PGP-UK Genomics Report for uk486E45

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	5013075
Variants filtered out	0
Novel / existing variants	507914 (10.2) / 4493782 (89.8)
Overlapped genes	56743
Overlapped transcripts	67535
Overlapped regulatory features	167879

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.
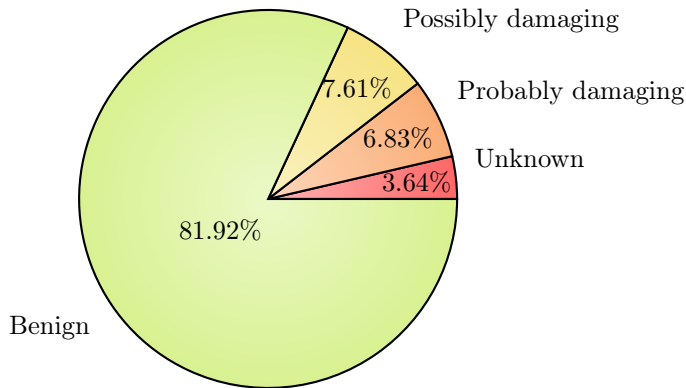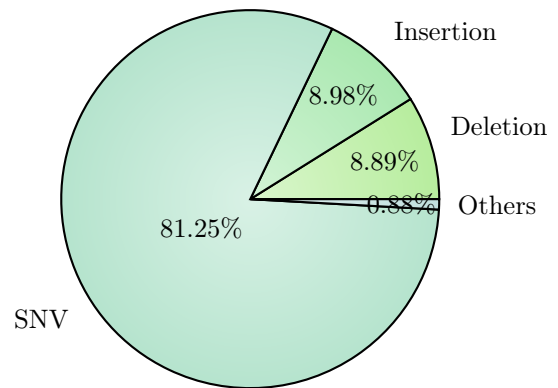
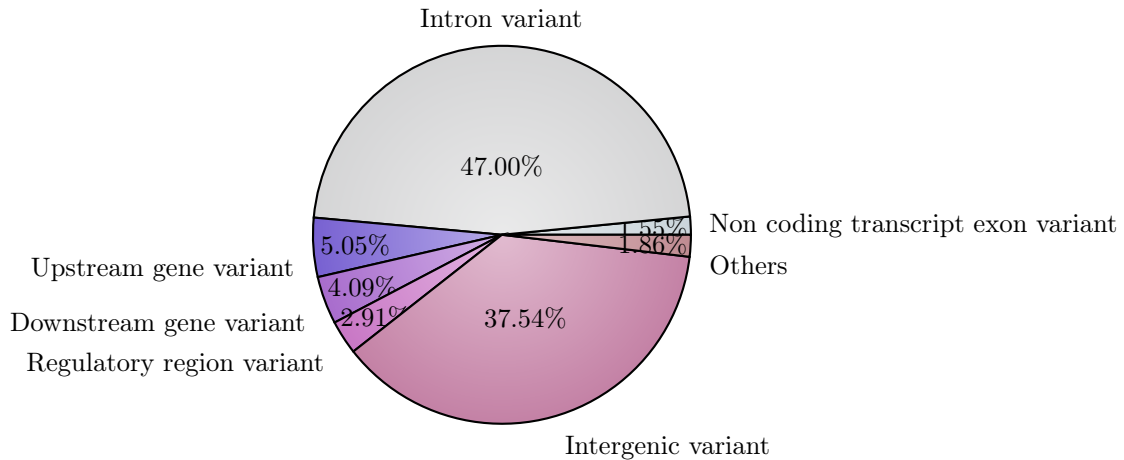Figure 2: PolyPhen Summary

