PGP-UK Genomics Report for uk37D41A

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.

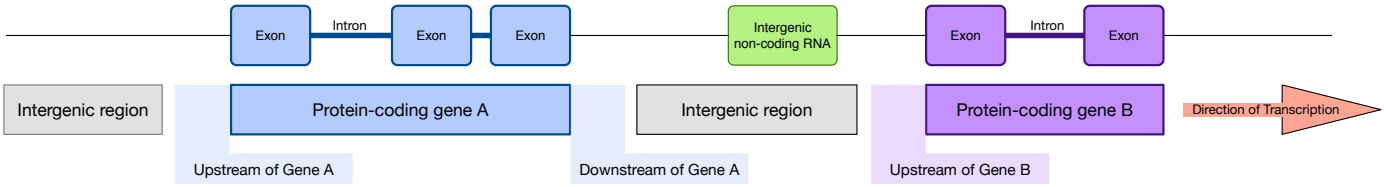

Figure 1: Diagram of gene structure indicating locations of potential variants

Feature	Count
Lines of input read	5026149
Variants filtered out	0
Novel / existing variants	511686 (10.2) / 4502842 (89.8)
Overlapped genes	56745
Overlapped transcripts	67609
Overlapped regulatory features	168055

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.

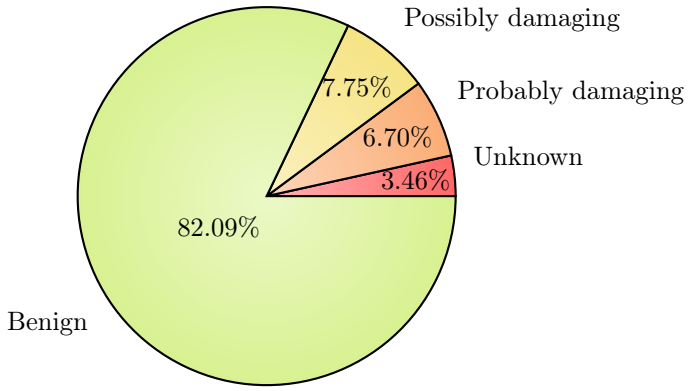

Figure 2: PolyPhen Summary

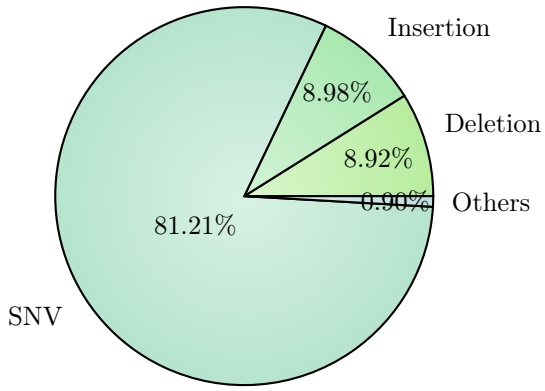

Figure 3: Variant Class

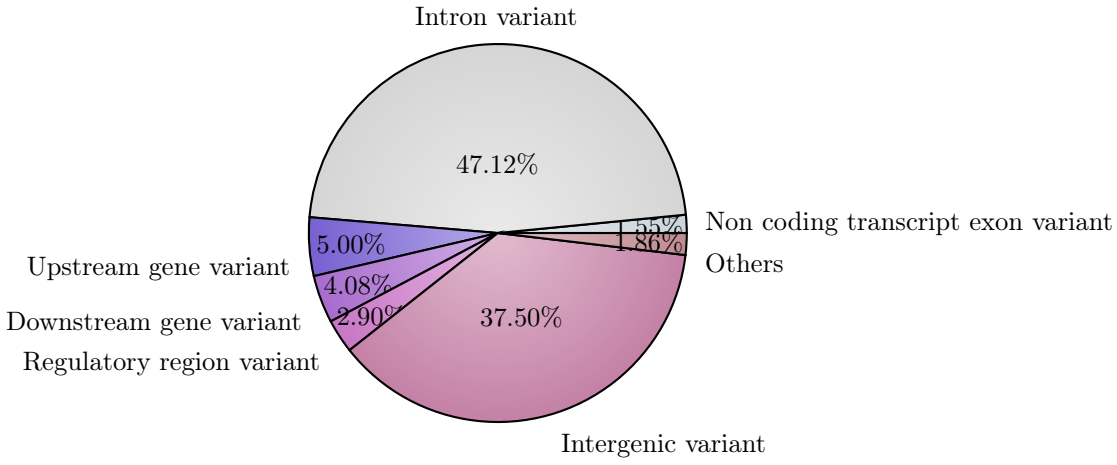

Figure 4: Consequence type

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 99.8 percent European [Toscani in Italia, Finnish in Finland, British in England and Scotland, Iberian Population in Spain].

