PGP-UK Genomics Report for uk58BACF

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.

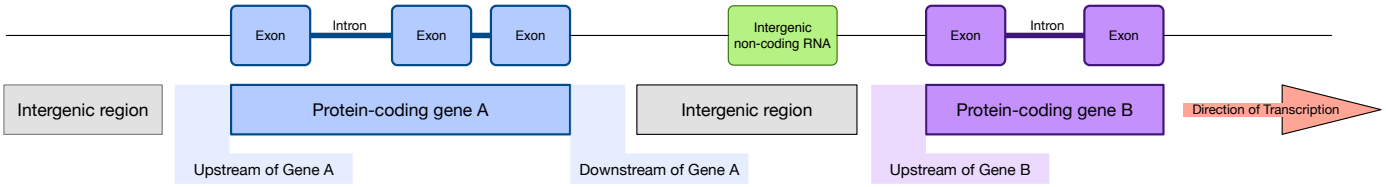

Figure 1: Diagram of gene structure indicating locations of potential variants

Feature	Count
Lines of input read	4965667
Variants filtered out	0
Novel / existing variants	482800 (9.7) / 4470841 (90.3)
Overlapped genes	56708
Overlapped transcripts	67521
Overlapped regulatory features	167494

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.

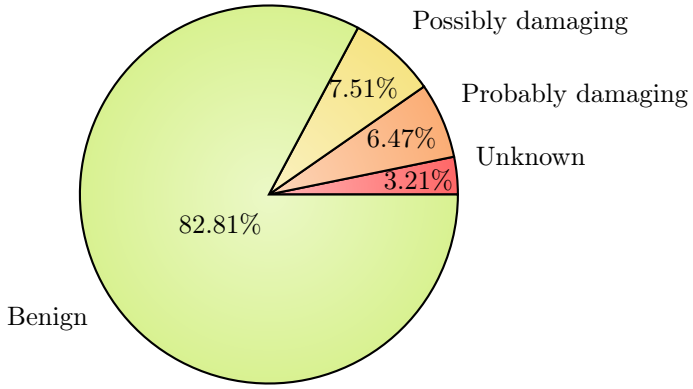

Figure 2: PolyPhen Summary

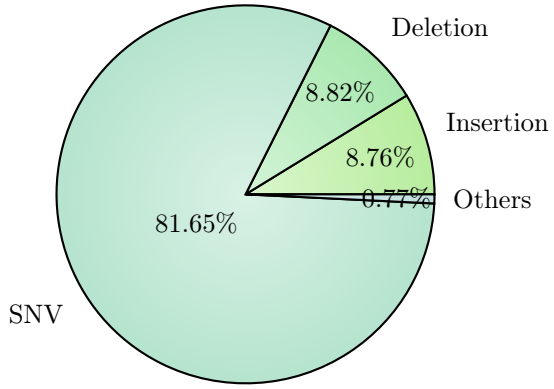

Figure 3: Variant Class

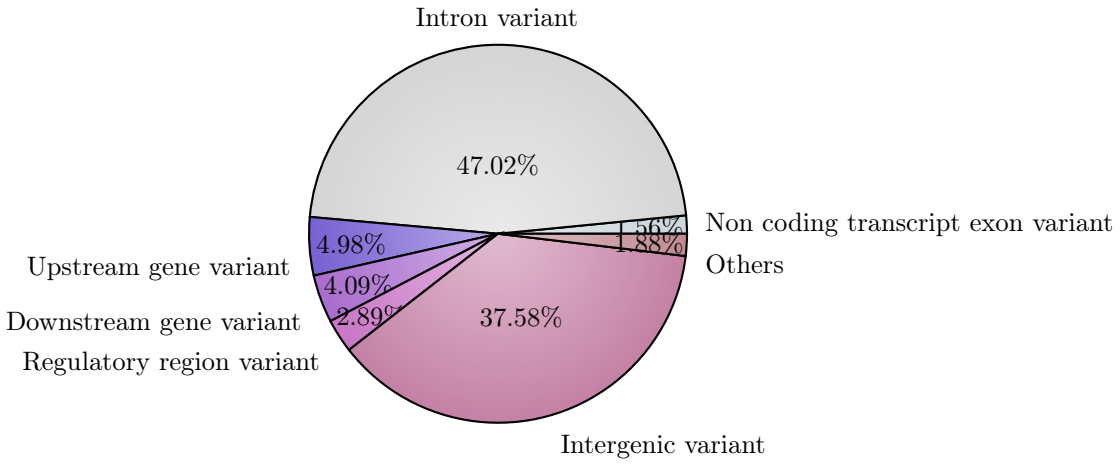

Figure 4: Consequence type

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).