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 uk486E45

Legend:
- Han Chinese (Bejing, China)
- Japanese (Tokyo, Japan)
- Southern Han Chinese
- Chinese Dai (Xishuangbanna, China)
- Kinh (Ho Chi Minh, Vietnam)
- NW–Europeans (Utah)
- Toscani (Italia)
- Finnish (Finland)
- British (England and Scotland)
- Iberian (Spain)
- Yoruba (Ibadan, Nigeria)
- Luhya (Webuye, Kenya)
- Gambian (Western Divisions, Gambia)
- Mende (Sierra Leone)
- Esan (Nigeria)
- African Americans (SW USA)
- African Caribbeans (Barbados)
- Mexican (Los Angeles, USA)
- Puerto Ricans (Puerto Rico)
- Colombians (Medellin, Colombia)
- Peruvians (Lima, Peru)
- Gujarati Indian (Houston, TX)
- Punjabi (Lahore, Pakistan)
- Bengali (Bangladesh)
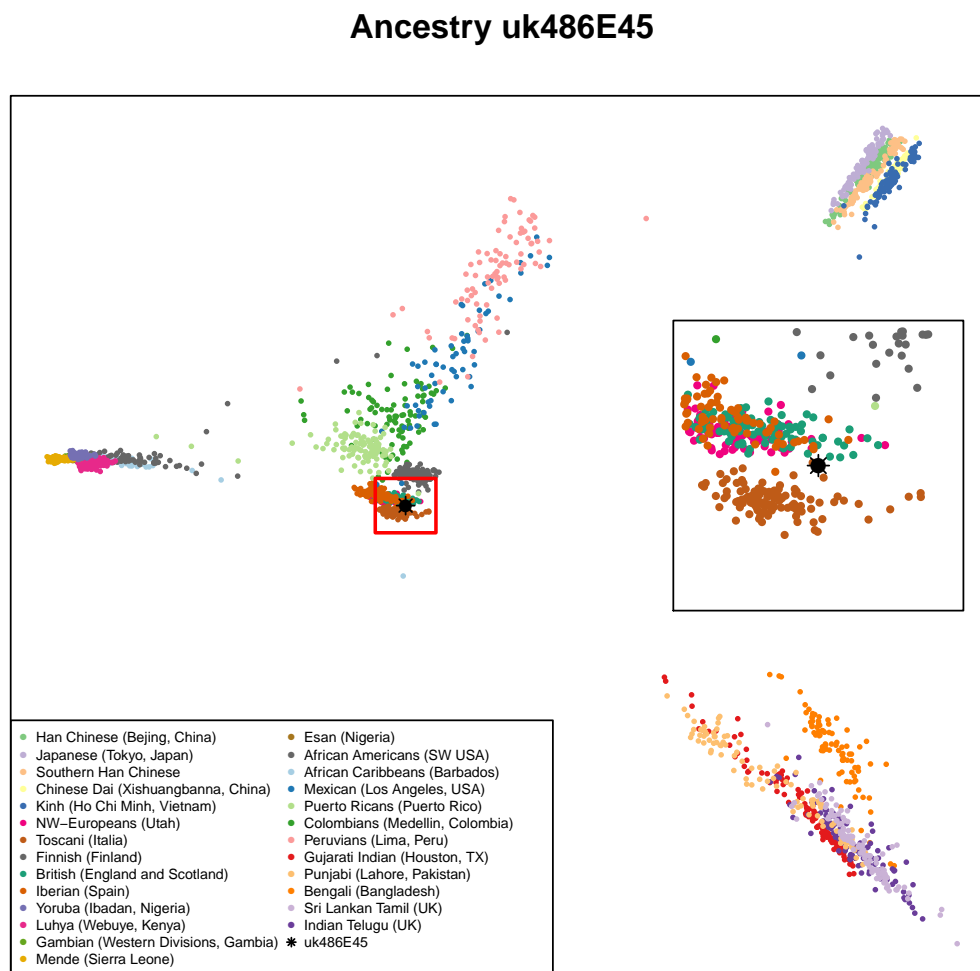- Sri Lankan Tamil (UK)
- Indian Telugu (UK)
- ✳ uk486E45

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
2.5	rs2943634	(A;A)	Lower risk of ischemic stroke		Link	
2.4	rs9272346	(G;G)	0.08x risk type-1 diabetes		Link	
2.1	rs2511989	(A;G)	0.63x decreased age-related macular degeneratio...		Link	
2.1	rs6505162	(A;A)	0.43x decreased risk for esophageal cancer	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	rs11045585	(A;A)	24% chance (lower than average) of docetaxel-in...		Link	
2	rs11635424	(A;A)	<0.70x risk for restless legs		Link	
2	rs12593813	(A;A)	<0.71x risk for restless legs		Link	
2	rs17070145	(C;T)	Increased memory performance			Link
2	rs1799884	(G;G)	Mothers have typical Birth-Weight babies. Sligh...			
2	rs2073963	(T;T)	Reduced risk of baldness			
2	rs2241423	(A;A)	0.79 decreased risk for obesity			
2	rs2542052	(C;C)	Better odds of living to 100			
2	rs261332	(A;G)	Associated with higher HDL cholesterol			
2	rs3738579	(C;T)	0.5x decreased risk for cervical cancer: HNSCC:...			
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	rs4149268	(A;G)	Associated with higher HDL cholesterol		Link	
2	rs4585	(G;G)	Slightly higher (1.35x) odds of good metformin ...			
2	rs7776725	(T;T)	Stronger bones		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	rs1800588	(T;T)	Higher HDL-C levels	Link	Link	
1.8	rs4714156	(C;C)	<0.61x risk for restless legs			

