Genomics Report for uk4CA868

1 Summary

This is the genome report for participant uk4CA868 . It was produced using collaborative research tools, including SNPedia and GetEvidence. This summary shows an overview of all the variants which were found in the genome for this individual. They have been compared with a reference genome.

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. "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. The diagram in Figure 1 is a simplification of the usual gene structure.

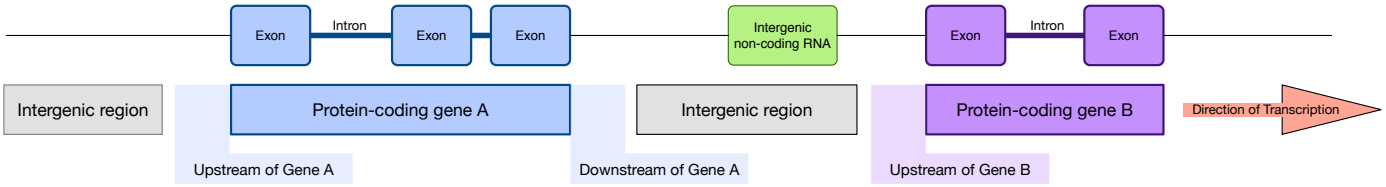

Figure 1: Diagram of gene structure indicating locations of potential variants

Feature	Count
Lines of input read	3921241
Variants remaining after filtering	3896926
Novel / existing variants	56212 (1.4%) / 3840714 (98.6%)
Overlapped genes	54278
Overlapped transcripts	63774
Overlapped regulatory features	206105

Table 1: Variant calling summary

There are several different types of genomic variants. The most common are single nucleotide variants (SNV) that correspond to the change of a single nucleotide in the DNA. 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.

