PGP-UK Genomics Report for uk65E564

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.

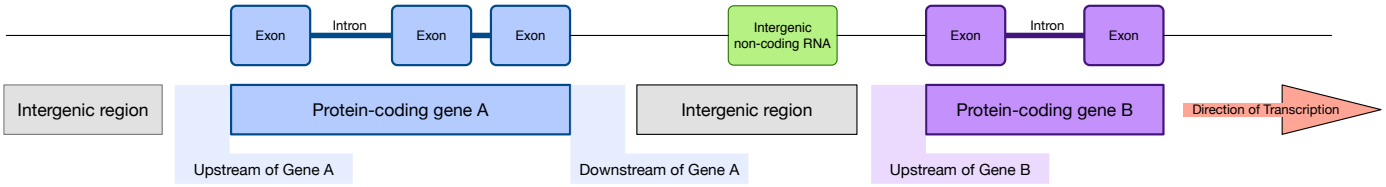

Figure 1: Diagram of gene structure indicating locations of potential variants

Feature	Count
Lines of input read	4655473
Variants filtered out	3662933
Novel / existing variants	0 (0.0) / 992540 (100.0)
Overlapped genes	52160
Overlapped transcripts	59599
Overlapped regulatory features	47782

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.

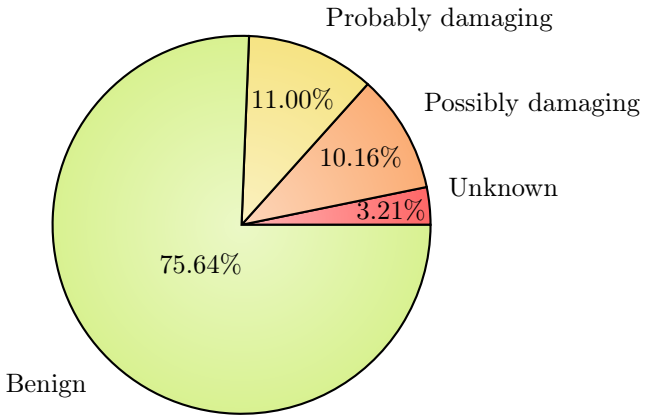

Figure 2: PolyPhen Summary

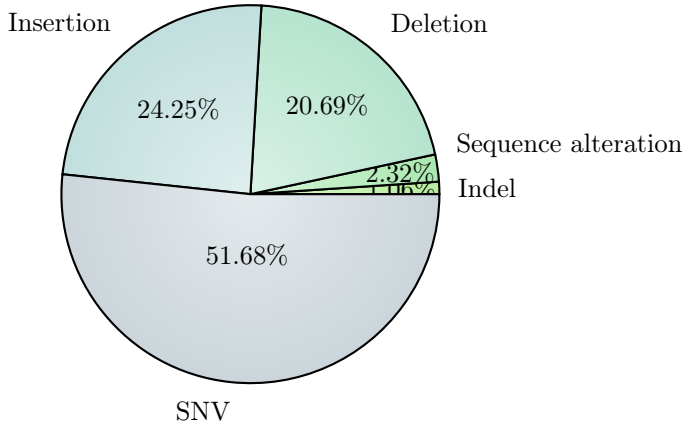

Figure 3: Variant Class

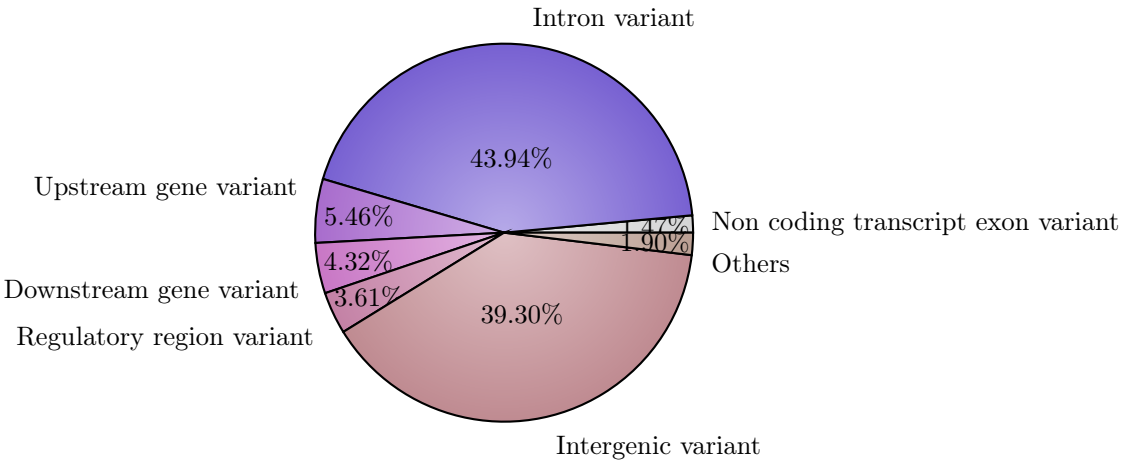

Figure 4: Consequence type

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).

