

PGP-UK Genomics Report for ukA41CB7

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.

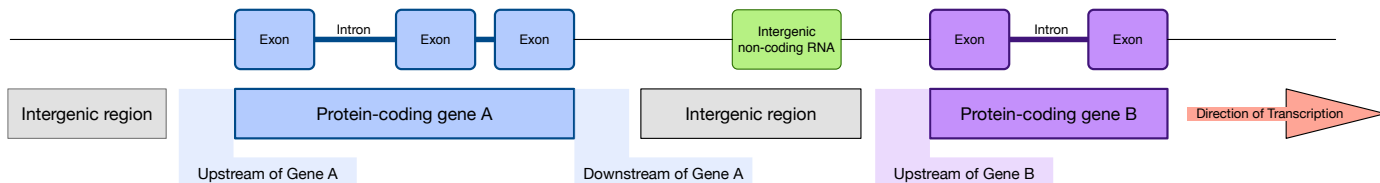


Figure 1: Diagram of gene structure indicating locations of potential variants

Feature	Count
Lines of input read	4947763
Variants filtered out	0
Novel / existing variants	495423 (10.0) / 4439640 (90.0)
Overlapped genes	56795
Overlapped transcripts	67649
Overlapped regulatory features	166681

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.

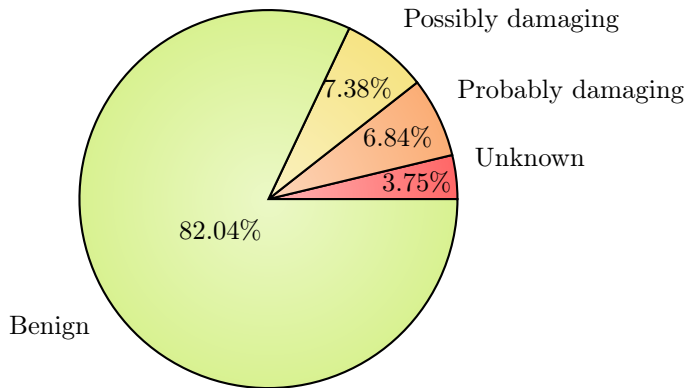


Figure 2: PolyPhen Summary

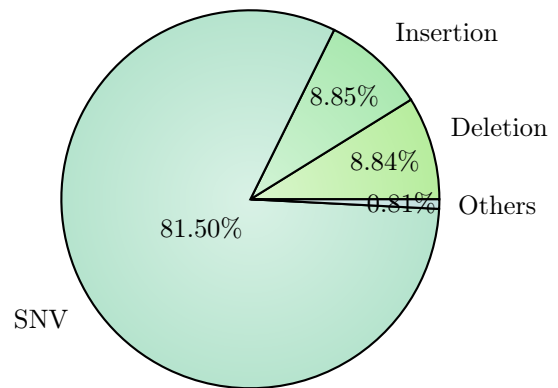


Figure 3: Variant Class

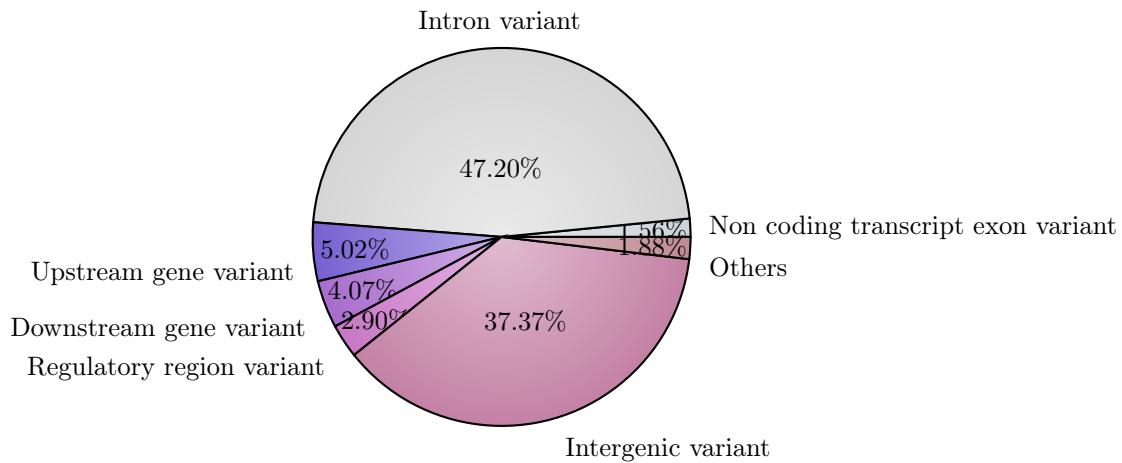


Figure 4: Consequence type

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).

