

PGP-UK Genomics Report for uk305BA4

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.

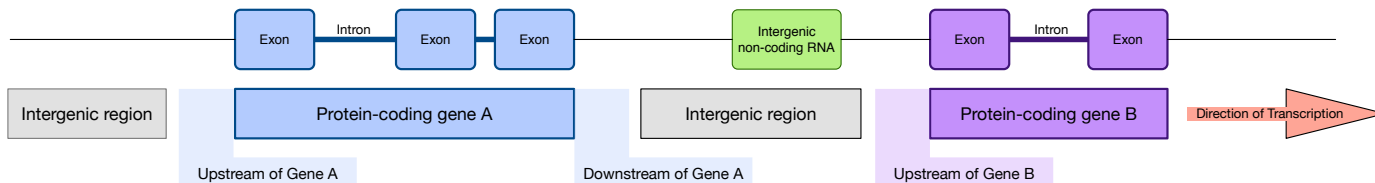


Figure 1: Diagram of gene structure indicating locations of potential variants

Feature	Count
Lines of input read	4958078
Variants filtered out	0
Novel / existing variants	497600 (10.1) / 4448397 (89.9)
Overlapped genes	56670
Overlapped transcripts	67483
Overlapped regulatory features	166868

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.

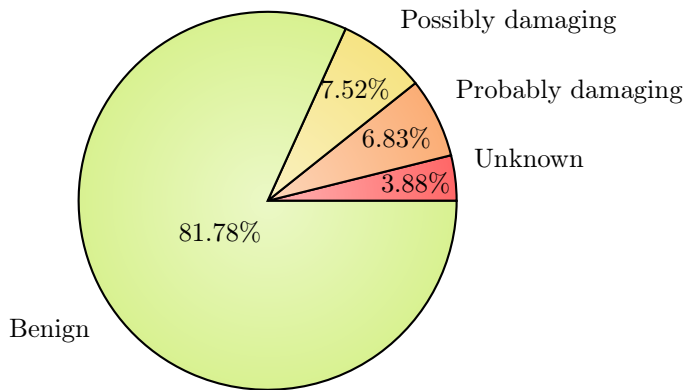


Figure 2: PolyPhen Summary

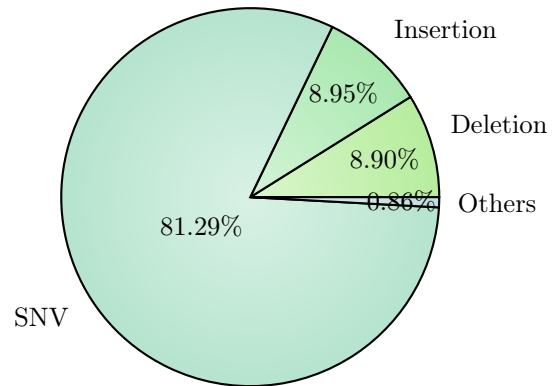


Figure 3: Variant Class

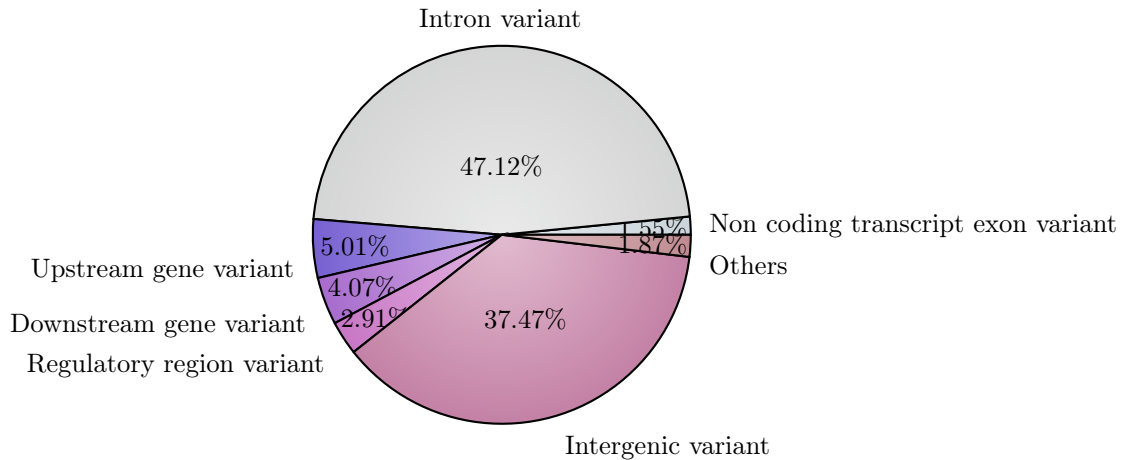


Figure 4: Consequence type

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).