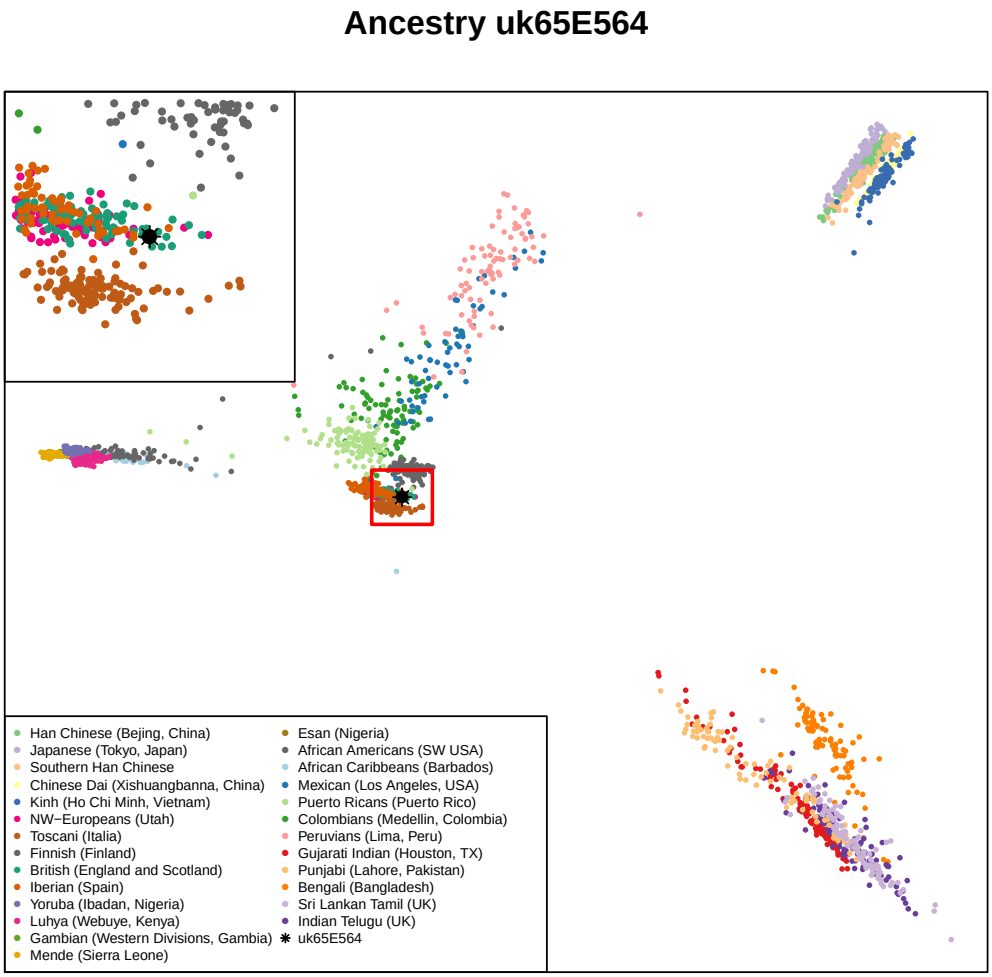

Figure 5: Ancestry Principal Component Analysis

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	rs925391	(T;T)	Unlikely to go bald	Link		
2.5	rs11649743	(A;A)	Lower prostate cancer risk?	Link	Link	
2.4	rs2802288	(A;A)	Longer lifespan	Link		
2.1	rs2511989	(A;G)	0.63x decreased age-related macular degeneratio...	Link	Link	
2.1	rs3775291	(A;G)	0.71x decreased risk for dry age related macula...	Link	Link	Link
2.1	rs6505162	(A;A)	0.43x decreased risk for esophageal cancer	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	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	rs25487	(A;A)	0.7x lower risk for skin cancer	Link	Link	Link
2	rs261332	(A;G)	Associated with higher HDL cholesterol	Link		
2	rs2764264	(C;C)	Greater odds of living to 95	Link		
2	rs3218536	(A;G)	Lower risk for breast: ovarian cancer	Link	Link	Link
2	rs3764261	(G;T)	Associated with higher HDL cholesterol	Link	Link	Link
2	rs37973	(A;A)	Possibly better response to inhaled corticoster...	Link		Link
2	rs3819331	(T;T)	Lower risk of autism	Link		Link
2	rs3914132	(C;T)	Lower otosclerosis risk	Link	Link	
2	rs4149268	(A;G)	Associated with higher HDL cholesterol	Link	Link	
2	rs6511720	(G;T)	Slightly lower odds of developing CHD.	Link	Link	Link
2	rs7216389	(C;C)	0.69x lower risk of Childhood Asthma.	Link	Link	
2	rs763110	(C;T)	~0.80x reduced cancer risk	Link		Link
2	rs800292	(T;T)	5% decreased risk of macular degeneration	Link	Link	Link
2	rs801114	(T;T)	0.78x decreased Basal Cell Carcinoma risk.	Link	Link	
2	rs887829	(A;A)	Higher levels of serum bilirubin: associated wi...	Link	Link	Link

