PGP-UK Genomics Report for ukF3D20F

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.

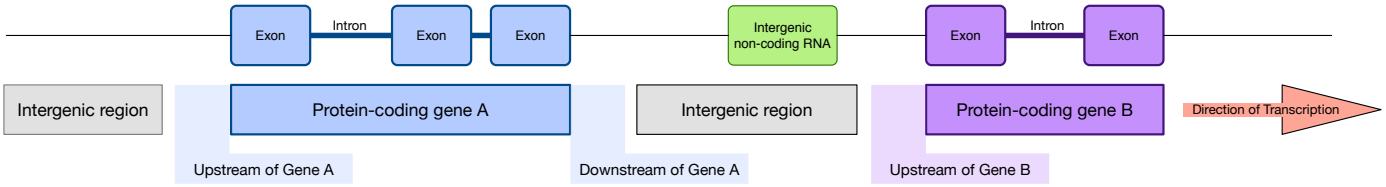

Figure 1: Diagram of gene structure indicating locations of potential variants

Feature	Count
Lines of input read	4671076
Variants filtered out	3697456
Novel / existing variants	0 (0.0) / 973620 (100.0)
Overlapped genes	51892
Overlapped transcripts	59351
Overlapped regulatory features	47304

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.

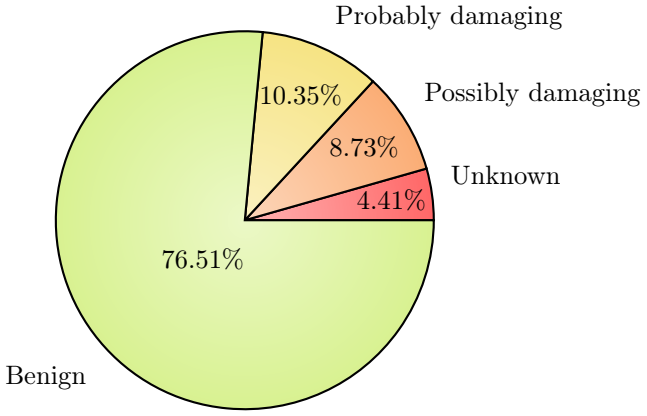

Figure 2: PolyPhen Summary

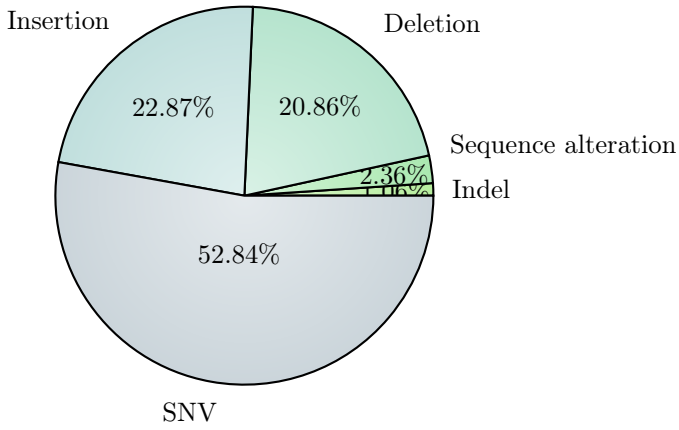

Figure 3: Variant Class

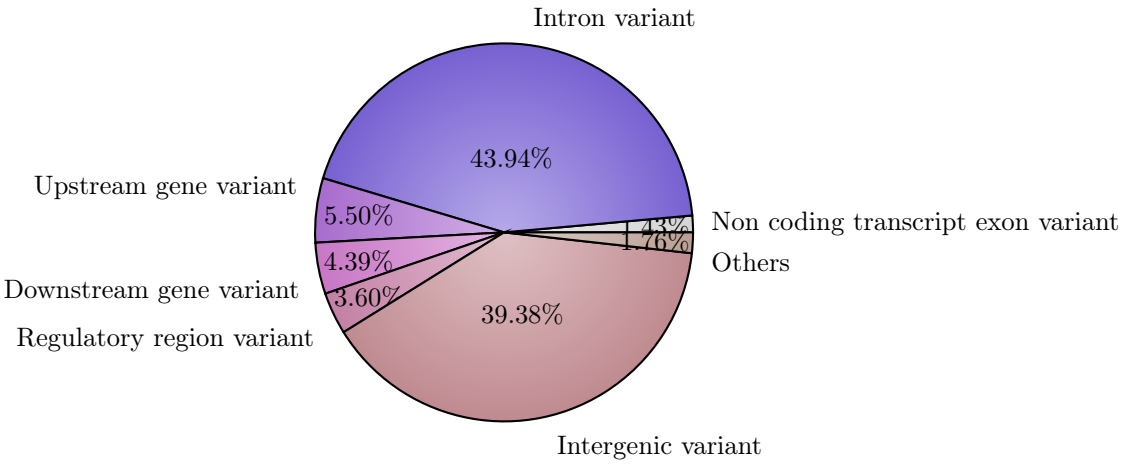

Figure 4: Consequence type

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).

