PGP-UK Genomics Report for uk48E359

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.

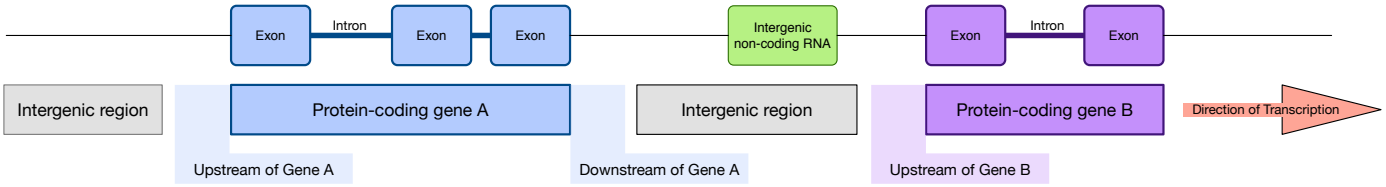

Figure 1: Diagram of gene structure indicating locations of potential variants

Feature	Count
Lines of input read	4864553
Variants filtered out	3310347
Novel / existing variants	0 (0.0) / 1554206 (100.0)
Overlapped genes	55463
Overlapped transcripts	64367
Overlapped regulatory features	144684

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.

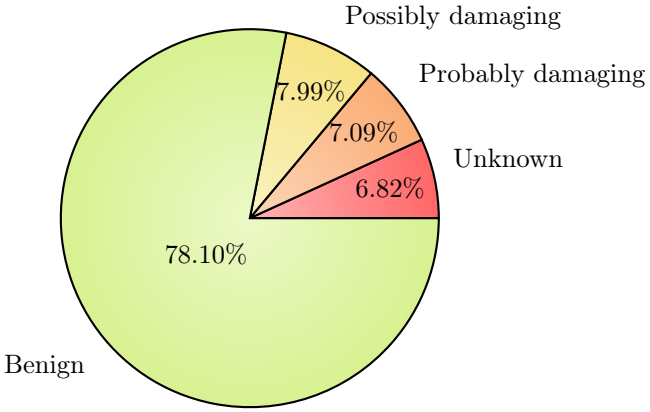

Figure 2: PolyPhen Summary

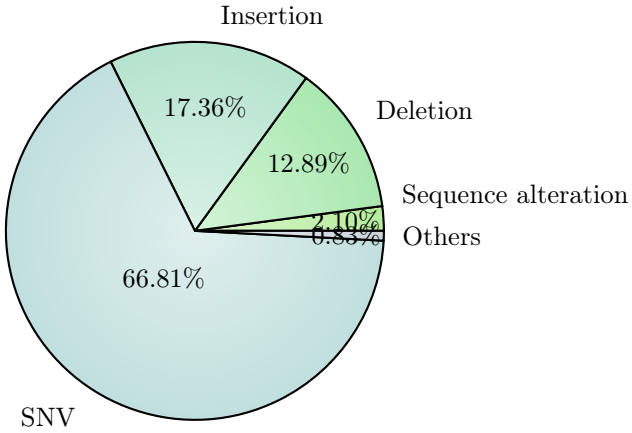

Figure 3: Variant Class

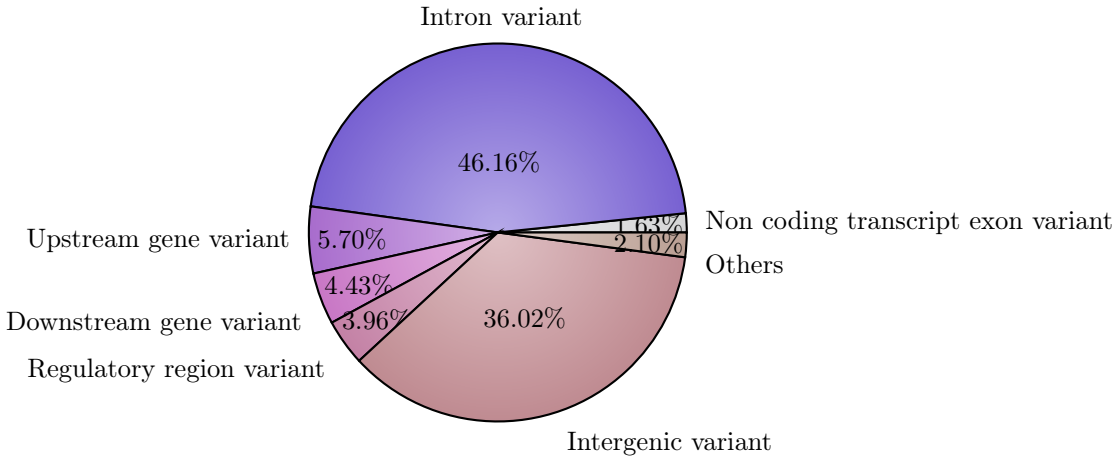

Figure 4: Consequence type

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).

