PGP-UK Genomics Report for ukC97BE5

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.

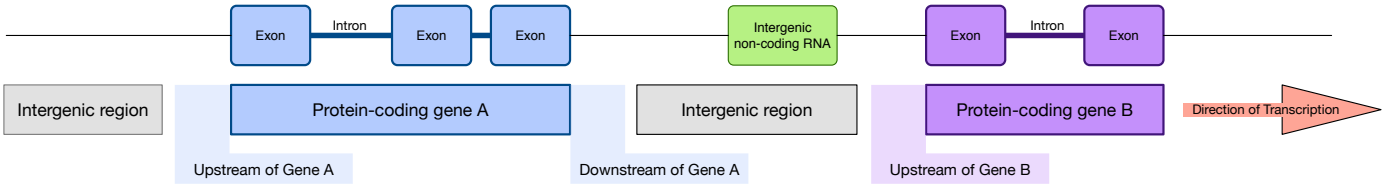

Figure 1: Diagram of gene structure indicating locations of potential variants

Feature	Count
Lines of input read	5023525
Variants filtered out	0
Novel / existing variants	516709 (10.3) / 4495374 (89.7)
Overlapped genes	56699
Overlapped transcripts	67535
Overlapped regulatory features	167457

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.

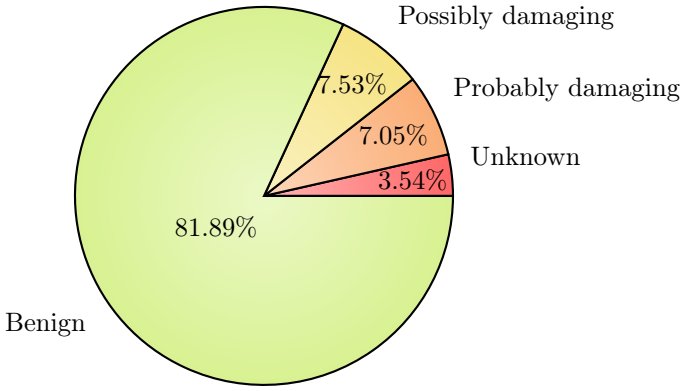

Figure 2: PolyPhen Summary

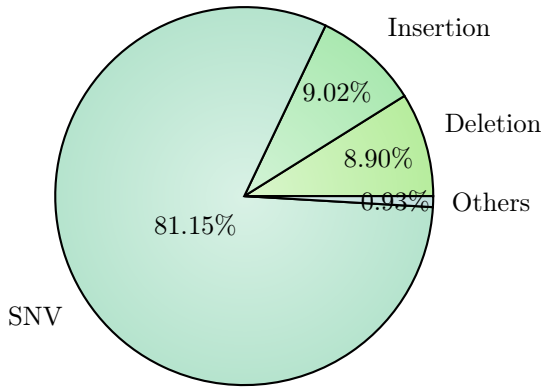

Figure 3: Variant Class

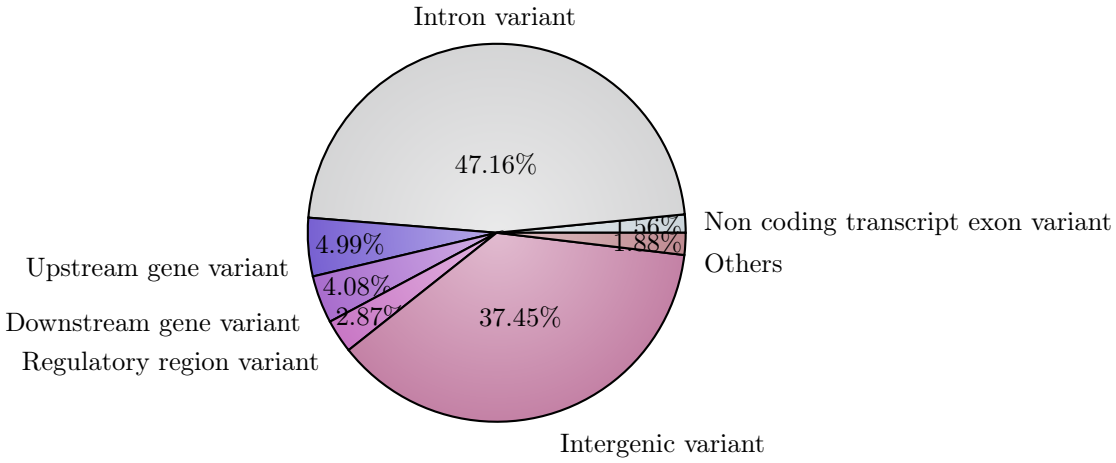

Figure 4: Consequence type

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).