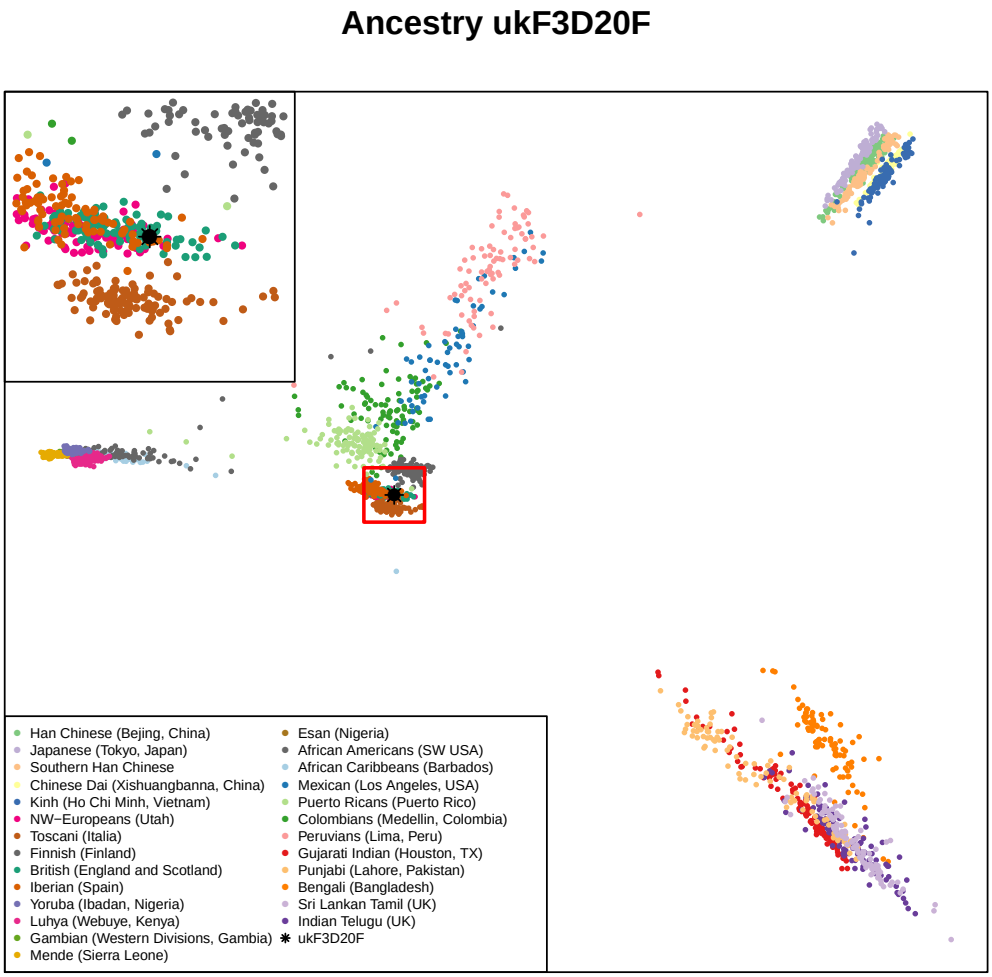

Figure 5: Ancestry Principal Component Analysis

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
2.5	rs11649743	(A;A)	Lower prostate cancer risk?	Link	Link	
2.1	rs3775291	(A;G)	0.71x decreased risk for dry age related macula...	Link	Link	Link
2.1	rs806380	(G;G)	Uncommon. lowest odds of cannabis dependence	Link		
2	rs11045585	(A;A)	24% chance (lower than average) of docetaxel-in...	Link	Link	
2	rs1136410	(C;T)	0.80x reduced risk for glioblastoma	Link	Link	
2	rs12979860	(C;C)	~80% of such hepatitis C patients respond to tr...	Link	Link	Link
2	rs17070145	(C;T)	Increased memory performance	Link		Link
2	rs1799884	(G;G)	Mothers have typical Birth-Weight babies. Sligh...	Link		
2	rs1864163	(G;G)	Associated with higher HDL cholesterol	Link	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	rs3782179	(C;T)	3x lower odds of testicular cancer risk for men...	Link		
2	rs37973	(A;A)	Possibly better response to inhaled corticoster...	Link		Link
2	rs3819331	(T;T)	Lower risk of autism	Link		Link
2	rs4149268	(G;G)	Associated with higher HDL cholesterol	Link	Link	
2	rs4307059	(C;C)	Reduced Autism risk	Link	Link	
2	rs763110	(C;T)	~0.80x reduced cancer risk	Link		Link
1.8	rs1128535	(A;G)	0.77x risk for Crohn's disease	Link		
1.8	rs187238	(C;G)	Hypertension not a risk factor for sudden cardi...	Link		
1.8	rs3814113	(C;T)	0.8x decreased risk for ovarian cancer	Link	Link	
1.6	rs1061170	(T;T)	Lower risk for AMD: generally longer live than ...	Link	Link	Link
1.6	rs2278206	(C;C)	Possibly less susceptible to asthma	Link	Link	