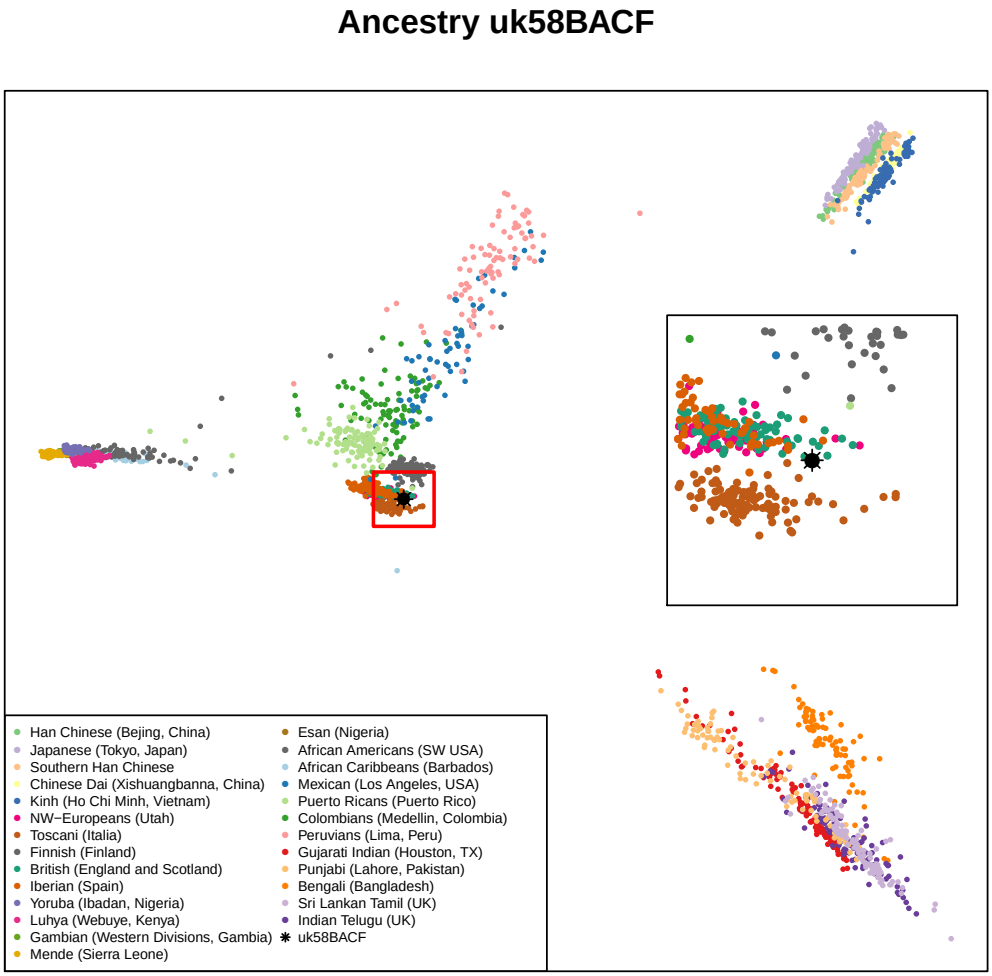

Figure 5: Ancestry Principal Component Analysis

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	rs7294919	(C;T)	Moderately enhanced hippocampal volume			
2.5	rs8070723	(G;G)	~0.05x (thus greatly) reduced risk for developi...			
2.4	rs2802288	(A;A)	Longer lifespan			
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	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	rs2241423	(A;G)	0.79 decreased risk for obesity			
2	rs2707466	(A;A)	Stronger bones	Link	Link	
2	rs2764264	(C;C)	Greater odds of living to 95			
2	rs2908004	(T;T)	Stronger bones	Link	Link	
2	rs3218536	(A;G)	Lower risk for breast: ovarian cancer	Link	Link	
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	rs7105934	(A;G)	0.69 times lower odds of developing renal cell ...			
2	rs763110	(C;T)	~0.80x reduced cancer risk			Link
2	rs801114	(T;T)	0.78x decreased Basal Cell Carcinoma risk.		Link	
2	rs925391	(C;T)	Lower odds of going bald			
2	rs9525638	(C;C)	Stronger bones			
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	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;C)	0.8x decreased risk for ovarian cancer		Link	
1.8	rs4714156	(C;C)	<0.61x risk for restless legs			

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
1.6	rs10801935	(C;C)	0.3x decreased risk of breast cancer			
1.6	rs3025786	(C;T)	Slightly decreased Alzheimer's disease risk amo...	Link		
1.5	rs1026732	(A;G)	0.70x risk for restless legs		Link	
1.5	rs1063192	(C;C)	0.71x reduced risk of myocardial infarction			
1.5	rs11136000	(C;T)	0.84x decreased risk for Alzheimer's disease		Link	
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	rs16991615	(A;G)	Slight increase (11 months) in avg age at menop...	Link	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	rs5888	(C;C)	Higher HDL cholesterol but lower risk for age-r...	Link		