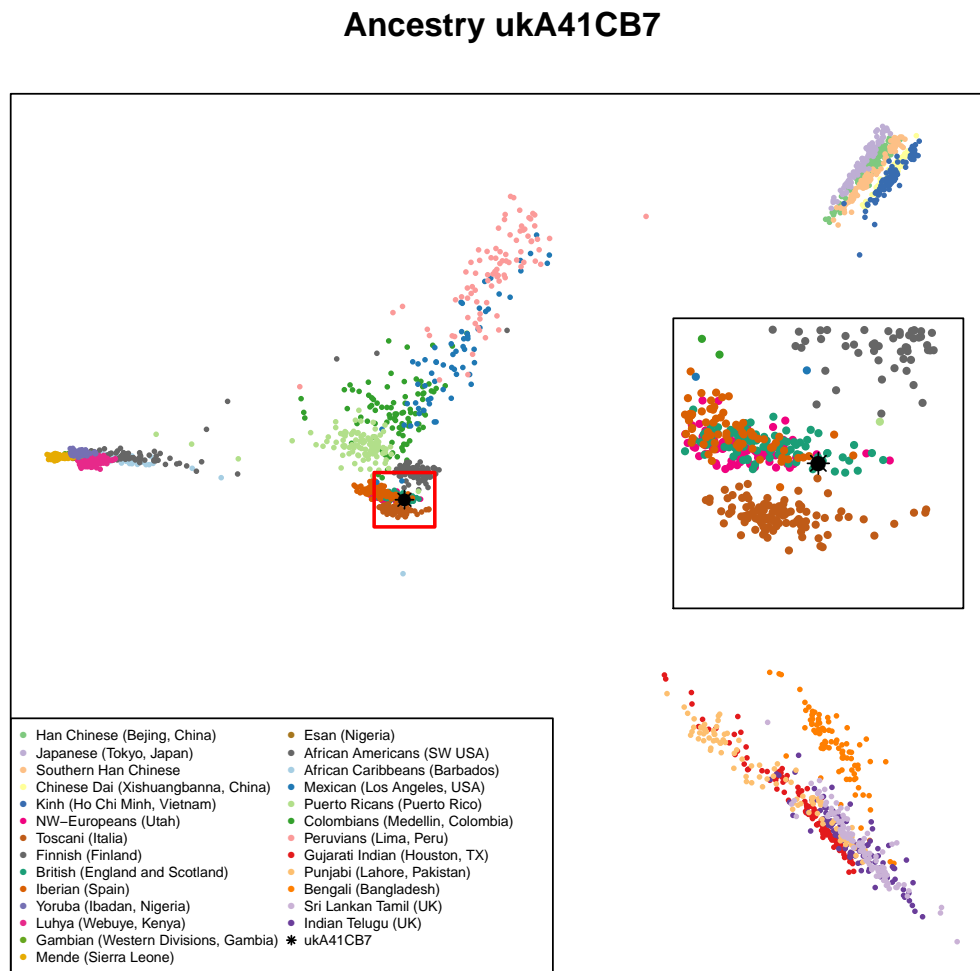


Figure 5: Ancestry Principal Component Analysis

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	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	rs11045585	(A;A)	24% chance (lower than average) of docetaxel-in...		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	rs2235015	(T;T)	Somewhat more likely to respond to certain anti...	Link	Link	
2	rs2542052	(C;C)	Better odds of living to 100			
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	rs4149268	(A;G)	Associated with higher HDL cholesterol		Link	
2	rs4585	(G;G)	Slightly higher (1.35x) odds of good metformin ...			
2	rs6807362	(G;G)	Decreased autism risk	Link	Link	
2	rs6855911	(A;G)	0.62x decreased risk for gout		Link	
2	rs7216389	(C;C)	0.69x lower risk of Childhood Asthma.		Link	
2	rs7776725	(T;T)	Stronger bones		Link	
2	rs800292	(T;T)	5% decreased risk of macular degeneration	Link	Link	Link
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	rs1746048	(C;T)	0.94 decreased risk for coronary heart disease		Link	
1.8	rs187238	(C;G)	Hypertension not a risk factor for sudden cardi...			
1.6	rs1061170	(T;T)	Lower risk for AMD: generally longer live than ...	Link	Link	Link
1.6	rs2278206	(C;C)	Possibly less susceptible to asthma	Link	Link	
1.5	rs1063192	(C;T)	0.71x reduced risk of myocardial infarction			

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
1.5	rs11136000	(T;T)	0.84x decreased risk for Alzheimer's disease		Link	
1.5	rs11212617	(C;C)	Somewhat increased likelihood of treatment succ...			Link
1.5	rs4149274	(C;T)	Associated with higher HDL (good) cholesterol			
1.5	rs464049	(C;C)	Decreased risk of schizophrenia in limited stud...			
1.5	rs4939883	(C;T)	Associated with higher HDL cholesterol		Link	
1.5	rs729302	(A;C)	0.89x decreased risk of developing rheumatoid a...			