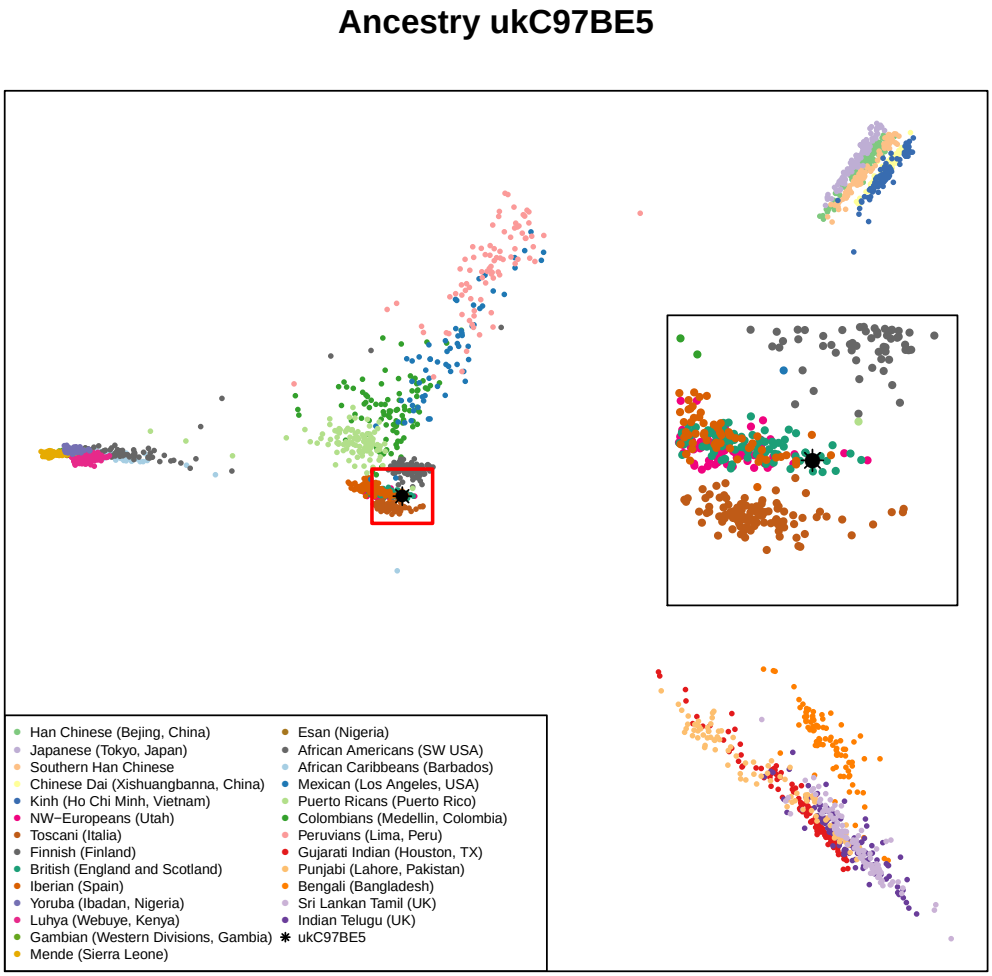

Figure 5: Ancestry Principal Component Analysis

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
2.1	rs6505162	(A;A)	0.43x decreased risk for esophageal cancer	Link		
2	rs1012053	(A;C)	0.625x reduced risk of Bipolar Disorder.		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	rs174537	(T;T)	Lower LDL-C and total cholesterol			
2	rs1800972	(G;G)	Reduced risk for Crohn's disease; reduced risk ...	Link		
2	rs261332	(A;G)	Associated with higher HDL cholesterol			
2	rs2707466	(A;A)	Stronger bones	Link	Link	
2	rs2908004	(T;T)	Stronger bones	Link	Link	
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	rs6807362	(G;G)	Decreased autism risk	Link	Link	
2	rs763110	(C;T)	~0.80x reduced cancer risk			Link
2	rs925391	(C;T)	Lower odds of going bald			
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	rs1800588	(C;T)	Higher HDL-C levels	Link	Link	
1.8	rs4714156	(C;C)	<0.61x risk for restless legs			
1.6	rs10801935	(C;C)	0.3x decreased risk of breast cancer			
1.5	rs1063192	(C;T)	0.71x reduced risk of myocardial infarction			
1.5	rs11212617	(A;C)	Somewhat increased likelihood of treatment succ...			Link
1.5	rs3790844	(C;T)	Slightly reduced risk (0.77x) for pancreatic ca...			
1.5	rs4149274	(C;T)	Associated with higher HDL (good) cholesterol			

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
1.5	rs4939883	(C;C)	Associated with higher HDL cholesterol		Link	
1.5	rs610932	(A;A)	A allele associated with reduced risk of Alzhei...			
1.5	rs729302	(A;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	rs1165205	(A;T)	0.85x decreased gout risk		Link	
1.4	rs6495446	(C;T)	0.8x reduced risk for chronic kidney disease			
1.2	rs6048	(G;G)	Slightly lower risk (10-20%) of deep vein throm...	Link	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	rs2293347	(G;G)	Among NSCLC patients: better Gefitinib response...	Link		Link
1.1	rs7568369	(G;T)	0.90x reduced risk of obesity			
1	rs182549	(C;T)	Can digest milk.			Link
1	rs2494732	(T;T)	Lower odds of psychosis	Link	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
1.0	rs11246226	(C;C)	Decreased risk of schizophrenia in limited stud...		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	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	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	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

