PGP-UK Genomics Report for uk2200DC

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 <u>personal and research purposes</u> 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.

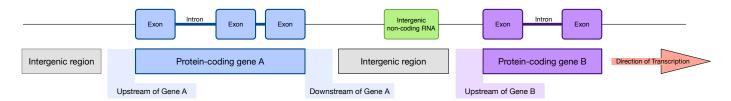


Figure 1: Diagram of gene structure indicating locations of potential variants

Feature	Count
Lines of input read	4915511
Variants filtered out	0
Novel / existing variants	$488970\ (10.0)\ /\ 4414452\ (90.0)$
Overlapped genes	56651
Overlapped transcripts	67411
Overlapped regulatory features	166193

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.

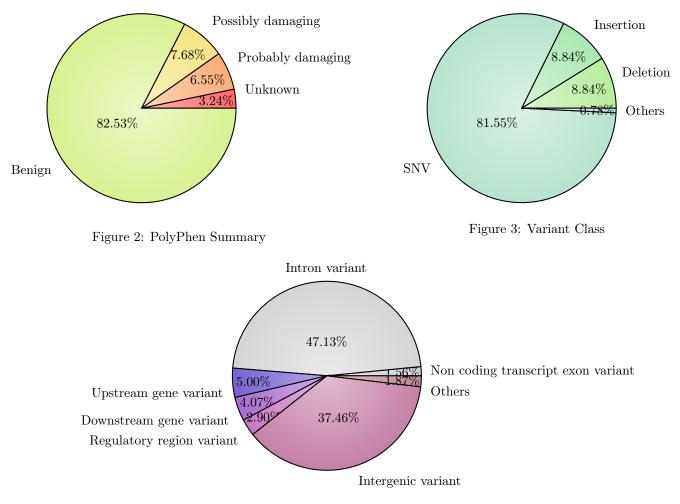


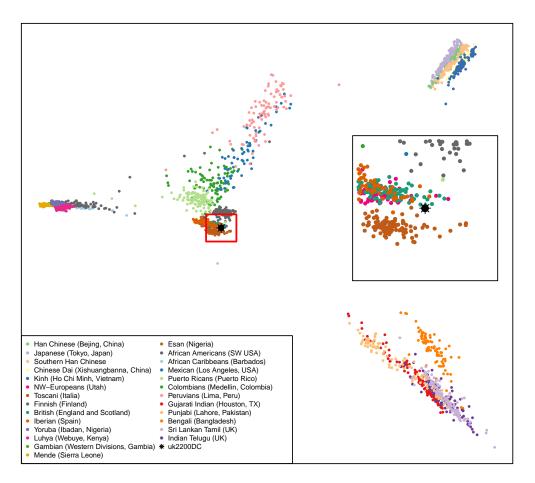
Figure 4: Consequence type

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 uk2200DC

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.

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
2.4	rs9272346	(G;G)	0.08x risk type-1 diabetes	LAITO	Link	Chirvar
2.1	rs6505162	(A;A)	0.43x decreased risk for esophageal cancer	Link		
2	rs10468017	(C;T)	Associated with higher HDL cholesterol		Link	
2	rs11045585	(A;A)	24% chance (lower than average) of docetaxel-in		Link	
2	rs11132186	(T;T)	0.5x decreased risk for bladder cancer			
2	rs12979860	(C;C)	$\sim 80\%$ of such hepatitis C patients respond to tr		Link	Link
2	rs13207033	(A;A)	Reduced risk of rheumatoid arthritis			
2	rs1799884	(G;G)	Mothers have typical Birth-Weight babies. Sligh			
2	rs1864163	(A;G)	Associated with higher HDL cholesterol		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	rs3764261	(G;T)	Associated with higher HDL cholesterol		Link	Link
2	rs3819331	(T;T)	Lower risk of autism	Link		
2	rs4307059	(C;C)	Reduced Autism risk		Link	
2	rs6855911	(G;G)	Rare: but 0.62x decreased risk for gout		Link	
2	rs7105934	(A;G)	0.69 times lower odds of developing renal cell			
2	rs763110	(C;T)	~ 0.80 x reduced cancer risk			Link
2	rs7776725	(T;T)	Stronger bones		Link	
1.8	rs1128535	(A;G)	0.77x risk for Crohn's disease			
1.8	rs187238	(C;G)	Hypertension not a risk factor for sudden cardi			
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
1.6	rs1061170	(T;T)	Lower risk for AMD: generally longer live than	Link	Link	Link
1.5	rs11136000	(C;T)	0.84x decreased risk for Alzheimer's disease		Link	
1.5	rs3851179	(A;G)	0.85x decreased risk for Alzheimer's disease		Link	
1.5	rs4939883	(C;C)	Associated with higher HDL cholesterol		Link	

