PGP-UK Genomics Report for ukD768D4

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.
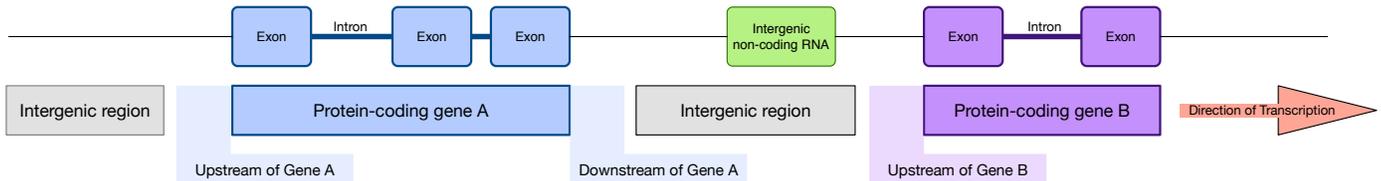

Figure 1: Diagram of gene structure indicating locations of potential variants

Feature	Count
Lines of input read	4903292
Variants filtered out	0
Novel / existing variants	464182 (9.5) / 4427199 (90.5)
Overlapped genes	56862
Overlapped transcripts	67595
Overlapped regulatory features	165554

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.
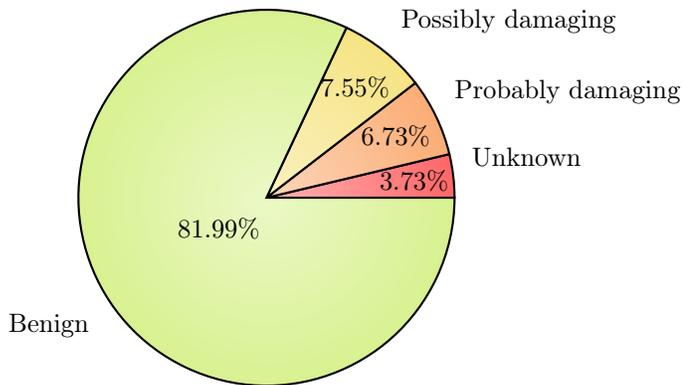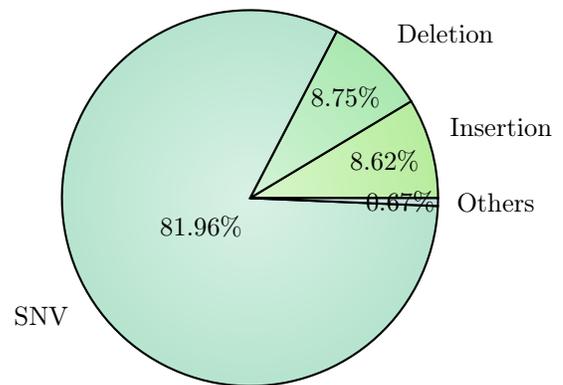
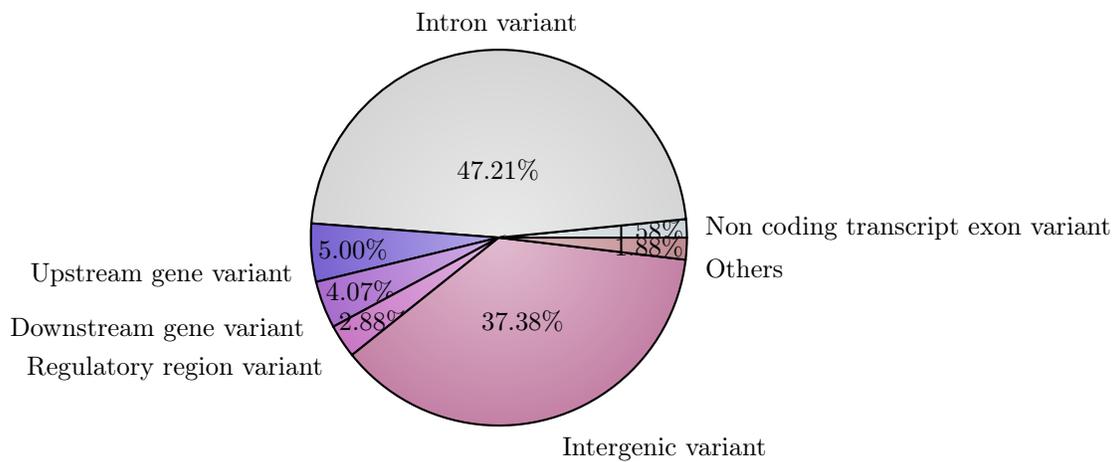Figure 2: PolyPhen Summary

