PGP-UK Genomics Report for ukE52AAC

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.
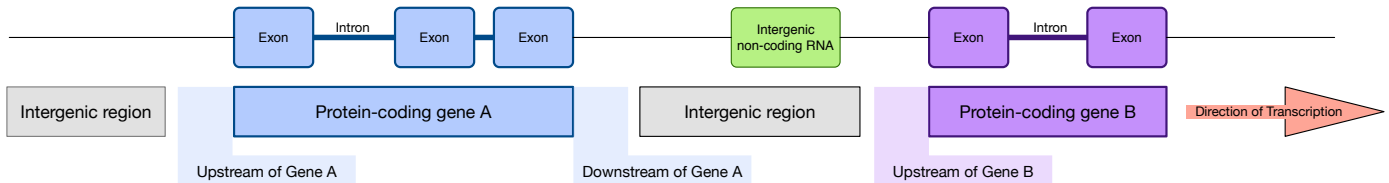

Figure 1: Diagram of gene structure indicating locations of potential variants

Feature	Count
Lines of input read	5017307
Variants filtered out	0
Novel / existing variants	513298 (10.3) / 4491747 (89.7)
Overlapped genes	56726
Overlapped transcripts	67573
Overlapped regulatory features	168062

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.
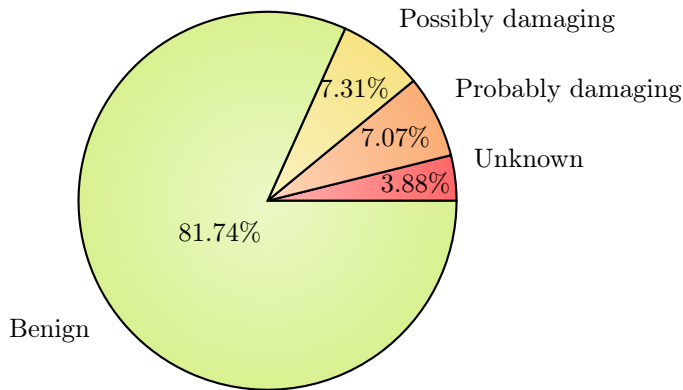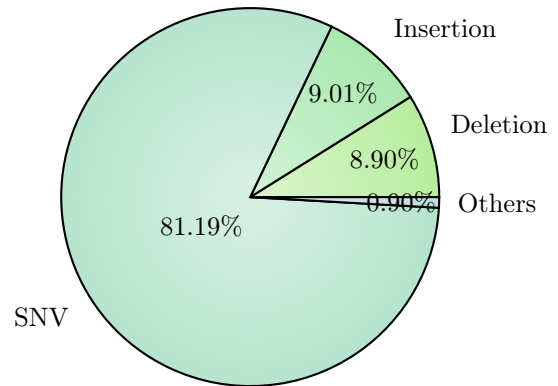
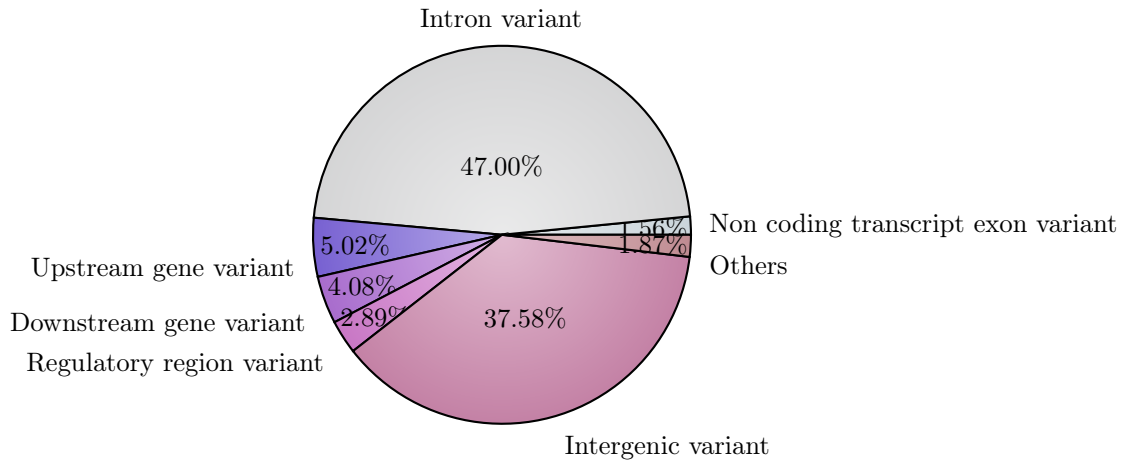Figure 2: PolyPhen Summary

