PGP-UK Genomics Report for uk0A0D88

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.

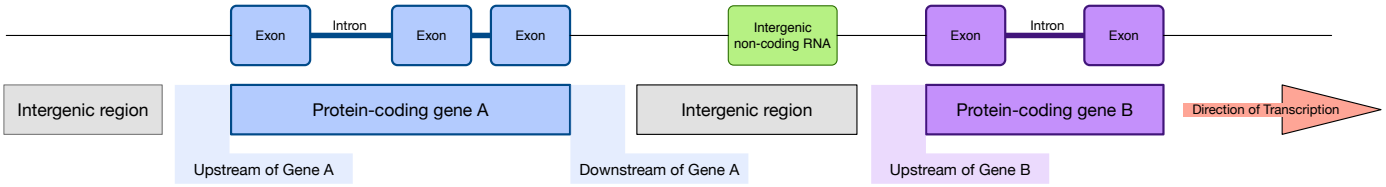

Figure 1: Diagram of gene structure indicating locations of potential variants

Feature	Count
Lines of input read	4972624
Variants filtered out	0
Novel / existing variants	485026 (9.8) / 4476002 (90.2)
Overlapped genes	56673
Overlapped transcripts	67431
Overlapped regulatory features	166985

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.

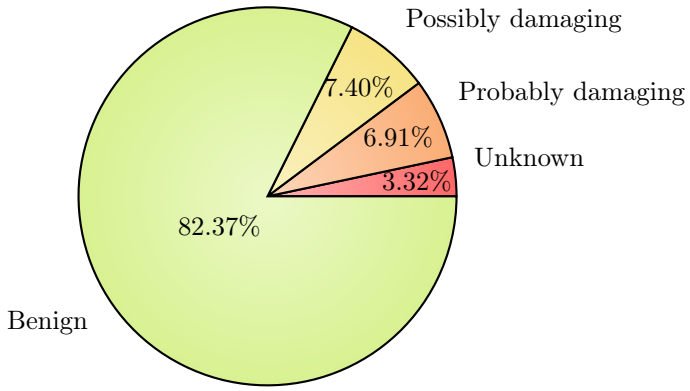

Figure 2: PolyPhen Summary

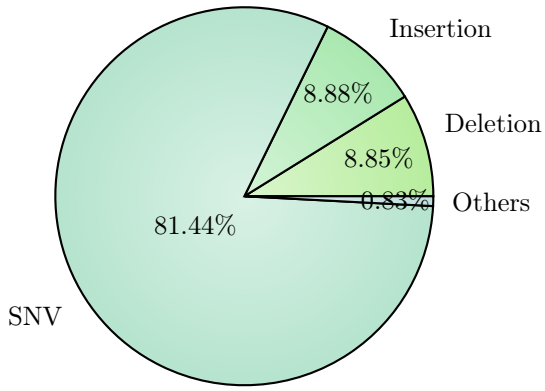

Figure 3: Variant Class

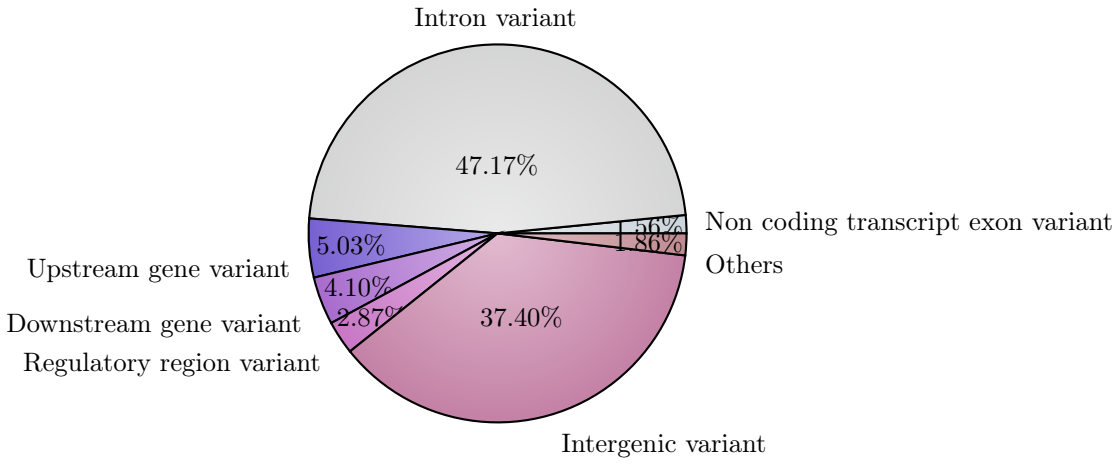

Figure 4: Consequence type

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).