1.5	rs1050631	(C;C)	Mean Survival Time of 32 months for esophageal ...	Link		
1.5	rs1063192	(C;T)	0.71x reduced risk of myocardial infarction	Link		
1.5	rs11136000	(C;T)	0.84x decreased risk for Alzheimer's disease	Link	Link	
1.5	rs11212617	(A;C)	Somewhat increased likelihood of treatment succ...	Link		Link
1.5	rs3851179	(A;G)	0.85x decreased risk for Alzheimer's disease	Link	Link	

Mag.	Identifier	Genotype	Summary	GnomAD	GetEvidence	ClinVar
1.5	rs4149274	(C;C)	Associated with higher HDL (good) cholesterol.	Link		
1.5	rs4939883	(C;C)	Associated with higher HDL cholesterol	Link	Link	
1.5	rs9939609	(T;T)	Lower risk of obesity and Type-2 diabetes	Link	Link	
1.4	rs1165205	(A;T)	0.85x decreased gout risk	Link	Link	
1.4	rs6495446	(C;T)	0.8x reduced risk for chronic kidney disease	Link		
1.2	rs4320932	(A;G)	0.87x decreased risk for ovarian cancer	Link		
1.1	rs2235040	(A;G)	Possibly higher chances of remission only for i...	Link	Link	
1.1	rs2293347	(G;G)	Among NSCLC patients: better Gefitinib response...	Link		Link
1.1	rs7568369	(G;T)	0.90x reduced risk of obesity	Link		
1	rs182549	(C;T)	Can digest milk.	Link		Link
1	rs2952768	(C;T)	Slightly less drug dependence: decreased effect...	Link		Link
1	rs4148739	(A;G)	Possibly: inpatients more likely to remit on ce...	Link	Link	
1	rs4752566	(G;T)	Associated with thicker hair in Asians	Link		
1	rs4939827	(C;T)	0.86x decreased risk for colorectal cancer	Link	Link	Link
1	rs7850258	(A;A)	Slightly lower odds of developing primary hypot...	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	

