PGP-UK Genomics Report for uk20E91D

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.

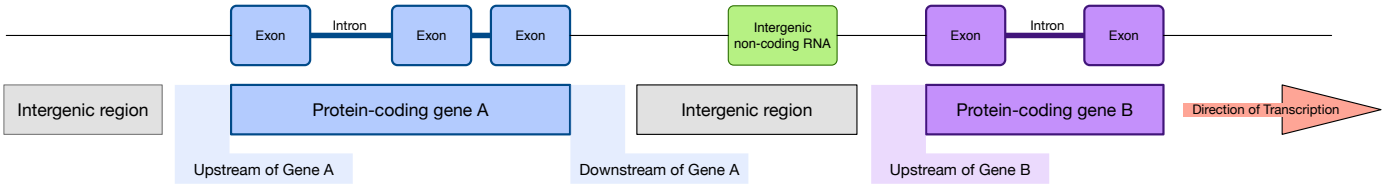

Figure 1: Diagram of gene structure indicating locations of potential variants

Feature	Count
Lines of input read	4721375
Variants filtered out	3732052
Novel / existing variants	0 (0.0) / 989323 (100.0)
Overlapped genes	52173
Overlapped transcripts	59688
Overlapped regulatory features	48538

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.

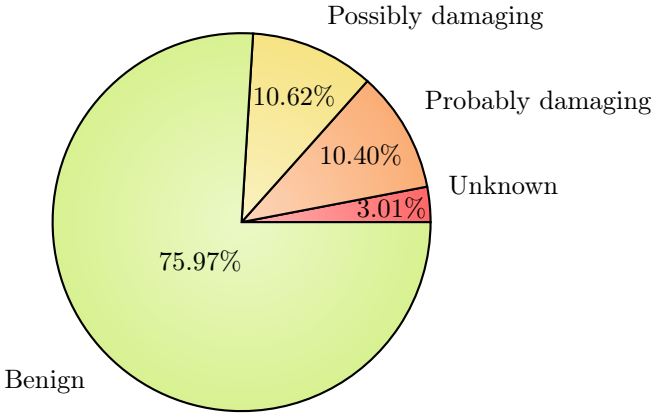

Figure 2: PolyPhen Summary

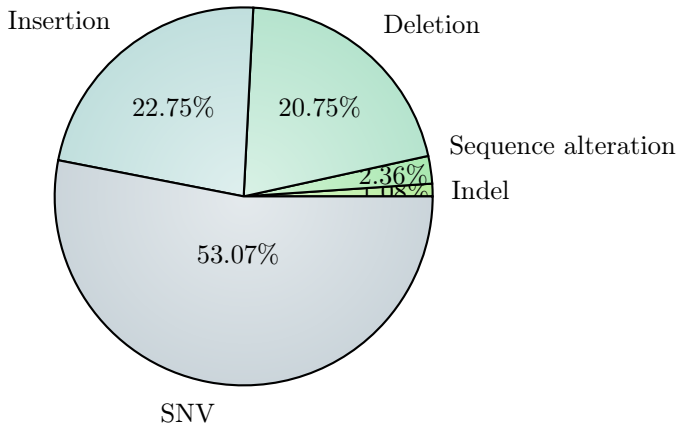

Figure 3: Variant Class

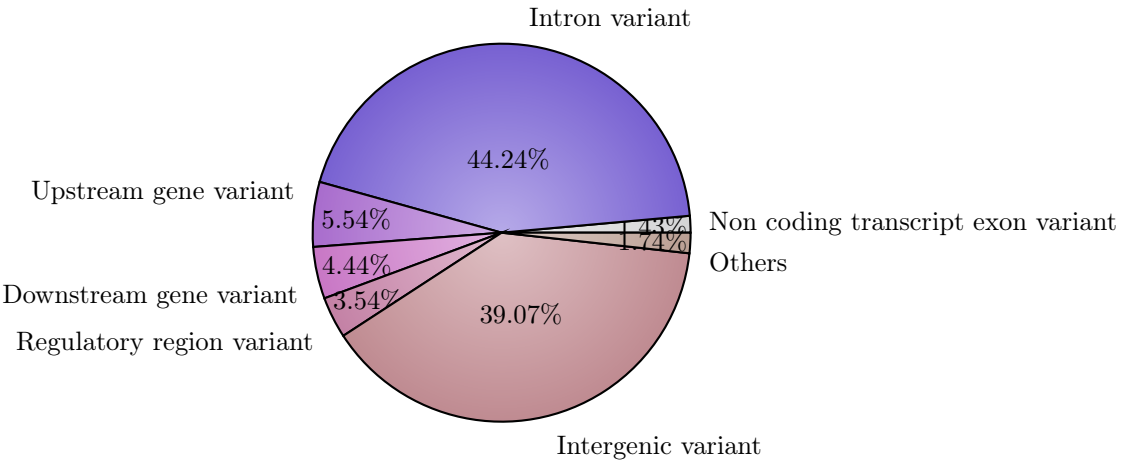

Figure 4: Consequence type

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.

Please note that this analysis is limited by the populations available in the 1000 genomes project (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).