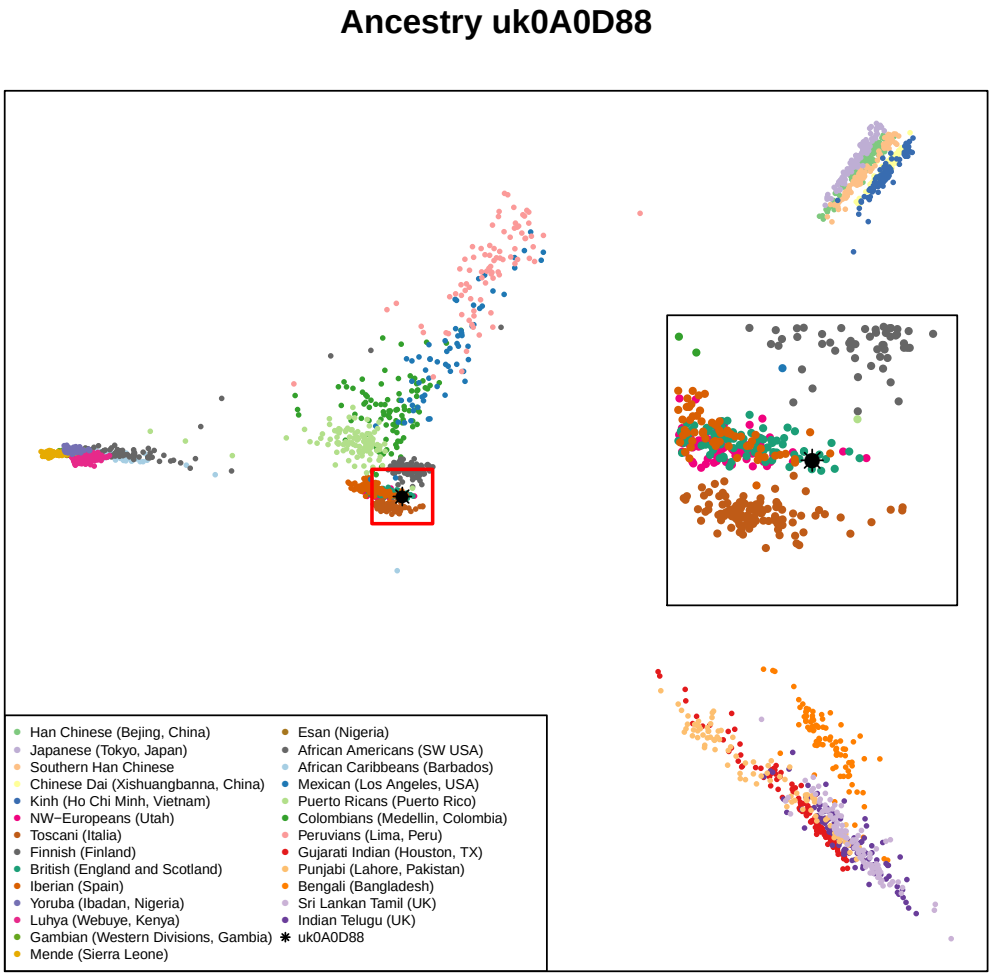

Figure 5: Ancestry Principal Component Analysis

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
2.4	rs2802288	(A;A)	Longer lifespan			
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	rs1160312	(G;G)	Reduced risk of Baldness.		Link	
2	rs12979860	(C;C)	~80% of such hepatitis C patients respond to tr...		Link	Link
2	rs1799884	(G;G)	Mothers have typical Birth-Weight babies. Sligh...			
2	rs1864163	(A;G)	Associated with higher HDL cholesterol		Link	
2	rs2542052	(C;C)	Better odds of living to 100			
2	rs261332	(A;A)	Associated with higher HDL cholesterol			
2	rs2707466	(A;A)	Stronger bones	Link	Link	
2	rs2908004	(T;T)	Stronger bones	Link	Link	
2	rs3218536	(A;G)	Lower risk for breast: ovarian cancer	Link	Link	
2	rs3738579	(C;T)	0.5x decreased risk for cervical cancer: HNSCC:...			
2	rs3764261	(G;T)	Associated with higher HDL cholesterol		Link	Link
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	rs4307059	(C;C)	Reduced Autism risk		Link	
2	rs4585	(G;G)	Slightly higher (1.35x) odds of good metformin ...			
2	rs6855911	(A;G)	0.62x decreased risk for gout		Link	
2	rs9272346	(A;G)	0.3x risk type-1 diabetes		Link	
2	rs9525638	(C;C)	Stronger bones			
1.9	rs1015362	(A;A)	Probably tans instead of freckles and sunburns....		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	rs266729	(C;G)	0.73x decreased risk for colorectal cancer		Link	