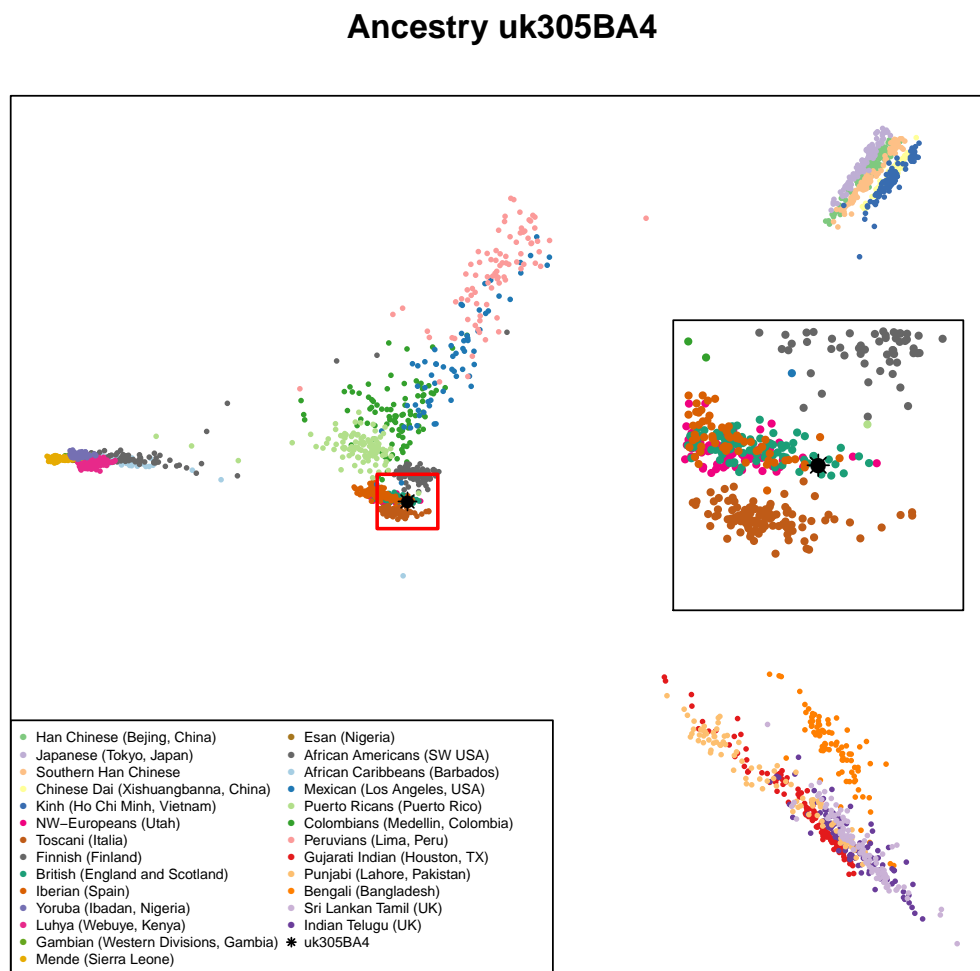


Figure 5: Ancestry Principal Component Analysis

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
2.2	rs3816873	(C;C)	Reduced risk of type-2 diabetes	Link	Link	Link
2.1	rs2511989	(A;G)	0.63x decreased age-related macular degeneratio...		Link	
2	rs1026732	(A;A)	<0.70x risk for restless legs		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	rs1128535	(G;G)	Reduced risk (0.77x) for Crohn's disease			
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	rs1544410	(G;G)	Decreased risk of low bone mineral density diso...		Link	
2	rs17070145	(C;T)	Increased memory performance			Link
2	rs1864163	(A;G)	Associated with higher HDL cholesterol		Link	
2	rs2241423	(A;A)	0.79 decreased risk for obesity			
2	rs261332	(A;G)	Associated with higher HDL cholesterol			
2	rs266729	(G;G)	0.73x decreased risk for colorectal cancer		Link	
2	rs3750817	(C;T)	0.78x reduced risk for breast cancer			
2	rs3782179	(C;T)	3x lower odds of testicular cancer risk for men...			
2	rs3819331	(T;T)	Lower risk of autism	Link		
2	rs3914132	(C;T)	Lower otosclerosis risk		Link	
2	rs4149268	(A;G)	Associated with higher HDL cholesterol		Link	
2	rs6505162	(A;C)	0.58x decreased risk for esophageal cancer	Link		
2	rs6807362	(G;G)	Decreased autism risk	Link	Link	
2	rs6855911	(A;G)	0.62x decreased risk for gout		Link	
2	rs763110	(C;T)	~0.80x reduced cancer risk			Link
2	rs7776725	(T;T)	Stronger bones		Link	
2	rs801114	(T;T)	0.78x decreased Basal Cell Carcinoma risk.		Link	