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).
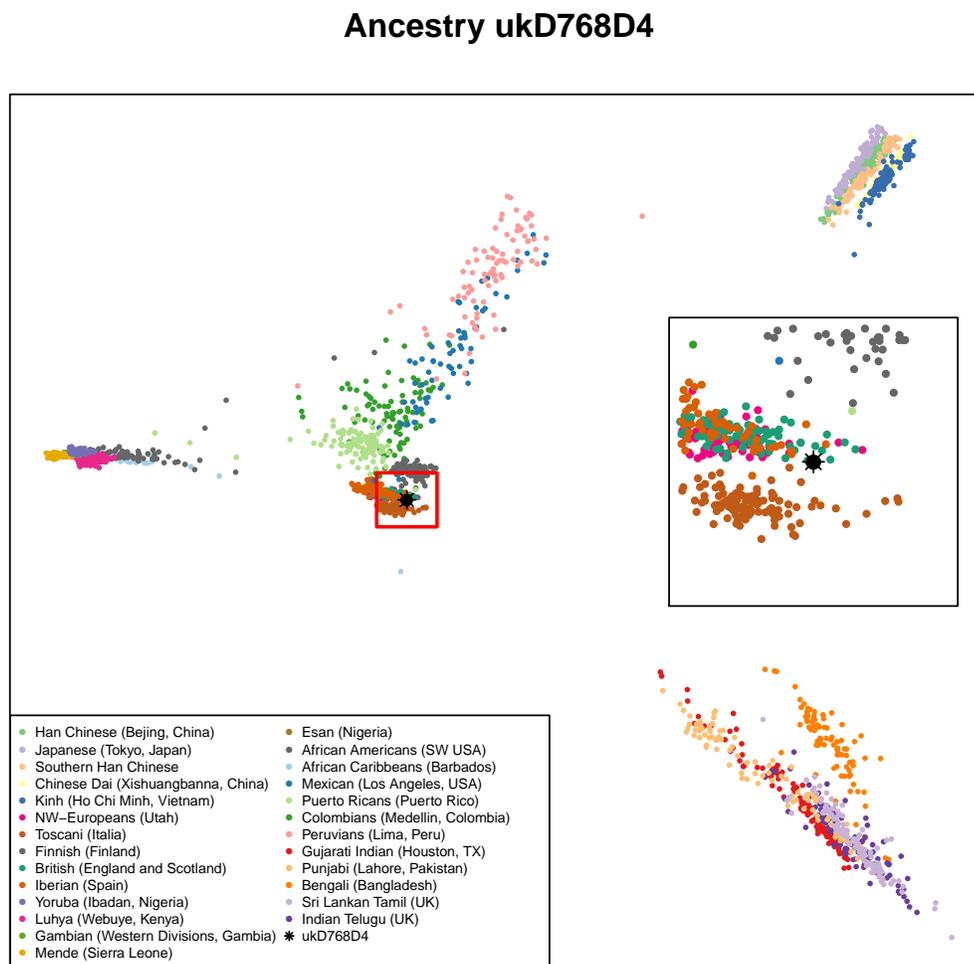
Ancestry ukD768D4

Figure 5: Ancestry Principal Component Analysis

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3 Traits (based on SNPedia information)

Existing research has associated many variants with phenotypic traits, some of which can be perceived as beneficial while others appear to have a harmful effect. Some traits are complex and can be affected by several variants. It is likely that some of these would confer a higher risk while others a lower risk of trait manifestation. These can not be combined linearly to produce an actual risk of disease.

It is important to note that in most cases genomic data is probabilistic, not deterministic- i.e. having a genetic predisposition for a disease is not a diagnosis; rather, it shows an increased likelihood of developing that disease. Also, one person can have both potentially beneficial and harmful variants in the same gene, or associated with the same disease.

Some variants can also affect certain populations more, or will only affect a particular gender. For example, a variant for higher risk of endometriosis in the sequence of a male will not directly affect that person, but can be passed on to descendants.

While many traits are the result of a unique variant, many are the combination of several variants throughout the genome. In SNPedia, these are called genosets. These can integrate some of the information already present in the single variant tables, or be the combination of variants that have no phenotypic effect on their own, but contribute to a trait when together.