3.2 Possibly Harmful Traits

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
3.5	rs875858	(C;T)	Docetaxel sensitive?			
3	rs10897346	(C;C)	If depressed: 2.6x more likely to not respond t...			
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	rs6920220	(A;G)	1.2x risk Rheumatoid Arthritis		Link	
3	rs7754840	(C;G)	1.3x increased risk for type-2 diabetes		Link	
2.5	rs10484554	(C;T)	2.8x increased risk for psoriasis		Link	
2.5	rs13266634	(C;T)	Increased risk for type-2 diabetes	Link	Link	Link
2.5	rs16969968	(A;G)	Slightly higher risk for nicotine dependence: l...	Link	Link	Link
2.5	rs187238	(G;G)	Hypertension increases risk 3.75x for sudden ca...			
2.5	rs2073963	(G;G)	Increased risk of baldness			
2.5	rs3780374	(A;G)	Substantially increased odds of developing V617...			
2.5	rs664143	(C;T)	Higher risk for number of cancers			
2.5	rs795484	(A;A)	Even more increased morphine dose requirement a...			
2.5	rs8034191	(C;T)	1.27x lung cancer risk		Link	
2.5	rs891512	(A;G)	Higher blood pressure than G;G	Link		
2.4	rs1143679	(A;G)	1.78x increased risk for SLE	Link	Link	
2.4	rs7966230	(G;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	
2.2	rs944289	(T;T)	1.69x increased thyroid cancer risk		Link	
2.1	rs10811661	(T;T)	1.2x increased risk for type-2 diabetes		Link	
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	rs2254958	(C;T)	1.24x increased risk for Alzheimer's			
2.1	rs4363657	(C;T)	4.5x increased myopathy risk for statin users		Link	
2.1	rs4444903	(G;G)	3.5x risk of hep-cancer in cirrhosis patients; ...			Link
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	rs10306114	(A;G)	Higher risk of bleeding during coronary angiogr...			Link
2	rs1042838	(G;T)	1.28x risk for endometrial ovarian cancer; over...	Link	Link	
2	rs1045642	(C;T)	Slower metaboliser for some drugs	Link	Link	Link
2	rs10488631	(C;T)	2x increased risk of developing SLE; 1.6x incre...		Link	
2	rs1051730	(C;T)	1.3x increased risk of lung cancer	Link	Link	Link
2	rs10937823	(C;T)	Some association with bipolar disorder			
2	rs10984447	(A;G)	1.17x increased risk for multiple sclerosis		Link	
2	rs11190870	(C;T)	Possibly increased risk of scoliosis			
2	rs1160312	(A;A)	1.6x increased risk of Male Pattern Baldness.		Link	
2	rs11983225	(T;T)	7x less likely to respond to certain antidepres...		Link	
2	rs12431733	(T;T)	Increased risk of developing Parkinson's Diseas...		Link	
2	rs12567232	(A;G)	Increased risk for Crohn's Disease		Link	
2	rs1265181	(C;G)	Increased risk for psoriasis		Link	
2	rs13254738	(A;C)	1.18x prostate cancer risk		Link	
2	rs1333048	(A;C)	1.3x increased coronary artery disease risk			
2	rs1544410	(A;A)	Increased risk of low bone mineral density diso...		Link	
2	rs1585215	(A;G)	2x increased risk for Hodgkin lymphoma			