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
2	rs9272346	(A;G)	0.3x risk type-1 diabetes		Link	
2	rs9642880	(G;G)	Slightly lower risk of Bladder Cancer.		Link	
1.8	rs1800588	(C;T)	Higher HDL-C levels	Link	Link	
1.8	rs3814113	(C;T)	0.8x decreased risk for ovarian cancer		Link	
1.5	rs1063192	(C;T)	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	rs1165205	(A;A)	0.85x decreased gout risk		Link	
1.5	rs2007153	(A;A)	Decreased risk of schizophrenia in limited stud...			
1.5	rs309375	(G;G)	Smaller mosquito bites			
1.5	rs4149274	(C;T)	Associated with higher HDL (good) cholesterol			
1.5	rs4939883	(C;C)	Associated with higher HDL cholesterol		Link	
1.5	rs729302	(C;C)	0.89x decreased risk of developing rheumatoid a...			
1.5	rs9939609	(T;T)	Lower risk of obesity and Type-2 diabetes		Link	
1.4	rs6495446	(C;T)	0.8x reduced risk for chronic kidney disease			
1.4	rs6700125	(C;C)	0.7x decreased risk for ALS			
1.2	rs11246226	(A;C)	Decreased risk of schizophrenia in limited stud...		Link	
1.2	rs4686484	(G;G)	Slightly decreased risk for celiac disease			
1.2	rs9306160	(C;T)	0.75x (reduced) risk for metastasis in LN-/ER+ ...	Link	Link	
1.1	rs13333226	(A;G)	Slightly lower risk for hypertension			Link
1.1	rs2293347	(G;G)	Among NSCLC patients: better Gefitinib response...	Link		Link
1.1	rs7568369	(G;T)	0.90x reduced risk of obesity			
1	rs182549	(T;T)	Can digest milk.			Link
1	rs800292	(C;T)	1% decreased risk of macular degeneration	Link	Link	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	rs1126809	(A;G)	Slight increase in skin cancer risk	Link	Link	Link
0	rs12252	(T;T)	More resistant to influenza	Link		Link
0	rs16990018	(A;A)	PrP Codon 171 Asn - Non-pathogenic variant	Link		Link
0	rs17244841	(A;A)	More responsive to statin treatment		Link	Link
0	rs1799782	(C;C)	Lower risk for skin cancer	Link	Link	
0	rs1799945	(C;C)	Not a H63D hemochromatosis carrier.	Link	Link	Link
0	rs1800562	(G;G)	Not a C282Y hemochromatosis carrier.	Link	Link	Link
0	rs28933385	(G;G)	Prion protein Codon 200 (E) - Non pathogenic va...			Link
0	rs312481	(C;C)	Better response to certain calcium channel bloc...			
0	rs5065	(A;A)	1.12x risk on diuretic; if hypertensive: better...	Link	Link	Link
0	rs74315403	(G;G)	PrP codon 178 (D) - non pathogenic variant			Link
0	rs7495174	(A;A)	Blue/gray eyes more likely		Link	
0	rs7997012	(A;A)	~18% more likely to respond to citalopram		Link	Link