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			
1.2	rs11172113	(C;C)	0.8x lower risk for migraines			
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	rs6048	(G;G)	Slightly lower risk (10-20%) of deep vein throm...	Link	Link	Link
1.2	rs9306160	(C;T)	0.75x (reduced) risk for metastasis in LN-/ER+ ...	Link	Link	
1.1	rs10166942	(C;T)	0.85x lower risk for migraines			
1.1	rs2293347	(G;G)	Among NSCLC patients: better Gefitinib response...	Link		Link
1.1	rs4988235	(T;T)	Can digest milk			Link
1	rs10248420	(A;G)	7x more likely to respond to certain antidepres...		Link	
1	rs11983225	(C;T)	7x more likely to respond to certain antidepres...		Link	
1	rs182549	(T;T)	Can digest milk.			Link
1	rs2235040	(A;G)	7x more likely to respond to certain antidepres...	Link	Link	
1	rs2235067	(A;G)	7x more likely to respond to certain antidepres...			
1	rs2351299	(G;T)	Possible reduced risk of Autism			
1	rs4148739	(A;G)	7x more likely to respond to certain antidepres...		Link	
1	rs7850258	(A;G)	Typical odds of developing primary hypothyroidi...			
1.0	rs2283123	(C;T)	Decreased risk of schizophrenia in limited stud...			
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	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	rs1799945	(C;C)	Not a H63D hemochromatosis carrier.	Link	Link	Link
0	rs1800562	(G;G)	Not a C282Y hemochromatosis carrier.	Link	Link	Link
0	rs242941	(G;G)	Better response to inhaled corticosteroid in pa...		Link	
0	rs28933385	(G;G)	Prion protein Codon 200 (E) - Non pathogenic va...			Link
0	rs312481	(C;C)	Better response to certain calcium channel bloc...			
0	rs5065	(A;A)	1.12x risk on diuretic; if hypertensive: better...	Link	Link	Link
0	rs6259	(G;G)	Best inverse correlation between tea-drinking: ...	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	