The variants in the following tables are sorted by magnitude. This is an subjective measure defined in SNPedia to highlight the perceived importance of the genotype described. At the moment this scale goes from 0 to 10. You can read more about it by visiting their explanatory webpage.

As our knowledge grows, the interpretation of the effect of certain variants might change. Clicking on the links in the genome report tables will take you to websites containing more information about each variant.

3.1 Possibly Beneficial Traits

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
2.8	rs6046	(T;T)	Better blood pressure: lower risk of myocardial...	Link	Link	Link
2.5	rs2943634	(A;A)	Lower risk of ischemic stroke		Link	
2.1	rs2511989	(A;G)	0.63x decreased age-related macular degeneratio...		Link	
2.1	rs547154	(A;C)	0.47x decreased risk for AMD			Link
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	rs10504861	(A;G)	Reduced risk of migraine without aura			
2	rs1056836	(G;G)	0.3x decreased risk for prostate cancer	Link	Link	Link
2	rs1160312	(G;G)	Reduced risk of Baldness.		Link	
2	rs1544410	(G;G)	Decreased risk of low bone mineral density diso...		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;G)	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	rs6807362	(G;G)	Decreased autism risk	Link	Link	
2	rs7776725	(T;T)	Stronger bones		Link	
2	rs8070723	(A;G)	0.18x reduced risk of developing progressive su...			
2	rs9272346	(A;G)	0.3x risk type-1 diabetes		Link	
2	rs9642880	(G;G)	Slightly lower risk of Bladder Cancer.		Link	
1.8	rs1746048	(C;T)	0.94 decreased risk for coronary heart disease		Link	
1.8	rs1800588	(C;T)	Higher HDL-C levels	Link	Link	
1.8	rs4714156	(C;C)	<0.61x risk for restless legs			
1.8	rs7101429	(A;G)	0.70x reduced risk for Alzheimer's risk			
1.5	rs1026732	(A;G)	0.70x risk for restless legs		Link	
1.5	rs1063192	(C;T)	0.71x reduced risk of myocardial infarction			

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
1.5	rs11212617	(A;C)	Somewhat increased likelihood of treatment succ...			Link
1.5	rs11635424	(A;G)	0.70x risk for restless legs		Link	
1.5	rs12593813	(A;G)	0.71x risk for restless legs		Link	
1.5	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		
1.4	rs1165205	(A;T)	0.85x decreased gout risk		Link	
1.2	rs9306160	(C;T)	0.75x (reduced) risk for metastasis in LN-/ER+ ...	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	rs182549	(T;T)	Can digest milk.			Link
1	rs800292	(C;T)	1% decreased risk of macular degeneration	Link	Link	Link
0.1	rs1726866	(C;C)	Can taste bitter	Link	Link	Link
0	rs10427255	(T;T)	Lowest odds of photic sneeze reflex			
0	rs1047781	(A;A)	ABH blood group "Secretor" status if Japanese	Link	Link	Link
0	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	rs6259	(G;G)	Best inverse correlation between tea-drinking: ...	Link	Link	
0	rs7305115	(A;A)	Individuals showed a significantly lower risk o...	Link	Link	
0	rs74315403	(G;G)	PrP codon 178 (D) - non pathogenic variant			Link
0	rs7495174	(A;A)	Blue/gray eyes more likely		Link	
0	rs9394492	(C;C)	<0.76x risk for restless legs			