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).
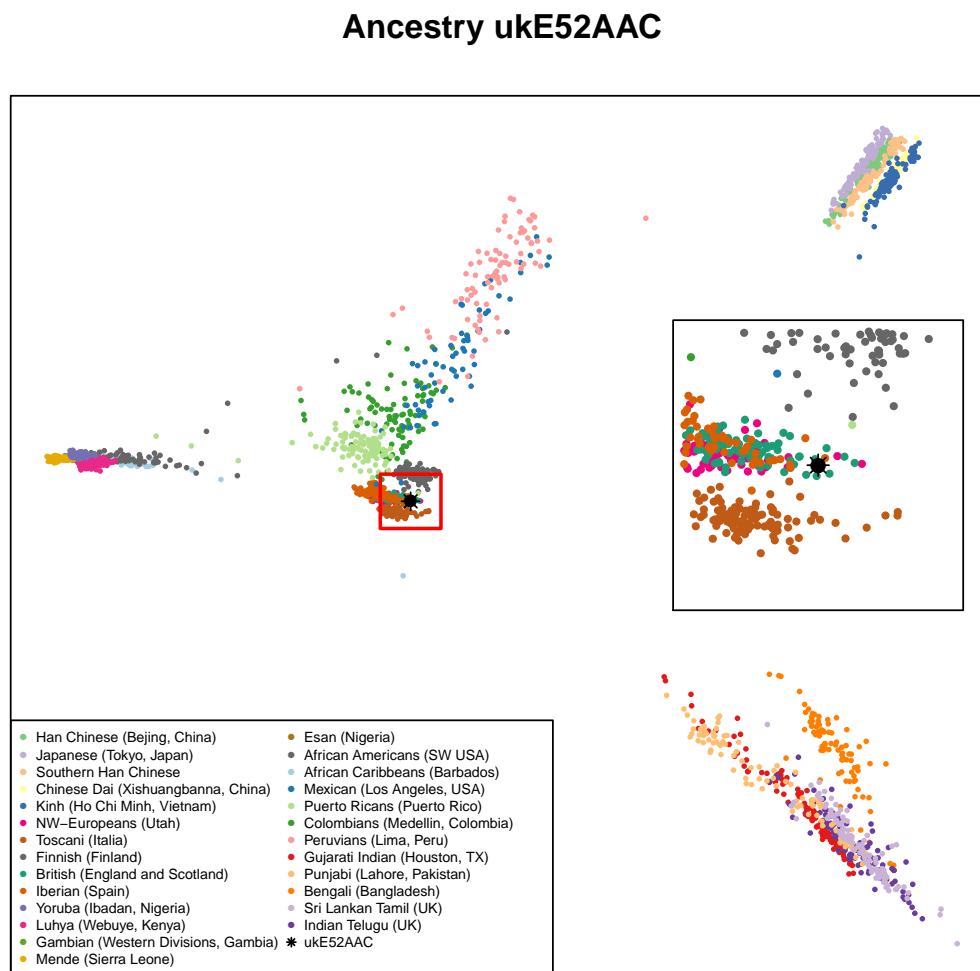
Ancestry ukE52AAC