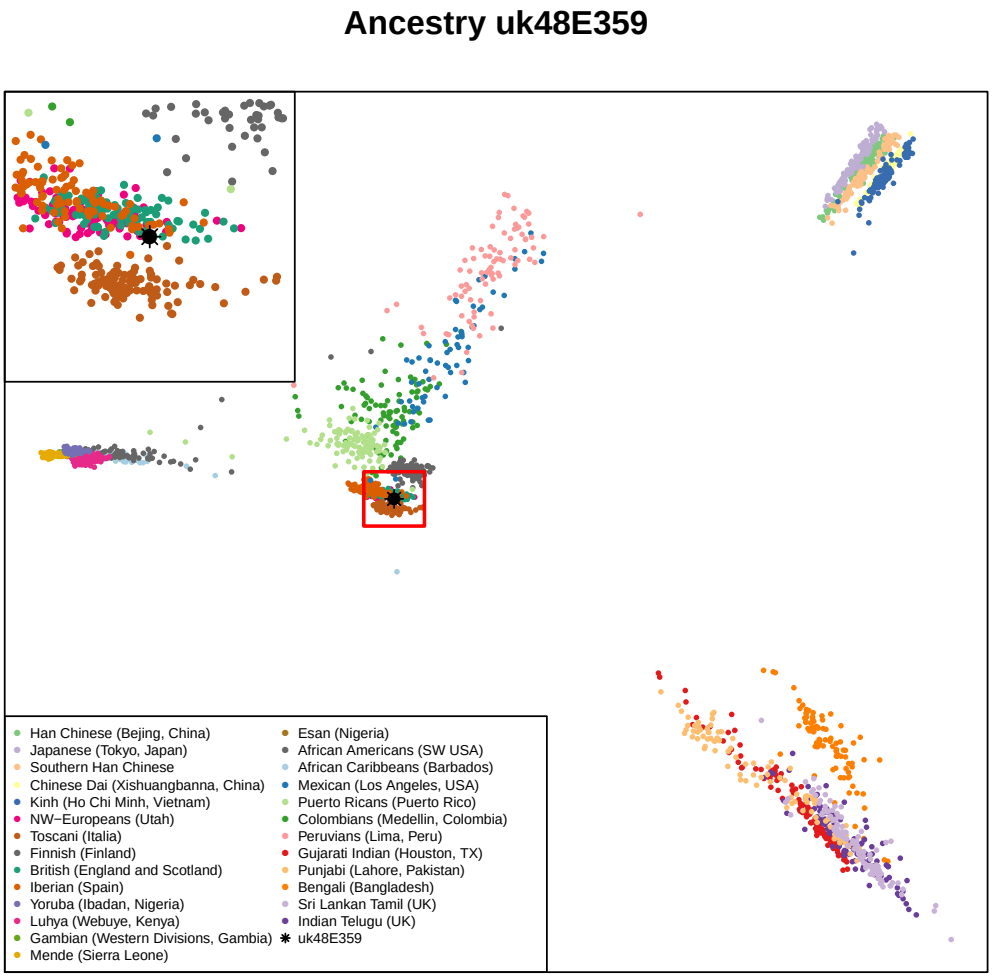

Figure 5: Ancestry Principal Component Analysis

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	
2.4	rs3750817	(T;T)	0.64x reduced risk for breast cancer: and highe...	Link		
2	rs10088218	(A;A)	0.5x decreased risk for ovarian cancer	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	rs10936599	(C;C)	Longer telomeres: longer life?	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	rs12979860	(C;C)	~80% of such hepatitis C patients respond to tr...	Link	Link	Link
2	rs1799884	(G;G)	Mothers have typical Birth-Weight babies. Sligh...	Link		
2	rs2056202	(T;T)	Rare decreased risk of autism	Link		
2	rs2241423	(A;G)	0.79 decreased risk for obesity	Link		
2	rs2241766	(G;G)	Slightly lower risk of breast cancer	Link		
2	rs2292813	(C;T)	Decreased risk of autism	Link		
2	rs2542052	(C;C)	Better odds of living to 100	Link		
2	rs261332	(A;G)	Associated with higher HDL cholesterol	Link		
2	rs2707466	(A;A)	Stronger bones	Link	Link	
2	rs2908004	(T;T)	Stronger bones	Link	Link	
2	rs3914132	(C;T)	Lower otosclerosis risk	Link	Link	
2	rs763110	(C;T)	~0.80x reduced cancer risk	Link		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	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
1.5	rs1026732	(A;G)	0.70x risk for restless legs	Link	Link	
1.5	rs1063192	(C;T)	0.71x reduced risk of myocardial infarction	Link		

Mag.	Identifier	Genotype	Summary	GnomAD	GetEvidence	ClinVar
1.5	rs11635424	(A;G)	0.70x risk for restless legs	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	rs3851179	(A;G)	0.85x decreased risk for Alzheimer's disease	Link	Link	
1.5	rs4149274	(C;T)	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	
1.4	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	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	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	rs11601907	(C;T)	Variant allele is designated benign in ClinVar	Link		Link
1	rs2494732	(T;T)	Lower odds of psychosis	Link	Link	
1	rs2546890	(G;G)	Lower risk of multiple sclerosis	Link		
1	rs2952768	(C;C)	Less drug dependence: decreased effectiveness o...	Link		Link
1	rs4148739	(A;G)	Possibly: inpatients more likely to remit on ce...	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	

