risk.		Link	
2	rs9272346	(A;G)	0.3x risk type-1 diabetes		Link	
2	rs9642880	(G;G)	Slightly lower risk of Bladder Cancer.		Link	

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
1.8	rs1128535	(A;G)	0.77x risk for Crohn's disease			
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	
1.8	rs4714156	(C;C)	<0.61x risk for restless legs			
1.8	rs9402571	(G;G)	0.85x decreased risk for type-2 diabetes			
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			
1.5	rs1026732	(A;G)	0.70x risk for restless legs		Link	
1.5	rs11136000	(T;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	rs3851179	(A;G)	0.85x decreased risk for Alzheimer's disease		Link	
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	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	rs6700125	(C;C)	0.7x decreased risk for ALS			
1.2	rs6048	(A;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	rs11172113	(C;T)	0.9x lower risk for migraines			
1.1	rs2293347	(G;G)	Among NSCLC patients: better Gefitinib response...	Link		Link
1	rs182549	(C;T)	Can digest milk.			Link
1	rs2494732	(T;T)	Lower odds of psychosis	Link	Link	
1	rs2952768	(C;T)	Slightly less drug dependence: decreased effect...			Link
1	rs800292	(C;T)	1% decreased risk of macular degeneration	Link	Link	Link
0.1	rs891512	(G;G)	Lower blood pressure than those with an A allel...	Link		
0	rs10427255	(T;T)	Lowest odds of photic sneeze reflex			
0	rs1047781	(A;A)	ABH blood group "Secretor" status if Japanese	Link	Link	Link
0	rs1126809	(A;G)	Slight increase in skin cancer risk	Link	Link	Link
0	rs12252	(T;T)	More resistant to influenza	Link		Link
0	rs16990018	(A;A)	PrP Codon 171 Asn - Non-pathogenic variant	Link		Link
0	rs17244841	(A;A)	More responsive to statin treatment		Link	Link
0	rs1799782	(C;C)	Lower risk for skin cancer	Link	Link	
0	rs1799945	(C;C)	Not a H63D hemochromatosis carrier.	Link	Link	Link
0	rs1800562	(G;G)	Not a C282Y hemochromatosis carrier.	Link	Link	Link
0	rs28933385	(G;G)	Prion protein Codon 200 (E) - Non pathogenic va...			Link
0	rs312481	(C;C)	Better response to certain calcium channel bloc...			
0	rs5065	(A;A)	1.12x risk on diuretic; if hypertensive: better...	Link	Link	Link
0	rs6259	(G;G)	Best inverse correlation between tea-drinking: ...	Link	Link	
0	rs74315403	(G;G)	PrP codon 178 (D) - non pathogenic variant			Link
0	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	
4	rs4363657	(C;C)	17x increased myopathy risk for statin users		Link	
3	rs10897346	(C;C)	If depressed: 2.6x more likely to not respond t...			
3	rs121965063	(G;T)	Carrier of factor XI mutation	Link		Link
3	rs2237717	(T;T)	Reduced abilities related to neurocognition and...			
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	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	
2.5	rs10484554	(C;T)	2.8x increased risk for psoriasis		Link	
2.5	rs11190870	(T;T)	Possibly even more increased risk of scoliosis			
2.5	rs12803066	(A;G)	Increased risk of myopia			
2.5	rs13266634	(C;T)	Increased risk for type-2 diabetes	Link	Link	Link
2.5	rs339331	(T;T)	Prostate cancer risk			
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	rs613872	(G;T)	~5 fold higher risk for Fuchs' dystrophy: a cor...			
2.3	rs3798220	(C;T)	2-3x higher risk for cardiovascular events: whi...	Link	Link	
2.2	rs1052133	(G;G)	2x increased bladder cancer risk; 4.5x increase...	Link	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	rs2305089	(T;T)	Higher risk for chordoma	Link	Link	
2.2	rs283413	(G;T)	3x higher risk for PD	Link	Link	Link
2.2	rs964184	(G;G)	Increased risk of hypertriglyceridemia		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	rs2254958	(C;T)	1.24x increased risk for Alzheimer's			
2.1	rs2383207	(G;G)	Increased risk for heart disease			
2.1	rs241448	(C;C)	2.14x increased risk for Alzheimer's	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	
2.1	rs795484	(A;G)	Increased morphine dose requirement and postope...			
2	rs10086908	(C;T)	1.7x increased risk for prostate cancer			
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	rs1050152	(C;T)	2.1x increased risk of Crohn's disease	Link	Link	Link
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;G)	1.17x increased risk for multiple sclerosis		Link	
2	rs11045585	(A;G)	63% chance (higher than average) of docetaxel-i...		Link	
2	rs11171739	(C;C)	1.75x risk of developing Type-1 diabetes		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	rs12567232	(A;G)	Increased risk for Crohn's Disease		Link	
2	rs1265181	(C;G)	Increased risk for psoriasis		Link	
2	rs12696304	(C;G)	Prone to aging faster: at least in European pop...			
2	rs13254738	(C;C)	1.18x prostate cancer risk		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	(G;G)	Increased risk of mental disorders		Link	
2	rs1734791	(A;T)	1.4x increased risk for lupus			
2	rs17435	(A;T)	1.4x increased risk for lupus			
2	rs17576	(A;G)	Higher risk for MI and lung cancer: and COPD in...	Link	Link	

