PGP-UK Genomics Report for uk401869

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.

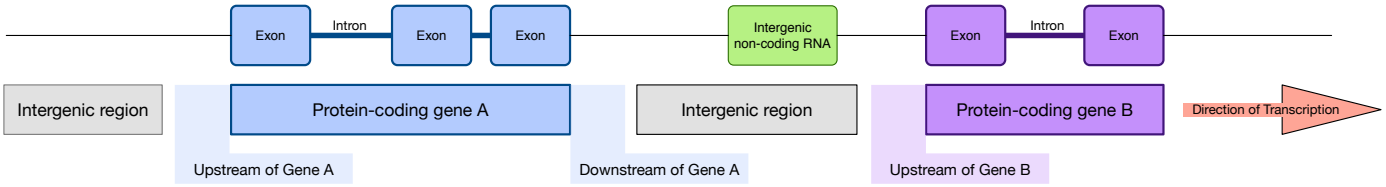

Figure 1: Diagram of gene structure indicating locations of potential variants

Feature	Count
Lines of input read	4936059
Variants filtered out	0
Novel / existing variants	468088 (9.5) / 4455333 (90.5)
Overlapped genes	56648
Overlapped transcripts	67548
Overlapped regulatory features	166519

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.

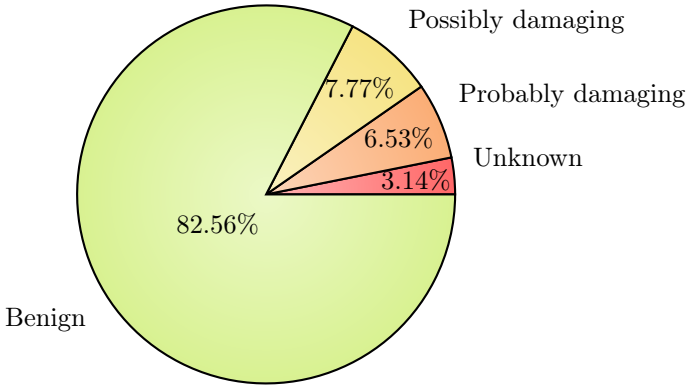

Figure 2: PolyPhen Summary

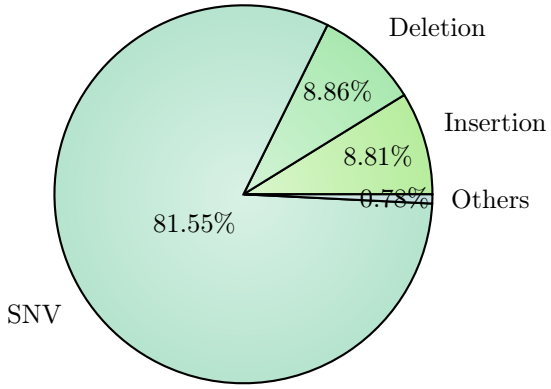

Figure 3: Variant Class

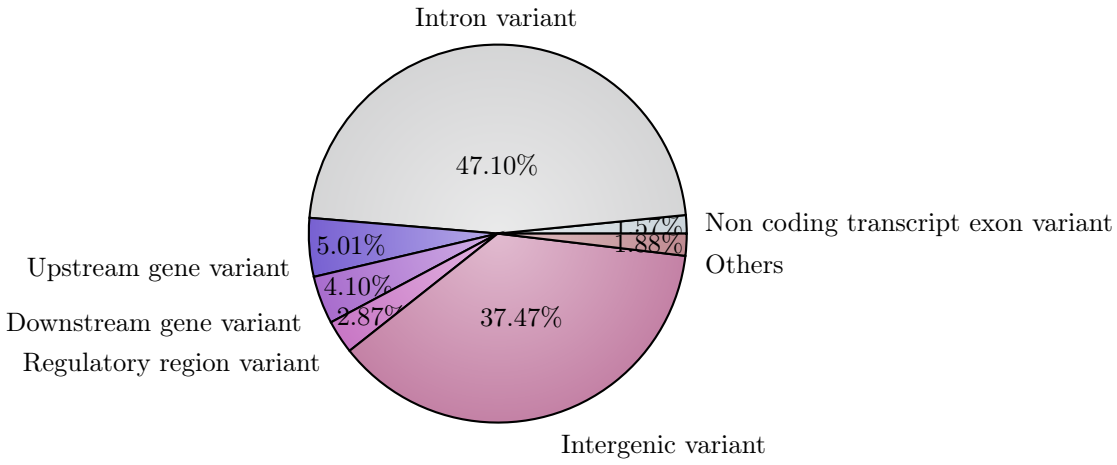

Figure 4: Consequence type

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).