3.2 Possibly Harmful Traits

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
4	rs7158782	(G;G)	4x higher risk of adverse side-effect if taking...			
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...			
3	rs3738579	(T;T)	1.5x - 2x increased risk for cervical cancer: H...			
3	rs891512	(A;A)	Higher blood pressure than G;G	Link		
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	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	rs339331	(T;T)	Prostate cancer risk			
2.5	rs3780374	(A;G)	Substantially increased odds of developing V617...			
2.5	rs613872	(G;T)	~5 fold higher risk for Fuchs' dystrophy: a cor...			
2.5	rs664143	(C;T)	Higher risk for number of cancers			
2.3	rs7966230	(C;G)	Slightly lower levels of plasma VWF			
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	rs1585215	(G;G)	3.5x increased risk for Hodgkin lymphoma			
2.1	rs2231137	(A;G)	~1.5-3x increased risk for ischemic stroke	Link	Link	Link
2.1	rs2254958	(C;T)	1.24x increased risk for Alzheimer's			
2.1	rs2294008	(T;T)	Increased risk of gastric and bladder cancer	Link	Link	
2.1	rs4430796	(A;A)	1.38x increased risk for prostate cancer		Link	
2.1	rs4693596	(C;C)	2x odds of myopathy if taking statins			
2.1	rs5186	(A;C)	~1.4x increased risk of hypertension	Link	Link	Link
2.1	rs7837688	(G;T)	1.7x increased risk for prostate cancer			
2.1	rs944289	(C;T)	1.3x increased thyroid cancer risk		Link	
2	rs10090154	(C;T)	1.4x increased risk for prostate cancer			
2	rs10248420	(A;A)	7x less likely to respond to certain antidepres...		Link	
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	rs10937823	(C;T)	Some association with bipolar disorder			
2	rs10984447	(A;G)	1.17x increased risk for multiple sclerosis		Link	
2	rs11045585	(A;G)	63% chance (higher than average) of docetaxel-i...		Link	
2	rs11171739	(C;C)	1.75x risk of developing Type-1 diabetes		Link	
2	rs11190870	(C;T)	Possibly increased risk of scoliosis			
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	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	rs17435	(T;T)	1.4x increased risk for lupus			
2	rs17696736	(A;G)	1.34x risk of type-1 diabetes		Link	
2	rs17782313	(C;T)	Adults likely to be 0.22 BMI units higher		Link	Link
2	rs1800896	(A;G)	1.6x increased prostate cancer risk			
2	rs2143340	(C;T)	Increased risk of dyslexia and poor reading per...			
2	rs2156921	(A;G)	1.29x increased risk for depression			
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	

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
2	rs2235067	(G;G)	7x less likely to respond to certain antidepres...			
2	rs2274223	(A;G)	1.5x increased risk for stomach and esophageal ...	Link	Link	Link
2	rs2305480	(C;T)	3.5x increase in risk of asthma for Han Chinese...	Link	Link	
2	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	rs241448	(C;T)	1.51x increased risk for Alzheimer's	Link		Link
2	rs25487	(A;G)	2x higher risk for skin cancer; possibly other ...	Link	Link	Link
2	rs2707466	(G;G)	Weaker bones	Link	Link	
2	rs2736990	(C;C)	Increased risk of developing Parkinson's Diseas...		Link	
2	rs27388	(A;A)	Increased risk of developing schizophrenia			
2	rs2908004	(C;C)	Weaker bones	Link	Link	
2	rs3025039	(C;T)	2.6x increased risk for ARMD in a Taiwanese pop...			
2	rs3129934	(C;T)	Increased risk of Multiple Sclerosis.		Link	
2	rs351855	(C;T)	1.2x increased risk for prostate cancer	Link	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	rs3775948	(G;G)	Slightly higher risk for gout			
2	rs3793784	(C;G)	1.5x risk for ARMD		Link	Link
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	rs4444903	(A;G)	3.5x risk of hep-cancer in cirrhosis patients; ...			Link
2	rs4464148	(C;C)	1.35x increased risk for colorectal cancer			
2	rs4792311	(A;G)	Increased risk of prostate cancer	Link	Link	Link
2	rs493258	(G;G)	1.15x risk of Age Related Macular Degeneration			
2	rs5174	(A;A)	1.3x increased risk for heart disease	Link	Link	Link
2	rs520354	(A;G)	Increased risk in men for biliary conditions			
2	rs638405	(G;G)	2x increased ALZ risk in ApoE4 carriers	Link		
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	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	rs6997709	(G;G)	1.5x 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	rs7774434	(C;C)	Increased risk of developing primary biliary ci...			
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	rs854560	(A;T)	Higher risk for heart disease: diabetic retinop...	Link	Link	Link
2	rs9525638	(T;T)	Weaker bones			
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	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	rs143383	(T;T)	1.3x increased risk for osteoarthritis		Link	Link
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
1.8	rs6700125	(C;T)	1.2x increased risk for ALS			
1.6	rs1537415	(C;G)	1.6x increased risk for periodontitis		Link	
1.6	rs2046210	(T;T)	1.6x increased breast cancer risk in certain wo...		Link	Link
1.6	rs3764880	(A;A)	1.2 - 1.8x increased tuberculosis risk	Link	Link	