1.5	rs729302	(A;C)	0.89x decreased risk of developing rheumatoid a...			
1.4	rs1165205	(A;T)	0.85x decreased gout risk		Link	
1.4	rs6495446	(C;T)	0.8x reduced risk for chronic kidney disease			
1.4	rs6700125	(C;C)	0.7x decreased risk for ALS			
1.25	rs10088218	(A;G)	0.76x decreased risk for ovarian cancer			
1.2	rs4320932	(A;G)	0.87x decreased risk for ovarian cancer			
1.2	rs4686484	(G;G)	Slightly decreased risk for celiac disease			
1.2	rs6048	(A;G)	Slightly lower risk (10-20%) of deep vein throm...	Link	Link	Link
1.1	rs11172113	(C;T)	0.9x lower risk for migraines			
1.1	rs2293347	(G;G)	Among NSCLC patients: better Gefitinib response...	Link		Link
1.1	rs4988235	(T;T)	Can digest milk			Link
1	rs1800547	(G;G)	MAPT H2/H2 diplotype	Link		Link
1	rs182549	(T;T)	Can digest milk.			Link
1	rs2494732	(T;T)	Lower odds of psychosis	Link	Link	
1	rs7850258	(A;G)	Typical odds of developing primary hypothyroidi...			
1	rs800292	(C;T)	1% decreased risk of macular degeneration	Link	Link	Link
1.0	rs11246226	(C;C)	Decreased risk of schizophrenia in limited stud...		Link	
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	rs1047781	(A;A)	ABH blood group "Secretor" status if Japanese	Link	Link	Link
0	rs12252	(T;T)	More resistant to influenza	Link		Link
0	rs16990018	(A;A)	PrP Codon 171 Asn - Non-pathogenic variant	Link		Link
0	rs17244841	(A;A)	More responsive to statin treatment		Link	Link
0	rs1799782	(C;C)	Lower risk for skin cancer	Link	Link	
0	rs1799945	(C;C)	Not a H63D hemochromatosis carrier.	Link	Link	Link
0	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	rs41303129	(C;T)	Likely to be a benign variant according to Clin...	Link		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	
0	rs9394492	(C;C)	<0.76x risk for restless legs			
0	rs9951307	(A;G)	0.10 decreased risk for brain edema after a str...			