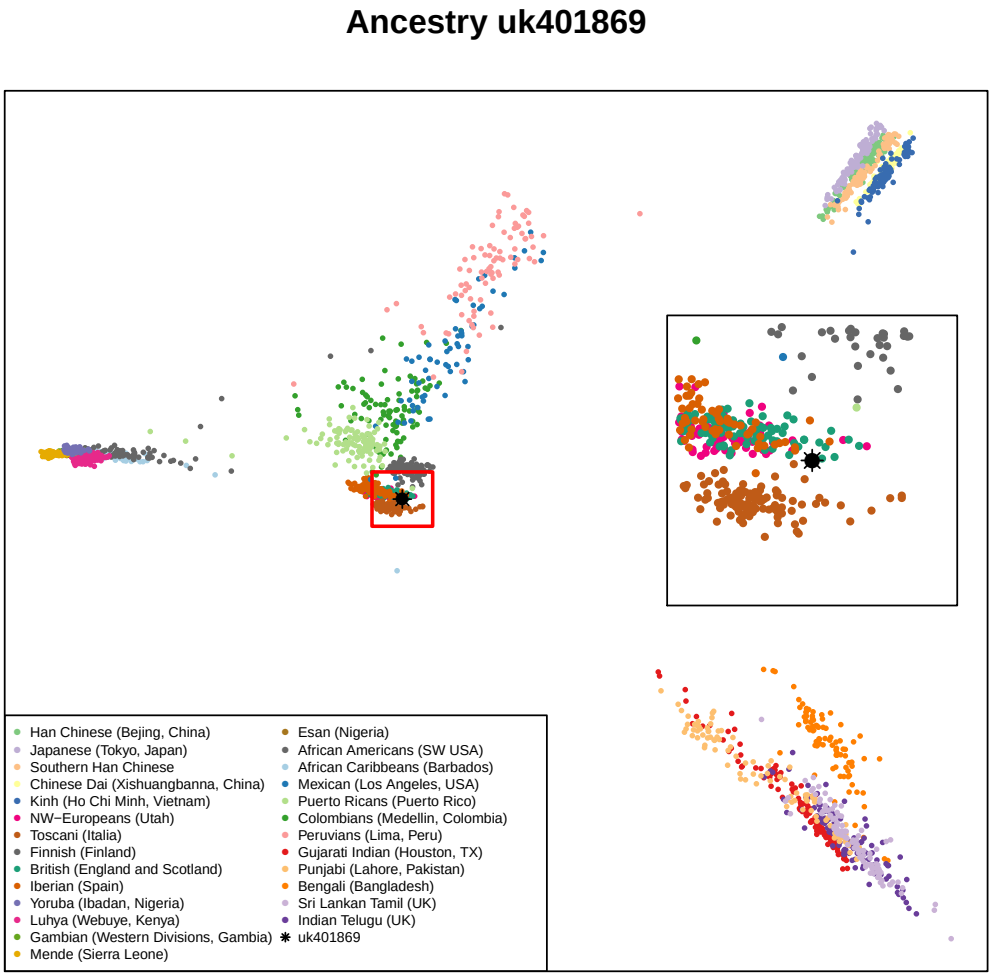

Figure 5: Ancestry Principal Component Analysis

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	rs3775291	(A;G)	0.71x decreased risk for dry age related macula...	Link	Link	Link
2.1	rs547154	(A;C)	0.47x decreased risk for AMD			Link
2	rs1026732	(A;A)	<0.70x risk for restless legs		Link	
2	rs10468017	(C;T)	Associated with higher HDL cholesterol		Link	
2	rs1128535	(G;G)	Reduced risk (0.77x) for Crohn's disease			
2	rs1136410	(C;T)	0.80x reduced risk for glioblastoma	Link	Link	
2	rs11635424	(A;A)	<0.70x risk for restless legs		Link	
2	rs1229984	(A;A)	0.56x decreased risk of oral/throat cancers	Link	Link	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	rs1799884	(G;G)	Mothers have typical Birth-Weight babies. Sligh...			
2	rs1864163	(A;G)	Associated with higher HDL cholesterol		Link	
2	rs2241423	(A;A)	0.79 decreased risk for obesity			
2	rs2243250	(C;T)	0.6x decreased risk for myocardial infarction i...			
2	rs2292813	(C;T)	Decreased risk of autism			
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	rs3819331	(T;T)	Lower risk of autism	Link		
2	rs3914132	(C;T)	Lower otosclerosis risk		Link	
2	rs4073582	(A;A)	Lower risk for gout	Link		
2	rs4149268	(A;G)	Associated with higher HDL cholesterol		Link	
2	rs6511720	(G;T)	Slightly lower odds of developing CHD.		Link	Link