3.2 Possibly Harmful Traits

Mag.	Identifier	Genotype	Summary	GnomAD	GetEvidence	ClinVar
3.5	rs875858	(C;T)	Docetaxel sensitive?	Link		
3	rs10974944	(G;G)	2 - 4 fold increased odds of V617F-associated M...	Link	Link	
3	rs12343867	(C;C)	2 - 4 fold increased odds of developing V617F-a...	Link		
3	rs13266634	(C;C)	Increased risk for type-2 diabetes	Link	Link	Link
3	rs1983132	(C;T)	2 - 3x higher prostate cancer risk if routinely...	Link		
3	rs4495487	(C;C)	2 - 4 fold increased odds of developing V617F-a...	Link		
2.8	rs3780374	(A;A)	Substantially increased odds of developing V617...	Link		
2.5	rs12803066	(A;G)	Increased risk of myopia	Link		
2.5	rs17696736	(G;G)	1.94x risk of type-1 diabetes	Link	Link	
2.5	rs2073963	(G;G)	Increased risk of baldness	Link		
2.5	rs2241880	(C;C)	2x-3x increased risk for Crohn's disease in Cau...	Link	Link	Link
2.5	rs2943634	(C;C)	Slightly higher risk of ischemic stroke	Link	Link	
2.5	rs339331	(T;T)	Prostate cancer risk	Link		
2.5	rs3738919	(C;C)	1.94x risk of developing rheumatoid arthritis	Link		
2.5	rs664143	(C;T)	Higher risk for number of cancers	Link		
2.5	rs891512	(A;G)	Higher blood pressure than G;G	Link		Link
2.4	rs1143679	(A;G)	1.78x increased risk for SLE	Link	Link	
2.4	rs2274223	(G;G)	1.9x increased risk for stomach and esophageal ...	Link	Link	Link
2.2	rs1024611	(C;C)	Increased risk of exercise induced ischemia: In...	Link		Link
2.2	rs2004640	(G;T)	1.4x increased risk for SLE	Link	Link	
2.2	rs944289	(T;T)	1.69x increased thyroid cancer risk	Link	Link	
2.1	rs1050152	(T;T)	2.1x increased risk of Crohn's disease	Link	Link	Link
2.1	rs10811661	(T;T)	1.2x increased risk for type-2 diabetes	Link	Link	
2.1	rs2306402	(C;C)	1.18x increased risk for late-onset Alzheimer's...	Link		
2.1	rs2494732	(C;C)	Greater odds of cannabis-associated psychosis	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	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	rs10086908	(C;C)	1.7x increased risk for prostate cancer	Link		
2	rs10260404	(C;C)	1.60x risk of developing ALS	Link	Link	
2	rs10883365	(G;G)	1.62x increased risk for developing Crohn's dis...	Link	Link	
2	rs10889677	(A;C)	1.5x increased risk for certain autoimmune dise...	Link	Link	
2	rs10984447	(A;A)	>1.17x increased risk for multiple sclerosis	Link	Link	
2	rs11171739	(C;C)	1.75x risk of developing Type-1 diabetes	Link	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	rs12037606	(A;A)	1.52x risk of developing Crohn's disease	Link		
2	rs12567232	(A;G)	Increased risk for Crohn's Disease	Link	Link	
2	rs12696304	(C;G)	Prone to aging faster: at least in European pop...	Link		
2	rs13254738	(C;C)	1.18x prostate cancer risk	Link	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	rs1361600	(G;G)	~2x increased risk for adult-onset asthma in Ja...	Link		
2	rs16942	(A;G)	Very slightly increased breast cancer risk	Link	Link	Link
2	rs16944	(G;G)	Slightly increased (~2x or less) risk for certa...	Link	Link	
2	rs1734791	(A;A)	1.4x increased risk for lupus	Link		
2	rs1800896	(A;G)	1.6x increased prostate cancer risk	Link		
2	rs1801160	(A;G)	Possible 5-fluorouracil toxicity	Link	Link	Link
2	rs2156921	(A;G)	1.29x increased risk for depression	Link		
2	rs2201841	(C;T)	1.5x increased risk for Crohn's disease; 2x inc...	Link	Link	
2	rs2230201	(G;G)	>1.4x risk of lupus	Link		Link
2	rs2305480	(C;T)	3.5x increase in risk of asthma for Han Chinese...	Link	Link	