3.2 Possibly Harmful Traits

Mag.	Identifier	Genotype	Summary	GnomAD	GetEvidence	ClinVar
3.8	rs5186	(C;C)	7.3x increased risk of hypertension	Link	Link	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	rs3738579	(T;T)	1.5x - 2x increased risk for cervical cancer: H...	Link		
3	rs4495487	(C;C)	2 - 4 fold increased odds of developing V617F-a...	Link		
2.6	rs4958847	(A;A)	2.6x increased risk for Crohn's disease	Link		
2.5	rs12803066	(A;G)	Increased risk of myopia	Link		
2.5	rs1421085	(C;T)	~1.3x increased obesity risk	Link	Link	Link
2.5	rs17696736	(G;G)	1.94x risk of type-1 diabetes	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	rs3780374	(A;G)	Substantially increased odds of developing V617...	Link		
2.5	rs4143094	(G;T)	Slightly (17%) higher risk of colorectal cancer...	Link		
2.5	rs5888	(C;T)	3x higher risk for age-related macular degenera...	Link		
2.5	rs6441286	(G;G)	3.08x chance of developing primary biliary cirr...	Link	Link	
2.5	rs891512	(A;G)	Higher blood pressure than G;G	Link		Link
2.3	rs7966230	(C;G)	Slightly lower levels of plasma VWF	Link		
2.2	rs2004640	(G;T)	1.4x increased risk for SLE	Link	Link	
2.1	rs10811661	(T;T)	1.2x increased risk for type-2 diabetes	Link	Link	
2.1	rs1360780	(T;T)	1.3x increased risk for depression	Link	Link	Link
2.1	rs17070145	(C;C)	Reduced memory abilities	Link		Link
2.1	rs17077540	(A;G)	1.6x major depressive disorder risk	Link		
2.1	rs17563	(C;C)	Risk for otosclerosis	Link	Link	Link
2.1	rs4402960	(T;T)	1.2x increased risk for type-2 diabetes: 1.5x r...	Link	Link	Link
2.1	rs944289	(C;T)	1.3x increased thyroid cancer risk	Link	Link	
2	rs10086908	(C;T)	1.7x increased risk for prostate cancer	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	rs10488631	(C;T)	2x increased risk of developing SLE; 1.6x incre...	Link	Link	
2	rs1050631	(C;T)	Mean Survival Time of 25 months for esophageal ...	Link		
2	rs10889677	(C;C)	Baseline (average) risk for certain autoimmune ...	Link	Link	
2	rs10984447	(A;G)	1.17x increased risk for multiple sclerosis	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	rs1265181	(C;G)	Increased risk for psoriasis	Link	Link	
2	rs13254738	(A;C)	1.18x prostate cancer risk	Link	Link	
2	rs1333048	(A;C)	1.3x increased coronary artery disease risk	Link		
2	rs16942	(G;G)	Very slightly increased breast cancer risk	Link	Link	Link
2	rs17228212	(C;T)	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	rs1800629	(A;G)	Complex; generally higher risk for certain dise...	Link	Link	Link
2	rs1800896	(A;G)	1.6x increased prostate cancer risk	Link		
2	rs2073963	(G;T)	Increased risk of baldness	Link		
2	rs2143340	(C;T)	Increased risk of dyslexia and poor reading per...	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	rs2274223	(A;G)	1.5x increased risk for stomach and esophageal ...	Link	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		

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	rs241448	(C;T)	1.51x increased risk for Alzheimer's	Link		Link
