

PGP-UK Genomics Report for ukE8DC4E

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	4954831
Variants filtered out	0
Novel / existing variants	493548 (10.0) / 4449721 (90.0)
Overlapped genes	56696
Overlapped transcripts	67537
Overlapped regulatory features	166340

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.

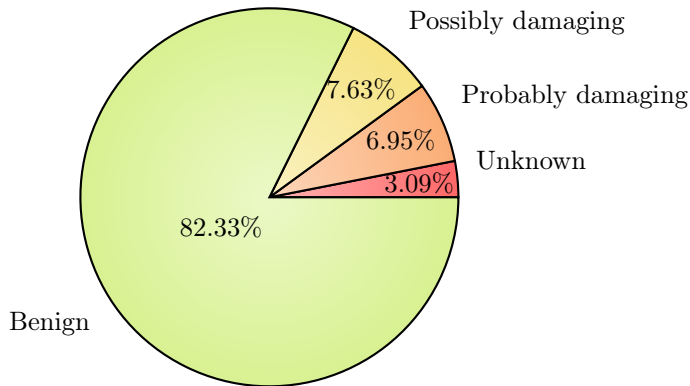


Figure 2: PolyPhen Summary

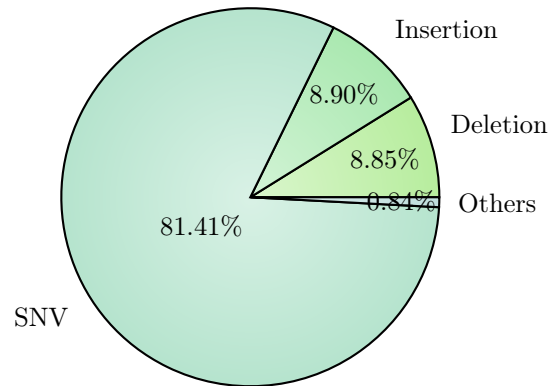


Figure 3: Variant Class

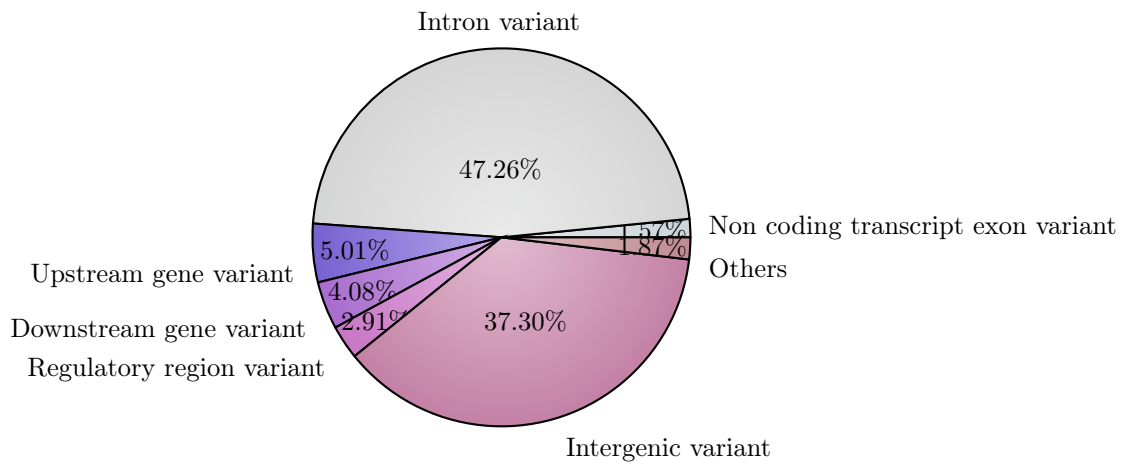


Figure 4: Consequence type

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).