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
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	rs6897932	(C;T)	0.91x decreased risk for multiple sclerosis	Link	Link	Link
1.8	rs7101429	(A;G)	0.70x reduced risk for Alzheimer's risk			
1.8	rs854560	(T;T)	0.5x lower risk of ovarian cancer	Link	Link	Link
1.5	rs1063192	(C;T)	0.71x reduced risk of myocardial infarction			
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;C)	Associated with higher HDL (good) cholesterol.			
1.5	rs4939883	(C;T)	Associated with higher HDL cholesterol		Link	
1.5	rs610932	(A;A)	A allele associated with reduced risk of Alzhei...			
1.4	rs1165205	(A;T)	0.85x decreased gout risk		Link	
1.4	rs2294008	(C;C)	Lower risk of gastric and bladder cancer	Link	Link	
1.4	rs9402571	(G;T)	Slightly decreased risk for type-2 diabetes			
1.3	rs9306160	(T;T)	0.75x (reduced) risk for metastasis in LN-/ER+ ...	Link	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	rs7850258	(A;G)	Typical odds of developing primary hypothyroidi...			
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	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	rs1799883	(A;A)	Two copies of the Thr allele in the FABP2 is as...	Link	Link	Link
0	rs1799945	(C;C)	Not a H63D hemochromatosis carrier.	Link	Link	Link
0	rs242941	(G;G)	Better response to inhaled corticosteroid in pa...		Link	
0	rs28933385	(G;G)	Prion protein Codon 200 (E) - Non pathogenic va...			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	rs9394492	(C;C)	<0.76x risk for restless legs			

3.2 Possibly Harmful Traits

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
3.8	rs5186	(C;C)	7.3x increased risk of hypertension	Link	Link	Link
3.1	rs10830963	(G;G)	Increased type-2 diabetes risk; higher gestatio...		Link	
3.1	rs1421085	(C;C)	~1.7x increased obesity risk		Link	Link
3	rs10897346	(C;C)	If depressed: 2.6x more likely to not respond t...			
3	rs1121980	(T;T)	2.76x risk for obesity		Link	
3	rs13266634	(C;C)	Increased risk for type-2 diabetes	Link	Link	Link
3	rs2306402	(C;C)	1.18x increased risk for late-onset Alzheimer's...			
3	rs2981582	(C;T)	1.3x higher risk of ER+ breast cancer		Link	
3	rs3848519	(A;C)	Carrier for an erythropoietic protoporphyria mu...	Link	Link	Link
3	rs3903239	(C;C)	Higher frequency of atrial fibrillation			
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	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	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	rs5888	(C;T)	3x higher risk for age-related macular degenera...	Link		
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		
2.4	rs7966230	(G;G)	Slightly lower levels of plasma VWF			
2.3	rs1859962	(G;G)	1.28x increased risk for prostate cancer		Link	
2.3	rs37973	(G;G)	Among asthmatics: 2.3x more likely to show less...			Link
2.2	rs2231137	(G;G)	~1.5-3x increased risk for ischemic stroke	Link	Link	Link
2.2	rs944289	(T;T)	1.69x increased thyroid cancer risk		Link	
2.1	rs10411210	(T;T)	1.15x increased risk of colorectal cancer		Link	
2.1	rs11887534	(C;G)	2x increased risk for gallstones	Link	Link	Link
2.1	rs17070145	(C;C)	Reduced memory abilities			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	rs4402960	(T;T)	1.2x increased risk for type-2 diabetes: 1.5x r...		Link	Link
2.1	rs4430796	(A;A)	1.38x increased risk for prostate cancer		Link	
2.1	rs4444903	(G;G)	3.5x risk of hep-cancer in cirrhosis patients; ...			Link
2.1	rs4961	(T;T)	1.8x increased risk for high blood pressure	Link	Link	Link
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	(G;T)	1.7x increased risk for prostate cancer			
2	rs10086908	(C;T)	1.7x increased risk for prostate cancer			
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	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	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	rs11983225	(T;T)	7x less likely to respond to certain antidepres...		Link	
2	rs1219648	(A;G)	1.20x risk for breast cancer		Link	
2	rs12567232	(A;G)	Increased risk for Crohn's Disease		Link	
2	rs1265181	(C;G)	Increased risk for psoriasis		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	rs16942	(G;G)	Very slightly increased breast cancer risk	Link	Link	Link
2	rs16944	(A;A)	Increased risk for osteoarthritis		Link	
2	rs17001266	(-;C)	1.58x increased risk for schizophrenia in males...			