3.2 Possibly Harmful Traits

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
3.5	rs875858	(C;T)	Docetaxel sensitive?			
3	rs1021737	(T;T)	Significantly higher plasma total homocysteine ...	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	rs2066845	(C;G)	3x higher risk for Crohn's disease	Link		Link
3	rs2306402	(C;T)	1.18x increased risk for late-onset Alzheimer's...			
3	rs2981582	(C;T)	1.3x higher risk of ER+ breast cancer		Link	
2.5	rs1121980	(C;T)	1.67x risk for obesity		Link	
2.5	rs1421085	(C;T)	~1.3x increased obesity risk		Link	Link
2.5	rs16969968	(A;G)	Slightly higher risk for nicotine dependence: l...	Link	Link	Link
2.5	rs2241880	(C;C)	2x-3x increased risk for Crohn's disease in Cau...	Link	Link	Link
2.5	rs2254958	(C;C)	1.61x increased risk for Alzheimer's			
2.5	rs2943634	(C;C)	Slightly higher risk of ischemic stroke		Link	
2.5	rs339331	(T;T)	Prostate cancer risk			
2.5	rs5219	(T;T)	2.5x increased risk for type-2 diabetes	Link	Link	Link
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...			
2.5	rs664143	(T;T)	Higher risk for number of cancers			
2.5	rs7574865	(G;T)	1.3x risk of rheumatoid arthritis; 1.55x risk o...		Link	Link
2.5	rs8034191	(C;T)	1.27x lung cancer risk		Link	
2.5	rs891512	(A;G)	Higher blood pressure than G;G	Link		
2.4	rs7966230	(G;G)	Slightly lower levels of plasma VWF			
2.2	rs2231137	(G;G)	~1.5-3x increased risk for ischemic stroke	Link	Link	Link
2.2	rs3129934	(T;T)	3.3x increased risk for multiple sclerosis		Link	
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	rs2494732	(C;C)	Greater odds of cannabis-associated psychosis	Link	Link	
2.1	rs4693596	(C;C)	2x odds of myopathy if taking statins			
2	rs10086908	(C;T)	1.7x increased risk for prostate cancer			
2	rs10260404	(C;C)	1.60x risk of developing ALS		Link	
2	rs10455872	(A;G)	1:51x increased Coronary Heart disease risk			Link
2	rs1050152	(C;T)	2.1x increased risk of Crohn's disease	Link	Link	Link
2	rs1051730	(C;T)	1.3x increased risk of lung cancer	Link	Link	Link
2	rs10980705	(T;T)	3.7x increased risk for knee osteoarthritis			
2	rs10984447	(A;A)	>1.17x increased risk for multiple sclerosis		Link	
2	rs10994336	(C;T)	1.45x increased odds of developing bipolar diso...		Link	
2	rs11190870	(C;T)	Possibly increased risk of scoliosis			
2	rs1160312	(A;G)	1.6x increased risk of Male Pattern Baldness.		Link	
2	rs1219648	(A;G)	1.20x risk for breast cancer		Link	
2	rs12431733	(T;T)	Increased risk of developing Parkinson's Diseas...		Link	
2	rs12567232	(A;A)	Increased risk for Crohn's Disease		Link	
2	rs12696304	(C;G)	Prone to aging faster: at least in European pop...			
2	rs1333048	(A;C)	1.3x increased coronary artery disease risk			
2	rs1360780	(C;T)	1.3x increased risk for depression		Link	
2	rs1544410	(A;A)	Increased risk of low bone mineral density diso...		Link	
2	rs1585215	(A;G)	2x increased risk for Hodgkin lymphoma			
2	rs1691053	(A;G)	Increased risk of developing prostate cancer			
2	rs16942	(A;G)	Very slightly increased breast cancer risk	Link	Link	Link
2	rs16944	(G;G)	Increased risk of mental disorders		Link	
2	rs17228212	(C;C)	>1.26x increased risk for heart disease		Link	
2	rs1734791	(A;A)	1.4x increased risk for lupus			
2	rs17576	(A;G)	Higher risk for MI and lung cancer: and COPD in...	Link	Link	
2	rs1800896	(A;G)	1.6x increased prostate cancer risk			