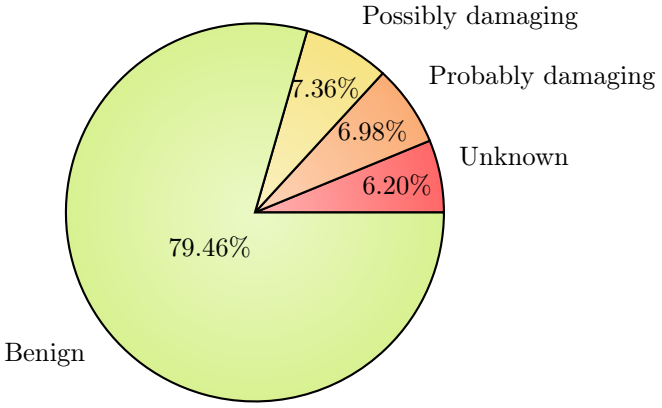

Figure 2: PolyPhen Summary

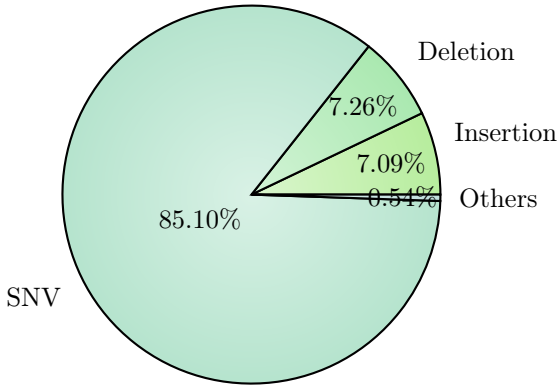

Figure 3: Variant Class

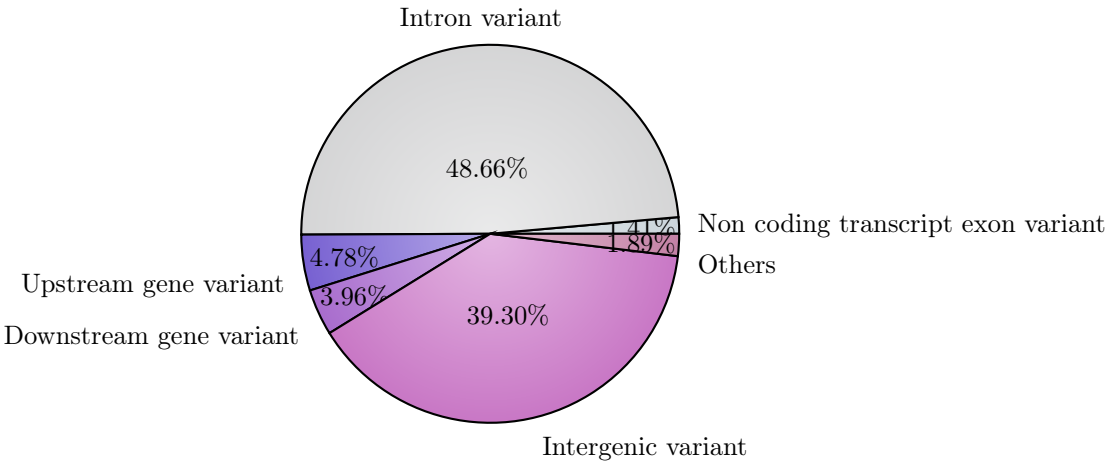

Figure 4: Consequence type

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.