Figure 5: Ancestry Principal Component Analysis

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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	
2.4	rs2802288	(A;A)	Longer lifespan			
2.4	rs3750817	(T;T)	0.64x reduced risk for breast cancer: and highe...			
2	rs10504861	(A;G)	Reduced risk of migraine without aura			
2	rs1056836	(G;G)	0.3x decreased risk for prostate cancer	Link	Link	Link
2	rs12979860	(C;C)	~80% of such hepatitis C patients respond to tr...		Link	Link
2	rs1799884	(G;G)	Mothers have typical Birth-Weight babies. Sligh...			
2	rs1864163	(A;G)	Associated with higher HDL cholesterol		Link	
2	rs2060793	(A;A)	Lower serum levels of vitamin D			
2	rs2073963	(T;T)	Reduced risk of baldness			
2	rs2241423	(A;G)	0.79 decreased risk for obesity			
2	rs2542052	(C;C)	Better odds of living to 100			
2	rs2764264	(C;C)	Greater odds of living to 95			
2	rs3764261	(G;T)	Associated with higher HDL cholesterol		Link	Link
2	rs3914132	(C;T)	Lower otosclerosis risk		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	rs7776725	(T;T)	Stronger bones		Link	
2	rs9272346	(A;G)	0.3x risk type-1 diabetes		Link	
2	rs9642880	(G;G)	Slightly lower risk of Bladder Cancer.		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	
1.8	rs4714156	(C;C)	<0.61x risk for restless legs			
1.8	rs7101429	(A;G)	0.70x reduced risk for Alzheimer's risk			
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	

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
1.5	rs11212617	(A;C)	Somewhat increased likelihood of treatment succ...			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	rs3790844	(C;T)	Slightly reduced risk (0.77x) for pancreatic ca...			
1.5	rs3851179	(A;A)	0.85x decreased risk for Alzheimer's disease		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	rs729302	(A;C)	0.89x decreased risk of developing rheumatoid a...			
1.4	rs6495446	(C;T)	0.8x reduced risk for chronic kidney disease			
1.4	rs6700125	(C;C)	0.7x decreased risk for ALS			
1.3	rs9306160	(T;T)	0.75x (reduced) risk for metastasis in LN-/ER+ ...	Link	Link	
1.2	rs11172113	(C;C)	0.8x lower risk for migraines			
1.2	rs11246226	(A;C)	Decreased risk of schizophrenia in limited stud...		Link	
1.1	rs2293347	(G;G)	Among NSCLC patients: better Gefitinib response...	Link		Link
1.1	rs4988235	(T;T)	Can digest milk			Link
1	rs182549	(T;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	rs6583817	(T;T)	~0.80x (lower) risk for late onset Alzheimer's ...			
1	rs7850258	(A;G)	Typical odds of developing primary hypothyroidi...			
1.0	rs2283123	(C;T)	Decreased risk of schizophrenia in limited stud...			
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	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	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	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	rs9394492	(C;C)	<0.76x risk for restless legs			

3.2 Possibly Harmful Traits

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
4	rs1333049	(C;C)	1.9x increased risk for coronary artery disease...		Link	
3.8	rs5186	(C;C)	7.3x increased risk of hypertension	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	rs16969968	(A;A)	Higher risk for nicotine dependence: lower risk...	Link	Link	Link
3	rs1801282	(C;G)	Unconfirmed higher risk of cardiovascular disea...	Link	Link	Link
3	rs1983132	(C;T)	2 - 3x higher prostate cancer risk if routinely...			
3	rs2306402	(C;C)	1.18x increased risk for late-onset Alzheimer's...			
3	rs3738579	(T;T)	1.5x - 2x increased risk for cervical cancer: H...			
2.6	rs8034191	(C;C)	1.80x lung cancer risk; decreased response to a...		Link	
2.5	rs1051730	(T;T)	1.8x increased risk of lung cancer; reduced res...	Link	Link	Link
2.5	rs1057910	(A;C)	CYP2C9*3 carrier; average 40% reduction in warf...	Link	Link	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	rs187238	(G;G)	Hypertension increases risk 3.75x for sudden ca...			
2.5	rs2254958	(C;C)	1.61x increased risk for Alzheimer's			
2.5	rs3738919	(C;C)	1.94x risk of developing rheumatoid arthritis			
2.5	rs664143	(C;T)	Higher risk for number of cancers			
2.5	rs7574865	(G;T)	1.3x risk of rheumatoid arthritis; 1.55x risk o...		Link	Link
2.3	rs37973	(G;G)	Among asthmatics: 2.3x more likely to show less...			Link
2.3	rs7966230	(C;G)	Slightly lower levels of plasma VWF			
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	
2.1	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	rs17070145	(C;C)	Reduced memory abilities			Link
2.1	rs17563	(C;C)	Risk for otosclerosis	Link	Link	Link
2.1	rs2231142	(A;C)	1.74x increased gout risk; gefinitib takers 4x ...	Link	Link	Link
2.1	rs2383207	(G;G)	Increased risk for heart disease			
2.1	rs646776	(A;A)	1.2x risk of coronary artery disease		Link	
2.1	rs795484	(A;G)	Increased morphine dose requirement and postope...			
2.1	rs944289	(C;T)	1.3x increased thyroid cancer risk		Link	
2	rs10248420	(A;A)	7x less likely to respond to certain antidepres...		Link	
2	rs10492519	(G;G)	Increased risk of developing prostate cancer			
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	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	rs1160312	(A;A)	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	rs12696304	(C;G)	Prone to aging faster: at least in European pop...			
2	rs12720461	(C;T)	Carrier of one CYP1A2*1K_-729C>T allele: decrea...			
2	rs1360780	(C;T)	1.3x increased risk for depression		Link	
2	rs1585215	(A;G)	2x increased risk for Hodgkin lymphoma			
2	rs1691053	(A;G)	Increased risk of developing prostate cancer			
2	rs17228212	(C;T)	1.26x increased risk for heart disease		Link	
2	rs1734791	(A;A)	1.4x increased risk for lupus			
2	rs1800629	(A;G)	Complex; generally higher risk for certain dise...	Link	Link	Link
2	rs2201841	(T;T)	2.4x increased risk for Graves' disease		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	