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
2	rs2073963	(G;T)	Increased risk of baldness			
2	rs2143340	(C;T)	Increased risk of dyslexia and poor reading per...			
2	rs2201841	(C;C)	1.5x increased risk for Crohn's disease		Link	
2	rs2230201	(G;G)	>1.4x risk of lupus	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	rs2383206	(A;G)	1.4x increased risk for heart disease			
2	rs2383207	(A;G)	Increased risk for heart disease			
2	rs2420946	(C;T)	1.20x risk for breast cancer			
2	rs25487	(A;G)	2x higher risk for skin cancer; possibly other ...	Link	Link	Link
2	rs2697962	(A;A)	Increased risk of developing Parkinson's Diseas...			
2	rs351855	(C;T)	1.2x increased risk for prostate cancer	Link	Link	Link
2	rs3738919	(A;C)	1.94x risk of developing rheumatoid arthritis			
2	rs3745516	(A;A)	Increased risk of developing primary biliary ci...			
2	rs4027132	(A;A)	1.51x increased risk of developing bipolar diso...			
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	rs4961	(G;T)	1.8x increased risk for high blood pressure	Link	Link	Link
2	rs5174	(A;G)	1.3x increased risk for heart disease	Link	Link	Link
2	rs6498169	(A;A)	>1.14x risk of multiple sclerosis		Link	
2	rs6897932	(C;C)	1.08x increased risk for multiple sclerosis	Link	Link	Link
2	rs6997709	(G;T)	1.2x higher risk for hypertension			
2	rs7190458	(A;G)	Slightly higher pancreatic cancer risk	Link		
2	rs744373	(C;T)	1.17x risk of Alzheimer's			
2	rs7639618	(C;T)	1.45x increased osteoarthritis risk	Link		
2	rs7807268	(C;G)	1.3x risk for Crohn's disease		Link	
2	rs7961152	(A;C)	1.2x higher risk for hypertension			
2	rs828907	(G;T)	Slightly increased risk of bladder cancer and 2...			
2	rs854560	(A;T)	Higher risk for heart disease: diabetic retinop...	Link	Link	Link
2	rs9303277	(T;T)	1.46x Increased risk of developing primary bili...			
2	rs9525638	(T;T)	Weaker bones			
2	rs965513	(A;G)	1.77x increased thyroid cancer risk		Link	
2	rs9954153	(G;T)	~2.5x higher risk for Fuchs' dystrophy: a corne...			
2.0	rs2156921	(G;G)	1.29x increased risk for depression			
2.0	rs9642880	(T;T)	1.5x increased bladder cancer risk		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	rs143383	(T;T)	1.3x increased risk for osteoarthritis		Link	Link
1.8	rs6700125	(C;T)	1.2x increased risk for ALS			
1.8	rs733618	(A;G)	1.87x risk for myasthenia gravis			
1.7	rs2024513	(A;A)	1.7x higher risk for schizophrenia (among Han C...			
1.6	rs11523871	(A;C)	1.6x increased breast cancer risk for women ove...	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	
1.6	rs3775948	(C;G)	Slightly higher risk for gout			
1.5	rs10492519	(A;G)	Slightly increased risk of developing prostate ...			
1.5	rs10757272	(C;T)	1.30x increased risk for Coronary artery diseas...			
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	rs1223271	(A;G)	Slightly increased risk of developing Parkinson...		Link	
1.5	rs13149290	(C;C)	Slightly increased risk of developing prostate ...			
1.5	rs1360517	(A;G)	Higher susceptibility for AIDS		Link	
1.5	rs140701	(A;A)	Increased risk for anxiety disorders			

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
1.5	rs144848	(G;T)	Very slightly increased breast cancer risk	Link	Link	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	rs2007153	(G;G)	Increased risk of schizophrenia in limited stud...			
1.5	rs2240340	(A;G)	Slightly increased (1.5x) risk for RA	Link		
1.5	rs2272127	(C;C)	Associated with herpes and schizophrenia			
1.5	rs2280714	(A;G)	1.4x increased risk of SLE			
1.5	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	rs27388	(A;G)	Slightly increased risk of developing schizophr...			
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	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	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	rs419788	(A;A)	2.3x risk for lupus	Link		
1.5	rs4464148	(C;T)	1.10x increased risk for colorectal cancer			
1.5	rs4626664	(A;G)	1.44x increased risk of developing restless leg...		Link	
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	rs4982731	(C;C)	Possible higher risk of childhood acute lymphob...			
1.5	rs5746059	(A;G)	Slightly higher fat mass			
1.5	rs6896702	(C;T)	Slightly increased risk of developing Parkinson...			
1.5	rs699473	(C;T)	~1.5x increased brain tumor risk			
1.5	rs7341475	(G;G)	1.58x increased schizophrenia risk for women		Link	
1.5	rs807701	(C;T)	Slightly increased dyslexia risk			
1.5	rs9652490	(A;G)	Slightly increased risk of developing Parkinson...		Link	
1.5	rs966221	(C;C)	1.5x increased stroke risk certain populations			
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	rs1545843	(A;A)	1.4x increased risk for depression (for those u...			
1.4	rs2046210	(C;T)	1.4x increased breast cancer risk		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	
1.3	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	rs1260326	(C;T)	Slightly higher risk for gout	Link	Link	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	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	rs501120	(A;G)	1.3x increased risk for heart disease		Link	
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	rs2072590	(G;T)	1.2x increased risk for ovarian cancer			

