PGP-UK Genomics Report for uk421E54

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.

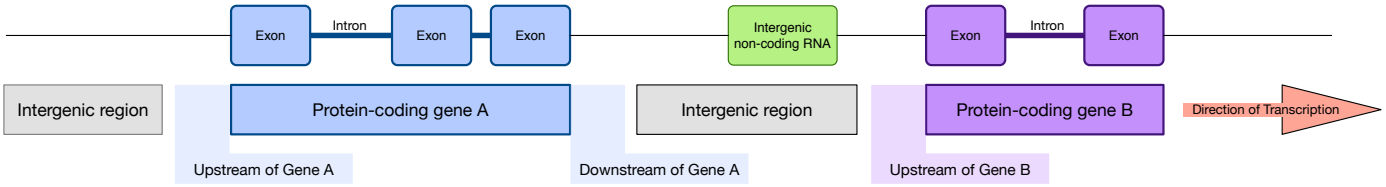

Figure 1: Diagram of gene structure indicating locations of potential variants

Feature	Count
Lines of input read	4957602
Variants filtered out	0
Novel / existing variants	480600 (9.7) / 4465428 (90.3)
Overlapped genes	56766
Overlapped transcripts	67560
Overlapped regulatory features	167199

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.

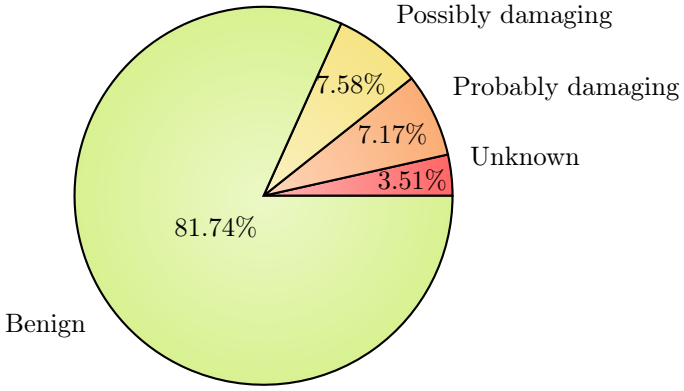

Figure 2: PolyPhen Summary

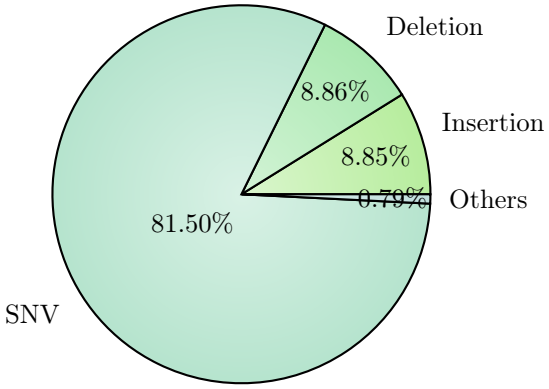

Figure 3: Variant Class

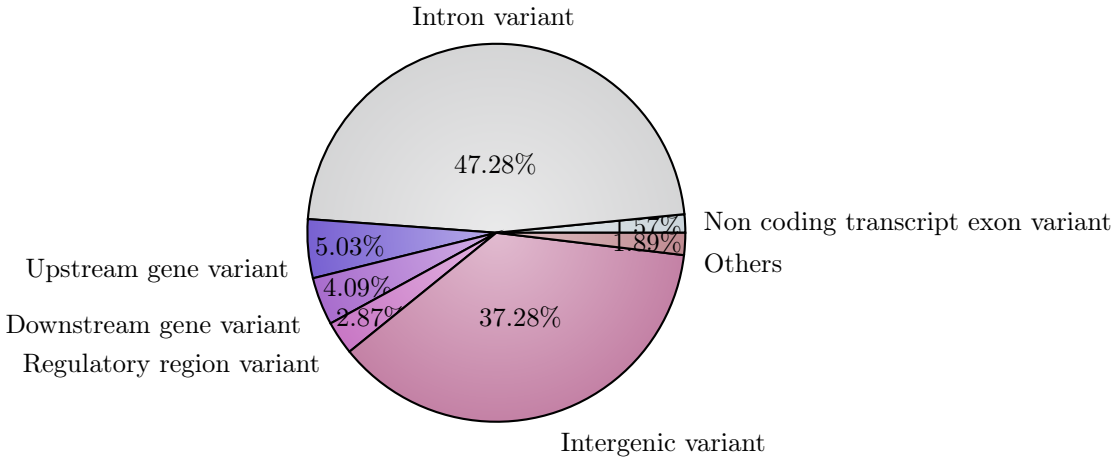

Figure 4: Consequence type

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 [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).