3.2 Possibly Harmful Traits

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
3.2	rs2981582	(T;T)	1.7x higher risk of ER+ breast cancer		Link	
3	rs10897346	(C;C)	If depressed: 2.6x more likely to not respond t...			
3	rs2306402	(C;C)	1.18x increased risk for late-onset Alzheimer's...			
2.7	rs10830963	(C;G)	Increased type-2 diabetes risk; higher gestatio...		Link	
2.5	rs1121980	(C;T)	1.67x risk for obesity		Link	
2.5	rs13266634	(C;T)	Increased risk for type-2 diabetes	Link	Link	Link
2.5	rs1421085	(C;T)	~1.3x increased obesity risk		Link	Link
2.5	rs17696736	(G;G)	1.94x risk of type-1 diabetes		Link	
2.5	rs1799971	(A;G)	Stronger cravings for alcohol. if alcoholic: na...	Link	Link	Link
2.5	rs2073963	(G;G)	Increased risk of baldness			
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	rs3738919	(C;C)	1.94x risk of developing rheumatoid arthritis			
2.5	rs3780374	(A;G)	Substantially increased odds of developing V617...			
2.5	rs664143	(C;T)	Higher risk for number of cancers			
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	rs2305089	(T;T)	Higher risk for chordoma	Link	Link	
2.2	rs944289	(T;T)	1.69x increased thyroid cancer risk		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	rs1219648	(G;G)	1.64x risk for breast cancer		Link	
2.1	rs1329428	(G;G)	2x increased risk for macular degeneration			
2.1	rs1360780	(T;T)	1.3x increased risk for depression		Link	
2.1	rs2294008	(T;T)	Increased risk of gastric and bladder cancer	Link	Link	
2.1	rs2420946	(T;T)	1.64x risk for breast cancer			
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	rs10248420	(A;A)	7x less likely to respond to certain antidepres...		Link	
2	rs10455872	(A;G)	1:51x increased Coronary Heart disease risk			Link
2	rs1045642	(C;T)	Slower metaboliser for some drugs	Link	Link	Link
2	rs10513789	(G;T)	Increased risk of Parkinson's disease			
2	rs10871777	(A;G)	Adults likely to be 0.22 BMI units higher			
2	rs10883365	(G;G)	1.62x increased risk for developing Crohn's dis...		Link	
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	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	rs1544410	(A;A)	Increased risk of low bone mineral density diso...		Link	
2	rs1585215	(A;G)	2x increased risk for Hodgkin lymphoma			
2	rs16942	(A;G)	Very slightly increased breast cancer risk	Link	Link	Link
2	rs17001266	(-;C)	1.58x increased risk for schizophrenia in males...			
2	rs17228212	(C;T)	1.26x increased risk for heart disease		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	
2	rs17782313	(C;T)	Adults likely to be 0.22 BMI units higher		Link	Link
2	rs1800629	(A;G)	Complex; generally higher risk for certain dise...	Link	Link	Link