3.1 Possibly Beneficial Traits

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
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	rs6700125	(C;C)	0.7x decreased risk for ALS			
1.3	rs9306160	(T;T)	0.75x (reduced) risk for metastasis in LN-/ER+	Link	Link	
1.2	rs11172113	(C;C)	0.8x lower risk for migraines			
1.2	rs11246226	(A;C)	Decreased risk of schizophrenia in limited stud		Link	
1.1	rs13333226	(A;G)	Slightly lower risk for hypertension			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
0.1	rs1726866	(C;C)	Can taste bitter	Link	Link	Link
0.1	rs891512	(G;G)	Lower blood pressure than those with an A allel	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	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	rs5065	(A;A)	1.12x risk on diuretic; if hypertensive: better	Link	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	rs10897346	(C;C)	If depressed: 2.6x more likely to not respond t			
3	rs2306402	(C;T)	1.18x increased risk for late-onset Alzheimer's			
3	rs2981582	(C;T)	1.3x higher risk of ER+ breast cancer		Link	
3	rs3903239	(C;C)	Higher frequency of atrial fibrillation			
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	rs12803066	(A;G)	Increased risk of myopia			
2.5	rs13266634	(C;T)	Increased risk for type-2 diabetes	Link	Link	Link
2.5	rs17595731	(C;G)	~ 5 fold higher risk for Fuchs' dystrophy: a cor			
2.5	rs339331	(T;T)	Prostate cancer risk			
2.5	rs3780374	(A;G)	Substantially increased odds of developing V617			
2.5	rs5888	(C;T)	3x higher risk for age-related macular degenera	Link		
2.5	rs613872	(G;T)	~ 5 fold higher risk for Fuchs' dystrophy: a cor			
2.4	rs7966230	(G;G)	Slightly lower levels of plasma VWF			
2.2	rs2231137	(G;G)	\sim 1.5-3x increased risk for ischemic stroke	Link	Link	Link
2.2	rs2305089	(T;T)	Higher risk for chordoma	Link	Link	
2.1	rs10427255	(C;C)	Highest odds of photic sneeze reflex			
2.1	rs1050152	(T;T)	2.1x increased risk of Crohn's disease	Link	Link	Link
2.1	rs1329428	(G;G)	2x increased risk for macular degeneration			
2.1	rs17070145	(C;C)	Reduced memory abilities			Link
2.1	rs17077540	(A;G)	1.6x major depressive disorder risk			
2.1	rs2231142	(A;C)	$1.74 \mathrm{x}$ increased gout risk; gefinitib takers $4 \mathrm{x} \ldots$	Link	Link	Link
2.1	rs2254958	(C;T)	1.24x increased risk for Alzheimer's			
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	TA 1	Link	
2.1	rs5751876	(T;T)	Significantly higher anxiety levels after moder	Link		
2.1	rs629242	(T;T)	Somewhat higher risk for prostate cancer			
2.1	rs795484	(A;G)	Increased morphine dose requirement and postope		T · 1	
2.1	rs944289	(C;T)	1.3x increased thyroid cancer risk		Link	
2	rs10086908	(C;C)	1.7x increased risk for prostate cancer			T · 1
2	rs1024611	(C;T)	Increased risk of exercise induced ischemia		T : 1-	Link
$\frac{2}{2}$	rs10248420	(A;A)	7x less likely to respond to certain antidepres		Link	Link
2	rs10306114	(A;G) (G;T)	Higher risk of bleeding during coronary angiogr Increased risk of Parkinson's disease			LIIIK
2	rs10513789 rs10811661	(G;T) (C;T)	1.2x increased risk for type-2 diabetes		Link	
2	rs10811001 rs10883365	$(C; \Gamma)$ (G;G)	1.62x increased risk for developing Crohn's dis		Link	
2	rs10984447	(G;G) (A;G)	1.17x increased risk for multiple sclerosis		Link	
2	rs11171739	$(\mathbf{A};\mathbf{G})$ $(\mathbf{C};\mathbf{C})$	1.75x risk of developing Type-1 diabetes		Link	
2	rs11190870	(C,C) (C;T)	Possibly increased risk of scoliosis			
2	rs1160312	(\mathbf{C},\mathbf{I}) (A;G)	1.6x increased risk of Male Pattern Baldness.		Link	
2	rs11983225	(T;T)	7x less likely to respond to certain antidepres		Link	
2	rs1219648	$(\mathbf{A};\mathbf{G})$	1.20x risk for breast cancer		Link	
2	rs1265181	(C;G)	Increased risk for psoriasis		Link	
2	rs12696304	(C;G)	Prone to aging faster: at least in European pop			
2	rs12770228	(A;A)	2x increased risk for meningioma			
2	rs1333048	(A;C)	1.3x increased coronary artery disease risk			
2	rs16942	(G;G)	Very slightly increased breast cancer risk	Link	Link	Link
2	rs17228212	(C;T)	1.26x increased risk for heart disease		Link	
2	rs1734791	(A;A)	1.4x increased risk for lupus			
2	rs17696736	(A;G)	1.34x risk of type-1 diabetes		Link	
2	rs1800896	(A;A)	1.8x increased prostate cancer risk			