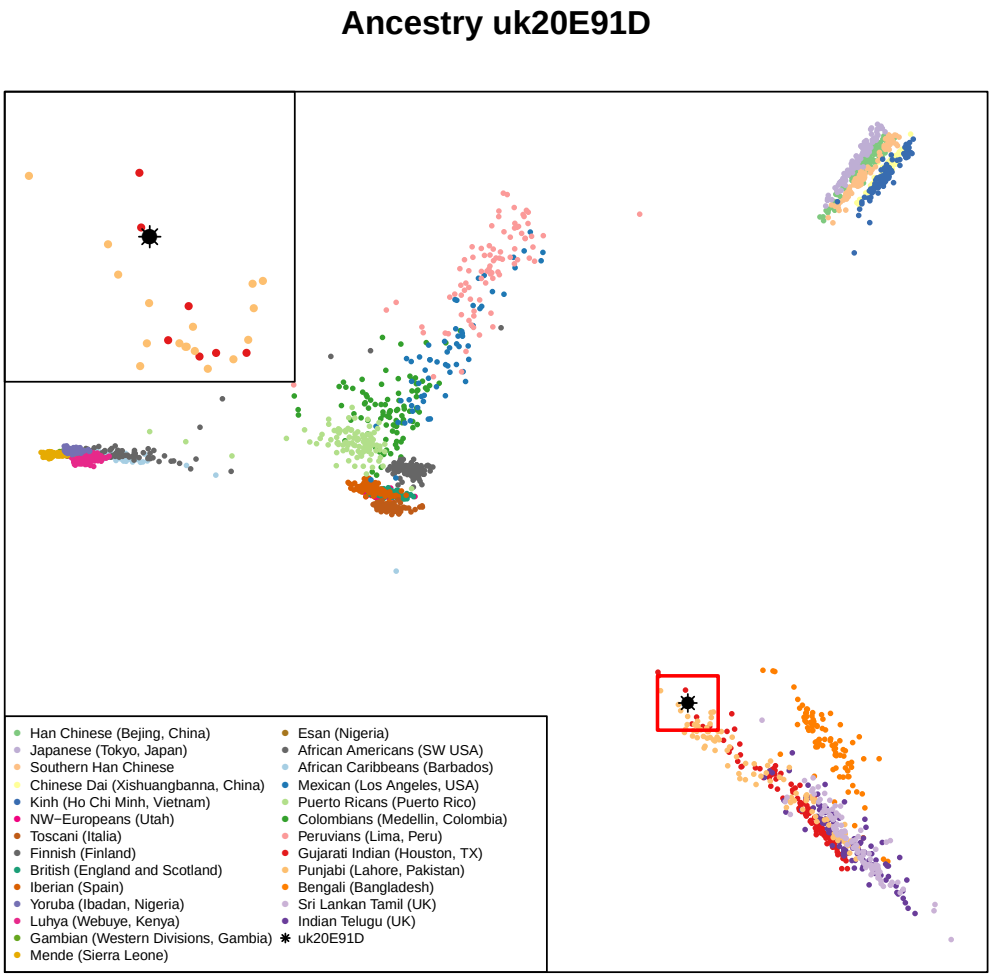

Figure 5: Ancestry Principal Component Analysis

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	GnomAD	GetEvidence	ClinVar
3	rs8177374	(C;T)	Resistance to several diseases	Link	Link	Link
2.5	rs2943634	(A;A)	Lower risk of ischemic stroke	Link	Link	
2.2	rs2511989	(A;A)	0.44x decreased age-related macular degeneratio...	Link	Link	
2.1	rs3775291	(A;G)	0.71x decreased risk for dry age related macula...	Link	Link	Link
2	rs1012053	(A;C)	0.625x reduced risk of Bipolar Disorder.	Link	Link	
2	rs10468017	(C;T)	Associated with higher HDL cholesterol	Link	Link	
2	rs11045585	(A;A)	24% chance (lower than average) of docetaxel-in...	Link	Link	
2	rs1128535	(G;G)	Reduced risk (0.77x) for Crohn's disease	Link		
2	rs1544410	(G;G)	Decreased risk of low bone mineral density diso...	Link	Link	
2	rs1799884	(G;G)	Mothers have typical Birth-Weight babies. Sligh...	Link		
2	rs1864163	(A;G)	Associated with higher HDL cholesterol	Link	Link	
2	rs2060793	(A;A)	Lower serum levels of vitamin D	Link		
2	rs2241423	(A;G)	0.79 decreased risk for obesity	Link		
2	rs2243250	(C;T)	0.6x decreased risk for myocardial infarction i...	Link		
2	rs2542052	(C;C)	Better odds of living to 100	Link		
2	rs25487	(A;A)	0.7x lower risk for skin cancer	Link	Link	Link
2	rs266729	(G;G)	0.73x decreased risk for colorectal cancer	Link	Link	
2	rs3178250	(C;C)	Lower otosclerosis risk	Link		
2	rs3750817	(C;T)	0.78x reduced risk for breast cancer	Link		
2	rs3764261	(G;T)	Associated with higher HDL cholesterol	Link	Link	Link
2	rs3819331	(T;T)	Lower risk of autism	Link		Link
2	rs4149268	(A;G)	Associated with higher HDL cholesterol	Link	Link	
2	rs7216389	(C;C)	0.69x lower risk of Childhood Asthma.	Link	Link	
2	rs801114	(T;T)	0.78x decreased Basal Cell Carcinoma risk.	Link	Link	
2	rs9642880	(G;G)	Slightly lower risk of Bladder Cancer.	Link	Link	
1.8	rs1746048	(C;T)	0.94 decreased risk for coronary heart disease	Link	Link	
1.8	rs187238	(C;G)	Hypertension not a risk factor for sudden cardi...	Link		