Mag.	Identifier	Genotype	Summary	GnomAD	GetEvidence	ClinVar
1.8	rs1128535	(A;G)	0.77x risk for Crohn's disease	Link		
1.8	rs1746048	(C;T)	0.94 decreased risk for coronary heart disease	Link	Link	
1.8	rs1800588	(C;T)	Higher HDL-C levels	Link	Link	
1.8	rs187238	(C;G)	Hypertension not a risk factor for sudden cardi...	Link		
1.8	rs266729	(C;G)	0.73x decreased risk for colorectal cancer	Link	Link	
1.8	rs3814113	(C;T)	0.8x decreased risk for ovarian cancer	Link	Link	
1.8	rs6897932	(C;T)	0.91x decreased risk for multiple sclerosis	Link	Link	Link
1.6	rs1061170	(T;T)	Lower risk for AMD: generally longer live than ...	Link	Link	Link
1.5	rs11136000	(C;T)	0.84x decreased risk for Alzheimer's disease	Link	Link	
1.5	rs1165205	(A;A)	0.85x decreased gout risk	Link	Link	
1.5	rs2007153	(A;A)	Decreased risk of schizophrenia in limited stud...	Link		
1.5	rs3790844	(C;T)	Slightly reduced risk (0.77x) for pancreatic ca...	Link		
1.5	rs3851179	(A;A)	0.85x decreased risk for Alzheimer's disease	Link	Link	
1.5	rs4149274	(C;T)	Associated with higher HDL (good) cholesterol	Link		
1.5	rs4939883	(C;C)	Associated with higher HDL cholesterol	Link	Link	
1.5	rs729302	(A;C)	0.89x decreased risk of developing rheumatoid a...	Link		
1.3	rs2361502	(C;C)	Possible higher levels of serum bilirubin and l...	Link		
1.2	rs4867568	(T;T)	Decreased risk for knee osteoporosis	Link		
1.2	rs9306160	(C;T)	0.75x (reduced) risk for metastasis in LN-/ER+ ...	Link	Link	
1.1	rs13333226	(A;G)	Slightly lower risk for hypertension	Link		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	rs2351299	(G;T)	Possible reduced risk of Autism	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;G)	Typical odds of developing primary hypothyroidi...	Link		
0.1	rs1726866	(C;C)	Can taste bitter	Link	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	rs1333049	(C;C)	1.9x increased risk for coronary artery disease...	Link	Link	
3.5	rs875858	(C;T)	Docetaxel sensitive?	Link		
3.2	rs2981582	(T;T)	1.7x higher risk of ER+ breast cancer	Link	Link	
3	rs1021737	(T;T)	Significantly higher plasma total homocysteine ...	Link	Link	Link
3	rs3738579	(T;T)	1.5x - 2x increased risk for cervical cancer: H...	Link		
3	rs3903239	(C;C)	Higher frequency of atrial fibrillation	Link		
3	rs4244285	(A;G)	Poorer metabolizer of several popular medicines...	Link	Link	Link
3	rs7754840	(C;G)	1.3x increased risk for type-2 diabetes	Link	Link	