3.2 Possibly Harmful Traits

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
3.5	rs7574865	(T;T)	1.69x risk of rheumatoid arthritis; 2.4x risk o...		Link	Link
3	rs10897346	(C;C)	If depressed: 2.6x more likely to not respond t...			
3	rs12735723	(C;G)	Variegate porphyria carrier	Link		Link
3	rs13266634	(C;C)	Increased risk for type-2 diabetes	Link	Link	Link
3	rs2306402	(C;C)	1.18x increased risk for late-onset Alzheimer's...			
3	rs2981582	(C;T)	1.3x higher risk of ER+ breast cancer		Link	
3	rs3738579	(T;T)	1.5x - 2x increased risk for cervical cancer: H...			
3	rs55705857	(A;G)	6x increased risk of glioma of IDH1/IDH2 subtyp...			
3	rs6920220	(A;G)	1.2x risk Rheumatoid Arthritis		Link	
2.7	rs10830963	(C;G)	Increased type-2 diabetes risk; higher gestatio...		Link	
2.5	rs11190870	(T;T)	Possibly even more increased risk of scoliosis			
2.5	rs16969968	(A;G)	Slightly higher risk for nicotine dependence: l...	Link	Link	Link
2.5	rs1799971	(A;G)	Stronger cravings for alcohol. if alcoholic: na...	Link	Link	Link
2.5	rs187238	(G;G)	Hypertension increases risk 3.75x for sudden ca...			
2.5	rs2241880	(C;C)	2x-3x increased risk for Crohn's disease in Cau...	Link	Link	Link
2.5	rs2943634	(C;C)	Slightly higher risk of ischemic stroke		Link	
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	rs664143	(C;T)	Higher risk for number of cancers			
2.5	rs8034191	(C;T)	1.27x lung cancer risk		Link	
2.4	rs7966230	(G;G)	Slightly lower levels of plasma VWF			
2.3	rs37973	(G;G)	Among asthmatics: 2.3x more likely to show less...			Link
2.2	rs1052133	(G;G)	2x increased bladder cancer risk; 4.5x increase...	Link	Link	Link
2.2	rs2231137	(G;G)	~1.5-3x increased risk for ischemic stroke	Link	Link	Link
2.2	rs944289	(T;T)	1.69x increased thyroid cancer risk		Link	
2.1	rs1050152	(T;T)	2.1x increased risk of Crohn's disease	Link	Link	Link
2.1	rs1695	(G;G)	3.5x asthma risk in certain populations	Link	Link	Link
2.1	rs2254958	(C;T)	1.24x increased risk for Alzheimer's			
2.1	rs4430796	(A;A)	1.38x increased risk for prostate cancer		Link	
2	rs10086908	(C;T)	1.7x increased risk for prostate cancer			
2	rs1024611	(C;T)	Increased risk of exercise induced ischemia			Link
2	rs10248420	(A;A)	7x less likely to respond to certain antidepres...		Link	
2	rs10455872	(A;G)	1:51x increased Coronary Heart disease risk			Link
2	rs1045642	(C;T)	Slower metaboliser for some drugs	Link	Link	Link
2	rs1051730	(C;T)	1.3x increased risk of lung cancer	Link	Link	Link
2	rs10811661	(C;T)	1.2x increased risk for type-2 diabetes		Link	
2	rs10883365	(G;G)	1.62x increased risk for developing Crohn's dis...		Link	
2	rs10984447	(A;A)	>1.17x increased risk for multiple sclerosis		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	rs1219648	(A;G)	1.20x risk for breast cancer		Link	
2	rs12567232	(A;G)	Increased risk for Crohn's Disease		Link	
2	rs13254738	(A;C)	1.18x prostate cancer risk		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	rs16942	(A;G)	Very slightly increased breast cancer risk	Link	Link	Link
2	rs17001266	(-;C)	1.58x increased risk for schizophrenia in males...			
2	rs17435	(T;T)	1.4x increased risk for lupus			
2	rs1800896	(A;A)	1.8x increased prostate cancer risk			
2	rs2070600	(A;G)	1.5x increased risk for gastric cancer	Link	Link	
2	rs2073963	(G;T)	Increased risk of baldness			
2	rs2156921	(A;G)	1.29x increased risk for depression			