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
2	rs2201841	(C;T)	1.5x increased risk for Crohn's disease; 2x inc...		Link	
2	rs2235015	(G;G)	Somewhat less likely to respond to certain anti...	Link	Link	
2	rs2235040	(G;G)	7x less likely to respond to certain antidepres...	Link	Link	
2	rs2235067	(G;G)	7x less likely to respond to certain antidepres...			
2	rs2274223	(A;G)	1.5x increased risk for stomach and esophageal ...	Link	Link	Link
2	rs2305795	(A;G)	1.28x higher risk of narcolepsy compared to (G;...			Link
2	rs25487	(A;G)	2x higher risk for skin cancer; possibly other ...	Link	Link	Link
2	rs3738579	(C;C)	0.6x decreased risk for cervical cancer: but 1....			
2	rs3775948	(G;G)	Slightly higher risk for gout			
2	rs3793784	(C;G)	1.5x risk for ARMD		Link	Link
2	rs3802842	(C;C)	>1.17x increased risk of colorectal cancer		Link	
2	rs4027132	(A;A)	1.51x increased risk of developing bipolar diso...			
2	rs4148739	(A;A)	7x less likely to respond to certain antidepres...		Link	
2	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	rs493258	(A;G)	1.15x risk of Age Related Macular Degeneration			
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	rs6441286	(G;T)	1.54x chance of developing primary biliary cirr...		Link	
2	rs6457617	(C;T)	2.3x risk of rheumatoid arthritis		Link	
2	rs6532197	(G;G)	Increased risk of developing Parkinson's Diseas...		Link	
2	rs662799	(A;G)	1.4x higher early heart attack risk; less weigh...		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	rs699	(C;C)	Increased risk of hypertension	Link	Link	Link
2	rs7442295	(A;A)	~4x higher risk for hyperuracemia		Link	
2	rs7794745	(A;T)	Slightly increased risk for autism		Link	Link
2	rs828907	(G;T)	Slightly increased risk of bladder cancer and 2...			
2	rs854560	(A;A)	Higher risk for heart disease: diabetic retinop...	Link	Link	Link
2	rs9652490	(A;A)	~2x increased risk for Parkinson's disease: and...		Link	
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	rs1044396	(C;C)	Increased risk of Nicotine dependence among mal...	Link	Link	Link
2.0	rs2156921	(G;G)	1.29x increased risk for depression			
2.0	rs4911414	(G;T)	2-4x higher risk of sun sensitivity if part of ...		Link	
1.9	rs7923837	(A;G)	1.6x risk for T2D			
1.8	rs2278206	(T;T)	1.16x increased risk for asthma	Link	Link	
1.8	rs37973	(A;G)	Among asthmatics: 1.5x more likely to show less...			Link
1.6	rs11523871	(A;C)	1.6x increased breast cancer risk for women ove...	Link	Link	
1.6	rs2059693	(T;T)	1.6x increased risk for testicular cancer			
1.6	rs2981745	(C;T)	1.6x increased risk for breast cancer in female...			
1.6	rs3764880	(A;A)	1.2 - 1.8x increased tuberculosis risk	Link	Link	
1.5	rs10464059	(A;G)	Slightly increased risk of developing Parkinson...			
1.5	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	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	rs12469063	(A;G)	Slightly increased risk of developing restless ...			
1.5	rs12498742	(A;A)	1.25 increased risk for gout			
1.5	rs13149290	(C;T)	Slightly increased risk of developing prostate ...			
1.5	rs13376333	(C;T)	1.5x higher risk of atrial fibrillation		Link	
1.5	rs140701	(A;G)	Increased risk for anxiety disorders			
1.5	rs16944	(A;G)	Minorly increased risk of mental illness and os...		Link	
1.5	rs1801274	(T;T)	Complex; generally greater risk for cancer prog...	Link	Link	Link