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
2	rs1867277	(A;A)	2x increased risk for thyroid cancer			
2	rs2073963	(G;T)	Increased risk of baldness			
2	rs2156921	(A;G)	1.29x increased risk for depression			
2	rs2201841	(T;T)	2.4x increased risk for Graves' disease		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	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	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	rs2707466	(G;G)	Weaker bones	Link	Link	
2	rs2736100	(T;T)	Higher risk of Interstitial lung disease: and t		Link	
2	rs2908004	(C;C)	Weaker bones	Link	Link	
2	rs3212227	(A;C)	Significantly increased risk of developing cerv			
2	rs3793784	(C;G)	1.5x risk for ARMD		Link	Link
2	rs3825776	(G;G)	>1.3x increased risk for ALS		Link	
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	rs493258	(A;G)	1.15x risk of Age Related Macular Degeneration			
2	rs4961	(G;T)	1.8x increased risk for high blood pressure	Link	Link	Link
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	rs5759167	(T;T)	Higher prostate cancer risk		Link	
2	rs6457617	(C;T)	2.3x risk of rheumatoid arthritis		Link	
2	rs6498169	(A;A)	>1.14x risk of multiple sclerosis		Link	
2	rs6601764	(C;C)	1.52x increased risk of developing Crohn's dise		Link	
2	rs662799	(A;G)	1.4x higher early heart attack risk; less weigh		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	rs6997709	(G;T)	1.2x higher risk for hypertension			
2	rs7250872	(T;T)	Increased risk of developing bipolar disorder	Link	Link	
2	rs7807268	(C;G)	1.3x risk for Crohn's disease		Link	
2	rs7923837	(G;G)	3.2x risk for T2D			
2	rs7961152	(A;C)	1.2x higher risk for hypertension			
2	rs828907	(T;T)	Increased risk of bladder cancer and 2x risk of			
2	rs854560	(A;T)	Higher risk for heart disease: diabetic retinop	Link	Link	Link
2	rs9525638	(T;T)	Weaker bones			
2	rs965513	(A;G)	1.77x increased thyroid cancer risk		Link	
2	rs9954153	(G;T)	~ 2.5 x higher risk for Fuchs' dystrophy: a corne			
2.0	rs9642880	(T;T)	1.5x increased bladder cancer risk		Link	
1.8	rs1136287	(C;T)	1.5x increased risk of wet ARMD in a Taiwanese	Link	Link	
1.8	rs37973	(A;G)	Among asthmatics: 1.5x more likely to show less			Link
1.7	rs2024513	(A;A)	1.7x higher risk for schizophrenia (among Han C			
1.6	rs1260326	(T;T)	Slightly higher risk for gout	Link	Link	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
1.6	rs3775948	(C;G)	Slightly higher risk for gout			
1.5	rs10260404	(C;T)	1.20x risk of developing ALS		Link	
1.5	rs10757272	(C;T)	1.30x increased risk for Coronary artery diseas			
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;C)	Slightly increased risk of developing prostate			
1.5	rs13376333	(C;T)	1.5x higher risk of atrial fibrillation		Link	
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	rs17115100	(G;T)	Slightly increased risk of developing Parkinson	Link	Link	
1.5	rs1801020	(C;T)	1.31x increased risk of heart disease	Link		Link
1.5	rs1994090	(G;T)	Slightly increased risk of developing Parkinson		Link	
1.5	rs2241880	(C;T)	1.4x increased risk for Crohn's disease in Cauc	Link	Link	Link
1.5	rs2286812	(C;T)	$\sim 2x$ higher risk for Fuchs' dystrophy: a corneal			
1.5	rs2697962	(A;G)	Slightly increased risk of developing Parkinson			
1.5	rs2736990	(C;T)	Slightly increased risk of developing Parkinson		Link	
1.5	rs27388	(A;G)	Slightly increased risk of developing schizophr			
1.5	rs2881766	(G;T)	Slightly increased risk for pregnancy-induced h			
1.5	rs358806	(A;C)	0.86x increased risk of developing Type-2 diabe		Link	
1.5	rs3745516	(A;G)	Slightly increased risk of developing primary b			
1.5	rs393152	(A;A)	Increased risk of both PD and AD	Link	Link	
1.5	rs4464148	(C;T)	1.10x increased risk for colorectal cancer			
1.5	rs4585	(T;T)	Slightly poorer $(0.75x)$ response to metformin i			
1.5	rs4626664	(A;G)	1.44x increased risk of developing restless leg		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	rs5746059	(A;A)	Slightly higher fat mass			
1.5	rs6435862	(G;T)	1.7x higher risk of aggressive neuroblastoma		Link	
1.5	rs6908425	(C;T)	1.63x increased risk of developing Crohn's dise		Link	
1.5	rs699473	(C;T)	\sim 1.5x increased brain tumor risk			
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)	\sim 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	
1.4	rs1126497	(T;T)	1.4x increased risk for breast cancer	Link	Link	Link
1.4	rs3131296	(G;G)	1.4x increased risk for schizophrenia	T 1 1	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	T 1 1	T * 1	
1.34	rs17465637	(C;C)	1.34x higher risk for myocardial infarction	Link	Link	T · 1
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		Link	
1.3	rs1746048	(C;C)	1.03 increased risk for coronary heart disease		Link	
1.3	rs356219	(A;G)	1.3x increased risk for Parkinson's disease		Link	
1.25	rs13387042	(A;A)	1.24x increased risk for breast cancer		Link	
1.25	rs748404	(T;T)	Slightly increased risk (1.25) for lung cancer		Link	
1.2	rs1344706	(T;T)	1.2x increased risk for schizophrenia		Link	T in la
1.2	rs143383	(C;T)	1.1x increased risk for osteoarthritis		Link	Link
1.2	rs2056116	(A;G)	1.18x risk for breast 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		T ' 1	
1.2	rs2814707	(A;G)	1.2x increased risk for ALS		Link	