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 uk37D41A

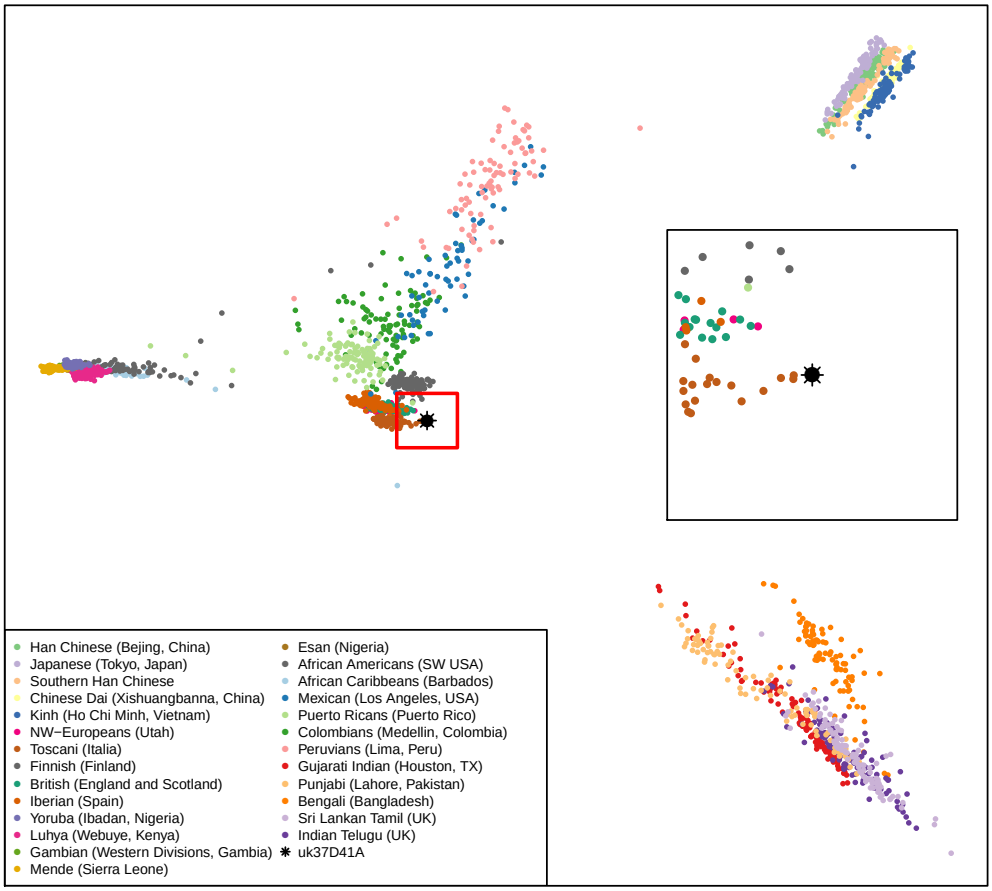

Figure 5: Ancestry Principal Component Analysis

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
3	rs925391	(T;T)	Unlikely to go bald			
2.4	rs9272346	(G;G)	0.08x risk type-1 diabetes		Link	
2.1	rs2511989	(A;G)	0.63x decreased age-related macular degeneratio...		Link	
2.1	rs3775291	(A;G)	0.71x decreased risk for dry age related macula...	Link	Link	Link
2	rs10468017	(T;T)	Associated with higher HDL cholesterol		Link	
2	rs11045585	(A;A)	24% chance (lower than average) of docetaxel-in...		Link	
2	rs12979860	(C;C)	~80% of such hepatitis C patients respond to tr...		Link	Link
2	rs1864163	(G;G)	Associated with higher HDL cholesterol		Link	
2	rs2235015	(G;T)	Somewhat more likely to respond to certain anti...	Link	Link	
2	rs2542052	(C;C)	Better odds of living to 100			
2	rs261332	(A;G)	Associated with higher HDL cholesterol			
2	rs3750817	(C;T)	0.78x reduced risk for breast cancer			
2	rs3819331	(T;T)	Lower risk of autism	Link		
2	rs4149268	(A;G)	Associated with higher HDL cholesterol		Link	
2	rs6505162	(A;C)	0.58x decreased risk for esophageal cancer	Link		
2	rs9642880	(G;G)	Slightly lower risk of Bladder Cancer.		Link	
1.8	rs1800588	(C;T)	Higher HDL-C levels	Link	Link	
1.8	rs187238	(C;C)	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;C)	0.8x decreased risk for ovarian cancer		Link	
1.8	rs4714156	(C;C)	<0.61x risk for restless legs			
1.5	rs1063192	(C;T)	0.71x reduced risk of myocardial infarction			
1.5	rs1165205	(A;A)	0.85x decreased gout risk		Link	
1.5	rs4149274	(C;T)	Associated with higher HDL (good) cholesterol			
1.5	rs4939883	(C;C)	Associated with higher HDL cholesterol		Link	
1.4	rs6495446	(C;T)	0.8x reduced risk for chronic kidney disease			
1.2	rs4686484	(G;G)	Slightly decreased risk for celiac disease			