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
1.8	rs854560	(T;T)	0.5x lower risk of ovarian cancer	Link	Link	Link
1.6	rs1061170	(T;T)	Lower risk for AMD: generally longer live than ...	Link	Link	Link
1.5	rs11136000	(C;T)	0.84x decreased risk for Alzheimer's disease		Link	
1.5	rs11212617	(C;C)	Somewhat increased likelihood of treatment succ...			Link
1.5	rs3851179	(A;G)	0.85x decreased risk for Alzheimer's disease		Link	
1.5	rs4149274	(C;T)	Associated with higher HDL (good) cholesterol			
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	rs9939609	(T;T)	Lower risk of obesity and Type-2 diabetes		Link	
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	rs9306160	(C;T)	0.75x (reduced) risk for metastasis in LN-/ER+ ...	Link	Link	
1.1	rs10166942	(C;T)	0.85x lower risk for migraines			
1.1	rs11172113	(C;T)	0.9x lower risk for migraines			
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	rs4988235	(T;T)	Can digest milk			Link
1.1	rs7568369	(G;T)	0.90x reduced risk of obesity			
1	rs182549	(T;T)	Can digest milk.			Link
1	rs2952768	(C;T)	Slightly less drug dependence: decreased effect...			Link
1	rs7850258	(A;A)	Slightly lower odds of developing primary hypot...			
1	rs800292	(C;T)	1% decreased risk of macular degeneration	Link	Link	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	rs16947	(A;A)	Homozygous for CYP2D6 variants (non-CYP2D6*1)	Link	Link	Link
0	rs16990018	(A;A)	PrP Codon 171 Asn - Non-pathogenic variant	Link		Link
0	rs17244841	(A;A)	More responsive to statin treatment		Link	Link
0	rs1799782	(C;C)	Lower risk for skin cancer	Link	Link	
0	rs28933385	(G;G)	Prion protein Codon 200 (E) - Non pathogenic va...			Link
0	rs312481	(C;C)	Better response to certain calcium channel bloc...			
0	rs5065	(A;A)	1.12x risk on diuretic; if hypertensive: better...	Link	Link	Link
0	rs6259	(G;G)	Best inverse correlation between tea-drinking: ...	Link	Link	
0	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
0	rs9394492	(C;C)	<0.76x risk for restless legs			

3.2 Possibly Harmful Traits

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
3.1	rs10830963	(G;G)	Increased type-2 diabetes risk; higher gestatio...		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	rs1801282	(C;G)	Unconfirmed higher risk of cardiovascular disea...	Link	Link	Link
3	rs2306402	(C;T)	1.18x increased risk for late-onset Alzheimer's...			
3	rs3803662	(T;T)	1.6x increased risk for breast cancer		Link	
3	rs4151667	(A;T)	Age related macular degeneration	Link	Link	Link
3	rs6920220	(A;G)	1.2x risk Rheumatoid Arthritis		Link	
2.5	rs10484554	(C;T)	2.8x increased risk for psoriasis		Link	
2.5	rs10490924	(G;T)	2.7x risk for age related macular degeneration	Link	Link	Link
2.5	rs17696736	(G;G)	1.94x risk of type-1 diabetes		Link	
2.5	rs187238	(G;G)	Hypertension increases risk 3.75x for sudden ca...			
2.5	rs2004640	(T;T)	1.4x increased risk for SLE		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	rs664143	(T;T)	Higher risk for number of cancers			
2.5	rs8034191	(C;T)	1.27x lung cancer risk		Link	
2.5	rs891512	(A;G)	Higher blood pressure than G;G	Link		
2.3	rs7966230	(C;G)	Slightly lower levels of plasma VWF			
2.2	rs1052133	(G;G)	2x increased bladder cancer risk; 4.5x increase...	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	
2.2	rs964184	(G;G)	Increased risk of hypertriglyceridemia		Link	
2.1	rs17563	(C;C)	Risk for otosclerosis	Link	Link	Link
2.1	rs2383207	(G;G)	Increased risk for heart disease			
2.1	rs5751876	(T;T)	Significantly higher anxiety levels after moder...	Link		
2.1	rs646776	(A;A)	1.2x risk of coronary artery disease		Link	
2.1	rs7837688	(T;T)	1.7x increased risk for prostate cancer			
2.1	rs795484	(A;G)	Increased morphine dose requirement and postope...			
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	rs1042838	(G;T)	1.28x risk for endometrial ovarian cancer; over...	Link	Link	
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	rs10811661	(C;T)	1.2x increased risk for type-2 diabetes		Link	
2	rs10984447	(A;A)	>1.17x increased risk for multiple sclerosis		Link	
2	rs11190870	(C;T)	Possibly increased risk of scoliosis			
2	rs11229030	(C;C)	Higher odds of Crohn's disease			
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;A)	Increased risk for Crohn's Disease		Link	
2	rs12770228	(A;A)	2x increased risk for meningioma			
2	rs1544410	(A;A)	Increased risk of low bone mineral density diso...		Link	
2	rs1585215	(A;G)	2x increased risk for Hodgkin lymphoma			
2	rs16944	(A;A)	Increased risk for osteoarthritis		Link	
2	rs17228212	(C;C)	>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	rs1800896	(A;G)	1.6x increased prostate cancer risk			
2	rs2201841	(C;C)	1.5x increased risk for Crohn's disease		Link	