Mag.	Identifier	Genotype	Summary	GnomAD	GetEvidence	ClinVar
1.8	rs3814113	(C;T)	0.8x decreased risk for ovarian cancer	Link	Link	
1.8	rs4714156	(C;C)	<0.61x risk for restless legs	Link		
1.8	rs9402571	(G;G)	0.85x decreased risk for type-2 diabetes	Link		
1.5	rs1026732	(A;G)	0.70x risk for restless legs	Link	Link	
1.5	rs11136000	(C;T)	0.84x decreased risk for Alzheimer's disease	Link	Link	
1.5	rs11635424	(A;G)	0.70x risk for restless legs	Link	Link	
1.5	rs1165205	(A;A)	0.85x decreased gout risk	Link	Link	
1.5	rs12593813	(A;G)	0.71x risk for restless legs	Link	Link	
1.5	rs2229169	(C;C)	1.5x decreased risk of heart attack and stroke ...	Link		
1.5	rs3784709	(C;T)	0.71x risk of developing restless legs syndrome...	Link	Link	
1.5	rs4149274	(C;C)	Associated with higher HDL (good) cholesterol.	Link		
1.5	rs4489954	(G;T)	0.69x risk risk of developing restless legs syn...	Link	Link	
1.5	rs4939883	(C;C)	Associated with higher HDL cholesterol	Link	Link	
1.5	rs5968255	(C;C)	Slower AIDS progression (8 years)	Link		
1.5	rs610932	(A;A)	A allele associated with reduced risk of Alzhei...	Link		
1.5	rs6427528	(A;G)	For rheumatoid arthritis patients: better respo...	Link		
1.4	rs10513789	(G;T)	0.8x decreased risk of Parkinson's disease	Link		
1.4	rs6495446	(C;T)	0.8x reduced risk for chronic kidney disease	Link		
1.3	rs2361502	(C;C)	Possible higher levels of serum bilirubin and l...	Link		
1.2	rs4320932	(A;G)	0.87x decreased risk for ovarian cancer	Link		
1.2	rs6048	(G;G)	Slightly lower risk (10-20%) of deep vein throm...	Link	Link	Link
1.2	rs9306160	(C;T)	0.75x (reduced) risk for metastasis in LN-/ER+ ...	Link	Link	
1.1	rs10166942	(C;T)	0.85x lower risk for migraines	Link		
1.1	rs11172113	(C;T)	0.9x lower risk for migraines	Link		
1.1	rs7568369	(G;T)	0.90x reduced risk of obesity	Link		
1	rs10784502	(C;T)	Slightly higher intracranial volume	Link		
1	rs182549	(C;T)	Can digest milk.	Link		Link
1	rs2351299	(G;T)	Possible reduced risk of Autism	Link		
1	rs2494732	(T;T)	Lower odds of psychosis	Link	Link	
1	rs2546890	(G;G)	Lower risk of multiple sclerosis	Link		
1	rs4939827	(C;C)	0.73x decreased risk for colorectal cancer	Link	Link	Link
1	rs800292	(C;T)	1% decreased risk of macular degeneration	Link	Link	Link
1.0	rs11246226	(C;C)	Decreased risk of schizophrenia in limited stud...	Link	Link	
0.1	rs891512	(G;G)	Lower blood pressure than those with an A allel...	Link		Link

3.2 Possibly Harmful Traits

Mag.	Identifier	Genotype	Summary	GnomAD	GetEvidence	ClinVar
4	rs2272783	(C;C)	Likely erythropoietic protoporphyria symptoms	Link		Link
3.1	rs10830963	(G;G)	Increased type-2 diabetes risk; higher gestatio...	Link	Link	
3.1	rs1421085	(C;C)	~1.7x increased obesity risk	Link	Link	Link
3	rs1121980	(T;T)	Moderate increase (2.76x) in risk for obesity	Link	Link	
3	rs1983132	(C;T)	2 - 3x higher prostate cancer risk if routinely...	Link		
3	rs2237717	(T;T)	Reduced abilities related to neurocognition and...	Link		
3	rs2981582	(C;T)	1.3x higher risk of ER+ breast cancer	Link	Link	