2	rs1691053	(A;G)	Increased risk of developing prostate cancer			
2	rs16942	(A;G)	Very slightly increased breast cancer risk	Link	Link	Link
2	rs16944	(G;G)	Increased risk of mental disorders		Link	
2	rs17001266	(-;C)	1.58x increased risk for schizophrenia in males...			

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
2	rs17228212	(C;T)	1.26x increased risk for heart disease		Link	
2	rs1734791	(A;A)	1.4x increased risk for lupus			
2	rs17576	(A;G)	Higher risk for MI and lung cancer: and COPD in...	Link	Link	
2	rs1800896	(A;A)	1.8x increased prostate cancer risk			
2	rs1867277	(A;A)	2x increased risk for thyroid cancer			
2	rs2201841	(C;T)	1.5x increased risk for Crohn's disease; 2x inc...		Link	
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	rs2383206	(A;G)	1.4x increased risk for heart disease			
2	rs2383207	(A;G)	Increased risk for heart disease			
2	rs25487	(A;G)	2x higher risk for skin cancer; possibly other ...	Link	Link	Link
2	rs27388	(A;A)	Increased risk of developing schizophrenia			
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	rs3738919	(A;C)	1.94x risk of developing rheumatoid arthritis			
2	rs3746444	(C;T)	~1.2x increased risk for cancer	Link		
2	rs3775948	(G;G)	Slightly higher risk for gout			
2	rs3842787	(C;T)	Higher risk of bleeding during coronary angiogr...	Link	Link	
2	rs4148739	(A;A)	7x less likely to respond to certain antidepres...		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;G)	Increased risk of prostate cancer	Link	Link	Link
2	rs4825476	(G;G)	1.9x higher risk of suicidal thoughts when taki...		Link	
2	rs493258	(A;G)	1.15x risk of Age Related Macular Degeneration			
2	rs4968451	(A;C)	1.61x increased risk for meningioma			
2	rs520354	(A;A)	Increased risk in men for biliary conditions			
2	rs587776825	(-;C)	Associated with MODY3; maturity onset of diabet...	Link		Link
2	rs6457617	(C;T)	2.3x risk of rheumatoid arthritis		Link	
2	rs6498169	(A;A)	>1.14x risk of multiple sclerosis		Link	
2	rs663048	(G;T)	Significantly increased risk of developing lung...	Link	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	rs6922269	(A;A)	1.6x risk of coronary artery disease		Link	
2	rs6997709	(G;T)	1.2x higher risk for hypertension			
2	rs699	(C;T)	Increased risk of hypertension	Link	Link	Link
2	rs7442295	(A;A)	~4x higher risk for hyperuracemia		Link	
2	rs7776725	(C;C)	Weaker bones		Link	
2	rs7807268	(C;C)	1.4x 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	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;A)	3.1x increased thyroid cancer risk		Link	
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
1.8	rs4474514	(A;G)	3x increased testicular cancer risk for men		Link	
1.8	rs6700125	(C;T)	1.2x increased risk for ALS			
1.6	rs1537415	(C;G)	1.6x increased risk for periodontitis		Link	