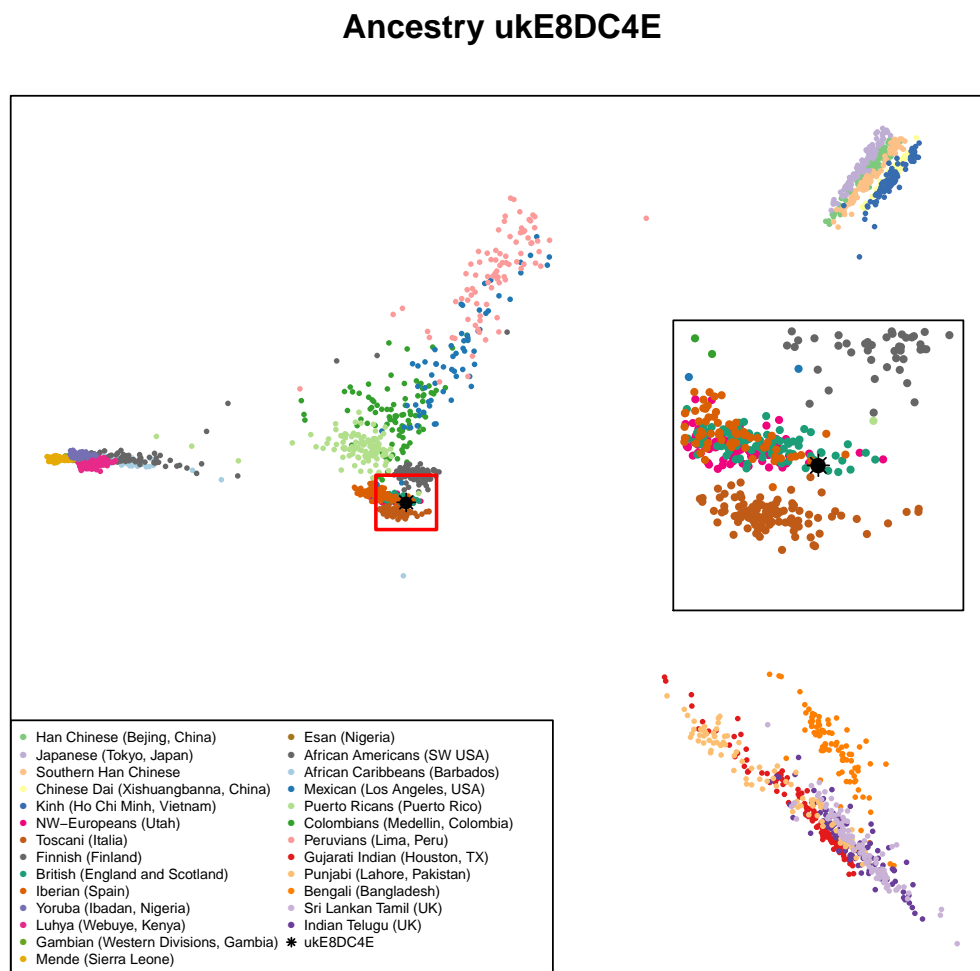


Figure 5: Ancestry Principal Component Analysis

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	rs925391	(T;T)	Unlikely to go bald			
2.4	rs3750817	(T;T)	0.64x reduced risk for breast cancer: and highe...			
2.4	rs9272346	(G;G)	0.08x risk type-1 diabetes		Link	
2.2	rs2511989	(A;A)	0.44x decreased age-related macular degeneratio...		Link	
2.1	rs3775291	(A;G)	0.71x decreased risk for dry age related macula...	Link	Link	Link
2.1	rs6897932	(T;T)	0.70x decreased risk for multiple sclerosis	Link	Link	Link
2.1	rs9332739	(C;G)	0.47x decreased risk for AMD	Link	Link	Link
2	rs1026732	(A;A)	<0.70x risk for restless legs		Link	
2	rs10468017	(C;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	rs1136410	(C;T)	0.80x reduced risk for glioblastoma	Link	Link	
2	rs11635424	(A;A)	<0.70x risk for restless legs		Link	
2	rs12593813	(A;A)	<0.71x risk for restless legs		Link	
2	rs12979860	(C;C)	~80% of such hepatitis C patients respond to tr...		Link	Link
2	rs1864163	(G;G)	Associated with higher HDL cholesterol		Link	
2	rs2229616	(A;G)	Lower risk of metabolic syndrome	Link	Link	Link
2	rs2241423	(A;A)	0.79 decreased risk for obesity			
2	rs2707466	(A;A)	Stronger bones	Link	Link	
2	rs2908004	(T;T)	Stronger bones	Link	Link	
2	rs3819331	(T;T)	Lower risk of autism	Link		
2	rs4073582	(A;A)	Lower risk for gout	Link		
2	rs4149268	(G;G)	Associated with higher HDL cholesterol		Link	
2	rs6505162	(A;C)	0.58x decreased risk for esophageal cancer	Link		
2	rs6511720	(G;T)	Slightly lower odds of developing CHD.		Link	Link
2	rs801114	(T;T)	0.78x decreased Basal Cell Carcinoma risk.		Link	
1.8	rs1128535	(A;G)	0.77x risk for Crohn's disease			

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
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...			