3	rs3738579	(T;T)	1.5x - 2x increased risk for cervical cancer: H...	Link		
3	rs4244285	(A;G)	Poorer metabolizer of several popular medicines...	Link	Link	Link
3	rs6920220	(A;G)	1.2x risk Rheumatoid Arthritis	Link	Link	
3	rs7754840	(C;G)	1.3x increased risk for type-2 diabetes	Link	Link	
2.9	rs16901979	(A;C)	1.5x increased risk for prostate cancer	Link	Link	
2.5	rs12803066	(A;G)	Increased risk of myopia	Link		
2.5	rs13266634	(C;T)	Increased risk for type-2 diabetes	Link	Link	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	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	Link
2.5	rs795484	(A;A)	Even more increased morphine dose requirement a...	Link		
2.3	rs7966230	(C;G)	Slightly lower levels of plasma VWF	Link		
2.1	rs10811661	(T;T)	1.2x increased risk for type-2 diabetes	Link	Link	
2.1	rs17070145	(C;C)	Reduced memory abilities	Link		Link
2.1	rs4430796	(A;A)	1.38x increased risk for prostate cancer	Link	Link	
2.1	rs646776	(A;A)	1.2x risk of coronary artery disease	Link	Link	
2.1	rs944289	(C;T)	1.3x increased thyroid cancer risk	Link	Link	
2	rs1024611	(C;T)	Increased risk of exercise induced ischemia	Link		Link
2	rs1045642	(C;T)	Slower metaboliser for some drugs	Link	Link	Link
2	rs1050152	(C;T)	2.1x increased risk of Crohn's disease	Link	Link	Link
2	rs1050631	(C;T)	Mean Survival Time of 25 months for esophageal ...	Link		
2	rs10871777	(A;G)	Adults likely to be 0.22 BMI units higher	Link		
2	rs10984447	(A;G)	1.17x increased risk for multiple sclerosis	Link	Link	
2	rs11123857	(A;G)	1.44-fold increased risk of bipolar disorder or...	Link		
2	rs11190870	(C;T)	Possibly increased risk of scoliosis	Link		
2	rs1143699	(C;C)	In men: 2.19x risk of type 2 diabetes	Link		
2	rs1160312	(A;G)	1.6x increased risk of Male Pattern Baldness.	Link	Link	
2	rs1219648	(A;G)	1.20x risk for breast cancer	Link	Link	
2	rs12567232	(A;A)	Increased risk for Crohn's Disease	Link	Link	
2	rs12696304	(C;G)	Prone to aging faster: at least in European pop...	Link		
2	rs1333048	(A;C)	1.3x increased coronary artery disease risk	Link		
2	rs1360780	(C;T)	1.3x increased risk for depression	Link	Link	Link
2	rs1537415	(G;G)	2x increased risk for periodontitis	Link	Link	
2	rs17228212	(C;C)	>1.26x increased risk for heart disease	Link	Link	
2	rs1734791	(A;A)	1.4x increased risk for lupus	Link		
2	rs17576	(A;G)	Higher risk for MI and lung cancer: and COPD in...	Link	Link	Link
2	rs17696736	(A;G)	1.34x risk of type-1 diabetes	Link	Link	
2	rs17782313	(C;T)	Adults likely to be 0.22 BMI units higher	Link	Link	Link
2	rs1800896	(A;A)	1.8x increased prostate cancer risk	Link		
2	rs2073963	(G;T)	Increased risk of baldness	Link		
2	rs2201841	(C;C)	1.5x increased risk for Crohn's disease	Link	Link	
2	rs2230199	(C;G)	1.6x+ risk of ARMD	Link	Link	Link
2	rs2230201	(G;G)	>1.4x risk of lupus	Link		Link
2	rs2305480	(T;T)	If 4 years old or younger: ~3x increased asthma...	Link	Link	
2	rs2306402	(C;T)	1.18x increased risk for late-onset Alzheimer's...	Link		