2.5	rs1057910	(A;C)	CYP2C9*3 carrier; average 40% reduction in warf...	Link	Link	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	Link
2.5	rs16969968	(A;G)	Slightly higher risk for nicotine dependence: l...	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	rs5888	(C;T)	3x higher risk for age-related macular degenera...	Link		
2.5	rs613872	(G;T)	~5 fold higher risk for Fuchs' dystrophy: a cor...	Link		
2.5	rs7574865	(G;T)	1.3x risk of rheumatoid arthritis; 1.55x risk o...	Link	Link	Link
2.5	rs8034191	(C;T)	1.27x lung cancer risk	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	rs10811661	(T;T)	1.2x increased risk for type-2 diabetes	Link	Link	
2.1	rs1219648	(G;G)	1.64x risk for breast cancer	Link	Link	
2.1	rs17070145	(C;C)	Reduced memory abilities	Link		Link
2.1	rs17563	(C;C)	Risk for otosclerosis	Link	Link	Link
2.1	rs2270641	(G;G)	3.7x higher risk for schizophrenia	Link	Link	
2.1	rs2383207	(G;G)	Increased risk for heart disease	Link		
2.1	rs2420946	(T;T)	1.64x risk for breast cancer	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	rs6742078	(T;T)	+16% bilirubin levels increased risk of gallsto...	Link	Link	Link
2.1	rs795484	(A;G)	Increased morphine dose requirement and postope...	Link		
2	rs1024611	(C;T)	Increased risk of exercise induced ischemia	Link		Link
2	rs1042838	(G;T)	1.28x risk for endometrial ovarian cancer; over...	Link	Link	
2	rs1050631	(C;T)	Mean Survival Time of 25 months for esophageal ...	Link		
2	rs1051730	(C;T)	1.3x increased risk of lung cancer	Link	Link	Link
2	rs1064395	(A;G)	Having any copies of A at this SNP heightens yo...	Link		
2	rs10757272	(T;T)	1.54x increased risk for Coronary artery diseas...	Link		
2	rs10889677	(C;C)	Baseline (average) risk for certain autoimmune ...	Link	Link	
2	rs10984447	(A;A)	>1.17x increased risk for multiple sclerosis	Link	Link	
2	rs110419	(A;A)	1.7x increased risk for neuroblastoma	Link		
2	rs11123857	(A;G)	1.44-fold increased risk of bipolar disorder or...	Link		
2	rs11171739	(C;C)	1.75x risk of developing Type-1 diabetes	Link	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;A)	1.6x increased risk of Male Pattern Baldness.	Link	Link	
2	rs12696304	(C;G)	Prone to aging faster: at least in European pop...	Link		
2	rs1360780	(C;T)	1.3x increased risk for depression	Link	Link	Link
2	rs16944	(A;A)	Increased risk (~3x) for osteoarthritis	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	rs17435	(T;T)	1.4x increased risk for lupus	Link		
2	rs17487223	(C;T)	Higher lung cancer risk?	Link		