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
2	rs6807362	(G;G)	Decreased autism risk	Link	Link	
2	rs6855911	(A;G)	0.62x decreased risk for gout		Link	
2	rs8070723	(A;G)	0.18x reduced risk of developing progressive su...			
1.8	rs1800588	(C;T)	Higher HDL-C levels	Link	Link	
1.8	rs187238	(C;G)	Hypertension not a risk factor for sudden cardi...			
1.8	rs266729	(C;G)	0.73x decreased risk for colorectal cancer		Link	
1.8	rs3814113	(C;T)	0.8x decreased risk for ovarian cancer		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			
1.8	rs9402571	(G;G)	0.85x decreased risk for type-2 diabetes			
1.6	rs3775948	(C;C)	Slightly lower risk for gout			
1.5	rs11136000	(T;T)	0.84x decreased risk for Alzheimer's disease		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			
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	rs6427528	(A;G)	For rheumatoid arthritis patients: better respo...			
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	rs6700125	(C;C)	0.7x decreased risk for ALS			
1.3	rs10166942	(C;C)	0.7x lower risk for migraines			
1.2	rs11246226	(A;C)	Decreased risk of schizophrenia in limited stud...		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	rs182549	(C;T)	Can digest milk.			Link
1	rs2494732	(T;T)	Lower odds of psychosis	Link	Link	
1	rs2546890	(G;G)	Lower risk of multiple sclerosis			
0.1	rs891512	(G;G)	Lower blood pressure than those with an A allel...	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	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	rs6259	(G;G)	Best inverse correlation between tea-drinking: ...	Link	Link	
0	rs74315403	(G;G)	PrP codon 178 (D) - non pathogenic variant			Link
0	rs9394492	(C;C)	<0.76x risk for restless legs			

3.2 Possibly Harmful Traits

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
3.3	rs7270101	(C;C)	Causative. Ribavirin-induced anemia iRibavirin-...	Link		Link
3	rs10897346	(C;C)	If depressed: 2.6x more likely to not respond t...			
3	rs2981582	(C;T)	1.3x higher risk of ER+ breast cancer		Link	
3	rs6920220	(A;G)	1.2x risk Rheumatoid Arthritis		Link	
2.6	rs110419	(A;A)	2.6x increased risk for neuroblastoma			
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	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	rs1799971	(A;G)	Stronger cravings for alcohol. if alcoholic: na...	Link	Link	Link
2.5	rs2004640	(T;T)	1.4x increased risk for SLE		Link	Link
2.5	rs339331	(T;T)	Prostate cancer risk			
2.5	rs3738919	(C;C)	1.94x risk of developing rheumatoid arthritis			
2.5	rs3780374	(A;G)	Substantially increased odds of developing V617...			
2.5	rs660895	(G;G)	6x higher risk of rheumatoid arthritis		Link	
2.5	rs7574865	(G;T)	1.3x risk of rheumatoid arthritis; 1.55x risk o...		Link	Link
2.5	rs8034191	(C;T)	1.27x lung cancer risk		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	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	rs1329428	(G;G)	2x increased risk for macular degeneration			
2.1	rs17563	(C;C)	Risk for otosclerosis	Link	Link	Link
2.1	rs1800730	(A;T)	Carrier for a mild form of hemochromatosis	Link		Link
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	rs4149056	(C;T)	Reduced breakdown of some drugs; 5x increased m...	Link	Link	Link
2.1	rs4363657	(C;T)	4.5x increased myopathy risk for statin users		Link	
2.1	rs4430796	(A;A)	1.38x increased risk for prostate cancer		Link	
2.1	rs5186	(A;C)	~1.4x increased risk of hypertension	Link	Link	Link
2.1	rs6457617	(T;T)	5.2x risk of rheumatoid arthritis		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	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	rs1050152	(C;T)	2.1x increased risk of Crohn's disease	Link	Link	Link
2	rs1051730	(C;T)	1.3x increased risk of lung cancer	Link	Link	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;A)	>1.17x increased risk for multiple sclerosis		Link	
2	rs11045585	(A;G)	63% chance (higher than average) of docetaxel-i...		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	rs12696304	(C;G)	Prone to aging faster: at least in European pop...			
2	rs13254738	(A;C)	1.18x prostate cancer risk		Link	
2	rs1333048	(A;C)	1.3x increased coronary artery disease risk			
2	rs1585215	(A;G)	2x increased risk for Hodgkin lymphoma			
2	rs16942	(A;G)	Very slightly increased breast cancer risk	Link	Link	Link
2	rs17228212	(C;T)	1.26x increased risk for heart disease		Link	