Mag.	Identifier	Genotype	Summary	GnomAD	GetEvidence	ClinVar
2	rs2383206	(A;G)	1.4x increased risk for heart disease	Link		
2	rs2383207	(A;G)	Increased risk for heart disease	Link		
2	rs2420946	(C;T)	1.20x risk for breast cancer	Link		
2	rs2697962	(A;A)	Increased risk of developing Parkinson's Diseas...	Link		
2	rs3025039	(T;T)	2.6x increased risk for ARMD in a Taiwanese pop...	Link		
2	rs3212227	(A;C)	Significantly increased risk of developing cerv...	Link		Link
2	rs326	(A;A)	Lower HDL cholesterol	Link	Link	Link
2	rs351855	(C;T)	1.2x increased risk for prostate cancer	Link	Link	Link
2	rs358806	(C;C)	1.78x increased risk of developing Type-2 diabe...	Link	Link	
2	rs3775948	(G;G)	Slightly higher risk for gout	Link		
2	rs4129148	(C;G)	3x risk of schizophrenia.	Link	Link	
2	rs4402960	(G;T)	1.2x increased risk for type-2 diabetes: ~1x ri...	Link	Link	Link
2	rs4420638	(A;G)	~3x increased Alzheimer's risk; 1.4x increased ...	Link	Link	Link
2	rs4633	(T;T)	Higher risk for endometrial cancer	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	Link		
2	rs638405	(G;G)	2x increased ALZ risk in ApoE4 carriers	Link		
2	rs6435862	(G;G)	2.8x higher risk of aggressive neuroblastoma	Link	Link	
2	rs6498169	(A;A)	>1.14x risk of multiple sclerosis	Link	Link	
2	rs663048	(G;T)	Significantly increased risk of developing lung...	Link	Link	
2	rs6807362	(C;C)	Increased autism risk	Link	Link	
2	rs6896702	(T;T)	Increased risk of developing Parkinson's Diseas...	Link		
2	rs6897932	(C;C)	1.08x increased risk for multiple sclerosis	Link	Link	Link
2	rs6997709	(G;G)	1.5x higher risk for hypertension	Link		
2	rs699	(C;C)	Increased risk of hypertension	Link	Link	Link
2	rs738409	(C;G)	Increased liver fat: odds of alcoholic liver di...	Link	Link	Link
2	rs7442295	(A;A)	~4x higher risk for hyperuracemia	Link	Link	Link
2	rs744373	(C;T)	1.17x risk of Alzheimer's	Link		
2	rs763361	(T;T)	Increased risk for multiple autoimmune diseases...	Link	Link	
2	rs7794745	(A;T)	Slightly increased risk for autism	Link	Link	Link
2	rs7807268	(C;C)	1.4x risk for Crohn's disease	Link	Link	
2	rs828907	(G;T)	Slightly increased risk of bladder cancer and 2...	Link		
2	rs854560	(A;A)	Higher risk for heart disease: diabetic retinop...	Link	Link	Link
2	rs9303277	(T;T)	1.46x Increased risk of developing primary bili...	Link		
2	rs9525638	(T;T)	Weaker bones	Link		
2	rs9652490	(A;A)	~2x increased risk for Parkinson's disease: and...	Link	Link	
2.0	rs1434536	(A;A)	1.94x increased breast cancer risk	Link		Link
2.0	rs2305795	(A;A)	1.64x higher risk of narcolepsy compared to (G;...	Link		Link
1.9	rs7923837	(A;G)	1.6x risk for T2D	Link		
1.8	rs1136287	(C;T)	1.5x increased risk of wet ARMD in a Taiwanese ...	Link	Link	Link
1.8	rs2278206	(T;T)	1.16x increased risk for asthma	Link	Link	
1.8	rs4807015	(C;C)	>1.74x risk of type 2 diabetes	Link		
1.7	rs10181656	(C;G)	1.7x increased SLE risk	Link		
1.7	rs2024513	(A;A)	1.7x higher risk for schizophrenia (among Han C...	Link		
1.7	rs8055236	(G;T)	1.9x risk for heart disease	Link	Link	
1.6	rs2059693	(T;T)	1.6x increased risk for testicular cancer	Link		
1.6	rs33980500	(C;T)	1.6x increase in risk for psoriatic arthritis	Link	Link	Link
1.6	rs3764880	(A;A)	1.2 - 1.8x increased tuberculosis risk	Link	Link	
1.6	rs4712653	(C;C)	Slightly (~1.6x) increased risk for neuroblasto...	Link		
1.5	rs10464059	(A;G)	Slightly increased risk of developing Parkinson...	Link		
1.5	rs10492519	(A;G)	Slightly increased risk of developing prostate ...	Link		
1.5	rs10757272	(C;T)	1.30x increased risk for Coronary artery diseas...	Link		
1.5	rs10859871	(A;C)	Slight (~1.2x) increase in endometriosis risk	Link		
1.5	rs10889677	(A;A)	1.5x increased risk for certain autoimmune dise...	Link	Link	
1.5	rs1154155	(G;T)	1.94x increased risk for narcolepsy	Link	Link	