Mag.	Identifier	Genotype	Summary	GnomAD	GetEvidence	ClinVar
2	rs17576	(G;G)	Higher risk for lung cancer: and COPD in smoker...	Link	Link	Link
2	rs17696736	(A;G)	1.34x risk of type-1 diabetes	Link	Link	
2	rs2073963	(G;T)	Increased risk of baldness	Link		
2	rs2143340	(C;T)	Increased risk of dyslexia and poor reading per...	Link		
2	rs2156921	(A;G)	1.29x increased risk for depression	Link		
2	rs2201841	(T;T)	2.4x increased risk for Graves' 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	(C;T)	3.5x increase in risk of asthma for Han Chinese...	Link	Link	
2	rs2305795	(A;G)	1.28x higher risk of narcolepsy compared to (G;...	Link		Link
2	rs2306402	(C;T)	1.18x increased risk for late-onset Alzheimer's...	Link		
2	rs2736100	(T;T)	Higher risk of Interstitial lung disease: and t...	Link	Link	Link
2	rs27388	(A;A)	Increased risk of developing schizophrenia	Link		
2	rs326	(A;A)	Lower HDL cholesterol	Link	Link	Link
2	rs358806	(C;C)	1.78x increased risk of developing Type-2 diabe...	Link	Link	
2	rs3746444	(C;T)	~1.2x increased risk for cancer	Link		
2	rs3775948	(G;G)	Slightly higher risk for gout	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	rs4792311	(A;G)	Increased risk of prostate cancer	Link	Link	Link
2	rs493258	(G;G)	1.15x risk of Age Related Macular Degeneration	Link		
2	rs4961	(G;T)	1.8x increased risk for high blood pressure	Link	Link	Link
2	rs520354	(A;A)	Increased risk in men for biliary conditions	Link		
2	rs6601764	(C;C)	1.52x increased risk of developing Crohn's dise...	Link	Link	
2	rs6896702	(T;T)	Increased risk of developing Parkinson's Diseas...	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	rs7774434	(C;C)	Increased risk of developing primary biliary ci...	Link		
2	rs7794745	(A;T)	Slightly increased risk for autism	Link	Link	Link
2	rs7961152	(A;C)	1.2x higher risk for hypertension	Link		
2	rs854560	(A;T)	Higher risk for heart disease: diabetic retinop...	Link	Link	Link
2	rs9303277	(T;T)	1.46x Increased risk of developing primary bili...	Link		
2	rs965513	(A;G)	1.77x increased thyroid cancer risk	Link	Link	
2	rs9954153	(G;T)	~2.5x higher risk for Fuchs' dystrophy: a corne...	Link		
2.0	rs1434536	(A;A)	1.94x increased breast cancer risk	Link		Link
1.9	rs7923837	(A;G)	1.6x risk for T2D	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	rs11523871	(C;C)	>1.6x increased breast cancer risk for women ov...	Link	Link	
1.6	rs1260326	(T;T)	Slightly higher risk for gout	Link	Link	Link
1.6	rs1537415	(C;G)	1.6x increased risk for periodontitis	Link	Link	
1.6	rs2981745	(T;T)	>1.6x increased risk for breast cancer in femal...	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	rs10260404	(C;T)	1.20x risk of developing ALS	Link	Link	
1.5	rs10464059	(A;G)	Slightly increased risk of developing Parkinson...	Link		
1.5	rs10784502	(T;T)	Less intracranial volume?	Link		
1.5	rs10859871	(A;C)	Slight (~1.2x) increase in endometriosis risk	Link		
1.5	rs10883365	(A;G)	1.2x increased risk for developing Crohn's dise...	Link	Link	
1.5	rs12037606	(A;G)	1.22x risk of developing Crohn's disease	Link		