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
1.6	rs2059693	(T;T)	1.6x increased risk for testicular cancer			
1.5	rs10260404	(C;T)	1.20x risk of developing ALS		Link	
1.5	rs10492519	(A;G)	Slightly increased risk of developing prostate ...			
1.5	rs10757272	(C;T)	1.30x increased risk for Coronary artery diseas...			
1.5	rs10859871	(A;C)	Slight (~1.2x) increase in endometriosis risk			
1.5	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	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	rs12469063	(A;G)	Slightly increased risk of developing restless ...			
1.5	rs12498742	(A;A)	1.25 increased risk for gout			
1.5	rs13149290	(C;C)	Slightly increased risk of developing prostate ...			
1.5	rs1360517	(A;G)	Higher susceptibility for AIDS		Link	
1.5	rs144848	(G;T)	Very slightly increased breast cancer risk	Link	Link	Link
1.5	rs17221417	(C;G)	1.3x higher risk for Crohn's disease		Link	
1.5	rs1975197	(C;T)	1.3x increased risk of developing restless legs...		Link	
1.5	rs1994090	(G;T)	Slightly increased risk of developing Parkinson...		Link	
1.5	rs2177369	(C;C)	1.5x increased risk for Alzheimer's disease			
1.5	rs2240340	(A;G)	Slightly increased (1.5x) risk for RA	Link		
1.5	rs2241880	(C;T)	1.4x increased risk for Crohn's disease in Cauc...	Link	Link	Link
1.5	rs2272127	(C;C)	Associated with herpes and schizophrenia			
1.5	rs2280714	(A;A)	1.4x increased risk of SLE			
1.5	rs2464196	(C;T)	~1.5x increased lung cancer risk	Link	Link	Link
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	rs3764880	(A;G)	Possible 1.2 - 1.8x increased tuberculosis susc...	Link	Link	
1.5	rs3814570	(C;T)	1.3x increased risk for Crohn's disease with il...			
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	rs4538475	(A;G)	Slightly increased risk of developing Parkinson...		Link	
1.5	rs464049	(T;T)	Increased risk of schizophrenia in limited stud...			
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	rs5746059	(A;A)	Slightly higher fat mass			
1.5	rs6435862	(G;T)	1.7x higher risk of aggressive neuroblastoma		Link	
1.5	rs6601764	(C;T)	1.16x increased risk of developing Crohn's dise...		Link	
1.5	rs699473	(C;T)	~1.5x increased brain tumor risk			
1.5	rs7341475	(G;G)	1.58x increased schizophrenia risk for women		Link	
1.5	rs7454108	(C;T)	Single HLA-DQ8 haplotype			
1.5	rs7536563	(A;G)	1.12x risk of multiple sclerosis		Link	
1.5	rs7774434	(C;T)	Slightly increased risk of developing primary b...			
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...			
1.4	rs1126497	(T;T)	1.4x increased risk for breast cancer	Link	Link	Link
1.4	rs1545843	(A;A)	1.4x increased risk for depression (for those u...			
1.4	rs1800693	(G;G)	Slight (1.4x) increase in risk for multiple scl...	Link	Link	Link
1.4	rs1801157	(A;G)	1.4x higher risk for breast cancer			
1.4	rs3131296	(G;G)	1.4x increased risk for schizophrenia		Link	
1.4	rs4959039	(A;G)	1.4x higher risk for multiple sclerosis			
1.34	rs17465637	(C;C)	1.34x higher risk for myocardial infarction	Link	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			