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
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	(C;C)	Slight (~1.4x) increase in endometriosis risk			
1.5	rs1169300	(A;G)	~1.5x increased lung cancer risk			
1.5	rs1223271	(A;G)	Slightly increased risk of developing Parkinson...		Link	
1.5	rs12498742	(A;A)	1.25 increased risk for gout			
1.5	rs13149290	(C;T)	Slightly increased risk of developing prostate ...			
1.5	rs140701	(A;G)	Increased risk for anxiety disorders			
1.5	rs144848	(G;T)	Very slightly increased breast cancer risk	Link	Link	Link
1.5	rs16944	(A;G)	Minorly increased risk of mental illness and os...		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	rs199533	(C;T)	Slightly increased risk of developing Parkinson...	Link		
1.5	rs2076295	(G;G)	Slightly increased risk for pulmonary fibrosis ...			
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	rs2272127	(C;C)	Associated with herpes and schizophrenia			
1.5	rs2280714	(A;A)	1.4x increased risk of SLE			
1.5	rs2286812	(C;T)	~2x higher risk for Fuchs' dystrophy: a corneal...			
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	rs2881766	(G;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	rs3814570	(C;T)	1.3x increased risk for Crohn's disease with il...			
1.5	rs3825776	(A;G)	1.3x increased risk for ALS		Link	
1.5	rs401681	(C;C)	~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	rs464049	(C;T)	Increased risk of schizophrenia in limited stud...			
1.5	rs4785763	(A;A)	2x higher risk for melanoma		Link	
1.5	rs5219	(C;T)	1.3x increased risk for type-2 diabetes	Link	Link	Link
1.5	rs5746059	(A;G)	Slightly higher fat mass			
1.5	rs619203	(C;G)	Increases susceptibility to Myocardial Infarcti...	Link	Link	
1.5	rs6435862	(G;T)	1.7x higher risk of aggressive neuroblastoma		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	rs7850258	(G;G)	Slightly higher odds of developing primary hypo...			
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	rs995030	(G;G)	Non-protective against testicular cancer		Link	
1.4	rs1126497	(C;T)	1.4x increased risk for breast cancer	Link	Link	Link
1.4	rs1447295	(A;C)	1.4x increased risk of prostate cancer		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	rs3184504	(C;T)	Slightly increased risk for celiac disease	Link	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	rs1042713	(A;G)	1.3x increased risk that pediatric inhaler use ...	Link	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			
1.3	rs2024513	(A;G)	1.3x higher risk for schizophrenia (among Han C...			

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
1.3	rs2059693	(C;T)	1.3x increased risk for testicular cancer			
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	rs356219	(A;G)	1.3x increased risk for Parkinson's disease			
1.3	rs501120	(A;G)	1.3x increased risk for heart disease		Link	
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	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	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	rs3740878	(A;A)	1.46x type II diabetes risk; common	Link		Link
1.2	rs393152	(A;G)	Slight increased risk of both PD and AD	Link	Link	
1.2	rs4324715	(C;C)	>1.5x increased testicular cancer risk for men			
1.2	rs449647	(A;T)	Possibly lower levels of ApoE			
1.2	rs4496877	(T;T)	For type-1 diabetics: 1.6x increased nephropath...			
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	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	rs8050136	(A;C)	1.2x increased risk for T2D in some populations...		Link	
1.17	rs3802842	(A;C)	1.17x increased risk of colorectal cancer		Link	
1.1	rs11110912	(C;C)	1.3x high blood pressure risk			
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	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	rs3818361	(C;T)	1.15x increased risk for late-onset Alzheimer's...			
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			
1.07	rs2291834	(C;C)	Very slightly higher risk for myocardial infarc...			
1	rs1143674	(A;A)	1.3x increased autism risk	Link		
1	rs2282679	(A;C)	Somewhat lower vitamin D levels			
1	rs2546890	(A;G)	Higher risk of multiple sclerosis			
1	rs3194051	(A;A)	>1.1x risk of type-1 diabetes	Link	Link	Link
1	rs6932590	(C;T)	1.1x increased risk for schizophrenia		Link	
1.0	rs11246226	(A;A)	Increased risk of schizophrenia in limited stud...		Link	
0.1	rs2070744	(C;C)	Increased prostate cancer risk		Link	Link
0.1	rs601338	(A;G)	Susceptible to Norovirus infections	Link	Link	Link
0	rs10761659	(A;A)	1.5x risk of Crohn's disease		Link	
0	rs1128503	(T;T)	Likely to require more methadone during heroin ...	Link	Link	Link
0	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	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
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	gs101	Probably able to digest milk
2	gs156	NAT2 Rapid metabolizer.
2	gs173	CYP2D6*10
2	gs246	APOE3/APOE3
1.5	gs185	The beta blocker metoprolol is effective with 1...
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 ERS1176601 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/ERS1176601

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.