Mag.	Identifier	Genotype	Summary	GnomAD	GetEvidence	ClinVar
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	rs12469063	(A;G)	Slightly increased risk of developing restless ...	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	rs17115100	(G;T)	Slightly increased risk of developing Parkinson...	Link	Link	
1.5	rs17221417	(C;G)	1.3x higher risk for Crohn's disease	Link	Link	
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	Link		
1.5	rs1994090	(G;T)	Slightly increased risk of developing Parkinson...	Link	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	rs2254958	(C;C)	1.61x reported increased risk for Alzheimer's; ...	Link		
1.5	rs2280714	(A;A)	1.4x increased risk of SLE	Link		
1.5	rs2736990	(C;T)	Slightly increased risk of developing Parkinson...	Link	Link	
1.5	rs28694718	(A;G)	2x higher risk for schizophrenia	Link		
1.5	rs2881766	(G;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	rs3212227	(A;A)	1.43x increased risk of developing psoriasis an...	Link		Link
1.5	rs3814570	(C;T)	1.3x increased risk for Crohn's disease with il...	Link		
1.5	rs3825776	(A;G)	1.3x increased risk for ALS	Link	Link	
1.5	rs4027132	(A;G)	1.39x increased risk of developing bipolar diso...	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	rs4656461	(A;G)	1.5x increased risk for open angle glaucoma	Link		
1.5	rs4785763	(A;C)	1.5x higher risk for melanoma	Link	Link	
1.5	rs4977574	(G;G)	Most studies find a somewhat elevated (~1.5x) r...	Link	Link	
1.5	rs5219	(C;T)	1.3x increased risk for type-2 diabetes	Link	Link	Link
1.5	rs5746059	(A;A)	Slightly higher fat mass	Link		
1.5	rs619203	(C;G)	Increases susceptibility to Myocardial Infarcti...	Link	Link	
1.5	rs6710341	(A;G)	Slightly increased risk of developing restless ...	Link		
1.5	rs6908425	(C;T)	1.63x increased risk of developing Crohn's dise...	Link	Link	
1.5	rs7341475	(G;G)	1.58x increased schizophrenia risk for women	Link	Link	
1.5	rs7536563	(A;G)	1.12x risk of multiple sclerosis	Link	Link	
1.5	rs872071	(A;G)	~1.5x increased risk for chronic lymphocytic le...	Link	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	
1.5	rs966221	(C;C)	1.5x increased stroke risk certain populations	Link		
1.5	rs995030	(G;G)	Non-protective against testicular cancer	Link	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	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	rs2228314	(C;G)	1.48x risk of osteoarthritis	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	rs1047286	(C;T)	1.3x increased risk for age-related macular deg...	Link	Link	Link
1.3	rs1375144	(C;T)	1.32x increased risk of developing bipolar diso...	Link		
1.3	rs2059693	(C;T)	1.3x increased risk for testicular cancer	Link		
1.3	rs2295490	(A;G)	1.32x increased risk of early-onset type-2 diab...	Link	Link	
1.3	rs34330	(T;T)	1.2x higher breast cancer risk; 1.3x higher ris...	Link		Link
1.3	rs356219	(A;G)	1.3x increased risk for Parkinson's disease	Link		
1.3	rs501120	(A;G)	1.3x increased risk for heart disease	Link	Link	