1.8	rs3814113	(C;T)	0.8x decreased risk for ovarian cancer		Link	
1.8	rs4714156	(C;C)	<0.61x risk for restless legs			
1.5	rs1063192	(C;C)	0.71x reduced risk of myocardial infarction			
1.5	rs11136000	(T;T)	0.84x decreased risk for Alzheimer's disease		Link	
1.5	rs2007153	(A;A)	Decreased risk of schizophrenia in limited stud...			
1.5	rs309375	(G;G)	Smaller mosquito bites			
1.5	rs3790844	(C;T)	Slightly reduced risk (0.77x) for pancreatic ca...			
1.5	rs4149274	(C;C)	Associated with higher HDL (good) cholesterol.			
1.5	rs4939883	(C;T)	Associated with higher HDL cholesterol		Link	
1.4	rs9402571	(G;T)	Slightly decreased risk for type-2 diabetes			
1.25	rs10088218	(A;G)	0.76x decreased risk for ovarian cancer			
1.2	rs11246226	(A;C)	Decreased risk of schizophrenia in limited stud...		Link	
1.2	rs9306160	(C;T)	0.75x (reduced) risk for metastasis in LN-/ER+ ...	Link	Link	
1.1	rs2293347	(G;G)	Among NSCLC patients: better Gefitinib response...	Link		Link
1	rs182549	(C;T)	Can digest milk.			Link
1	rs2952768	(C;T)	Slightly less drug dependence: decreased effect...			Link
1	rs33927012	(C;T)	Currently evaluated as benign in ClinVar	Link	Link	Link
1	rs7850258	(A;G)	Typical odds of developing primary hypothyroidi...			
0.1	rs891512	(G;G)	Lower blood pressure than those with an A allel...	Link		
0	rs1047781	(A;A)	ABH blood group "Secretor" status if Japanese	Link	Link	Link
0	rs1126809	(A;G)	Slight increase in skin cancer risk	Link	Link	Link
0	rs12252	(T;T)	More resistant to influenza	Link		Link
0	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	rs5065	(A;A)	1.12x risk on diuretic; if hypertensive: better...	Link	Link	Link
0	rs6259	(G;G)	Best inverse correlation between tea-drinking: ...	Link	Link	
0	rs74315403	(G;G)	PrP codon 178 (D) - non pathogenic variant			Link
0	rs7495174	(A;A)	Blue/gray eyes more likely		Link	