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
2	rs17696736	(A;G)	1.34x risk of type-1 diabetes		Link	
2	rs17782313	(C;T)	Adults likely to be 0.22 BMI units higher		Link	Link
2	rs1799732	(-;C)	1.3x increased adenoma recurrence risk		Link	
2	rs1800629	(A;G)	Complex; generally higher risk for certain dise...	Link	Link	Link
2	rs1800896	(A;G)	1.6x increased prostate cancer risk			
2	rs1867277	(A;A)	2x increased risk for thyroid cancer			
2	rs2073963	(G;T)	Increased risk of baldness			
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	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	rs2305795	(A;G)	1.28x higher risk of narcolepsy compared to (G;...			Link
2	rs2352028	(T;T)	Increased risk of lung cancer in non-smokers an...		Link	
2	rs2420946	(C;T)	1.20x risk for breast cancer			
2	rs2464196	(T;T)	~2x increased lung cancer risk	Link	Link	Link
2	rs25487	(G;G)	2x higher risk for skin cancer; possibly other ...	Link	Link	Link
2	rs2736990	(C;C)	Increased risk of developing Parkinson's Diseas...		Link	
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	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	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	rs4444903	(A;G)	3.5x risk of hep-cancer in cirrhosis patients; ...			Link
2	rs5174	(A;G)	1.3x increased risk for heart disease	Link	Link	Link
2	rs520354	(A;A)	Increased risk in men for biliary conditions			
2	rs629242	(C;T)	Somewhat higher risk for prostate cancer			
2	rs6441286	(G;T)	1.54x chance of developing primary biliary cirr...		Link	
2	rs6457617	(C;T)	2.3x risk of rheumatoid arthritis		Link	
2	rs6498169	(A;A)	>1.14x risk of multiple sclerosis		Link	
2	rs6807362	(C;C)	Increased autism risk	Link	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	rs6908425	(C;C)	1.95x increased risk of developing Crohn's dise...		Link	
2	rs6997709	(G;T)	1.2x higher risk for hypertension			
2	rs699	(C;T)	Increased risk of hypertension	Link	Link	Link
2	rs7442295	(A;A)	~4x higher risk for hyperuracemia		Link	
2	rs763361	(T;T)	Increased risk for multiple autoimmune diseases...	Link	Link	
2	rs7774434	(C;C)	Increased risk of developing primary biliary ci...			
2	rs7776725	(C;C)	Weaker bones		Link	
2	rs7794745	(T;T)	Slightly increased risk for autism		Link	Link
2	rs7807268	(C;G)	1.3x risk for Crohn's disease		Link	
2	rs7923837	(G;G)	3.2x risk for T2D			
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	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	rs9954153	(G;T)	~2.5x higher risk for Fuchs' dystrophy: a corne...			
2.0	rs4911414	(T;T)	2-4x higher risk of sun sensitivity if part of ...		Link	