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
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	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	rs2073963	(G;T)	Increased risk of baldness			
2	rs2201841	(T;T)	2.4x increased risk for Graves' disease		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	(A;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	rs2736990	(C;C)	Increased risk of developing Parkinson's Diseas...		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	rs3790565	(C;C)	Increased risk of developing primary biliary ci...			
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	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	rs493258	(A;G)	1.15x risk of Age Related Macular Degeneration			
2	rs4968451	(A;C)	1.61x increased risk for meningioma			
2	rs5174	(A;G)	1.3x increased risk for heart disease	Link	Link	Link
2	rs520354	(A;G)	Increased risk in men for biliary conditions			
2	rs587776825	(-;C)	Associated with MODY3; maturity onset of diabet...	Link		Link
2	rs629242	(C;T)	Somewhat higher risk for prostate cancer			
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	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	rs763361	(T;T)	Increased risk for multiple autoimmune diseases...	Link	Link	
2	rs7639618	(C;T)	1.45x increased osteoarthritis risk	Link		
2	rs7776725	(C;C)	Weaker bones		Link	
2	rs7807268	(C;G)	1.3x risk for Crohn's disease		Link	
2	rs7961152	(A;A)	1.5x higher risk for hypertension			
2	rs800292	(C;C)	5% higher risk of Age related macular degenerat...	Link	Link	Link
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	rs9303277	(T;T)	1.46x Increased risk of developing primary bili...			
2	rs9525638	(T;T)	Weaker bones			
2	rs9652490	(A;A)	~2x increased risk for Parkinson's disease: and...		Link	
2	rs9954153	(G;T)	~2.5x higher risk for Fuchs' dystrophy: a corne...			
2.0	rs1044396	(C;C)	Increased risk of Nicotine dependence among mal...	Link	Link	Link
2.0	rs2156921	(G;G)	1.29x increased risk for depression			
2.0	rs4911414	(G;T)	2-4x higher risk of sun sensitivity if part of ...		Link	
1.9	rs7923837	(A;G)	1.6x risk for T2D			
1.8	rs2278206	(T;T)	1.16x increased risk for asthma	Link	Link	
1.8	rs37973	(A;G)	Among asthmatics: 1.5x more likely to show less...			Link
1.6	rs11523871	(A;C)	1.6x increased breast cancer risk for women ove...	Link	Link	