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
2	rs2201841	(C;T)	1.5x increased risk for Crohn's disease; 2x inc...		Link	
2	rs2230201	(G;G)	>1.4x risk of lupus	Link		
2	rs2235015	(G;G)	Somewhat less likely to respond to certain anti...	Link	Link	
2	rs2235040	(G;G)	7x less likely to respond to certain antidepres...	Link	Link	
2	rs2235067	(G;G)	7x less likely to respond to certain antidepres...			
2	rs2305480	(C;T)	3.5x increase in risk of asthma for Han Chinese...	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	(G;G)	2x higher risk for skin cancer; possibly other ...	Link	Link	Link
2	rs2707466	(G;G)	Weaker bones	Link	Link	
2	rs2908004	(C;C)	Weaker bones	Link	Link	
2	rs3129934	(C;T)	Increased risk of Multiple Sclerosis.		Link	
2	rs326	(A;A)	Lower HDL cholesterol		Link	Link
2	rs358806	(C;C)	1.78x increased risk of developing Type-2 diabe...		Link	
2	rs3738919	(A;C)	1.94x risk of developing rheumatoid arthritis			
2	rs3746444	(C;T)	~1.2x increased risk for cancer	Link		
2	rs3793784	(C;G)	1.5x risk for ARMD		Link	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	(C;T)	Higher risk for endometrial cancer	Link	Link	Link
2	rs5174	(A;G)	1.3x increased risk for heart disease	Link	Link	Link
2	rs520354	(A;A)	Increased risk in men for biliary conditions			
2	rs629242	(C;T)	Somewhat higher risk for prostate cancer			
2	rs638405	(G;G)	2x increased ALZ risk in ApoE4 carriers	Link		
2	rs6457617	(C;T)	2.3x risk of rheumatoid arthritis		Link	
2	rs6897932	(C;C)	1.08x increased risk for multiple sclerosis	Link	Link	Link
2	rs6997709	(G;G)	1.5x higher risk for hypertension			
2	rs744373	(C;T)	1.17x risk of Alzheimer's			
2	rs7807268	(C;G)	1.3x risk for Crohn's disease		Link	
2	rs7923837	(G;G)	3.2x risk for T2D			
2	rs7961152	(A;C)	1.2x higher risk for hypertension			
2	rs828907	(T;T)	Increased risk of bladder cancer and 2x risk of...			
2	rs854560	(A;A)	Higher risk for heart disease: diabetic retinop...	Link	Link	Link
2	rs9525638	(T;T)	Weaker bones			
1.8	rs143383	(T;T)	1.3x increased risk for osteoarthritis		Link	Link
1.8	rs4474514	(A;G)	3x increased testicular cancer risk for men		Link	
1.7	rs2024513	(A;A)	1.7x higher risk for schizophrenia (among Han C...			
1.61	rs4295627	(G;G)	1.85x higher risk for glioma development		Link	
1.6	rs1260326	(T;T)	Slightly higher risk for gout	Link	Link	Link
1.6	rs2736100	(G;G)	1.6x higher risk for glioma development		Link	
1.6	rs3764880	(A;A)	1.2 - 1.8x increased tuberculosis risk	Link	Link	
1.6	rs3775948	(C;G)	Slightly higher risk for gout			
1.6	rs4959039	(G;G)	1.6x higher risk for multiple sclerosis			
1.5	rs10260404	(C;T)	1.20x risk of developing ALS		Link	
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	rs1154155	(G;T)	1.94x increased risk for narcolepsy		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	rs12210050	(C;T)	Slightly higher risk for basal cell carcinoma		Link	
1.5	rs1223271	(A;G)	Slightly increased risk of developing Parkinson...		Link	
1.5	rs12431733	(C;T)	Slightly increased risk of developing Parkinson...		Link	

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
1.5	rs13149290	(C;T)	Slightly increased risk of developing prostate ...			
1.5	rs13181	(G;T)	1.12x increased risk for cutaneous melanoma	Link	Link	Link
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	rs144848	(G;T)	Very slightly increased breast cancer risk	Link	Link	Link
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	rs1994090	(G;T)	Slightly increased risk of developing Parkinson...		Link	
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	rs2697962	(A;G)	Slightly increased risk of developing Parkinson...			
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	rs3745516	(A;G)	Slightly increased risk of developing primary b...			
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	rs464049	(C;T)	Increased risk of schizophrenia in limited stud...			
1.5	rs4982731	(C;C)	Possible higher risk of childhood acute lymphob...			
1.5	rs619203	(C;G)	Increases susceptibility to Myocardial Infarcti...	Link	Link	
1.5	rs6601764	(C;T)	1.16x increased risk of developing Crohn's dise...		Link	
1.5	rs6896702	(C;T)	Slightly increased risk of developing Parkinson...			
1.5	rs6908425	(C;T)	1.63x increased risk of developing Crohn's dise...		Link	
1.5	rs7536563	(A;G)	1.12x risk of multiple sclerosis		Link	
1.5	rs7850258	(G;G)	Slightly higher odds of developing primary hypo...			
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	rs9652490	(A;G)	Slightly increased risk of developing Parkinson...		Link	
1.5	rs966221	(C;C)	1.5x increased stroke risk certain populations			
1.4	rs1126497	(C;T)	1.4x increased risk for breast cancer	Link	Link	Link
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	rs3184504	(C;T)	Slightly increased risk for celiac disease	Link	Link	
1.4	rs498872	(T;T)	1.4x 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	rs1375144	(C;T)	1.32x increased risk of developing bipolar diso...			
1.3	rs1434536	(A;G)	1.29x increased breast cancer risk			
1.3	rs16847548	(C;T)	1.3x increased risk for sudden cardiac death in...			
1.3	rs1746048	(C;C)	1.03 increased risk for coronary heart disease		Link	
1.3	rs2295490	(A;G)	1.32x increased risk of early-onset type-2 diab...	Link	Link	
1.3	rs34330	(T;T)	1.2x higher breast cancer risk; 1.3x higher ris...			
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	rs1344706	(T;T)	1.2x increased risk for schizophrenia		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	rs2072590	(G;T)	1.2x increased risk for ovarian cancer			
1.2	rs2076295	(G;T)	One copy of the risk allele (G): slightly incre...			