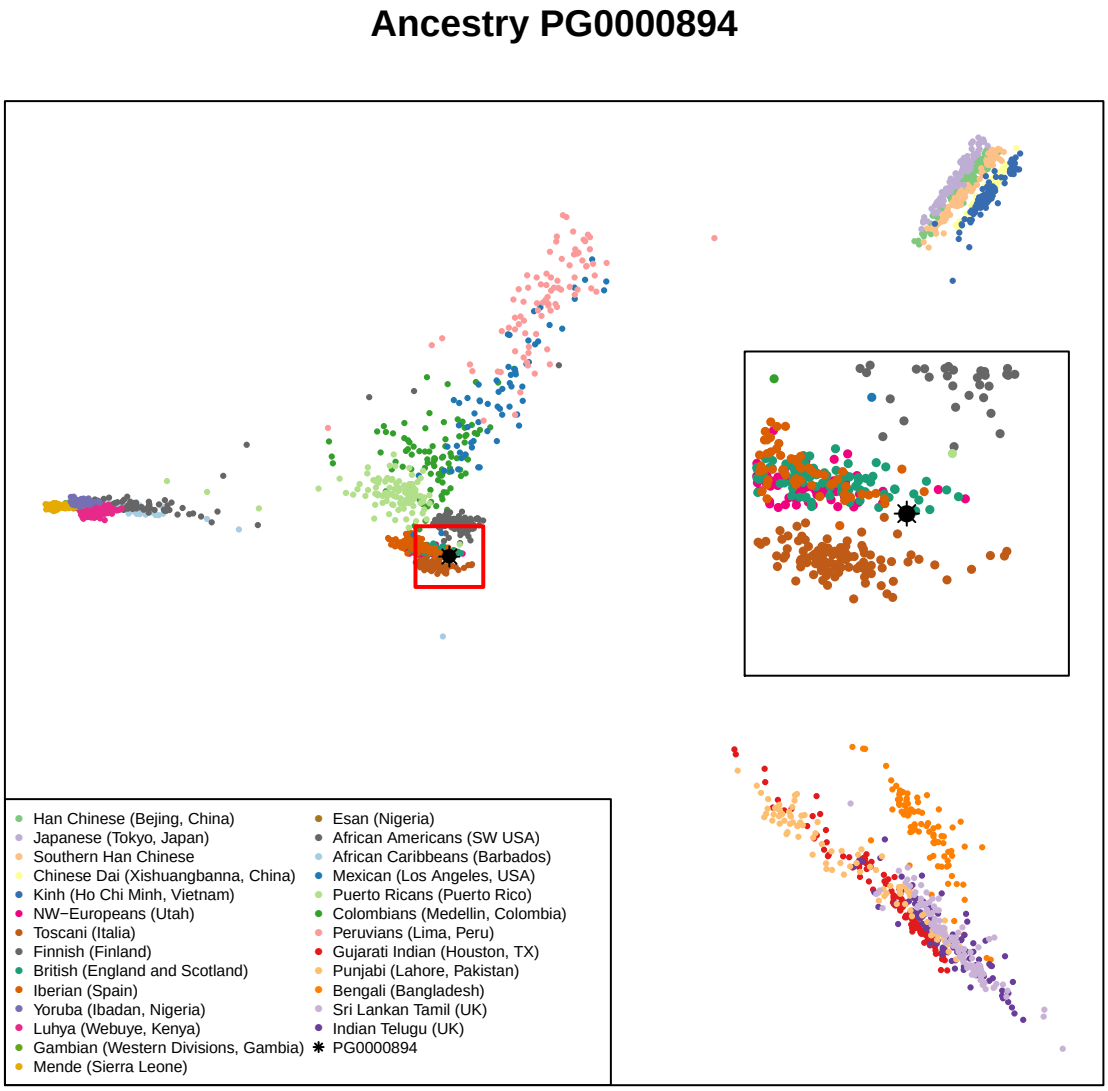

Figure 5: Ancestry Principal Component Analysis

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.

- Possibly Beneficial Traits

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
2.5	rs2943634	(A;A)	Lower risk of ischemic stroke		Link	
2.5	rs3764261	(T;T)	Associated with higher HDL cholesterol. HDL		Link	Link
2.1	rs3775291	(A;G)	0.71x decreased risk for dry age related macula...	Link	Link	Link
2.1	rs738409	(G;G)	Most common genotype; slightly less damage from...	Link	Link	
2	rs1026732	(A;A)	<0.70x risk for restless legs		Link	
2	rs10503669	(A;C)	Associated with higher HDL cholesterol		Link	
2	rs10504861	(A;A)	Reduced risk of migraine without aura			
2	rs11045585	(A;A)	24% chance (lower than average) of docetaxel-in...		Link	
2	rs11635424	(A;A)	<0.70x risk for restless legs		Link	
2	rs12593813	(A;A)	<0.71x risk for restless legs		Link	
2	rs12678919	(A;G)	Associated with higher HDL cholesterol		Link	
2	rs1544410	(G;G)	Decreased risk of low bone mineral density diso...		Link	
2	rs17070145	(C;T)	Increased memory performance			Link
2	rs1799884	(G;G)	Mothers have typical Birth-Weight babies. Sligh...			
2	rs1864163	(G;G)	Associated with higher HDL cholesterol		Link	
2	rs2542052	(C;C)	Better odds of living to 100			
2	rs3738579	(C;T)	0.5x decreased risk for cervical cancer: HNSCC:...			
2	rs3819331	(T;T)	Lower risk of autism	Link		
2	rs4143094	(G;G)	No increased risk of colorectal cancer correlat...			
2	rs4149268	(A;G)	Associated with higher HDL cholesterol		Link	
2	rs4307059	(C;C)	Reduced Autism risk		Link	
2	rs6505162	(A;C)	0.58x decreased risk for esophageal cancer	Link		
2	rs6511720	(G;T)	Slightly lower odds of developing CHD.		Link	
2	rs6855911	(A;G)	0.62x decreased risk for gout		Link	
2	rs763110	(T;T)	~0.80x reduced cancer risk			Link
2	rs7776725	(T;T)	Stronger bones		Link	
2	rs801114	(T;T)	0.78x decreased Basal Cell Carcinoma risk.		Link	
2	rs9642880	(G;G)	Slightly lower risk of Bladder Cancer.		Link	