2	rs25487	(A;G)	2x higher risk for skin cancer; possibly other ...	Link	Link	Link
2	rs268	(A;G)	3X increased risk for venous thromboembolism	Link	Link	Link
2	rs3212227	(C;C)	Significantly increased risk of developing cerv...	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	rs3738919	(A;C)	1.94x risk of developing rheumatoid arthritis	Link		
2	rs3746444	(C;T)	~1.2x increased risk for cancer	Link		
2	rs3775948	(G;G)	Slightly higher risk for gout	Link		
2	rs3793784	(C;G)	1.5x risk for ARMD	Link	Link	Link
2	rs4444903	(A;G)	3.5x risk of hep-cancer in cirrhosis patients; ...	Link		Link
2	rs4464148	(C;C)	1.35x increased risk for colorectal cancer	Link		
2	rs4633	(C;T)	Higher risk for endometrial cancer	Link	Link	Link
2	rs493258	(A;G)	1.15x risk of Age Related Macular Degeneration	Link		
2	rs520354	(A;A)	Increased risk in men for biliary conditions	Link		
2	rs629242	(C;T)	Somewhat higher risk for prostate cancer	Link		
2	rs6457617	(C;T)	2.3x risk of rheumatoid arthritis	Link	Link	
2	rs6603272	(G;T)	2.74x increased risk of developing schizophreni...	Link		
2	rs6807362	(C;C)	Increased autism risk	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;T)	Increased risk of hypertension	Link	Link	Link
2	rs7216389	(T;T)	1.5x increased risk for Childhood Asthma.	Link	Link	
2	rs7442295	(A;A)	~4x higher risk for hyperuracemia	Link	Link	Link
2	rs744373	(C;C)	1.17x risk of Alzheimer's	Link		
2	rs763361	(T;T)	Increased risk for multiple autoimmune diseases...	Link	Link	
2	rs828907	(T;T)	Increased risk of bladder cancer and 2x risk of...	Link		
2	rs854560	(A;T)	Higher risk for heart disease: diabetic retinop...	Link	Link	Link
2	rs9652490	(A;A)	~2x increased risk for Parkinson's disease: and...	Link	Link	
2	rs987525	(A;A)	6x increased risk for cleft lip	Link	Link	
2.0	rs1044396	(C;C)	Increased risk of Nicotine dependence among mal...	Link	Link	Link
2.0	rs4911414	(G;T)	2-4x higher risk of sun sensitivity if part of ...	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		
1.6	rs11523871	(A;C)	1.6x increased breast cancer risk for women ove...	Link	Link	
1.6	rs1537415	(C;G)	1.6x increased risk for periodontitis	Link	Link	
1.6	rs2736100	(G;G)	1.6x higher risk for glioma development	Link	Link	Link
1.6	rs2981745	(C;T)	1.6x increased risk for breast cancer in female...	Link		
1.5	rs10260404	(C;T)	1.20x risk of developing ALS	Link	Link	
1.5	rs10492519	(A;G)	Slightly increased risk of developing prostate ...	Link		
1.5	rs10784502	(T;T)	Less intracranial volume?	Link		
1.5	rs10883365	(A;G)	1.2x increased risk for developing Crohn's dise...	Link	Link	
1.5	rs11171739	(C;T)	1.34x risk of developing Type-1 diabetes	Link	Link	
1.5	rs1154155	(G;T)	1.94x increased risk for narcolepsy	Link	Link	
1.5	rs12210050	(C;T)	Slightly higher risk for basal cell carcinoma	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	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	rs17221417	(C;G)	1.3x higher risk for Crohn's disease	Link	Link	