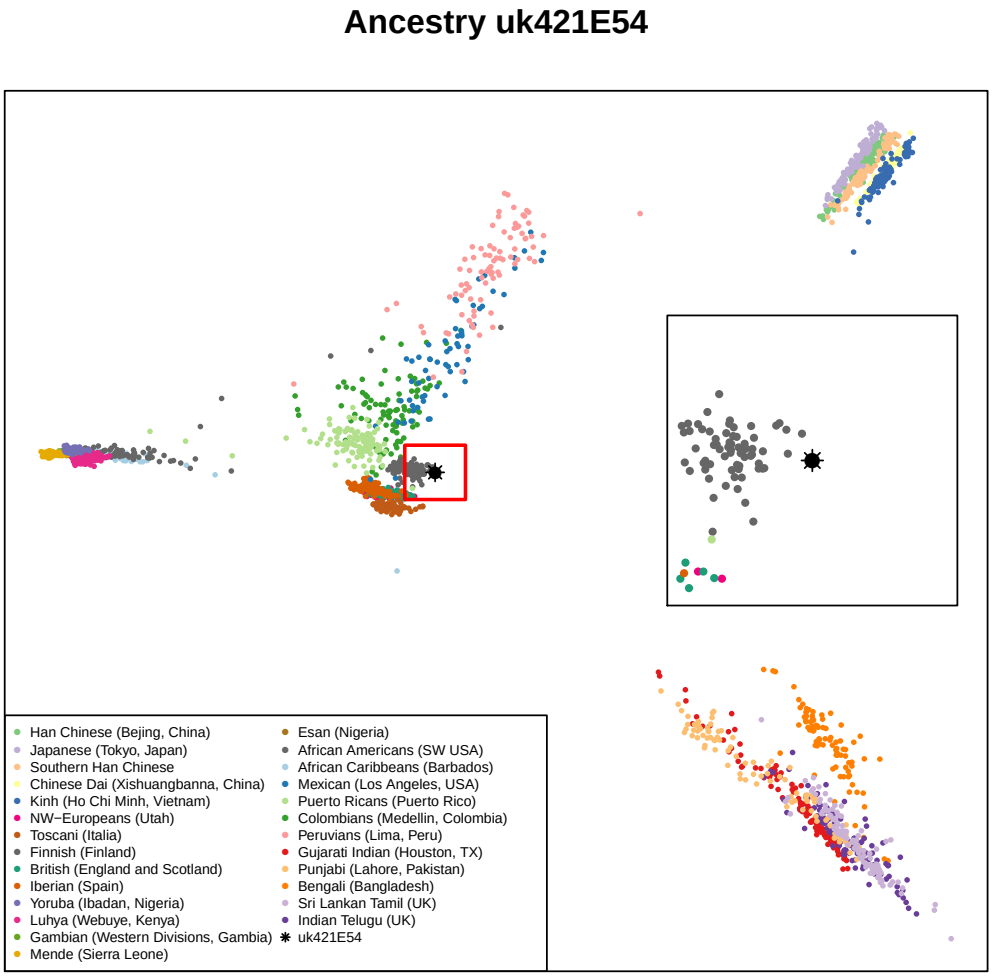

Figure 5: Ancestry Principal Component Analysis

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	rs10468017	(C;T)	Associated with higher HDL cholesterol		Link	
2	rs11045585	(A;A)	24% chance (lower than average) of docetaxel-in...		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	rs1160312	(G;G)	Reduced risk of Baldness.		Link	
2	rs12979860	(C;C)	~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	rs2073963	(T;T)	Reduced risk of baldness			
2	rs2229616	(A;G)	Lower risk of metabolic syndrome	Link	Link	Link
2	rs2241423	(A;G)	0.79 decreased risk for obesity			
2	rs2243250	(C;T)	0.6x decreased risk for myocardial infarction i...			
2	rs261332	(A;G)	Associated with higher HDL cholesterol			
2	rs3736309	(A;G)	0.44x decreased risk for chronic obstructive pu...			
2	rs3750817	(C;T)	0.78x reduced risk for breast cancer			
2	rs3819331	(T;T)	Lower risk of autism	Link		
2	rs4149268	(G;G)	Associated with higher HDL cholesterol		Link	
2	rs6505162	(A;C)	0.58x decreased risk for esophageal cancer	Link		
2	rs6855911	(A;G)	0.62x decreased risk for gout		Link	
2	rs7776725	(T;T)	Stronger bones		Link	
2	rs8070723	(A;G)	0.18x reduced risk of developing progressive su...			
1.8	rs1800588	(T;T)	Higher HDL-C levels	Link	Link	
1.8	rs187238	(C;G)	Hypertension not a risk factor for sudden cardi...			
1.8	rs3814113	(C;C)	0.8x decreased risk for ovarian cancer		Link	
1.6	rs10801935	(C;C)	0.3x decreased risk of breast cancer			
1.6	rs2278206	(C;C)	Possibly less susceptible to asthma	Link	Link	
1.6	rs3025786	(C;T)	Slightly decreased Alzheimer's disease risk amo...	Link		

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
1.5	rs1026732	(A;G)	0.70x risk for restless legs		Link	
1.5	rs1063192	(C;C)	0.71x reduced risk of myocardial infarction			
1.5	rs11136000	(C;T)	0.84x decreased risk for Alzheimer's disease		Link	
1.5	rs11212617	(A;C)	Somewhat increased likelihood of treatment succ...			Link
1.5	rs11635424	(A;G)	0.70x risk for restless legs		Link	
1.5	rs12593813	(A;G)	0.71x risk for restless legs		Link	
1.5	rs3784709	(C;T)	0.71x risk of developing restless legs syndrome...		Link	
1.5	rs3790844	(C;T)	Slightly reduced risk (0.77x) for pancreatic ca...			
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	rs4489954	(G;T)	0.69x risk risk of developing restless legs syn...		Link	
1.5	rs4939883	(C;C)	Associated with higher HDL cholesterol		Link	
1.5	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			
1.2	rs4320932	(A;G)	0.87x decreased risk for ovarian cancer			
1.2	rs4686484	(G;G)	Slightly decreased risk for celiac disease			
1.1	rs11172113	(C;T)	0.9x lower risk for migraines			
1.1	rs2293347	(G;G)	Among NSCLC patients: better Gefitinib response...	Link		Link
1.1	rs7568369	(G;T)	0.90x reduced risk of obesity			
1	rs182549	(C;T)	Can digest milk.			Link
1	rs7850258	(A;G)	Typical odds of developing primary hypothyroidi...			