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
2.0	rs3790844	(C;C)	Reduced risk (0.59x) of pancreatic cancer			
1.5	rs1063192	(C;T)	0.71x reduced risk of myocardial infarction			
1.5	rs11136000	(T;T)	0.84x decreased risk for Alzheimer's disease		Link	
1.5	rs4149274	(C;C)	Associated with higher HDL (good) cholesterol.			
1.5	rs4939883	(C;C)	Associated with higher HDL cholesterol		Link	
1.5	rs5888	(C;C)	Higher HDL cholesterol but lower risk for age-r...	Link		
1.5	rs6427528	(A;G)	For rheumatoid arthritis patients: better respo...			
1.5	rs729302	(C;C)	0.89x decreased risk of developing rheumatoid a...			
1.1	rs11172113	(C;T)	0.9x lower risk for migraines			
1.1	rs2293347	(G;G)	Among NSCLC patients: better Gefitinib response...	Link		
1.1	rs4988235	(T;T)	Can digest milk			Link
1.1	rs7568369	(T;T)	0.90x reduced risk of obesity			
1	rs182549	(T;T)	Can digest milk.			Link
1	rs2351299	(G;T)	Possible reduced risk of Autism			
1	rs4939827	(C;C)	0.73x decreased risk for colorectal cancer		Link	
1	rs7850258	(A;G)	Typical odds of developing primary hypothyroidi...			
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	rs10897346	(C;C)	If depressed: 2.6x more likely to not respond t...			
0	rs1126742	(T;T)	Higher hypertension risk	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	rs12593929	(A;A)	Blue eye color more likely			
0	rs16990018	(A;A)	PrP Codon 171 Asn - Non-pathogenic variant	Link		Link
0	rs17244841	(A;A)	More responsive to statin treatment		Link	
0	rs1800562	(G;G)	Not a C282Y hemochromatosis carrier.	Link	Link	Link
0	rs2240203	(A;A)	Blue eye color more likely			
0	rs2306402	(C;C)	1.18x increased risk for LOAD			
0	rs28933385	(G;G)	Prion protein Codon 200 (E) - Non pathogenic va...			Link
0	rs403016	(C;C)	2x risk for lupus		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	rs8028689	(T;T)	Blue eye color if part of blue eye color haplot...			

- Possibly Harmful Traits

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
3.2	rs2981582	(T;T)	1.7x higher risk of ER+ breast cancer		Link	
3	rs1800460	(A;G)	(TPMT*3B) impaired drug metabolism	Link	Link	Link
3	rs4244285	(A;G)	Poorer metabolizer of several popular medicines...	Link	Link	Link
3	rs6920220	(A;G)	1.2x risk Rheumatoid Arthritis		Link	
3	rs7754840	(C;G)	1.3x increased risk for type-2 diabetes		Link	
3.0	rs1142345	(A;G)	TPMT*3C . impaired drug metabolism	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	rs1154155	(G;G)	2.5x increased risk for narcolepsy		Link	
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	rs2254958	(C;C)	1.61x increased risk for Alzheimer's			
2.5	rs339331	(T;T)	Prostate cancer risk			
2.5	rs3780374	(A;G)	Substantially increased odds of developing V617...			
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	
2.4	rs7966230	(G;G)	Slightly lower levels of plasma VWF			
2.2	rs2231137	(G;G)	~1.5-3x increased risk for ischemic stroke	Link	Link	Link
2.2	rs944289	(T;T)	2.6x increased thyroid cancer risk		Link	
2.1	rs1219648	(G;G)	1.64x risk for breast cancer		Link	
2.1	rs1329428	(G;G)	2x increased risk for macular degeneration			
2.1	rs17077540	(A;G)	1.6x major depressive disorder risk			
2.1	rs2231142	(A;C)	1.74x increased gout risk; gefinitib takers 4x ...	Link	Link	Link
2.1	rs241448	(C;C)	2.14x increased risk for Alzheimer's	Link		Link
2.1	rs2420946	(T;T)	1.64x risk for breast cancer			
2.1	rs380390	(C;C)	Increased risk for ARMD		Link	
2.1	rs4402960	(T;T)	1.2x increased risk for type-2 diabetes		Link	Link
2.1	rs4693596	(C;C)	2x odds of myopathy if taking statins			
2.1	rs6457617	(T;T)	5.2x risk of rheumatoid arthritis		Link	
2.1	rs7837688	(G;G)	1.7x increased risk for prostate cancer			
2	rs10086908	(C;T)	1.7x increased risk for prostate cancer			
2	rs10248420	(A;A)	7x less likely to respond to certain antidepres...		Link	
2	rs1050631	(C;T)	Mean Survival Time of 25 months for esophageal ...	Link		
2	rs1064395	(A;G)	Having any copies of A at this SNP heightens yo...			
2	rs10811661	(C;T)	1.2x increased risk for type-2 diabetes		Link	
2	rs10871777	(A;G)	Adults likely to be 0.22 BMI units higher			
2	rs10889677	(C;C)	1x increased risk for certain autoimmune diseas...		Link	
2	rs10984447	(A;G)	1.17x increased risk for multiple sclerosis		Link	
2	rs11229030	(C;C)	Higher odds of Crohn's disease			
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	rs13254738	(A;C)	1.18x prostate cancer risk		Link	
2	rs1333048	(A;C)	1.3x increased coronary artery disease risk			
2	rs1360780	(C;T)	1.3x increased risk for depression		Link	
2	rs1585215	(A;G)	2x increased risk for Hodgkin lymphoma			
2	rs17228212	(C;T)	1.26x increased risk for heart disease		Link	
2	rs1734791	(A;A)	1.4x increased risk for lupus			
2	rs17782313	(C;T)	Adults likely to be 0.22 BMI units higher		Link	
2	rs1800896	(A;A)	1.8x increased prostate cancer risk			
2	rs2070600	(A;G)	1.5x increased risk for gastric cancer	Link	Link	
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	