Mag.	Identifier	Genotype	Summary	GnomAD	GetEvidence	ClinVar
1.5	rs1801274	(T;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	rs2254958	(C;C)	1.61x reported increased risk for Alzheimer's; ...	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	rs2305089	(T;T)	Higher risk for chordoma reported in one study;...	Link	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	rs309375	(T;T)	Larger mosquito bites	Link		
1.5	rs3764880	(A;G)	Possible 1.2 - 1.8x increased tuberculosis susc...	Link	Link	
1.5	rs3825776	(A;G)	1.3x increased risk for ALS	Link	Link	
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	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	rs4845618	(G;T)	1.7x increased melanoma risk	Link		
1.5	rs4939827	(T;T)	1x risk for colorectal cancer	Link	Link	Link
1.5	rs5746059	(A;G)	Slightly higher fat mass	Link		
1.5	rs6498169	(A;G)	1.14x risk of multiple sclerosis	Link	Link	
1.5	rs6601764	(C;T)	1.16x increased risk of developing Crohn's dise...	Link	Link	
1.5	rs6656401	(A;G)	1.18x increased risk for late-onset Alzheimer...	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	rs763035	(C;T)	1.2x increased risk for rosacea	Link		
1.5	rs7774434	(C;T)	Slightly increased risk of developing primary b...	Link		
1.5	rs7850258	(G;G)	Slightly higher odds of developing primary hypo...	Link		
1.5	rs872071	(A;G)	~1.5x increased risk for chronic lymphocytic le...	Link	Link	
1.5	rs9561778	(G;T)	~2x increased risk of adverse drug reactions fr...	Link	Link	
1.5	rs9642880	(G;T)	1.2x increased bladder cancer risk	Link	Link	
1.5	rs995030	(G;G)	Non-protective against testicular cancer	Link	Link	
1.4	rs10865331	(A;A)	1.4x higher risk for ankylosing spondylitis	Link		
1.4	rs1126497	(T;T)	1.4x increased risk for breast cancer	Link	Link	Link
1.4	rs1545843	(A;A)	1.4x increased risk for depression (for those u...	Link		
1.4	rs1893217	(C;T)	Slightly increased (1.4x) risk for Crohn's dise...	Link	Link	
1.4	rs4959039	(A;G)	1.4x higher risk for multiple sclerosis	Link		
1.4	rs498872	(T;T)	1.4x higher risk for glioma development	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	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	rs10947262	(C;C)	1.3x increased risk for osteoarthritis	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	rs13361189	(C;T)	1.3x increased risk for Crohn's disease	Link	Link	
1.3	rs1375144	(C;T)	1.32x increased risk of developing bipolar diso...	Link		
1.3	rs16847548	(C;T)	1.3x increased risk for sudden cardiac death in...	Link		
1.3	rs2024513	(A;G)	1.3x higher risk for schizophrenia (among Han C...	Link		
1.3	rs2542151	(G;T)	1.3x risk for Crohn's; 1.3x for T1D	Link	Link	
1.3	rs34330	(T;T)	1.2x higher breast cancer risk; 1.3x higher ris...	Link		Link
1.3	rs4712653	(C;T)	Very slightly (~1.3x) increased risk for neurob...	Link		
1.3	rs501120	(A;G)	1.3x increased risk for heart disease	Link	Link	