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
1.2	rs3849942	(A;G)	1.2x increased risk for ALS		Link	
1.2	rs4324715	(C;C)	>1.5x increased testicular cancer risk for men			
1.2	rs4795067	(A;G)	Slight increase in risk for psoriatic arthritis			
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	rs9960767	(A;C)	1.2x increased risk for schizophrenia		Link	
1.1	rs11110912	(C;C)	1.3x high blood pressure risk			
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	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	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	rs2282679	(A;C)	Somewhat lower vitamin D levels			
1	rs2546890	(A;G)	Higher risk of multiple sclerosis			
1	rs3194051	(A;G)	1.12x risk of type-1 diabetes	Link	Link	Link
1	rs6166	(G;G)	Females slightly more likely to be sterile	Link	Link	Link
1	rs6932590	(C;T)	1.1x increased risk for schizophrenia		Link	
1	rs761100	(G;G)	Higher risk for dyslexia			
0.1	rs601338	(A;G)	Susceptible to Norovirus infections	Link	Link	Link
0	rs1004819	(C;C)	1.5x risk of Crohn's disease		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	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	rs6277	(C;C)	1.6x higher schizophrenia risk	Link	Link	Link
0	rs6314	(C;C)	Higher risk for RA	Link	Link	
0	rs7652331	(T;T)	Somewhat higher risk for prostate cancer			
0	rs7787082	(G;G)	7x less likely to respond to certain antidepres		Link	
0	rs855791	(T;T)	0.2 g/dL lower hemoglobin on average	Link	Link	Link

3.3 Genosets (Multi-variant Phenotypes)

Magnitude	Identifier	Summary
3.1	gs191	Problem metabolizing NSAIDs
3	gs127	Intermediate warfarin metabolizer
2.5	gs155	CYP3A5 non-expressor
2.5	gs161	CYP2C9 Intermediate Metabolizers
2.5	gs281	Part of the 88% of the population claimed not t
2	gs101	Probably able to digest milk
2	gs154	NAT2 Slow metabolizer
2	gs246	APOE3/APOE3
2	gs249	Parkinson's Disease Risk
1.5	gs220	HLA-B*1502?
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 ERS1176615 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/ERS1176615

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.