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	rs2274223	(A;G)	1.5x increased risk for stomach and esophageal ...	Link	Link	
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	rs25487	(A;G)	2x higher risk for skin cancer; possibly other ...	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	rs3129934	(C;T)	Increased risk of Multiple Sclerosis.		Link	
2	rs3184504	(C;T)	Increased risk for celiac disease	Link	Link	
2	rs358806	(C;C)	1.78x increased risk of developing Type-2 diabe...		Link	
2	rs3802842	(C;C)	>1.17x increased risk of colorectal cancer		Link	
2	rs4129148	(C;G)	3x risk of schizophrenia.		Link	
2	rs4148739	(A;A)	7x less likely to respond to certain antidepres...		Link	
2	rs4444903	(A;G)	3.5x risk of hep-cancer in cirrhosis patients; ...			
2	rs4626664	(A;A)	>1.44x increased risk of developing restless le...		Link	
2	rs4633	(C;T)	Higher risk for endometrial cancer	Link	Link	
2	rs493258	(G;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;A)	Increased risk in men for biliary conditions			
2	rs5759167	(T;T)	Higher prostate cancer risk		Link	
2	rs629242	(C;T)	Somewhat higher risk for prostate cancer			
2	rs6441286	(G;T)	1.54x chance of developing primary biliary cirr...		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	rs6807362	(C;C)	Increased autism risk	Link	Link	
2	rs6897932	(C;C)	1.5x increased risk for multiple sclerosis	Link	Link	Link
2	rs7774434	(C;C)	Increased risk of developing primary biliary ci...			
2	rs7807268	(C;G)	1.3x risk for Crohn's disease		Link	
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	rs9652490	(A;A)	~2x increased risk for Parkinson's disease: and...		Link	
2	rs965513	(A;G)	1.7x increased thyroid cancer risk		Link	
2.0	rs1434536	(A;A)	1.94x increased breast cancer risk			
2.0	rs2305795	(A;A)	1.64x higher risk of narcolepsy compared to (G;...			Link
2.0	rs4911414	(G;T)	2-4x higher risk of sun sensitivity if part of ...		Link	
1.9	rs721048	(A;A)	Slightly increased prostate cancer risk		Link	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
1.6	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	
1.6	rs3775948	(C;G)	Slightly higher risk for gout			
1.5	rs10492519	(A;G)	Slightly increased risk of developing prostate ...			
1.5	rs10757272	(C;T)	1.30x increased risk for Coronary artery diseas...			
1.5	rs10883365	(A;G)	1.2x increased risk for developing Crohn's dise...		Link	
1.5	rs11171739	(C;T)	1.34x risk of developing Type-1 diabetes		Link	
1.5	rs1169300	(A;G)	~1.5x increased lung cancer risk			
1.5	rs12037606	(A;G)	1.22x risk of developing Crohn's disease			
1.5	rs12431733	(C;T)	Slightly increased risk of developing Parkinson...		Link	
1.5	rs12469063	(A;G)	Slightly increased risk of developing restless ...			