Mag.	Identifier	Genotype	Summary	GnomAD	GetEvidence	ClinVar
2	rs2305795	(A;G)	1.28x higher risk of narcolepsy compared to (G;...	Link		Link
2	rs2383206	(A;G)	1.4x increased risk for heart disease	Link		
2	rs2383207	(A;G)	Increased risk for heart disease	Link		
2	rs25487	(A;G)	2x higher risk for skin cancer; possibly other ...	Link	Link	Link
2	rs3212227	(A;C)	Significantly increased risk of developing cerv...	Link		Link
2	rs351855	(C;T)	1.2x increased risk for prostate cancer	Link	Link	Link
2	rs3738579	(C;C)	0.6x decreased risk for cervical cancer: but 1....	Link		
2	rs3775948	(G;G)	Slightly higher risk for gout	Link		
2	rs3793784	(C;G)	1.5x risk for ARMD	Link	Link	Link
2	rs4027132	(A;A)	1.51x increased risk of developing bipolar diso...	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	rs4444903	(A;G)	3.5x risk of hep-cancer in cirrhosis patients; ...	Link		Link
2	rs4633	(C;T)	Higher risk for endometrial cancer	Link	Link	Link
2	rs4792311	(A;G)	Increased risk of prostate cancer	Link	Link	Link
2	rs4825476	(G;G)	1.9x higher risk of suicidal thoughts when taki...	Link	Link	
2	rs486907	(A;A)	2x increased prostate cancer risk	Link	Link	Link
2	rs493258	(A;G)	1.15x risk of Age Related Macular Degeneration	Link		
2	rs4968451	(A;C)	1.61x increased risk for meningioma	Link		
2	rs520354	(A;G)	Increased risk in men for biliary conditions	Link		
2	rs6232	(A;G)	Higher risk of obesity and insulin sensitivity	Link	Link	Link
2	rs629242	(C;T)	Somewhat higher risk for prostate cancer	Link		
2	rs6441286	(G;T)	1.54x chance of developing primary biliary cirr...	Link	Link	
2	rs6498169	(A;A)	>1.14x risk of multiple sclerosis	Link	Link	
2	rs6601764	(C;C)	1.52x increased risk of developing Crohn's dise...	Link	Link	
2	rs663048	(G;T)	Significantly increased risk of developing lung...	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	rs6908425	(C;C)	1.95x increased risk of developing Crohn's dise...	Link	Link	
2	rs6997709	(G;T)	1.2x higher risk for hypertension	Link		
2	rs699	(C;T)	Increased risk of hypertension	Link	Link	Link
2	rs7190458	(A;G)	Slightly higher pancreatic cancer risk	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	rs7536563	(A;A)	>1.12x risk of multiple sclerosis	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	rs7923837	(G;G)	3.2x risk for T2D	Link		
2	rs7961152	(A;A)	1.5x higher risk for hypertension	Link		
2	rs854560	(A;T)	Higher risk for heart disease: diabetic retinop...	Link	Link	Link
2	rs965513	(A;A)	3.1x increased thyroid cancer risk	Link	Link	
2	rs9954153	(G;T)	~2.5x higher risk for Fuchs' dystrophy: a corne...	Link		
1.8	rs10210302	(T;T)	1.8x increased risk for Crohn's disease	Link	Link	
1.8	rs143383	(T;T)	1.3x increased risk for osteoarthritis	Link	Link	
1.8	rs4474514	(A;G)	3x increased testicular cancer risk for men	Link	Link	
1.7	rs1042713	(A;A)	1.7x increased risk that pediatric inhaler use ...	Link	Link	Link
1.7	rs2024513	(A;A)	1.7x higher risk for schizophrenia (among Han C...	Link		
1.6	rs1537415	(C;G)	1.6x increased risk for periodontitis	Link	Link	
1.6	rs1978237	(C;C)	>1.59x risk of Type 2 diabetes	Link		
1.6	rs2059693	(T;T)	1.6x increased risk for testicular cancer	Link		
1.6	rs2736100	(G;G)	1.6x higher risk for glioma development	Link	Link	Link
1.6	rs3764880	(A;A)	1.2 - 1.8x increased tuberculosis risk	Link	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		