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
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	rs25487	(A;G)	2x higher risk for skin cancer; possibly other ...	Link	Link	Link
2	rs2707466	(G;G)	Weaker bones	Link	Link	
2	rs2736100	(T;T)	Higher risk of Interstitial lung disease: and t...		Link	
2	rs27388	(A;A)	Increased risk of developing schizophrenia			
2	rs2908004	(C;C)	Weaker bones	Link	Link	
2	rs3025039	(C;T)	2.6x increased risk for ARMD in a Taiwanese pop...			
2	rs3129934	(C;T)	Increased risk of Multiple Sclerosis.		Link	
2	rs358806	(C;C)	1.78x increased risk of developing Type-2 diabe...		Link	
2	rs3775948	(G;G)	Slightly higher risk for gout			
2	rs4148739	(A;A)	7x less likely to respond to certain antidepres...		Link	
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	rs493258	(G;G)	1.15x risk of Age Related Macular Degeneration			
2	rs4961	(G;T)	1.8x increased risk for high blood pressure	Link	Link	Link
2	rs520354	(A;G)	Increased risk in men for biliary conditions			
2	rs5759167	(T;T)	Higher prostate cancer risk		Link	
2	rs6601764	(C;C)	1.52x increased risk of developing Crohn's dise...		Link	
2	rs669	(G;G)	3.8x or higher increased risk for Alzheimers	Link	Link	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	rs6997709	(G;G)	1.5x 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	rs763361	(T;T)	Increased risk for multiple autoimmune diseases...	Link	Link	
2	rs7639618	(C;T)	1.45x increased osteoarthritis risk	Link		
2	rs7794745	(T;T)	Slightly increased risk for autism		Link	Link
2	rs7807268	(C;C)	1.4x risk for Crohn's disease		Link	
2	rs800292	(C;C)	5% higher risk of Age related macular degenerat...	Link	Link	Link
2	rs854560	(A;A)	Higher risk for heart disease: diabetic retinop...	Link	Link	Link
2	rs9525638	(T;T)	Weaker bones			
2	rs9652490	(A;A)	~2x increased risk for Parkinson's disease: and...		Link	
2	rs965513	(A;G)	1.77x increased thyroid cancer risk		Link	
2.0	rs1434536	(A;A)	1.94x increased breast cancer risk			
2.0	rs2156921	(G;G)	1.29x increased risk for depression			
2.0	rs2305795	(A;A)	1.64x higher risk of narcolepsy compared to (G;...			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	
1.6	rs1537415	(C;G)	1.6x increased risk for periodontitis		Link	
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	rs10883365	(A;G)	1.2x increased risk for developing Crohn's dise...		Link	
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	rs12223271	(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	rs144848	(G;T)	Very slightly increased breast cancer risk	Link	Link	Link