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
1.2	rs9306160	(C;T)	0.75x (reduced) risk for metastasis in LN-/ER+ ...	Link	Link	
1.1	rs13333226	(A;G)	Slightly lower risk for hypertension			Link
1.1	rs2293347	(G;G)	Among NSCLC patients: better Gefitinib response...	Link		Link
1.1	rs7568369	(T;T)	0.90x reduced risk of obesity			
1	rs2351299	(G;T)	Possible reduced risk of Autism			
1	rs2952768	(C;C)	Less drug dependence: decreased effectiveness o...			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	rs6583817	(C;T)	~0.80x (lower) risk for late onset Alzheimer's ...			
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	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	rs1800562	(G;G)	Not a C282Y 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	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
3	rs10897346	(C;C)	If depressed: 2.6x more likely to not respond t...			
3	rs2237717	(T;T)	Reduced abilities related to neurocognition and...			
3	rs2306402	(C;T)	1.18x increased risk for late-onset Alzheimer's...			
3	rs3738579	(T;T)	1.5x - 2x increased risk for cervical cancer: H...			
2.7	rs10830963	(C;G)	Increased type-2 diabetes risk; higher gestatio...		Link	
2.5	rs1057910	(A;C)	CYP2C9*3 carrier; average 40% reduction in warf...	Link	Link	Link
2.5	rs11190870	(T;T)	Possibly even more increased risk of scoliosis			
2.5	rs1121980	(C;T)	1.67x risk for obesity		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	rs1421085	(C;T)	~1.3x increased obesity risk		Link	Link
2.5	rs17696736	(G;G)	1.94x risk of type-1 diabetes		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	rs2254958	(C;C)	1.61x increased risk for Alzheimer's			
2.5	rs3738919	(C;C)	1.94x risk of developing rheumatoid arthritis			
2.5	rs5888	(C;T)	3x higher risk for age-related macular degenera...	Link		
2.5	rs7574865	(G;T)	1.3x risk of rheumatoid arthritis; 1.55x risk o...		Link	Link
2.4	rs1143679	(A;G)	1.78x increased risk for SLE	Link	Link	
2.3	rs1859962	(G;G)	1.28x increased risk for prostate cancer		Link	
2.2	rs1024611	(C;C)	Increased risk of exercise induced ischemia: In...			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	rs10427255	(C;C)	Highest odds of photic sneeze reflex			
2.1	rs1695	(G;G)	3.5x asthma risk in certain populations	Link	Link	Link
2.1	rs17070145	(C;C)	Reduced memory abilities			Link
2.1	rs17077540	(A;G)	1.6x major depressive disorder risk			
2.1	rs17563	(C;C)	Risk for otosclerosis	Link	Link	Link
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	rs4444903	(G;G)	3.5x risk of hep-cancer in cirrhosis patients; ...			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	rs646776	(A;A)	1.2x risk of coronary artery disease		Link	
2.1	rs795484	(A;G)	Increased morphine dose requirement and postope...			
2	rs10248420	(A;A)	7x less likely to respond to certain antidepres...		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	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	rs12567232	(A;G)	Increased risk for Crohn's Disease		Link	
2	rs1333048	(A;C)	1.3x increased coronary artery disease risk			
2	rs1537415	(G;G)	2x increased risk for periodontitis		Link	
2	rs16942	(A;G)	Very slightly increased breast cancer risk	Link	Link	Link
2	rs16944	(G;G)	Increased risk of mental disorders		Link	
2	rs17001266	(-;C)	1.58x increased risk for schizophrenia in males...			
2	rs1734791	(A;T)	1.4x increased risk for lupus			
2	rs17435	(A;T)	1.4x increased risk for lupus			
2	rs17576	(G;G)	Higher risk for lung cancer: and COPD in smoker...	Link	Link	
2	rs1801160	(A;G)	Possible 5-fluorouracil toxicity	Link	Link	Link
2	rs2073963	(G;T)	Increased risk of baldness			
2	rs2143340	(C;T)	Increased risk of dyslexia and poor reading per...			