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
2	rs2230199	(C;G)	1.6x+ risk of ARMD	Link	Link	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	rs2352028	(T;T)	Increased risk of lung cancer in non-smokers an...		Link	
2	rs25487	(G;G)	2x higher risk for skin cancer; possibly other ...	Link	Link	Link
2	rs2736100	(T;T)	Higher risk of Interstitial lung disease: and t...		Link	
2	rs3129934	(C;T)	Increased risk of Multiple Sclerosis.		Link	
2	rs3738919	(A;C)	1.94x risk of developing rheumatoid arthritis			
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	rs4633	(C;T)	Higher risk for endometrial cancer	Link	Link	Link
2	rs4792311	(A;A)	Increased risk of prostate cancer	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	rs6441286	(G;T)	1.54x chance of developing primary biliary cirr...		Link	
2	rs662799	(A;G)	1.4x higher early heart attack risk; less weigh...		Link	
2	rs6700125	(T;T)	1.76x increased risk for ALS			
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	rs6997709	(G;G)	1.5x higher risk for hypertension			
2	rs7190458	(A;G)	Slightly higher pancreatic cancer risk	Link		
2	rs7442295	(A;A)	~4x higher risk for hyperuracemia		Link	
2	rs7639618	(C;T)	1.45x increased osteoarthritis risk	Link		
2	rs7794745	(A;T)	Slightly increased risk for autism		Link	Link
2	rs7807268	(C;G)	1.3x risk for Crohn's disease		Link	
2	rs7961152	(A;C)	1.2x higher risk for hypertension			
2	rs828907	(G;T)	Slightly increased risk of bladder cancer and 2...			
2	rs855913	(G;T)	Reduced survival with ALS		Link	
2	rs9525638	(T;T)	Weaker bones			
2	rs965513	(A;A)	3.1x increased thyroid cancer risk		Link	
2.0	rs1434536	(A;A)	1.94x increased breast cancer risk			
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	rs1136287	(C;T)	1.5x increased risk of wet ARMD in a Taiwanese ...	Link	Link	
1.8	rs143383	(T;T)	1.3x increased risk for osteoarthritis		Link	Link
1.8	rs4474514	(A;G)	3x increased testicular cancer risk for men		Link	
1.7	rs1042713	(A;A)	1.7x increased risk that pediatric inhaler use ...	Link	Link	Link
1.6	rs1260326	(T;T)	Slightly higher risk for gout	Link	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	
1.6	rs3775948	(C;G)	Slightly higher risk for gout			
1.5	rs10492519	(A;G)	Slightly increased risk of developing prostate ...			
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	rs12498742	(A;A)	1.25 increased risk for gout			
1.5	rs13181	(G;T)	1.12x increased risk for cutaneous melanoma	Link	Link	Link
1.5	rs144848	(G;T)	Very slightly increased breast cancer risk	Link	Link	Link
1.5	rs1801274	(C;T)	Complex; generally greater risk for cancer prog...	Link	Link	Link