1.0	rs11246226	(C;C)	Decreased risk of schizophrenia in limited stud...		Link	
0	rs10427255	(T;T)	Lowest odds of photic sneeze reflex			
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	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	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	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	

3.2 Possibly Harmful Traits

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
3.5	rs875858	(C;T)	Docetaxel sensitive?			
3	rs10897346	(C;C)	If depressed: 2.6x more likely to not respond t...			
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	rs3903239	(C;C)	Higher frequency of atrial fibrillation			
2.9	rs16901979	(A;C)	1.5x increased risk for prostate cancer		Link	
2.5	rs10490924	(G;T)	2.7x risk for age related macular degeneration	Link	Link	Link
2.5	rs12803066	(A;G)	Increased risk of myopia			
2.5	rs16969968	(A;G)	Slightly higher risk for nicotine dependence: l...	Link	Link	Link
2.5	rs17595731	(C;G)	~5 fold higher risk for Fuchs' dystrophy: a cor...			
2.5	rs2254958	(C;C)	1.61x increased risk for Alzheimer's			
2.5	rs2943634	(C;C)	Slightly higher risk of ischemic stroke		Link	
2.5	rs3780374	(A;G)	Substantially increased odds of developing V617...			
2.5	rs613872	(G;T)	~5 fold higher risk for Fuchs' dystrophy: a cor...			
2.5	rs664143	(C;T)	Higher risk for number of cancers			
2.5	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	
2.5	rs891512	(A;G)	Higher blood pressure than G;G	Link		
2.2	rs2231137	(G;G)	~1.5-3x increased risk for ischemic stroke	Link	Link	Link
2.2	rs2305089	(T;T)	Higher risk for chordoma	Link	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	rs17070145	(C;C)	Reduced memory abilities			Link
2.1	rs17077540	(A;G)	1.6x major depressive disorder risk			
2.1	rs1800730	(A;T)	Carrier for a mild form of hemochromatosis	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	rs5186	(A;C)	~1.4x increased risk of hypertension	Link	Link	Link
2.1	rs6457617	(T;T)	5.2x risk of rheumatoid arthritis		Link	
2.1	rs795484	(A;G)	Increased morphine dose requirement and postope...			
2.1	rs944289	(C;T)	1.3x increased thyroid cancer risk		Link	
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	rs10984447	(A;G)	1.17x increased risk for multiple sclerosis		Link	
2	rs11171739	(C;C)	1.75x risk of developing Type-1 diabetes		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	rs12696304	(G;G)	Prone to aging faster: at least in European pop...			
2	rs1585215	(A;G)	2x increased risk for Hodgkin lymphoma			
2	rs1691053	(A;G)	Increased risk of developing prostate cancer			
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	rs17435	(T;T)	1.4x increased risk for lupus			
2	rs17576	(A;G)	Higher risk for MI and lung cancer: and COPD in...	Link	Link	
2	rs17696736	(A;G)	1.34x risk of type-1 diabetes		Link	
2	rs1800896	(A;G)	1.6x increased prostate cancer risk			
2	rs2201841	(C;T)	1.5x increased risk for Crohn's disease; 2x inc...		Link	