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
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;A)	Slightly increased (1.5x) risk for RA	Link		
1.5	rs2272127	(C;C)	Associated with herpes and schizophrenia			
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	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	rs3745516	(A;G)	Slightly increased risk of developing primary b...			
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	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	rs486907	(A;G)	1.5x increased prostate cancer risk	Link	Link	Link
1.5	rs4979462	(C;T)	Slightly increased risk of developing primary b...			
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	rs6498169	(A;G)	1.14x risk of multiple sclerosis		Link	
1.5	rs6710341	(A;G)	Slightly increased risk of developing restless ...			
1.5	rs7454108	(C;T)	Single HLA-DQ8 haplotype			
1.5	rs763035	(C;T)	1.2x increased risk for rosacea			
1.5	rs7774434	(C;T)	Slightly increased risk of developing primary b...			
1.5	rs807701	(C;T)	Slightly increased dyslexia risk			
1.5	rs9303277	(C;T)	1.46x Slightly increased risk of developing pri...			
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	rs2230201	(A;G)	1.4x risk of lupus	Link		
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	
1.3	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	rs110419	(A;G)	1.3x increased risk for neuroblastoma			
1.3	rs13361189	(C;T)	1.3x increased risk for Crohn's disease		Link	
1.3	rs1746048	(C;C)	1.03 increased risk for coronary heart disease		Link	
1.3	rs2024513	(A;G)	1.3x higher risk for schizophrenia (among Han C...			
1.3	rs2295490	(A;G)	1.32x increased risk of early-onset type-2 diab...	Link	Link	
1.3	rs2736100	(G;T)	1.3x higher risk for glioma development: 2.1x r...		Link	
1.3	rs356219	(A;G)	1.3x increased risk for Parkinson's disease			
1.3	rs4958847	(A;G)	1.3x increased risk for Crohn's disease			
1.25	rs748404	(T;T)	Slightly increased risk (1.25) for lung 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	rs2076295	(G;T)	One copy of the risk allele (G): slightly incre...			
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	
1.2	rs419788	(A;G)	2.0x risk for lupus	Link		
1.2	rs4795067	(A;G)	Slight increase in risk for psoriatic arthritis...			

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
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;C)	1.3x high blood pressure risk			
1.1	rs11650354	(C;T)	Possible risk for allergic asthma	Link		
1.1	rs249954	(C;T)	Potentially increased risk of Breast Cancer			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	rs4324715	(C;T)	1.5x increased testicular cancer risk for men			
1.1	rs6800901	(T;T)	1.3x multiple myeloma risk			
1.1	rs6897876	(C;T)	Slight increase in testicular cancer risk for m...			
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	rs997669	(G;G)	Very slightly increased (1.18x) increased breas...			
1.05	rs2291834	(C;T)	Very slightly higher risk for myocardial infarc...			
1	rs10504861	(G;G)	Major allele: normal risk of migraine			
1	rs10761659	(A;G)	1.2x risk of Crohn's disease		Link	
1	rs1143674	(A;G)	1.3x increased autism risk	Link		
1	rs2228000	(T;T)	Statistically significant: but slight: increase...	Link	Link	Link
1	rs2546890	(A;G)	Higher risk of multiple sclerosis			
1	rs6932590	(T;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	rs1061646	(C;C)	1.16x increased risk for breast cancer	Link		Link
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	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
3.1	gs191	Problem metabolizing NSAIDs
3	gs127	Intermediate warfarin metabolizer
3	gs241	Lighter green: brown or hazel eye color
2.9	gs192	MTHFR polymorphisms affecting homocysteine
2.5	gs155	CYP3A5 non-expressor
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 ...
2.2	gs280	Light hair color for europeans
2	gs101	Probably able to digest milk
2	gs104	Restless legs syndrome risk
2	gs154	NAT2 Slow metabolizer
2	gs239	Reduced conversion of beta-carotene to retinol
2	gs246	APOE3/APOE3
2	gs249	Parkinson's Disease Risk
1.5	gs185	The beta blocker metoprolol is effective with 1...
1.5	gs186	HLA-B*5801 heterozygosity is possible: unfortun...
1.5	gs220	HLA-B*1502?
1.2	gs184	Able to taste bitterness.
1	gs182	CYP2D6*39
0	gs158	CYP1A2 normal metabolizer

4 Raw Data

The raw data used to create this report has been assigned the identifier ERS1176631 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/ERS1176631

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.