Mag.	Identifier	Genotype	Summary	GnomAD	GetEvidence	ClinVar
1.3	rs7234029	(A;G)	Slightly increased (1.36x) risk for Crohn's dis...	Link		
1.25	rs13387042	(A;A)	1.24x increased risk for breast cancer	Link	Link	
1.2	rs11842874	(A;G)	+17% increased risk for osteoarthritis	Link		
1.2	rs12050604	(A;A)	Slightly increased risk for lung cancer	Link		
1.2	rs2072590	(G;T)	1.2x increased risk for ovarian cancer	Link		
1.2	rs3131296	(A;G)	1.2x increased risk for schizophrenia	Link	Link	
1.2	rs419788	(A;G)	2.0x risk for lupus	Link		
1.2	rs4324715	(C;C)	>1.5x increased testicular cancer risk for men	Link		
1.2	rs4795067	(A;G)	Slight increase in risk for psoriatic arthritis...	Link		
1.2	rs4977756	(A;G)	1.39x higher risk for glioma development	Link	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	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	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	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	rs3818361	(C;T)	1.15x increased risk for late-onset Alzheimer's...	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		
1.1	rs889312	(A;C)	Very slightly higher risk for breast cancer	Link	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	rs1010	(A;G)	1.75x risk of MI	Link	Link	
1	rs11206244	(C;T)	Slight risk of decreased thyroid hormone metabo...	Link		
1	rs1143674	(A;G)	1.3x increased autism risk	Link		
1	rs1417066	(C;T)	Slightly increased risk of osteoarthritis	Link		
1	rs2273697	(A;G)	Adverse reaction more likely to carbamazepine i...	Link	Link	Link
1	rs3194051	(A;A)	>1.1x 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	rs6976	(C;T)	Slight risk of osteoarthritis	Link		
0.5	rs1566734	(G;T)	Somatic mutation: cancer associated	Link	Link	Link
0.1	rs2304256	(C;C)	1.6x increased risk for SLE	Link	Link	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		
0.1	rs601338	(A;G)	Susceptible to Norovirus infections	Link	Link	Link

3.3 Genosets (Multi-variant Phenotypes)

Magnitude	Identifier	Summary
6	gs216	2 copies of the APOE-Îµ4*allele*
4	gs145	Female
3.5	gs243	Increased risk of individuals with prostate can...
3.1	gs325	Reduced risk (0.64x) of breast cancer compared ...
3	gs273	Lowest risk (13% of white women) of Atrial Fibr...
2.5	gs100	Lactose intolerance risk
2.5	gs155	CYP3A5 non-expressor
2.5	gs242	Increased risk of individuals with prostate can...
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	gs154	NAT2 Slow metabolizer
2	gs159	CYP1A2 fast metabolizer
2	gs239	Reduced conversion of beta-carotene to retinol
2	gs244	2x increased risk for esophageal squamous cell ...
2	gs290	You might have two short form 5-HTTLPR.
2	gs313	Normal DPYD activity and thus 5-FU metabolism p...
2	gs315	Reduced risk of PD in a haplotype with the C2 v...
1.7	gs233	Normal pain sensitivity; APS/APS: LPS/APS: and ...
1.5	gs186	HLA-B*5801ââ heterozygosity is possible: un...
1.5	gs230	Possible Alzheimer's disease-related haplotype
1.5	gs247	Parkinson's Disease Risk
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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