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
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	rs2305795	(A;G)	1.28x higher risk of narcolepsy compared to (G;...			Link
2	rs2420946	(C;T)	1.20x risk for breast cancer			
2	rs25487	(G;G)	2x higher risk for skin cancer; possibly other ...	Link	Link	Link
2	rs2697962	(A;A)	Increased risk of developing Parkinson's Diseas...			
2	rs3197999	(T;T)	1.2x risk of Crohn's	Link	Link	
2	rs358806	(C;C)	1.78x increased risk of developing Type-2 diabe...		Link	
2	rs3738579	(C;C)	0.6x decreased risk for cervical cancer: but 1....			
2	rs3738919	(A;C)	1.94x risk of developing rheumatoid arthritis			
2	rs3746444	(C;T)	~1.2x increased risk for cancer	Link		
2	rs3793784	(C;G)	1.5x risk for ARMD		Link	Link
2	rs3802842	(C;C)	>1.17x increased risk of colorectal cancer		Link	
2	rs4148739	(A;A)	7x less likely to respond to certain antidepres...		Link	
2	rs4633	(T;T)	Higher risk for endometrial cancer	Link	Link	Link
2	rs4792311	(A;G)	Increased risk of prostate cancer	Link	Link	Link
2	rs486907	(A;A)	2x increased prostate cancer risk	Link	Link	Link
2	rs4961	(G;T)	1.8x increased risk for high blood pressure	Link	Link	Link
2	rs4968451	(A;C)	1.61x increased risk for meningioma			
2	rs5174	(A;A)	1.3x increased risk for heart disease	Link	Link	Link
2	rs520354	(A;A)	Increased risk in men for biliary conditions			
2	rs587776825	(-;C)	Associated with MODY3; maturity onset of diabet...	Link		Link
2	rs6441286	(G;T)	1.54x chance of developing primary biliary cirr...		Link	
2	rs6897932	(C;C)	1.08x increased risk for multiple sclerosis	Link	Link	Link
2	rs7216389	(T;T)	1.5x increased risk for Childhood Asthma.		Link	
2	rs7794745	(A;T)	Slightly increased risk for autism		Link	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	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	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	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	
1.6	rs3775948	(C;G)	Slightly higher risk for gout			
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	rs13149290	(C;T)	Slightly increased risk of developing prostate ...			
1.5	rs140701	(A;G)	Increased risk for anxiety disorders			
1.5	rs1799950	(A;G)	Very slightly increased breast cancer risk	Link	Link	Link
1.5	rs1801274	(C;T)	Complex; generally greater risk for cancer prog...	Link	Link	Link
1.5	rs1867277	(A;G)	1.5x increased risk for thyroid cancer			
1.5	rs1975197	(C;T)	1.3x increased risk of developing restless legs...		Link	
1.5	rs1994090	(G;T)	Slightly increased risk of developing Parkinson...		Link	
1.5	rs199533	(C;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	rs2272127	(C;C)	Associated with herpes and schizophrenia			
1.5	rs2286812	(C;T)	~2x higher risk for Fuchs' dystrophy: a corneal...			