3.2 Possibly Harmful Traits

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
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	rs2306402	(C;T)	1.18x increased risk for late-onset Alzheimer's...			
3	rs4151667	(A;T)	Age related macular degeneration	Link	Link	Link
3	rs7754840	(C;G)	1.3x increased risk for type-2 diabetes		Link	
2.6	rs110419	(A;A)	2.6x increased risk for neuroblastoma			
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	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	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...			
2.4	rs7966230	(G;G)	Slightly lower levels of plasma VWF			
2.3	rs1859962	(G;G)	1.28x increased risk for prostate cancer		Link	
2.2	rs2004640	(G;T)	1.4x increased risk for SLE		Link	Link
2.1	rs10427255	(C;C)	Highest odds of photic sneeze reflex			
2.1	rs1050152	(T;T)	2.1x increased risk of Crohn's disease	Link	Link	Link
2.1	rs10811661	(T;T)	1.2x increased risk for type-2 diabetes		Link	
2.1	rs1329428	(G;G)	2x increased risk for macular degeneration			
2.1	rs1585215	(G;G)	3.5x increased risk for Hodgkin lymphoma			
2.1	rs1695	(G;G)	3.5x asthma risk in certain populations	Link	Link	Link
2.1	rs17070145	(C;C)	Reduced memory abilities			Link
2.1	rs17563	(C;C)	Risk for otosclerosis	Link	Link	Link
2.1	rs2231137	(A;G)	~1.5-3x increased risk for ischemic stroke	Link	Link	Link
2.1	rs5186	(A;C)	~1.4x increased risk of hypertension	Link	Link	Link
2.1	rs646776	(A;A)	1.2x risk of coronary artery disease		Link	
2.1	rs7837688	(G;T)	1.7x increased risk for prostate cancer			
2.1	rs944289	(C;T)	1.3x increased thyroid cancer risk		Link	
2	rs10090154	(C;T)	1.4x 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	rs10260404	(C;C)	1.60x risk of developing ALS		Link	
2	rs10488631	(C;T)	2x increased risk of developing SLE; 1.6x incre...		Link	
2	rs10937823	(C;T)	Some association with bipolar disorder			
2	rs10984447	(A;G)	1.17x increased risk for multiple sclerosis		Link	
2	rs11190870	(C;T)	Possibly increased risk of scoliosis			
2	rs1160312	(A;G)	1.6x increased risk of Male Pattern Baldness.		Link	
2	rs11983225	(T;T)	7x less likely to respond to certain antidepres...		Link	
2	rs1265181	(C;G)	Increased risk for psoriasis		Link	
2	rs1333048	(A;C)	1.3x increased coronary artery disease risk			
2	rs1360780	(C;T)	1.3x increased risk for depression		Link	
2	rs1361600	(G;G)	~2x increased risk for adult-onset asthma in Ja...			
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	rs17001266	(-;C)	1.58x increased risk for schizophrenia in males...			
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	rs1800629	(A;G)	Complex; generally higher risk for certain dise...	Link	Link	Link
2	rs1800896	(A;G)	1.6x increased prostate cancer risk			
2	rs2073963	(G;T)	Increased risk of baldness			
2	rs2143340	(C;T)	Increased risk of dyslexia and poor reading per...			