Mag.	Identifier	Genotype	Summary	GnomAD	GetEvidence	ClinVar
1.5	rs10784502	(T;T)	Less intracranial volume?	Link		
1.5	rs12210050	(C;T)	Slightly higher risk for basal cell carcinoma	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;C)	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	rs1360517	(A;G)	Higher susceptibility for AIDS	Link	Link	
1.5	rs140701	(A;A)	Increased risk for anxiety disorders	Link		
1.5	rs1571801	(A;A)	>1.36x risk for prostate cancer	Link		
1.5	rs17221417	(C;G)	1.3x higher risk for Crohn's disease	Link	Link	
1.5	rs1867277	(A;G)	1.5x increased risk for thyroid cancer	Link		
1.5	rs1994090	(G;T)	Slightly increased risk of developing Parkinson...	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	rs2177369	(C;C)	1.5x increased risk for Alzheimer's disease	Link		
1.5	rs2229169	(A;A)	1.5x increased risk of heart attack and stroke ...	Link		
1.5	rs2240340	(A;G)	Slightly increased (1.5x) risk for RA	Link		
1.5	rs2272127	(C;C)	Associated with herpes and schizophrenia	Link		
1.5	rs2280714	(A;G)	1.4x increased risk of SLE	Link		
1.5	rs2282679	(C;C)	Lower vitamin D levels	Link		
1.5	rs2286812	(C;T)	~2x higher risk for Fuchs' dystrophy: a corneal...	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	rs358806	(A;C)	0.86x increased risk of developing Type-2 diabe...	Link	Link	
1.5	rs3790565	(C;T)	Slightly increased risk of developing primary b...	Link		
1.5	rs3814570	(T;T)	1.3x increased risk for Crohn's disease with il...	Link		
1.5	rs4506565	(A;T)	1.4x increased risk for type-2 diabetes	Link	Link	
1.5	rs4626664	(A;G)	1.44x increased risk of developing restless leg...	Link	Link	
1.5	rs464049	(T;T)	Increased risk of schizophrenia in limited stud...	Link		
1.5	rs4785763	(A;C)	1.5x higher risk for melanoma	Link	Link	
1.5	rs5746059	(A;A)	Slightly higher fat mass	Link		
1.5	rs642961	(A;G)	1.68x increased risk of cleft lip	Link	Link	
1.5	rs6435862	(G;T)	1.7x higher risk of aggressive neuroblastoma	Link	Link	
1.5	rs699473	(C;C)	~1.5x increased brain tumor risk	Link		
1.5	rs7341475	(G;G)	1.58x increased schizophrenia risk for women	Link	Link	
1.5	rs872071	(G;G)	~1.5x increased risk for chronic lymphocytic le...	Link	Link	
1.5	rs9303277	(C;T)	1.46x Slightly increased risk of developing pri...	Link		
1.5	rs9642880	(G;T)	1.2x increased bladder cancer risk	Link	Link	
1.5	rs9652490	(A;G)	Slightly increased risk of developing Parkinson...	Link	Link	
1.4	rs12770228	(A;G)	1.4x increased risk for meningioma	Link		
1.4	rs1545843	(A;A)	1.4x increased risk for depression (for those u...	Link		
1.4	rs1801157	(A;G)	1.4x higher risk for breast cancer	Link		
1.4	rs2046210	(C;T)	1.4x increased breast cancer risk	Link	Link	Link
1.4	rs6010620	(G;G)	1.4x higher risk for glioma development; but th...	Link	Link	
1.34	rs17465637	(C;C)	1.34x higher risk for myocardial infarction	Link	Link	
1.3	rs1746048	(C;C)	1.03 increased risk for coronary heart disease	Link	Link	
1.3	rs4295627	(G;T)	1.36x higher risk for glioma development	Link	Link	
1.25	rs13387042	(A;A)	1.24x increased risk for breast cancer	Link	Link	
1.25	rs748404	(T;T)	Slightly increased risk (1.25) for lung cancer...	Link	Link	
1.2	rs10865331	(A;G)	1.2x higher risk for ankylosing spondylitis	Link		