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
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	rs309375	(T;T)	Larger mosquito bites			
1.5	rs3212227	(A;A)	1.43x increased risk of developing psoriasis an...			
1.5	rs3745516	(A;G)	Slightly increased risk of developing primary b...			
1.5	rs3814570	(C;T)	1.3x increased risk for Crohn's disease with il...			
1.5	rs3825776	(A;G)	1.3x increased risk for ALS		Link	
1.5	rs401681	(C;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	rs4464148	(C;T)	1.10x increased risk for colorectal cancer			
1.5	rs4626664	(A;G)	1.44x increased risk of developing restless leg...		Link	
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	rs5746059	(A;G)	Slightly higher fat mass			
1.5	rs619203	(C;G)	Increases susceptibility to Myocardial Infarcti...	Link	Link	
1.5	rs642961	(A;G)	1.68x increased risk of cleft lip		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	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	rs699473	(C;T)	~1.5x increased brain tumor risk			
1.5	rs7341475	(G;G)	1.58x increased schizophrenia risk for women		Link	
1.5	rs7454108	(C;T)	Single HLA-DQ8 haplotype			
1.5	rs7774434	(C;T)	Slightly increased risk of developing primary b...			
1.5	rs872071	(A;G)	~1.5x increased risk for chronic lymphocytic le...		Link	
1.5	rs9561778	(G;T)	~2x increased risk of adverse drug reactions fr...		Link	
1.5	rs9642880	(G;T)	1.2x increased bladder cancer risk		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	rs1545843	(A;A)	1.4x increased risk for depression (for those u...			
1.4	rs1800693	(G;G)	Slight (1.4x) increase in risk for multiple scl...	Link	Link	Link
1.4	rs3131296	(G;G)	1.4x increased risk for schizophrenia		Link	
1.4	rs4977756	(G;G)	1.93x higher risk for glioma development		Link	
1.4	rs6010620	(G;G)	1.4x higher risk for glioma development; but th...		Link	
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	rs1434536	(A;G)	1.29x increased breast cancer risk			
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	rs2736100	(G;T)	1.3x higher risk for glioma development: 2.1x r...		Link	
1.3	rs9858542	(A;A)	1.8x risk of Crohn's disease	Link	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	rs2076295	(G;T)	One copy of the risk allele (G): slightly incre...			
1.2	rs3740878	(A;A)	1.46x type II diabetes risk; common	Link		Link
1.2	rs393152	(A;G)	Slight increased risk of both PD and AD	Link	Link	
1.2	rs419788	(A;G)	2.0x risk for lupus	Link		
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	

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
1.2	rs6897876	(C;C)	Slight increase in testicular cancer risk for m...			
1.2	rs9960767	(A;C)	1.2x increased risk for schizophrenia		Link	
1.17	rs17465637	(A;C)	1.17x higher risk for myocardial infarction	Link	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	rs249954	(C;T)	Potentially increased risk of Breast Cancer			Link
1.1	rs34516635	(G;G)	Less longevity for Ashkenazi Jewish women.	Link		Link
1.1	rs3818361	(C;T)	1.15x increased risk for late-onset Alzheimer's...			
1.1	rs7171755	(A;G)	Very slight decrease in cortical thickness and ...			
1.1	rs7412	(C;C)	More likely to gain weight if taking olanzapine...	Link	Link	Link
1.1	rs889312	(C;C)	Very slightly higher risk for breast cancer		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	rs2546890	(A;A)	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	
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	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	rs6277	(C;C)	1.6x higher schizophrenia risk	Link	Link	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	gs155	CYP3A5 non-expressor
2.5	gs157	More stimulated by coffee
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 ...
2.4	gs297	Lower heart attack risk than average
2.2	gs280	Light hair color for europeans
2	gs101	Probably able to digest milk
2	gs104	Restless legs syndrome risk
2	gs154	NAT2 Slow metabolizer
2	gs181	CYP2D6*2
2	gs239	Reduced conversion of beta-carotene to retinol
2	gs246	APOE3/APOE3
1.5	gs247	Parkinson's Disease Risk
1	gs163	CYP2D6*2A

4 Raw Data

The raw data used to create this report has been assigned the identifier ERS1176563 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/ERS1176563

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.