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
2	rs17228212	(C;C)	>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	rs1800629	(A;G)	Complex; generally higher risk for certain dise...	Link	Link	Link
2	rs1800888	(C;T)	Increased risk of coronary artery disease	Link	Link	Link
2	rs1800896	(A;G)	1.6x increased prostate cancer risk			
2	rs2073963	(G;T)	Increased risk of baldness			
2	rs2201841	(C;T)	1.5x increased risk for Crohn's disease; 2x inc...		Link	
2	rs2230201	(G;G)	>1.4x risk of lupus	Link		
2	rs2235015	(G;G)	Somewhat less likely to respond to certain anti...	Link	Link	
2	rs2235040	(G;G)	7x less likely to respond to certain antidepres...	Link	Link	
2	rs2235067	(G;G)	7x less likely to respond to certain antidepres...			
2	rs2305480	(C;T)	3.5x increase in risk of asthma for Han Chinese...	Link	Link	
2	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	rs27388	(A;A)	Increased risk of developing schizophrenia			
2	rs3129934	(C;T)	Increased risk of Multiple Sclerosis.		Link	
2	rs3212227	(A;C)	Significantly increased risk of developing cerv...			
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	rs4420638	(A;G)	~3x increased Alzheimer's risk; 1.4x increased ...		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	rs493258	(A;G)	1.15x risk of Age Related Macular Degeneration			
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	rs61753344	(G;T)	Trimethylaminuria: to varying degrees	Link		Link
2	rs6498169	(A;A)	>1.14x risk of multiple sclerosis		Link	
2	rs6807362	(C;C)	Increased autism risk	Link	Link	
2	rs6997709	(G;T)	1.2x higher risk for hypertension			
2	rs7250872	(T;T)	Increased risk of developing bipolar disorder	Link	Link	
2	rs7776725	(C;C)	Weaker bones		Link	
2	rs7794745	(A;T)	Slightly increased risk for autism		Link	Link
2	rs7961152	(A;C)	1.2x higher risk for hypertension			
2	rs9652490	(A;A)	~2x increased risk for Parkinson's disease: and...		Link	
2	rs965513	(A;G)	1.77x increased thyroid cancer risk		Link	
2.0	rs1044396	(C;C)	Increased risk of Nicotine dependence among mal...	Link	Link	Link
2.0	rs2305795	(A;A)	1.64x higher risk of narcolepsy compared to (G;...			Link
2.0	rs4911414	(T;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	rs4474514	(A;G)	3x increased testicular cancer risk for men		Link	
1.8	rs6700125	(C;T)	1.2x increased risk for ALS			
1.8	rs733618	(A;G)	1.87x risk for myasthenia gravis			
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	
1.5	rs10492519	(A;G)	Slightly increased risk of developing prostate ...			

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
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	rs10895068	(A;G)	2.5x increased odds of breast cancer among horm...			
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	rs12210050	(T;T)	Slighly higher risk for basal cell carcinoma		Link	
1.5	rs1223271	(A;G)	Slightly increased risk of developing Parkinson...		Link	
1.5	rs12469063	(A;G)	Slightly increased risk of developing restless ...			
1.5	rs13149290	(C;T)	Slightly increased risk of developing prostate ...			
1.5	rs13181	(G;T)	1.12x increased risk for cutaneous melanoma	Link	Link	Link
1.5	rs13376333	(C;T)	1.5x higher risk of atrial fibrillation		Link	
1.5	rs144848	(G;T)	Very slightly increased breast cancer risk	Link	Link	Link
1.5	rs1801274	(T;T)	Complex; generally greater risk for cancer prog...	Link	Link	Link
1.5	rs2240340	(A;G)	Slightly increased (1.5x) risk for RA	Link		
1.5	rs2272127	(C;C)	Associated with herpes and schizophrenia			
1.5	rs2280714	(A;A)	1.4x increased risk of SLE			
1.5	rs2464196	(C;T)	~1.5x increased lung cancer risk	Link	Link	Link
1.5	rs2697962	(A;G)	Slightly increased risk of developing Parkinson...			
1.5	rs2736990	(C;T)	Slightly increased risk of developing Parkinson...		Link	
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	rs309375	(T;T)	Larger mosquito bites			
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	rs419788	(A;A)	2.3x risk for lupus	Link		
1.5	rs4506565	(A;T)	1.4x increased risk for type-2 diabetes		Link	
1.5	rs464049	(C;T)	Increased risk of schizophrenia in limited stud...			
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	rs642961	(A;G)	1.68x increased risk of cleft lip		Link	
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	rs6896702	(C;T)	Slightly increased risk of developing Parkinson...			
1.5	rs6908425	(C;T)	1.63x increased risk of developing Crohn's dise...		Link	
1.5	rs6974491	(A;A)	Higher risk of coeliac and/or inflammatory bowe...			
1.5	rs7341475	(G;G)	1.58x increased schizophrenia risk for women		Link	
1.5	rs763035	(C;T)	1.2x increased risk for rosacea			
1.5	rs807701	(C;T)	Slightly increased dyslexia risk			
1.5	rs872071	(G;G)	~1.5x increased risk for chronic lymphocytic le...		Link	
1.5	rs9303277	(C;T)	1.46x Slightly increased risk of developing pri...			
1.5	rs9642880	(G;T)	1.2x increased bladder cancer risk		Link	
1.5	rs975278	(A;A)	1.5x higher risk for emphysema: higher in smoke...			
1.4	rs10865331	(A;A)	1.4x higher risk for ankylosing spondylitis			
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	rs6010620	(G;G)	1.4x higher risk for glioma development; but th...		Link	
1.4	rs8050136	(A;A)	1.4x increased risk for T2D in some populations...		Link	
1.34	rs17465637	(C;C)	1.34x higher risk for myocardial infarction	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	rs1260326	(C;T)	Slightly higher risk for gout	Link	Link	Link
1.3	rs1434536	(A;G)	1.29x increased breast cancer risk			