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
1.8	rs2278206	(T;T)	1.16x increased risk for asthma	Link	Link	
1.6	rs1260326	(T;T)	Slightly higher risk for gout	Link	Link	Link
1.6	rs356219	(G;G)	1.6x increased risk for Parkinson's disease			
1.6	rs3764880	(A;A)	1.2 - 1.8x increased tuberculosis risk	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	rs10859871	(A;C)	Slight (~1.2x) increase in endometriosis risk			
1.5	rs1169300	(A;G)	~1.5x increased lung cancer risk			
1.5	rs12037606	(A;G)	1.22x risk of developing Crohn's disease			
1.5	rs1223271	(A;G)	Slightly increased risk of developing Parkinson...		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	rs2007153	(G;G)	Increased risk of schizophrenia in limited stud...			
1.5	rs2272127	(C;C)	Associated with herpes and schizophrenia			
1.5	rs2280714	(A;A)	1.4x increased risk of SLE			
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	rs356220	(T;T)	Increased risk of Parkinson's Disease			
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	rs393152	(A;A)	Increased risk of both PD and AD	Link	Link	
1.5	rs401681	(C;C)	~1.2x increased risk for several types of cance...		Link	
1.5	rs4464148	(C;T)	1.10x increased risk for colorectal cancer			
1.5	rs4538475	(A;G)	Slightly increased risk of developing Parkinson...		Link	
1.5	rs4585	(T;T)	Slightly poorer (0.75x) response to metformin i...			
1.5	rs464049	(C;T)	Increased risk of schizophrenia in limited stud...			
1.5	rs4656461	(A;G)	1.5x increased risk for open angle glaucoma			
1.5	rs4785763	(A;C)	1.5x higher risk for melanoma		Link	
1.5	rs4845618	(G;T)	1.7x increased melanoma risk			
1.5	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	rs5746059	(A;A)	Slightly higher fat mass			
1.5	rs6532197	(A;G)	Slightly increased risk of developing Parkinson...		Link	
1.5	rs6601764	(C;T)	1.16x increased risk of developing Crohn's dise...		Link	
1.5	rs699473	(C;T)	~1.5x increased brain tumor risk			
1.5	rs7850258	(G;G)	Slightly higher odds of developing primary hypo...			
1.5	rs995030	(G;G)	Non-protective against testicular cancer		Link	
1.4	rs1126497	(T;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	rs3184504	(C;T)	Slightly increased risk for celiac disease	Link	Link	
1.4	rs3849942	(A;A)	1.4x increased risk for ALS		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	rs1047031	(A;A)	1.3x increased risk for periodontitis	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	rs1375144	(C;T)	1.32x increased risk of developing bipolar diso...			
1.3	rs1434536	(A;G)	1.29x increased breast cancer risk			
1.3	rs1746048	(C;C)	1.03 increased risk for coronary heart disease		Link	
1.3	rs2059693	(C;T)	1.3x increased risk for testicular cancer			
1.3	rs2736100	(G;T)	1.3x higher risk for glioma development: 2.1x r...		Link	
1.3	rs34330	(C;T)	1.3x higher risk for endometrial cancer (in Chi...			

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
1.25	rs13387042	(A;A)	1.24x increased risk for breast 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	rs1800693	(A;G)	Slight (1.2x) increase in risk for multiple scl...	Link	Link	Link
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	rs2651899	(G;G)	1.2x higher risk for migraines			
1.2	rs2814707	(A;A)	>1.2x increased risk for ALS		Link	
1.2	rs3740878	(A;A)	1.46x type II diabetes risk; common	Link		Link
1.2	rs419788	(A;G)	2.0x risk for lupus	Link		
1.2	rs4795067	(A;G)	Slight increase in risk for psoriatic arthritis...			
1.2	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...			
1.2	rs9858542	(A;G)	1.1x risk Crohn's Disease	Link	Link	
1.2	rs9960767	(A;C)	1.2x increased risk for schizophrenia		Link	
1.15	rs748404	(C;T)	Very slightly increased risk (1.15) for lung ca...		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	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	rs7171755	(A;G)	Very slight decrease in cortical thickness and ...			
1.1	rs7412	(C;C)	More likely to gain weight if taking olanzapine...	Link	Link	Link
1.1	rs889312	(A;C)	Very slightly higher risk for breast cancer		Link	
1.1	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	rs1143674	(A;G)	1.3x increased autism risk	Link		
1	rs2282679	(A;C)	Somewhat lower vitamin D levels			
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	rs987525	(A;C)	2.5x increased risk for cleft lip		Link	
1.0	rs11246226	(A;A)	Increased risk of schizophrenia in limited stud...		Link	
0.1	rs601338	(G;G)	Susceptible to Norovirus infections	Link	Link	Link
0	rs10761659	(A;A)	1.5x risk of Crohn's disease		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	rs4795400	(T;T)	If 4 years old or younger: ~2.5x increased asth...		Link	
0	rs6277	(C;C)	1.6x higher schizophrenia risk	Link	Link	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
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	gs101	Probably able to digest milk
2	gs173	CYP2D6*10
2	gs179	CYP2D6*41
2	gs239	Reduced conversion of beta-carotene to retinol
2	gs246	APOE3/APOE3
1.5	gs247	Parkinson's Disease Risk
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 ERS1176571 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/ERS1176571

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.