Mag.	Identifier	Genotype	Summary	GnomAD	GetEvidence	ClinVar
1.2	rs2056116	(A;G)	1.18x risk for breast cancer	Link		
1.2	rs2254958	(C;T)	1.24x reported increased risk for Alzheimer's; ...	Link		
1.2	rs249954	(T;T)	Potentially increased risk of Breast Cancer	Link		Link
1.2	rs3850641	(A;G)	Increased risk of myocardial infarction in wome...	Link		
1.2	rs449647	(A;T)	Possibly lower levels of ApoE	Link		
1.2	rs4977756	(A;G)	1.39x higher risk for glioma development	Link	Link	
1.2	rs6897876	(C;C)	Slight increase in testicular cancer risk for m...	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	
1.17	rs3802842	(A;C)	1.17x increased risk of colorectal cancer	Link	Link	
1.1	rs11037909	(C;T)	1.27x type II diabetes risk	Link		
1.1	rs11650494	(A;G)	Slightly higher prostate cancer risk	Link		
1.1	rs1344706	(G;T)	1.1x increased risk for schizophrenia	Link	Link	
1.1	rs1800450	(A;G)	Carrier of mannose binding deficiency but of lo...	Link	Link	Link
1.1	rs2295190	(G;T)	Slightly increased risk for ovarian cancer in w...	Link	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	rs4324715	(C;T)	1.5x increased testicular cancer risk for men	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	rs7531806	(A;G)	Very slightly increased risk of acne occurrence...	Link		
1.07	rs2291834	(C;C)	Very slightly higher risk for myocardial infarc...	Link		
1	rs1004819	(C;T)	1.5x risk of Crohn's disease: 1.2 for developin...	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	rs2546890	(A;A)	Higher risk of multiple sclerosis	Link		
1	rs6166	(G;G)	Females slightly more likely to be sterile	Link	Link	Link
1	rs6932590	(T;T)	1.1x increased risk for schizophrenia	Link	Link	
1	rs6976	(C;T)	Slight risk of osteoarthritis	Link		
0.1	rs11110912	(C;G)	Maybe some quite minor increase in high blood p...	Link		
0.1	rs3095870	(G;G)	1.7x increased risk for SLE (lupus)	Link		
0.1	rs3748079	(G;G)	1.9x 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
3	gs137	5x risk of thyroid cancer
3	gs273	Lowest risk (13% of white women) of Atrial Fibr...
2.7	gs311	Slow metabolizer of certain substances
2.5	gs102	ALS risk
2.5	gs155	CYP3A5 non-expressor
2.5	gs282	Claimed to be part of the 12% of the population...
2.5	gs284	Any diet works for you
2.5	gs298	Increased surveillance for colorectal cancer re...
2.4	gs297	Lower heart attack risk than average
2	gs101	Probably able to digest milk
2	gs129	Unable to classify the ABO blood type
2	gs154	NAT2 Slow metabolizer
2	gs159	CYP1A2 fast metabolizer
2	gs194	Myocardial Infarction Risk
2	gs213	Haplogroup R (Y-DNA)
2	gs244	2x increased risk for esophageal squamous cell ...
2	gs246	APOE E3/E3
2	gs289	You have one 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	gs247	Parkinson's Disease Risk
1.2	gs184	Able to taste bitterness.

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

Report generated on June 13, 2019.