Mag.	Identifier	Genotype	Summary	GnomAD	GetEvidence	ClinVar
1.5	rs1169300	(A;G)	~1.5x increased lung cancer risk	Link		
1.5	rs1223271	(A;G)	Slightly increased risk of developing Parkinson...	Link	Link	
1.5	rs12431733	(C;T)	Slightly increased risk of developing Parkinson...	Link	Link	
1.5	rs12498742	(A;A)	1.25 increased risk for gout	Link		
1.5	rs13149290	(C;T)	Slightly increased risk of developing prostate ...	Link		
1.5	rs13181	(G;T)	1.12x increased risk for cutaneous melanoma	Link	Link	Link
1.5	rs13376333	(C;T)	1.5x higher risk of atrial fibrillation	Link	Link	
1.5	rs140701	(A;A)	Increased risk for anxiety disorders	Link		
1.5	rs16944	(A;G)	Minorly increased risk of mental illness and os...	Link	Link	
1.5	rs1801020	(C;T)	1.31x increased risk of heart disease	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	Link		
1.5	rs1975197	(C;T)	1.3x increased risk of developing restless legs...	Link	Link	
1.5	rs2007153	(G;G)	Increased risk of schizophrenia in limited stud...	Link		
1.5	rs2076295	(G;G)	Slightly increased risk for pulmonary fibrosis ...	Link		
1.5	rs2240340	(A;G)	Slightly increased (1.5x) risk for RA	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	Link		
1.5	rs2280714	(A;A)	1.4x increased risk of SLE	Link		
1.5	rs2286812	(C;T)	~2x higher risk for Fuchs' dystrophy: a corneal...	Link		
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	Link	
1.5	rs27388	(A;G)	Slightly increased risk of developing schizophr...	Link		
1.5	rs28694718	(A;G)	2x higher risk for schizophrenia	Link		
1.5	rs2881766	(T;T)	Slightly increased risk for pregnancy-induced h...	Link		
1.5	rs3087243	(A;G)	Increased risk for auto-immune diseases	Link	Link	Link
1.5	rs309375	(T;T)	Larger mosquito bites	Link		
1.5	rs3825776	(A;G)	1.3x increased risk for ALS	Link	Link	
1.5	rs4538475	(A;G)	Slightly increased risk of developing Parkinson...	Link	Link	
1.5	rs4585	(T;T)	Slightly poorer (0.75x) response to metformin i...	Link		Link
1.5	rs464049	(T;T)	Increased risk of schizophrenia in limited stud...	Link		
1.5	rs4785763	(A;A)	2x higher risk for melanoma	Link	Link	
1.5	rs4845618	(G;T)	1.7x increased melanoma risk	Link		
1.5	rs5746059	(A;G)	Slightly higher fat mass	Link		
1.5	rs6601764	(C;T)	1.16x increased risk of developing Crohn's dise...	Link	Link	
1.5	rs6908425	(C;T)	1.63x increased risk of developing Crohn's dise...	Link	Link	
1.5	rs699473	(C;T)	~1.5x increased brain tumor risk	Link		
1.5	rs7341475	(G;G)	1.58x increased schizophrenia risk for women	Link	Link	
1.5	rs7850258	(G;G)	Slightly higher odds of developing primary hypo...	Link		
1.5	rs807701	(C;T)	Slightly increased dyslexia risk	Link		
1.5	rs995030	(G;G)	Non-protective against testicular cancer	Link	Link	
1.4	rs12770228	(A;G)	1.4x increased risk for meningioma	Link		
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	Link
1.4	rs2228314	(C;G)	1.48x risk of osteoarthritis	Link	Link	
1.4	rs3184504	(C;T)	Slightly increased risk for celiac disease	Link	Link	
1.34	rs17465637	(C;C)	1.34x higher risk for myocardial infarction	Link	Link	
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	rs110419	(A;G)	1.3x increased risk for neuroblastoma	Link		
1.3	rs1260326	(C;T)	Slightly higher risk for gout	Link	Link	Link
1.3	rs16847548	(C;T)	1.3x increased risk for sudden cardiac death in...	Link		
1.3	rs2736100	(G;T)	1.3x higher risk for glioma development: 2.1x r...	Link	Link	Link
1.3	rs4295627	(G;T)	1.36x higher risk for glioma development	Link	Link	
1.3	rs4958847	(A;G)	1.3x increased risk for Crohn's disease	Link		