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
1.6	rs1537415	(C;G)	1.6x increased risk for periodontitis		Link	
1.6	rs2981745	(C;T)	1.6x increased risk for breast cancer in female...			
1.6	rs356219	(G;G)	1.6x increased risk for Parkinson's disease			
1.5	rs10757272	(C;T)	1.30x increased risk for Coronary artery diseas...			
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	rs13149290	(C;C)	Slightly increased risk of developing prostate ...			
1.5	rs13181	(G;T)	1.12x increased risk for cutaneous melanoma	Link	Link	Link
1.5	rs140701	(A;A)	Increased risk for anxiety disorders			
1.5	rs1571801	(A;A)	>1.36x risk for prostate cancer			
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	rs1801274	(C;T)	Complex; generally greater risk for cancer prog...	Link	Link	Link
1.5	rs199533	(C;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	rs27388	(A;G)	Slightly increased risk of developing schizophr...			
1.5	rs2881766	(G;T)	Slightly increased risk for pregnancy-induced h...			
1.5	rs3087243	(G;G)	Increased risk for autoimmune diseases		Link	
1.5	rs3212227	(A;A)	1.43x increased risk of developing psoriasis an...			
1.5	rs356220	(T;T)	Increased risk of Parkinson's Disease			
1.5	rs3745516	(A;G)	Slightly increased risk of developing primary b...			
1.5	rs3764880	(A;G)	Possible 1.2 - 1.8x increased tuberculosis susc...	Link	Link	
1.5	rs3825776	(A;G)	1.3x increased risk for ALS		Link	
1.5	rs401681	(C;T)	~1.2x increased risk for several types of cance...		Link	
1.5	rs4464148	(C;T)	1.10x increased risk for colorectal cancer			
1.5	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	rs4585	(T;T)	Slightly poorer (0.75x) response to metformin i...			
1.5	rs464049	(C;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	rs6498169	(A;G)	1.14x risk of multiple sclerosis		Link	
1.5	rs6532197	(A;G)	Slightly increased risk of developing Parkinson...		Link	
1.5	rs6601764	(C;T)	1.16x increased risk of developing Crohn's dise...		Link	
1.5	rs6908425	(C;T)	1.63x increased risk of developing Crohn's dise...		Link	
1.5	rs699473	(C;T)	~1.5x increased brain tumor risk			
1.5	rs7454108	(C;T)	Single HLA-DQ8 haplotype			
1.5	rs7774434	(C;T)	Slightly increased risk of developing primary b...			
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	rs9642880	(G;T)	1.2x increased bladder cancer risk		Link	
1.5	rs995030	(G;G)	Non-protective against testicular cancer		Link	
1.4	rs1126497	(T;T)	1.4x increased risk for breast cancer	Link	Link	Link
1.4	rs12770228	(A;G)	1.4x increased risk for meningioma			
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	rs6010620	(G;G)	1.4x higher risk for glioma development; but th...		Link	

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
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	rs1375144	(C;T)	1.32x increased risk of developing bipolar diso...			
1.3	rs1746048	(C;C)	1.03 increased risk for coronary heart disease		Link	
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	rs12050604	(A;A)	Slightly increased risk for lung cancer			
1.2	rs143383	(C;T)	1.1x increased risk for osteoarthritis		Link	Link
1.2	rs1800693	(A;G)	Slight (1.2x) increase in risk for multiple scl...	Link	Link	Link
1.2	rs2252586	(A;G)	1.2x higher risk for glioma development			
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	rs498872	(C;T)	1.2x higher risk for glioma development		Link	
1.17	rs17465637	(A;C)	1.17x higher risk for myocardial infarction	Link	Link	
1.17	rs3802842	(A;C)	1.17x increased risk of colorectal cancer		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	rs1344706	(G;T)	1.1x increased risk for schizophrenia		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	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	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	rs1143674	(A;A)	1.3x increased autism risk	Link		
1	rs2282679	(A;C)	Somewhat lower vitamin D levels			
1	rs3194051	(A;G)	1.12x risk of type-1 diabetes	Link	Link	Link
1	rs6932590	(C;T)	1.1x increased risk for schizophrenia		Link	
1	rs6974491	(A;G)	Higher risk of coeliac and/or inflammatory bowe...			
0.1	rs601338	(G;G)	Susceptible to Norovirus infections	Link	Link	Link
0	rs1004819	(C;C)	1.5x risk of Crohn's disease		Link	
0	rs10239794	(T;T)	>1.3x risk for ALS			
0	rs1061646	(C;C)	1.16x increased risk for breast cancer	Link		Link
0	rs1495965	(A;A)	1.2x higher risk for spondylitis			
0	rs3761418	(A;A)	1.3x increased risk for depression			
0	rs3813929	(C;C)	Possible weight gain if taking olanzapine		Link	Link
0	rs4293393	(T;T)	1.25x Increased Risk of CKD for T allele in ...			
0	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
2.5	gs155	CYP3A5 non-expressor
2.5	gs281	Part of the 88% of the population claimed not t...
2.5	gs298	Increased surveillance for colorectal cancer re...
2	gs101	Probably able to digest milk
2	gs269	APOE E2/E3
1.5	gs247	Parkinson's Disease Risk
1.2	gs184	Able to taste bitterness.
1	gs182	CYP2D6*39
0	gs158	CYP1A2 normal metabolizer

4 Raw Data

The raw data used to create this report has been assigned the identifier ERS1176600 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/ERS1176600

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.