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
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	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	rs34330	(C;T)	1.3x higher risk for endometrial cancer (in Chi...			
1.3	rs4295627	(G;T)	1.36x higher risk for glioma development		Link	
1.25	rs748404	(T;T)	Slightly increased risk (1.25) for lung cancer...		Link	
1.2	rs10865331	(A;G)	1.2x higher risk for ankylosing spondylitis			
1.2	rs1344706	(T;T)	1.2x increased risk for schizophrenia		Link	
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...			
1.2	rs2814707	(A;G)	1.2x increased risk for ALS		Link	
1.2	rs3176336	(T;T)	Slightly higher (1.25x) higher risk for breast ...			
1.2	rs35677470	(A;G)	2x higher risk for scleroderma	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	rs4686484	(A;A)	Slightly increased risk for celiac disease			
1.2	rs4977756	(A;G)	1.39x higher risk for glioma development		Link	
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	
1.2	rs9858542	(A;G)	1.1x risk Crohn's Disease	Link	Link	
1.1	rs11037909	(C;T)	1.27x type II diabetes risk	Link		
1.1	rs11110912	(C;C)	1.3x high blood pressure risk			
1.1	rs11650354	(C;T)	Possible risk for allergic asthma	Link		
1.1	rs11650494	(A;G)	Slightly higher prostate cancer risk			
1.1	rs13387042	(A;G)	1.12x increased risk for breast cancer		Link	
1.1	rs249954	(C;T)	Potentially increased risk of Breast Cancer			Link
1.1	rs2651899	(A;G)	1.1x higher risk for migraines			
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	rs4324715	(C;T)	1.5x increased testicular cancer risk for men			
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;A)	Very slight descrease in cortical thickness and...			
1.1	rs7412	(C;C)	More likely to gain weight if taking olanzapine...	Link	Link	Link
1.07	rs2291834	(C;C)	Very slightly higher risk for myocardial infarc...			
1	rs10504861	(G;G)	Major allele: normal risk of migraine			
1	rs1143674	(A;G)	1.3x increased autism risk	Link		
1	rs2435357	(A;A)	Slightly higher (2x?) risk for Hirschsprung dis...			Link
1	rs2546890	(A;G)	Higher risk of multiple sclerosis			
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	
0.1	rs601338	(A;G)	Susceptible to Norovirus infections	Link	Link	Link
0	rs1042173	(T;T)	Among alcoholics: likely to be heavier drinkers...			
0	rs10761659	(A;A)	1.5x risk of Crohn's disease		Link	
0	rs3761418	(A;A)	1.3x increased risk for depression			
0	rs4293393	(T;T)	1.25x Increased Risk of CKD for T allele in ...			
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	gs137	5x risk of thyroid cancer
2.9	gs192	MTHFR polymorphisms affecting homocysteine
2.5	gs155	CYP3A5 non-expressor
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	gs101	Probably able to digest milk
2	gs154	NAT2 Slow metabolizer
2	gs239	Reduced conversion of beta-carotene to retinol
2	gs246	APOE3/APOE3
1.5	gs247	Parkinson's Disease Risk
1.2	gs184	Able to taste bitterness.
0	gs158	CYP1A2 normal metabolizer

4 Raw Data

The raw data used to create this report has been assigned the identifier ERS1176628 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/ERS1176628

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.