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
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	(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	rs25487	(G;G)	2x higher risk for skin cancer; possibly other ...	Link	Link	Link
2	rs2572886	(A;G)	1.4x increased risk of HIV infection			
2	rs2619522	(G;G)	Associated with lower attention capacity but al...			
2	rs2736100	(T;T)	Higher risk of Interstitial lung disease: and t...		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	rs351855	(C;T)	1.2x increased risk for prostate cancer	Link	Link	Link
2	rs3738579	(C;C)	0.6x decreased risk for cervical cancer: but 1....			
2	rs3738919	(A;C)	1.94x risk of developing rheumatoid arthritis			
2	rs3746444	(C;T)	~1.2x increased risk for cancer	Link		
2	rs3775948	(G;G)	Slightly higher risk for gout			
2	rs3793784	(C;G)	1.5x risk for ARMD		Link	Link
2	rs4148739	(A;A)	7x less likely to respond to certain antidepres...		Link	
2	rs4242382	(A;G)	1.7x increased risk for prostate cancer		Link	
2	rs4402960	(G;T)	1.2x increased risk for type-2 diabetes: ~1x ri...		Link	Link
2	rs4444903	(A;G)	3.5x risk of hep-cancer in cirrhosis patients; ...			Link
2	rs4633	(T;T)	Higher risk for endometrial cancer	Link	Link	Link
2	rs4792311	(A;G)	Increased risk of prostate cancer	Link	Link	Link
2	rs486907	(A;A)	2x increased prostate cancer risk	Link	Link	Link
2	rs493258	(A;G)	1.15x risk of Age Related Macular Degeneration			
2	rs520354	(A;G)	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;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	rs744373	(C;T)	1.17x risk of Alzheimer's			
2	rs763361	(T;T)	Increased risk for multiple autoimmune diseases...	Link	Link	
2	rs7776725	(C;C)	Weaker bones		Link	
2	rs7807268	(C;C)	1.4x risk for Crohn's disease		Link	
2	rs7923837	(G;G)	3.2x risk for T2D			
2	rs7961152	(A;A)	1.5x higher risk for hypertension			
2	rs800292	(C;C)	5% higher risk of Age related macular degenerat...	Link	Link	Link
2	rs828907	(T;T)	Increased risk of bladder cancer and 2x risk of...			
2	rs854560	(A;T)	Higher risk for heart disease: diabetic retinop...	Link	Link	Link
2	rs9543325	(C;C)	1.37x Slightly higher pancreatic cancer risk			
2	rs9652490	(A;A)	~2x increased risk for Parkinson's disease: and...		Link	
2	rs965513	(A;G)	1.77x increased thyroid cancer risk		Link	
2	rs987525	(A;A)	6x increased risk for cleft lip		Link	
2	rs9954153	(G;T)	~2.5x higher risk for Fuchs' dystrophy: a corne...			
2.0	rs1044396	(C;C)	Increased risk of Nicotine dependence among mal...	Link	Link	Link
2.0	rs4911414	(G;T)	2-4x higher risk of sun sensitivity if part of ...		Link	
1.8	rs143383	(T;T)	1.3x increased risk for osteoarthritis		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			

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
1.7	rs8055236	(G;T)	1.9x risk for heart disease		Link	
1.6	rs1537415	(C;G)	1.6x increased risk for periodontitis		Link	
1.6	rs33980500	(C;T)	1.6x increase in risk for psoriatic arthritis	Link	Link	Link
1.6	rs3764880	(A;A)	1.2 - 1.8x increased tuberculosis risk	Link	Link	
1.6	rs763035	(T;T)	1.4x increased risk for rosacea			
1.5	rs10757272	(C;T)	1.30x increased risk for Coronary artery diseas...			
1.5	rs10859871	(A;C)	Slight (~1.2x) increase in endometriosis risk			
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	rs1169300	(A;G)	~1.5x increased lung cancer risk			
1.5	rs12037606	(A;G)	1.22x risk of developing Crohn's disease			
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	rs17221417	(C;G)	1.3x higher risk for Crohn's disease		Link	
1.5	rs1801020	(T;T)	1.31x increased risk of heart disease	Link		Link
1.5	rs1801274	(C;T)	Complex; generally greater risk for cancer prog...	Link	Link	Link
1.5	rs1867277	(A;G)	1.5x increased risk for thyroid cancer			
1.5	rs1994090	(G;T)	Slightly increased risk of developing Parkinson...		Link	
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	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	(T;T)	Slightly increased risk for pregnancy-induced h...			
1.5	rs3087243	(A;G)	Increased risk for auto-immune diseases		Link	
1.5	rs358806	(A;C)	0.86x increased risk of developing Type-2 diabe...		Link	
1.5	rs3790565	(C;T)	Slightly increased risk of developing primary b...			
1.5	rs3814570	(C;T)	1.3x increased risk for Crohn's disease with il...			
1.5	rs3825776	(A;G)	1.3x increased risk for ALS		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	rs4464148	(C;T)	1.10x increased risk for colorectal cancer			
1.5	rs4506565	(A;T)	1.4x increased risk for type-2 diabetes		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;C)	1.5x higher risk for melanoma		Link	
1.5	rs4845618	(G;T)	1.7x increased melanoma risk			
1.5	rs5219	(C;T)	1.3x increased risk for type-2 diabetes	Link	Link	Link
1.5	rs5746059	(A;A)	Slightly higher fat mass			
1.5	rs6435862	(G;T)	1.7x higher risk of aggressive neuroblastoma		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;T)	~1.5x increased brain tumor risk			
1.5	rs7341475	(G;G)	1.58x increased schizophrenia risk for women		Link	
1.5	rs872071	(A;G)	~1.5x increased risk for chronic lymphocytic le...		Link	
1.5	rs9303277	(C;T)	1.46x Slightly increased risk of developing pri...			
1.5	rs9642880	(G;T)	1.2x increased bladder cancer risk		Link	
1.5	rs966221	(C;C)	1.5x increased stroke risk certain populations			
1.5	rs995030	(G;G)	Non-protective against testicular cancer		Link	
1.4	rs12770228	(A;G)	1.4x increased risk for meningioma			
1.4	rs1447295	(A;C)	1.4x increased risk of prostate cancer		Link	