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
1.5	rs16944	(A;G)	Minorly increased risk of mental illness and os...		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	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	rs2241880	(C;T)	1.4x increased risk for Crohn's disease in Cauc...	Link	Link	Link
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	rs2881766	(G;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	rs3212227	(A;A)	1.43x increased risk of developing psoriasis an...			
1.5	rs393152	(A;A)	Increased risk of both PD and AD	Link	Link	
1.5	rs4027132	(A;G)	1.39x increased risk of developing bipolar diso...			
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	rs4506565	(A;T)	1.4x increased risk for type-2 diabetes		Link	
1.5	rs4626664	(A;G)	1.44x increased risk of developing restless leg...		Link	
1.5	rs464049	(C;T)	Increased risk of schizophrenia in limited stud...			
1.5	rs4785763	(A;C)	1.5x higher risk for melanoma		Link	
1.5	rs5746059	(A;A)	Slightly higher fat mass			
1.5	rs619203	(C;G)	Increases susceptibility to Myocardial Infarcti...	Link	Link	
1.5	rs6498169	(A;G)	1.14x risk of multiple sclerosis		Link	
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	rs7536563	(A;G)	1.12x risk of multiple sclerosis		Link	
1.5	rs763035	(C;T)	1.2x increased risk for rosacea			
1.5	rs807701	(C;T)	Slightly increased dyslexia risk			
1.5	rs872071	(G;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	rs9561778	(G;T)	~2x increased risk of adverse drug reactions fr...		Link	
1.5	rs995030	(G;G)	Non-protective against testicular cancer		Link	
1.4	rs1126497	(C;T)	1.4x increased risk for breast cancer	Link	Link	Link
1.4	rs2230201	(A;G)	1.4x risk of lupus	Link		
1.4	rs4959039	(A;G)	1.4x higher risk for multiple sclerosis			
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	rs1746048	(C;C)	1.03 increased risk for coronary heart disease		Link	
1.3	rs34330	(T;T)	1.2x higher breast cancer risk; 1.3x higher ris...			
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	rs1344706	(T;T)	1.2x increased risk for schizophrenia		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	rs2056116	(A;G)	1.18x risk for breast cancer			
1.2	rs2076295	(G;T)	One copy of the risk allele (G): slightly incre...			
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;G)	1.2x increased risk for ALS		Link	
1.2	rs3131296	(A;G)	1.2x increased risk for schizophrenia		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	

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
1.2	rs4324715	(C;C)	>1.5x increased testicular cancer risk for men			
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	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	
1.15	rs748404	(C;T)	Very slightly increased risk (1.15) for lung ca...		Link	
1.1	rs11110912	(C;G)	1.3x high blood pressure risk			
1.1	rs2653349	(G;G)	2-6x increased risk for cluster headaches	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	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	(A;C)	Very slightly higher risk for breast cancer		Link	
1.1	rs925391	(C;C)	More likely to go bald; common			
1.05	rs2291834	(C;T)	Very slightly higher risk for myocardial infarc...			
1	rs2273697	(A;G)	Adverse reaction more likely to carbamazepine i...	Link	Link	Link
1	rs2546890	(A;A)	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	
1	rs6974491	(A;G)	Higher risk of coeliac and/or inflammatory bowe...			
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	rs10761659	(A;A)	1.5x risk of Crohn's disease		Link	
0	rs1128503	(T;T)	Likely to require more methadone during heroin ...	Link	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	rs7652331	(T;T)	Somewhat higher risk for prostate cancer			
0	rs7787082	(G;G)	7x less likely to respond to certain antidepres...		Link	
0	rs855791	(T;T)	0.2 g/dL lower hemoglobin on average	Link	Link	Link

3.3 Genosets (Multi-variant Phenotypes)

Magnitude	Identifier	Summary
3.1	gs191	Problem metabolizing NSAIDs
2.9	gs192	MTHFR polymorphisms affecting homocysteine
2.6	gs296	Lower heart attack risk than average
2.5	gs155	CYP3A5 non-expressor
2.5	gs157	More stimulated by coffee
2.5	gs161	CYP2C9 Intermediate Metabolizers
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	gs156	NAT2 Rapid metabolizer.
2	gs188	One copy of APOE4 is possible: but not certain
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.
1	gs182	CYP2D6*39

4 Raw Data

The raw data used to create this report has been assigned the identifier ERS1176612 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/ERS1176612

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 5, 2017.