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
1.5	rs13149290	(C;T)	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	rs16944	(A;G)	Minorly increased risk of mental illness and os...		Link	
1.5	rs17221417	(C;G)	1.3x higher risk for Crohn's disease		Link	
1.5	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	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;G)	1.4x increased risk of SLE			
1.5	rs2282679	(C;C)	Lower vitamin D levels			
1.5	rs2286812	(C;T)	~2x higher risk for Fuchs' dystrophy: a corneal...			
1.5	rs2464196	(C;T)	~1.5x increased lung cancer risk	Link	Link	Link
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	(T;T)	Slightly increased risk for pregnancy-induced h...			
1.5	rs3087243	(A;G)	Increased risk for auto-immune diseases		Link	
1.5	rs309375	(T;T)	Larger mosquito bites			
1.5	rs3212227	(A;A)	1.43x increased risk of developing psoriasis an...			
1.5	rs393152	(A;A)	Increased risk of both PD and AD	Link	Link	
1.5	rs401681	(C;C)	~1.2x increased risk for several types of cance...		Link	
1.5	rs4585	(T;T)	Slightly poorer (0.75x) response to metformin i...			
1.5	rs464049	(C;T)	Increased risk of schizophrenia in limited stud...			
1.5	rs4656461	(A;G)	1.5x increased risk for open angle glaucoma			
1.5	rs4785763	(A;C)	1.5x higher risk for melanoma		Link	
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	rs6435862	(G;T)	1.7x higher risk of aggressive neuroblastoma		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	rs6908425	(C;T)	1.63x increased risk of developing Crohn's dise...		Link	
1.5	rs700651	(A;G)	~1.18x increased risk of aneurysm		Link	
1.5	rs7341475	(G;G)	1.58x increased schizophrenia risk for women		Link	
1.5	rs7536563	(A;G)	1.12x risk of multiple sclerosis		Link	
1.5	rs807701	(C;T)	Slightly increased dyslexia risk			
1.5	rs9303277	(C;T)	1.46x Slightly increased risk of developing pri...			
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	rs3131296	(G;G)	1.4x increased risk for schizophrenia		Link	
1.4	rs4959039	(A;G)	1.4x higher risk for multiple sclerosis			
1.4	rs498872	(T;T)	1.4x higher risk for glioma development		Link	
1.4	rs6010620	(G;G)	1.4x higher risk for glioma development; but th...		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	rs2059693	(C;T)	1.3x increased risk for testicular cancer			
1.3	rs4295627	(G;T)	1.3x higher risk for glioma development		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	rs11037909	(T;T)	1.47x type II diabetes risk	Link		
1.2	rs1344706	(T;T)	1.2x increased risk for schizophrenia		Link	
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	rs2814707	(A;G)	1.2x increased risk for ALS		Link	
1.2	rs3740878	(A;A)	1.46x type II diabetes risk; common	Link		