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
1.4	rs1545843	(A;A)	1.4x increased risk for depression (for those u...			
1.4	rs1801157	(A;G)	1.4x higher risk for breast cancer			
1.4	rs2046210	(C;T)	1.4x increased breast cancer risk		Link	Link
1.4	rs4959039	(A;G)	1.4x higher risk for multiple sclerosis			
1.4	rs4977756	(G;G)	1.93x higher risk for glioma development		Link	
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	rs1260326	(C;T)	Slightly higher risk for gout	Link	Link	Link
1.3	rs1434536	(A;G)	1.29x increased breast cancer risk			
1.3	rs2024513	(A;G)	1.3x higher risk for schizophrenia (among Han C...			
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	rs2072590	(G;T)	1.2x increased risk for ovarian cancer			
1.2	rs2651899	(G;G)	1.2x higher risk for migraines			
1.2	rs2814707	(A;G)	1.2x increased risk for ALS		Link	
1.2	rs3131296	(A;G)	1.2x increased risk for schizophrenia		Link	
1.2	rs3849942	(A;G)	1.2x increased risk for ALS		Link	
1.2	rs419788	(A;G)	2.0x risk for lupus	Link		
1.2	rs4324715	(C;C)	>1.5x increased testicular cancer risk for men			
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	rs3802842	(A;C)	1.17x increased risk of colorectal cancer		Link	
1.1	rs11037909	(C;T)	1.27x type II diabetes risk	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	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	rs6897876	(C;T)	Slight increase in testicular cancer risk for m...			
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	
1.07	rs2291834	(C;C)	Very slightly higher risk for myocardial infarc...			
1.05	rs1800056	(C;T)	Very slightly increased risk (1.05) for breast...	Link	Link	Link
1	rs1143674	(A;A)	1.3x increased autism risk	Link		
1	rs2273697	(A;G)	Adverse reaction more likely to carbamazepine i...	Link	Link	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	rs6166	(G;G)	Females slightly more likely to be sterile	Link	Link	Link
1	rs6932590	(C;T)	1.1x increased risk for schizophrenia		Link	
0.1	rs601338	(A;G)	Susceptible to Norovirus infections	Link	Link	Link
0	rs1004819	(C;C)	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	rs4293393	(T;T)	1.25x Increased Risk of CKD for T allele in ...			
0	rs4712653	(T;T)	2x increased risk for neuroblastoma			
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	gs102	ALS risk
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
1.5	gs185	The beta blocker metoprolol is effective with 1...
1.5	gs186	HLA-B*5801 heterozygosity is possible: unfortun...
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 ERS1176575 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/ERS1176575>

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.