Mag.	Identifier	Genotype	Summary	GnomAD	GetEvidence	ClinVar
1.3	rs501120	(A;G)	1.3x increased risk for heart disease	Link	Link	
1.25	rs748404	(T;T)	Slightly increased risk (1.25) for lung cancer...	Link	Link	
1.2	rs10210302	(C;T)	1.2x increased risk for Crohn's disease	Link	Link	
1.2	rs10865331	(A;G)	1.2x higher risk for ankylosing spondylitis	Link		
1.2	rs2254958	(C;T)	1.24x reported increased risk for Alzheimer's; ...	Link		
1.2	rs2665390	(C;T)	1.2x increased risk for ovarian cancer	Link		
1.2	rs4795067	(A;G)	Slight increase in risk for psoriatic arthritis...	Link		
1.2	rs6010620	(A;G)	1.2x higher risk for glioma development: 1.17x ...	Link	Link	
1.2	rs6897876	(C;C)	Slight increase in testicular cancer risk for m...	Link		
1.2	rs7514229	(G;G)	Associated with early-onset autoimmune thyroid ...	Link		
1.2	rs8050136	(A;C)	1.2x increased risk for T2D in some populations...	Link	Link	
1.2	rs851715	(A;A)	Risk of nonsense-word repetition problems if sp...	Link		
1.2	rs9858542	(A;G)	1.1x risk Crohn's Disease	Link	Link	
1.2	rs9960767	(A;C)	1.2x increased risk for schizophrenia	Link	Link	
1.1	rs10248420	(A;A)	Possibly less likely to remit on certain antide...	Link	Link	
1.1	rs11037909	(C;T)	1.27x type II diabetes risk	Link		
1.1	rs1800450	(A;G)	Carrier of mannose binding deficiency but of lo...	Link	Link	Link
1.1	rs2235040	(G;G)	Possibly lesser chances of remission only for i...	Link	Link	
1.1	rs249954	(C;T)	Potentially increased risk of Breast Cancer	Link		Link
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	rs3740878	(A;G)	1.26x type II diabetes risk	Link		Link
1.1	rs4977574	(A;G)	Some studies - but not others - report a slight...	Link	Link	
1.1	rs688034	(C;T)	1.1x risk higher risk for coronary artery disea...	Link	Link	
1.1	rs7171755	(A;G)	Very slight decrease in cortical thickness and ...	Link		
1.1	rs889312	(C;C)	Very slightly higher risk for breast cancer	Link	Link	
1.07	rs2291834	(C;C)	Very slightly higher risk for myocardial infarc...	Link		
1	rs1010	(A;G)	1.75x risk of MI	Link	Link	
1	rs10761659	(A;G)	1.2x risk of Crohn's disease	Link	Link	
1	rs11206244	(C;T)	Slight risk of decreased thyroid hormone metabo...	Link		
1	rs1417066	(C;T)	Slightly increased risk of osteoarthritis	Link		
1	rs17300539	(G;G)	Increased risk of insulin resistance	Link		
1	rs2282679	(A;C)	Somewhat lower vitamin D levels	Link		
1	rs3194051	(A;A)	>1.1x risk of type-1 diabetes	Link	Link	Link
1	rs3735684	(C;T)	Associated with increased colorectal cancer ris...	Link	Link	
1	rs6932590	(T;T)	1.1x increased risk for schizophrenia	Link	Link	
1	rs6974491	(A;G)	Higher risk of coeliac and/or inflammatory bowe...	Link		
0.1	rs3095870	(A;G)	1.7x increased risk for SLE (lupus)	Link		