Mag.	Identifier	Genotype	Summary	GnomAD	GetEvidence	ClinVar
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	rs11037909	(T;T)	1.47x type II diabetes risk	Link		
1.2	rs143383	(C;T)	1.1x increased risk for osteoarthritis	Link	Link	
1.2	rs2072590	(G;T)	1.2x increased risk for ovarian cancer	Link		
1.2	rs2076295	(G;T)	One copy of the risk allele (G): slightly incre...	Link		
1.2	rs3176336	(T;T)	Slightly higher (1.25x) higher risk for breast ...	Link		
1.2	rs3740878	(A;A)	1.46x type II diabetes risk; common	Link		Link
1.2	rs449647	(A;T)	Possibly lower levels of ApoE	Link		
1.2	rs4686484	(A;A)	Slightly increased risk for celiac disease	Link		
1.2	rs4795067	(A;G)	Slight increase in risk for psoriatic arthritis...	Link		
1.2	rs6897876	(C;C)	Slight increase in testicular cancer risk for m...	Link		
1.2	rs8050136	(A;C)	1.2x increased risk for T2D in some populations...	Link	Link	
1.17	rs3802842	(A;C)	1.17x increased risk of colorectal cancer	Link	Link	
1.1	rs13387042	(A;G)	1.12x increased risk for breast cancer	Link	Link	
1.1	rs2651899	(A;G)	1.1x higher risk for migraines	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	rs4324715	(C;T)	1.5x increased testicular cancer risk for men	Link		
1.1	rs5030737	(C;T)	Carrier of mannose binding deficiency but of lo...	Link	Link	Link
1.1	rs889312	(A;C)	Very slightly higher risk for breast cancer	Link	Link	
1.09	rs12050604	(A;C)	Very slightly increased risk for lung cancer	Link		
1.07	rs2291834	(C;C)	Very slightly higher risk for myocardial infarc...	Link		
1	rs1004819	(C;C)	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	rs1143674	(A;A)	1.3x increased autism risk	Link		
1	rs17300539	(G;G)	Increased risk of insulin resistance	Link		
1	rs1804197	(A;C)	Increased risk of familial colorectal cancer an...	Link		Link
1	rs2273697	(A;G)	Adverse reaction more likely to carbamazepine i...	Link	Link	Link
1	rs2282679	(A;C)	Somewhat lower vitamin D levels	Link		
1	rs2546890	(A;A)	Higher risk of multiple sclerosis	Link		
1	rs3194051	(A;G)	1.12x risk of type-1 diabetes	Link	Link	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	rs6974491	(A;G)	Higher risk of coeliac and/or inflammatory bowe...	Link		
1	rs6976	(C;T)	Slight risk of osteoarthritis	Link		
1	rs761100	(G;G)	Higher risk for dyslexia	Link		
0.5	rs1566734	(G;T)	Somatic mutation: cancer associated	Link	Link	Link
0.1	rs601338	(G;G)	Susceptible to Norovirus infections	Link	Link	Link

3.3 Genosets (Multi-variant Phenotypes)

Magnitude	Identifier	Summary
4	gs144	Male
3.1	gs122	7x risk of male baldness
3.1	gs191	Impaired NSAID drug metabolism
3	gs214	Paternal ancestry associated with counties of N...
3	gs273	Lowest risk (13% of white women) of Atrial Fibr...
3.0	gs291	Lower heart attack risk than average
2.7	gs311	Slow metabolizer of certain substances
2.5	gs100	Lactose intolerance risk
2.5	gs155	CYP3A5 non-expressor
2.5	gs161	CYP2C9 Intermediate Metabolizers
2.5	gs281	Part of the 88% of the population claimed not t...
2.5	gs285	Claimed to lose 2.5x as much weight on a low fa...
2.1	gs215	R-L21
2.1	gs223	One copy of GCH1 variant associated with lower ...
2	gs104	Restless legs syndrome risk
2	gs154	NAT2 Slow metabolizer
2	gs159	CYP1A2 fast metabolizer
2	gs211	Ethanol biodisposition
2	gs213	Haplogroup R (Y-DNA)
2	gs290	You might have two short form 5-HTTLPR.
2	gs313	Normal DPYD activity and thus 5-FU metabolism p...
1.7	gs232	Possible low pain sensitivity; LPS/LPS
1.6	gs236	Alzheimer's disease-related haplotype
1.5	gs247	Parkinson's Disease Risk
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

Report generated on June 13, 2019.