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
1.5	rs1975197	(C;T)	1.3x increased risk of developing restless legs...		Link	
1.5	rs2007153	(G;G)	Increased risk of schizophrenia in limited stud...			
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	rs2280714	(A;A)	1.4x increased risk of SLE			
1.5	rs2697962	(A;G)	Slightly increased risk of developing Parkinson...			
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	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	rs3790565	(C;T)	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;T)	~1.2x increased risk for several types of cance...		Link	
1.5	rs4027132	(A;G)	1.39x increased risk of developing bipolar diso...			
1.5	rs4506565	(A;T)	1.4x increased risk for type-2 diabetes		Link	
1.5	rs4538475	(A;G)	Slightly increased risk of developing Parkinson...		Link	
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	rs4845618	(G;T)	1.7x increased melanoma risk			
1.5	rs486907	(A;G)	1.5x increased prostate cancer risk	Link	Link	Link
1.5	rs5219	(C;T)	1.3x increased risk for type-2 diabetes	Link	Link	Link
1.5	rs6498169	(A;G)	1.14x risk of multiple sclerosis		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	rs7536563	(A;G)	1.12x risk of multiple sclerosis		Link	
1.5	rs807701	(C;T)	Slightly increased dyslexia risk			
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	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	(T;T)	1.4x increased risk for breast cancer	Link	Link	Link
1.4	rs1447295	(A;C)	1.4x increased risk of prostate cancer		Link	
1.4	rs1545843	(A;A)	1.4x increased risk for depression (for those u...			
1.4	rs2046210	(C;T)	1.4x increased breast cancer risk		Link	Link
1.4	rs3131296	(G;G)	1.4x increased risk for schizophrenia		Link	
1.3	rs1047286	(C;T)	1.3x increased risk for age-related macular deg...	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	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	rs2024513	(A;G)	1.3x higher risk for schizophrenia (among Han C...			
1.3	rs2059693	(C;T)	1.3x increased risk for testicular cancer			
1.3	rs356219	(A;G)	1.3x increased risk for Parkinson's disease			
1.25	rs748404	(T;T)	Slightly increased risk (1.25) for lung cancer...		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	rs2252586	(A;G)	1.2x higher risk for glioma development			
1.2	rs2814707	(A;G)	1.2x increased risk for ALS		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	rs6010620	(A;G)	1.2x higher risk for glioma development: 1.17x ...		Link	

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
1.17	rs17465637	(A;C)	1.17x higher risk for myocardial infarction	Link	Link	
1.1	rs11037909	(C;T)	1.27x type II diabetes risk	Link		
1.1	rs11110912	(C;G)	1.3x high blood pressure risk			
1.1	rs13387042	(A;G)	1.12x increased risk for breast cancer		Link	
1.1	rs1800450	(A;G)	Carrier of mannose binding deficiency but of lo...	Link	Link	Link
1.1	rs2651899	(A;G)	1.1x higher risk for migraines			
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	rs7171755	(A;G)	Very slight decrease in cortical thickness and ...			
1.1	rs7412	(C;T)	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	rs10504861	(G;G)	Major allele: normal risk of migraine			
1	rs10761659	(A;G)	1.2x risk of Crohn's disease		Link	
1	rs12752888	(C;C)	Faster progression of mild cognitive impairment...			
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;G)	1.12x risk of type-1 diabetes	Link	Link	Link
1	rs5326	(A;G)	Possible psychiatric risks			
1	rs6932590	(T;T)	1.1x increased risk for schizophrenia		Link	
1	rs6974491	(A;G)	Higher risk of coeliac and/or inflammatory bowe...			
1	rs987525	(A;C)	2.5x increased risk for cleft lip		Link	
1.0	rs11246226	(A;A)	Increased risk of schizophrenia in limited stud...		Link	
0	rs1042173	(T;T)	Among alcoholics: likely to be heavier drinkers...			
0	rs1061646	(C;C)	1.16x increased risk for breast cancer	Link		Link
0	rs4712653	(T;T)	2x increased risk for neuroblastoma			
0	rs6277	(C;C)	1.6x higher schizophrenia risk	Link	Link	Link
0	rs6314	(C;C)	Higher risk for RA	Link	Link	
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
3	gs127	Intermediate warfarin metabolizer
3	gs137	5x risk of thyroid cancer
2.9	gs192	MTHFR polymorphisms affecting homocysteine
2.5	gs102	ALS risk
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	gs283	You will lose 2.5x as much weight on a low carb...
2.4	gs297	Lower heart attack risk than average
2	gs101	Probably able to digest milk
2	gs269	APOE E2/E3
1.5	gs220	HLA-B*1502?
1.5	gs247	Parkinson's Disease Risk
0	gs158	CYP1A2 normal metabolizer

4 Raw Data

The raw data used to create this report has been assigned the identifier ERS1176565 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/ERS1176565

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.