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
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	rs356219	(A;G)	1.3x increased risk for Parkinson's disease			
1.2	rs11037909	(T;T)	1.47x type II diabetes risk	Link		
1.2	rs1344706	(T;T)	1.2x increased risk for schizophrenia		Link	
1.2	rs1800693	(A;G)	Slight (1.2x) increase in risk for multiple scl...	Link	Link	Link
1.2	rs2056116	(A;G)	1.18x risk for breast cancer			
1.2	rs2072590	(G;T)	1.2x increased risk for ovarian cancer			
1.2	rs2076295	(G;T)	One copy of the risk allele (G): slightly incre...			
1.2	rs2252586	(A;G)	1.2x higher risk for glioma development			
1.2	rs3131296	(A;G)	1.2x increased risk for schizophrenia		Link	
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	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	rs6897876	(C;C)	Slight increase in testicular cancer risk for m...			
1.2	rs9858542	(A;G)	1.1x risk Crohn's Disease	Link	Link	
1.17	rs3802842	(A;C)	1.17x increased risk of colorectal cancer		Link	
1.15	rs748404	(C;T)	Very slightly increased risk (1.15) for lung ca...		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	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	rs2295190	(G;T)	Slightly increased risk for ovarian cancer in w...	Link	Link	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	rs3818361	(C;T)	1.15x increased risk for late-onset Alzheimer's...			
1.1	rs6800901	(T;T)	1.3x multiple myeloma risk			
1.1	rs688034	(C;T)	1.1x risk higher risk for coronary artery disea...		Link	
1.1	rs7412	(C;C)	More likely to gain weight if taking olanzapine...	Link	Link	Link
1.1	rs889312	(A;C)	Very slightly higher risk for breast cancer		Link	
1.1	rs925391	(C;C)	More likely to go bald; common			
1.09	rs12050604	(A;C)	Very slightly increased risk for lung cancer			
1.07	rs2291834	(C;C)	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	rs2273697	(A;G)	Adverse reaction more likely to carbamazepine i...	Link	Link	Link
1	rs2546890	(A;G)	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	
0	rs10239794	(T;T)	>1.3x risk for ALS			
0	rs1042173	(T;T)	Among alcoholics: likely to be heavier drinkers...			
0	rs10761659	(A;A)	1.5x risk of Crohn's disease		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	rs440446	(G;G)	Increased risk in men for biliary conditions	Link		
0	rs7787082	(G;G)	7x less likely to respond to certain antidepres...		Link	

3.3 Genosets (Multi-variant Phenotypes)

Magnitude	Identifier	Summary
3	gs241	Lighter green: brown or hazel eye color
2.5	gs281	Part of the 88% of the population claimed not t...
2.5	gs285	You will lose 2.5x as much weight on a low fat ...
2	gs101	Probably able to digest milk
2	gs188	One copy of APOE4 is possible: but not certain
1.5	gs186	HLA-B*5801 heterozygosity is possible: unfortun...
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 ERS1176622 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/ERS1176622

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.