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
1.2	rs3849942	(A;G)	1.2x increased risk for ALS		Link	
1.2	rs4795067	(A;G)	Slight increase in risk for psoriatic arthritis...			
1.2	rs4977756	(A;G)	1.2x higher risk for glioma development		Link	
1.2	rs8050136	(A;C)	1.2x increased risk for T2D in some populations...		Link	
1.2	rs9858542	(A;G)	1.1x risk Crohn's Disease	Link	Link	
1.1	rs11110912	(C;G)	1.3x high blood pressure risk			
1.1	rs1800450	(A;G)	Mannose binding deficiency but of low clinical ...	Link	Link	Link
1.1	rs249954	(C;T)	Slight if any increased risk of Breast Cancer			Link
1.1	rs2651899	(A;G)	1.1x higher risk for migraines			
1.1	rs34516635	(G;G)	Less longevity for Ashkenazi Jewish women.	Link		Link
1.1	rs4324715	(C;T)	1.5x increased testicular cancer risk for men			
1.1	rs4977574	(A;G)	Some studies - but not others - report a slight...		Link	
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	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
1.1	rs889312	(A;C)	Very slightly higher risk for breast cancer		Link	
1.1	rs925391	(C;C)	More likely to go bald; common			
1	rs1143674	(A;G)	1.3x increased autism risk	Link		
1	rs12718541	(A;A)	Nicotine dependence			
1	rs2273697	(A;G)	Adverse reaction more likely to carbamazepine i...	Link	Link	
1	rs2546890	(A;A)	Higher risk of multiple sclerosis			
1	rs3194051	(A;G)	1.12x risk of type-1 diabetes	Link	Link	Link
1	rs6932590	(C;T)	1.1x increased risk for schizophrenia		Link	
1	rs761100	(G;G)	Higher risk for dyslexia			
1.0	rs11246226	(A;A)	Increased risk of schizophrenia in limited stud...		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	rs1333040	(C;T)	1.24x increased myocardial infarction risk: 1.2...		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	rs4712653	(T;T)	2x increased risk for neuroblastoma			
0	rs6314	(C;C)	Higher risk for RA	Link	Link	
0	rs7787082	(G;G)	7x less likely to respond to certain antidepres...		Link	
0	rs9298506	(A;A)	Higher Risk Aneurysm		Link	

- Genosets (Multi-variant Phenotypes)

Magnitude	Identifier	Summary
4	gs144	Male
3.1	gs122	7x risk of baldness
3.1	gs191	Problem metabolizing NSAIDs
3.1	gs237	Blue eyes are much more likely
3	gs127	Intermediate warfarin metabolizer
3	gs273	Lowest risk (13% of white women) of Atrial Fibr...
2.5	gs155	CYP3A5 non-expressor
2.5	gs157	More stimulated by coffee
2.5	gs161	CYP2C9 Intermediate Metabolizers
2.5	gs259	Homozygous for eye color haplotype #3
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
2.3	gs255	Homozygous eye color haplotype #1
2	gs101	Probably able to digest milk
2	gs104	Restless legs syndrome risk
2	gs129	Unable to classify your ABO blood type
2	gs140	NAT2 slow metabolizer
2	gs154	NAT2 Slow metabolizer
2	gs213	Haplogroup R (Y-DNA)
2	gs246	APOE3/APOE3
1.5	gs1105	Mitochondrial Haplogroup U
1.5	gs186	HLA-B*5801 heterozygosity is possible: unfortun...
1.5	gs247	Parkinson's Disease Risk
1.5	gs299	Too common. Prone to Gout and possibly HLA-5801...
1	gs182	CYP2D6*39
0.1	gs233	Normal pain sensitivity

4 Report Metadata

Resource	Version	Website
Genome	GRCh37	Link
BWA	0.7.12	Link
SAMtools	1.2	Link
GATK	3.4-46	Link
PLINK	v1.90b3.35	Link
VEP	84	Link
SNPedia	8-Apr-2016	Link
ExAC	v0.3.1	Link
GetEvidence	8-Apr-2016	Link
ClinVar	4-Apr-2016	Link

Table 5: Analysis Pipeline Versions

Report generated on July 20, 2016 (using report generator version 16-174).