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
1.2	rs2252586	(A;G)	1.2x higher risk for glioma development			
1.2	rs2665390	(C;T)	1.2x increased risk for ovarian cancer			
1.2	rs3176336	(T;T)	Slightly higher (1.25x) higher risk for breast ...			
1.2	rs3740878	(A;A)	1.46x type II diabetes risk; common	Link		Link
1.2	rs419788	(A;G)	2.0x risk for lupus	Link		
1.2	rs449647	(A;T)	Possibly lower levels of ApoE			
1.2	rs4795067	(A;G)	Slight increase in risk for psoriatic arthritis...			
1.2	rs4977756	(A;G)	1.39x higher risk for glioma development		Link	
1.2	rs6897876	(C;C)	Slight increase in testicular cancer risk for m...			
1.2	rs9858542	(A;G)	1.1x risk Crohn's Disease	Link	Link	
1.17	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	rs2295190	(G;T)	Slightly increased risk for ovarian cancer in w...	Link	Link	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	rs5030737	(C;T)	Carrier of mannose binding deficiency but of lo...	Link	Link	Link
1.1	rs7171755	(A;G)	Very slight decrease in cortical thickness and ...			
1.1	rs7412	(C;C)	More likely to gain weight if taking olanzapine...	Link	Link	Link
1.1	rs889312	(A;C)	Very slightly higher risk for breast cancer		Link	
1.1	rs925391	(C;C)	More likely to go bald; common			
1.09	rs12050604	(A;C)	Very slightly increased risk for lung cancer			
1.05	rs2291834	(C;T)	Very slightly higher risk for myocardial infarc...			
1	rs10761659	(A;G)	1.2x risk of Crohn's disease		Link	
1	rs2282679	(A;C)	Somewhat lower vitamin D levels			
1	rs2546890	(A;G)	Higher risk of multiple sclerosis			
1	rs3194051	(A;G)	1.12x risk of type-1 diabetes	Link	Link	Link
1	rs6932590	(T;T)	1.1x increased risk for schizophrenia		Link	
1	rs987525	(A;C)	2.5x increased risk for cleft lip		Link	
0	rs1004819	(C;C)	1.5x risk of Crohn's disease		Link	
0	rs1061646	(C;C)	1.16x increased risk for breast cancer	Link		Link
0	rs3813929	(C;C)	Possible weight gain if taking olanzapine		Link	Link
0	rs6277	(C;C)	1.6x higher schizophrenia risk	Link	Link	Link
0	rs6314	(C;C)	Higher risk for RA	Link	Link	
0	rs7787082	(G;G)	7x less likely to respond to certain antidepres...		Link	
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	gs241	Lighter green: brown or hazel eye color
2.5	gs155	CYP3A5 non-expressor
2.5	gs281	Part of the 88% of the population claimed not t...
2.5	gs285	You will lose 2.5x as much weight on a low fat ...
2	gs101	Probably able to digest milk
2	gs154	NAT2 Slow metabolizer
2	gs181	CYP2D6*2
2	gs239	Reduced conversion of beta-carotene to retinol
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	gs220	HLA-B*1502?
1.5	gs247	Parkinson's Disease Risk
1.2	gs184	Able to taste bitterness.
1	gs163	CYP2D6*2A
0	gs158	CYP1A2 normal metabolizer

4 Raw Data

The raw data used to create this report has been assigned the identifier ERS1176597 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/ERS1176597>

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.