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
1.2	rs2076295	(G;T)	One copy of the risk allele (G): slightly incre...			
1.2	rs2814707	(A;G)	1.2x increased risk for ALS		Link	
1.2	rs3740878	(A;A)	1.46x type II diabetes risk; common	Link		Link
1.2	rs3849942	(A;G)	1.2x increased risk for ALS		Link	
1.2	rs4324715	(C;C)	>1.5x increased testicular cancer risk for men			
1.2	rs449647	(A;T)	Possibly lower levels of ApoE			
1.2	rs4496877	(T;T)	For type-1 diabetics: 1.6x increased nephropath...			
1.2	rs4977756	(A;G)	1.39x higher risk for glioma development		Link	
1.2	rs498872	(C;T)	1.2x higher risk for glioma development		Link	
1.2	rs8050136	(A;C)	1.2x increased risk for T2D in some populations...		Link	
1.2	rs9858542	(A;G)	1.1x risk Crohn's Disease	Link	Link	
1.17	rs17465637	(A;C)	1.17x higher risk for myocardial infarction	Link	Link	
1.1	rs11110912	(C;G)	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	rs1800451	(A;G)	Carrier of mannose binding deficiency but of lo...	Link	Link	Link
1.1	rs2828520	(G;G)	1.35x major depressive disorder risk			
1.1	rs34516635	(G;G)	Less longevity for Ashkenazi Jewish women.	Link		Link
1.1	rs7412	(C;C)	More likely to gain weight if taking olanzapine...	Link	Link	Link
1.1	rs925391	(C;C)	More likely to go bald; common			
1.1	rs997669	(G;G)	Very slightly increased (1.18x) increased breas...			
1.05	rs2291834	(C;T)	Very slightly higher risk for myocardial infarc...			
1	rs10761659	(A;G)	1.2x risk of Crohn's disease		Link	
1	rs1143674	(A;A)	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;A)	>1.1x 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	rs761100	(G;G)	Higher risk for dyslexia			
1	rs987525	(A;C)	2.5x increased risk for cleft lip		Link	
0.1	rs2070744	(C;C)	Increased prostate cancer risk		Link	Link
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	rs1495965	(A;A)	1.2x higher risk for spondylitis			
0	rs3761418	(A;A)	1.3x increased risk for depression			
0	rs3813929	(C;C)	Possible weight gain if taking olanzapine		Link	Link
0	rs4293393	(T;T)	1.25x Increased Risk of CKD for T allele in ...			
0	rs4795400	(T;T)	If 4 years old or younger: ~2.5x increased asth...		Link	

3.3 Genosets (Multi-variant Phenotypes)

Magnitude	Identifier	Summary
2.9	gs192	MTHFR polymorphisms affecting homocysteine
2.5	gs102	ALS risk
2.5	gs155	CYP3A5 non-expressor
2.5	gs281	Part of the 88% of the population claimed not t...
2.5	gs285	You will lose 2.5x as much weight on a low fat ...
2.4	gs297	Lower heart attack risk than average
2	gs101	Probably able to digest milk
2	gs154	NAT2 Slow metabolizer
2	gs188	One copy of APOE4 is possible: but not certain
1.5	gs220	HLA-B*1502?
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 ERS1176614 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/ERS1176614>

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.