3.3 Genosets (Multi-variant Phenotypes)

Magnitude	Identifier	Summary
4	gs144	Male
3.1	gs122	7x risk of male baldness
2.6	gs296	Lower heart attack risk than average
2.5	gs281	Part of the 88% of the population claimed not t...
2.5	gs285	Claimed to lose 2.5x as much weight on a low fa...
2	gs101	Probably able to digest milk
2	gs140	NAT2 slow metabolizer
2	gs154	NAT2 Slow metabolizer
2	gs159	CYP1A2 fast metabolizer
2	gs213	Haplogroup R (Y-DNA)
2	gs249	Parkinson's Disease Risk
2	gs290	You might have two short form 5-HTTLPR.
2	gs313	Normal DPYD activity and thus 5-FU metabolism p...
1.7	gs233	Normal pain sensitivity; APS/APS: LPS/APS: and ...
1.5	gs230	Possible Alzheimer's disease-related haplotype
1.2	gs184	Able to taste bitterness.
1	gs182	CYP2D6*39

4 Report Metadata

Resource	Version	Website
Genome	GRCh37	Link
BWA	0.7.12	Link
SAMtools	1.3	Link
GATK	3.4-46	Link
PLINK	v1.90b3.35	Link
SNPedia	02-May-2019	Link
GnomAD	v2.1.1	Link
GetEvidence	10-May-2019	Link
ClinVar	10-May-2019	Link

Table 5: Analysis Pipeline Versions

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