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
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	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	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	rs27388	(A;A)	Increased risk of developing schizophrenia			
2	rs358806	(C;C)	1.78x increased risk of developing Type-2 diabe...		Link	
2	rs3746444	(C;T)	~1.2x increased risk for cancer	Link		
2	rs3775948	(G;G)	Slightly higher risk for gout			
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	rs449647	(A;A)	Lower levels of ApoE			
2	rs4633	(C;T)	Higher risk for endometrial cancer	Link	Link	Link
2	rs5174	(A;G)	1.3x increased risk for heart disease	Link	Link	Link
2	rs5759167	(T;T)	Higher prostate cancer risk		Link	
2	rs6457617	(C;T)	2.3x risk of rheumatoid arthritis		Link	
2	rs6601764	(C;C)	1.52x increased risk of developing Crohn's dise...		Link	
2	rs663048	(G;T)	Significantly increased risk of developing lung...	Link	Link	
2	rs6700125	(T;T)	1.76x increased risk for ALS			
2	rs6807362	(C;C)	Increased autism risk	Link	Link	
2	rs6896702	(T;T)	Increased risk of developing Parkinson's Diseas...			
2	rs6897932	(C;C)	1.08x increased risk for multiple sclerosis	Link	Link	Link
2	rs6908425	(C;C)	1.95x increased risk of developing Crohn's dise...		Link	
2	rs6997709	(G;G)	1.5x higher risk for hypertension			
2	rs699	(C;T)	Increased risk of hypertension	Link	Link	Link
2	rs7216389	(T;T)	1.5x increased risk for Childhood Asthma.		Link	
2	rs7442295	(A;A)	~4x higher risk for hyperuracemia		Link	
2	rs7794745	(T;T)	Slightly increased risk for autism		Link	Link
2	rs7807268	(C;G)	1.3x risk for Crohn's disease		Link	
2	rs7961152	(A;C)	1.2x higher risk for hypertension			
2	rs854560	(A;A)	Higher risk for heart disease: diabetic retinop...	Link	Link	Link
2	rs965513	(A;G)	1.77x increased thyroid cancer risk		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	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
1.7	rs8055236	(G;T)	1.9x risk for heart disease		Link	
1.6	rs11523871	(A;C)	1.6x increased breast cancer risk for women ove...	Link	Link	
1.6	rs2981745	(C;T)	1.6x increased risk for breast cancer in female...			
1.6	rs3764880	(A;A)	1.2 - 1.8x increased tuberculosis risk	Link	Link	
1.5	rs10260404	(C;T)	1.20x risk of developing ALS		Link	
1.5	rs10464059	(A;G)	Slightly increased risk of developing Parkinson...			
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	rs11171739	(C;T)	1.34x risk of developing Type-1 diabetes		Link	
1.5	rs12037606	(A;G)	1.22x risk of developing Crohn's disease			
1.5	rs1223271	(A;G)	Slightly increased risk of developing Parkinson...		Link	
1.5	rs12431733	(C;T)	Slightly increased risk of developing Parkinson...		Link	
1.5	rs12469063	(A;G)	Slightly increased risk of developing restless ...			
1.5	rs12498742	(A;A)	1.25 increased risk for gout			
1.5	rs13149290	(C;C)	Slightly increased risk of developing prostate ...			

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
1.5	rs13181	(G;T)	1.12x increased risk for cutaneous melanoma	Link	Link	Link
1.5	rs144848	(G;T)	Very slightly increased breast cancer risk	Link	Link	Link
1.5	rs1799950	(A;G)	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	rs2007153	(G;G)	Increased risk of schizophrenia in limited stud...			
1.5	rs2177369	(C;C)	1.5x increased risk for Alzheimer's disease			
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	rs2736990	(C;T)	Slightly increased risk of developing Parkinson...		Link	
1.5	rs2881766	(T;T)	Slightly increased risk for pregnancy-induced h...			
1.5	rs309375	(T;T)	Larger mosquito bites			
1.5	rs3212227	(A;A)	1.43x increased risk of developing psoriasis an...			
1.5	rs3825776	(A;G)	1.3x increased risk for ALS		Link	
1.5	rs393152	(A;A)	Increased risk of both PD and AD	Link	Link	
1.5	rs4027132	(A;G)	1.39x increased risk of developing bipolar diso...			
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	rs4785763	(A;C)	1.5x higher risk for melanoma		Link	
1.5	rs4845618	(G;T)	1.7x increased melanoma risk			
1.5	rs4982731	(C;C)	Possible higher risk of childhood acute lymphob...			
1.5	rs5746059	(A;A)	Slightly higher fat mass			
1.5	rs619203	(C;G)	Increases susceptibility to Myocardial Infarcti...	Link	Link	
1.5	rs6498169	(A;G)	1.14x risk of multiple sclerosis		Link	
1.5	rs6710341	(A;G)	Slightly increased risk of developing restless ...			
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	rs9561778	(G;T)	~2x increased risk of adverse drug reactions fr...		Link	
1.5	rs9652490	(A;G)	Slightly increased risk of developing Parkinson...		Link	
1.5	rs966221	(C;C)	1.5x increased stroke risk certain populations			
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	rs12770228	(A;G)	1.4x increased risk for meningioma			
1.4	rs1800693	(G;G)	Slight (1.4x) increase in risk for multiple scl...	Link	Link	Link
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	rs4959039	(A;G)	1.4x higher risk for multiple sclerosis			
1.4	rs6010620	(G;G)	1.4x higher risk for glioma development; but th...		Link	
1.34	rs17465637	(C;C)	1.34x higher risk for myocardial infarction	Link	Link	
1.3	rs1047286	(C;T)	1.3x increased risk for age-related macular deg...	Link	Link	Link
1.3	rs10947262	(C;C)	1.3x increased risk for osteoarthritis			
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	rs1375144	(C;T)	1.32x increased risk of developing bipolar diso...			
1.3	rs1434536	(A;G)	1.29x increased breast cancer risk			
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	rs2059693	(C;T)	1.3x increased risk for testicular cancer			
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			
1.3	rs4295627	(G;T)	1.36x higher risk for glioma development		Link	

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
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	rs10865331	(A;G)	1.2x higher risk for ankylosing spondylitis			
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	rs143383	(C;T)	1.1x increased risk for osteoarthritis		Link	Link
1.2	rs2056116	(A;G)	1.18x risk for breast cancer			
1.2	rs2252586	(A;G)	1.2x higher risk for glioma development			
1.2	rs2814707	(A;G)	1.2x increased risk for ALS		Link	
1.2	rs3740878	(A;A)	1.46x type II diabetes risk; common	Link		Link
1.2	rs3849942	(A;G)	1.2x increased risk for ALS		Link	
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	rs8050136	(A;C)	1.2x increased risk for T2D in some populations...		Link	
1.1	rs11110912	(C;C)	1.3x high blood pressure risk			
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	rs4324715	(C;T)	1.5x increased testicular cancer risk for men			
1.1	rs688034	(C;T)	1.1x risk higher risk for coronary artery disea...		Link	
1.1	rs6897876	(C;T)	Slight increase in testicular cancer risk for m...			
1.1	rs7171755	(A;G)	Very slight decrease in cortical thickness and ...			
1.1	rs7412	(C;C)	More likely to gain weight if taking olanzapine...	Link	Link	Link
1.07	rs2291834	(C;C)	Very slightly higher risk for myocardial infarc...			
1	rs10504861	(G;G)	Major allele: normal risk of migraine			
1	rs10761659	(A;G)	1.2x risk of Crohn's disease		Link	
1	rs1143674	(A;A)	1.3x increased autism risk	Link		
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	(T;T)	1.1x increased risk for schizophrenia		Link	
1.0	rs11246226	(A;A)	Increased risk of schizophrenia in limited stud...		Link	
0.1	rs601338	(G;G)	Susceptible to Norovirus infections	Link	Link	Link
0	rs1061646	(C;C)	1.16x increased risk for breast cancer	Link		Link
0	rs1800860	(A;A)	10% smaller kidneys as newborns	Link		Link
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	rs7652331	(T;T)	Somewhat higher risk for prostate cancer			
0	rs7787082	(G;G)	7x less likely to respond to certain antidepres...		Link	

3.3 Genosets (Multi-variant Phenotypes)

Magnitude	Identifier	Summary
3.1	gs191	Problem metabolizing NSAIDs
2.6	gs296	Lower heart attack risk than average
2.5	gs100	Lactose intolerance risk
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...
2.5	gs285	You will lose 2.5x as much weight on a low fat ...
2	gs104	Restless legs syndrome risk
2	gs156	NAT2 Rapid metabolizer.
2	gs239	Reduced conversion of beta-carotene to retinol
2	gs246	APOE3/APOE3
1.5	gs247	Parkinson's Disease Risk
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 ERS1176605 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/ERS1176605

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.