

# Genomics Report for PGP-UK4/uk740176

## 1 Summary

This is the genome report for participant PGP-UK4/uk740176 . It was produced using collaborative research tools, including SNPedia and GetEvidence. This summary shows an overview of all the variants which were found in the genome for this individual. They have been compared with a reference genome.

This report was generated automatically and is not clinically approved. It is provided for personal and research purposes only.

This document contains hyperlinks, shown in grey, that will take you to external websites where you can find more detailed explanations. Some of the technical terms are also explained in more detail in the [Ensembl Glossary](#). We would welcome your feedback about this report, for example, if you would like more information about anything or if any of the links have become inactive. You can contact us on: [pgp-uk@ucl.ac.uk](mailto:pgp-uk@ucl.ac.uk).

This summary shows an overview of all the variants which were found in the genome for this individual. The "variants remaining after filtering" refers to any differences in the DNA identified when compared to the reference genome. Of these, the majority will have already been found in some other sequenced individual and put on a database (existing variants) while others have not yet been annotated (novel variants).

"Overlapped genes" refers to the number of times where a variant was found in a region of the genome containing a gene. "Exon" refers to the part of the gene which goes on to form a protein, and variants in this part of the gene are more likely to cause changes in the shape of the protein. Upstream, downstream, intronic and intergenic variants are more likely to alter the regulation of that gene but will not change the protein itself.

A transcript for a protein-coding gene can include the exons, introns and other gene features that are transcribed and important for gene function but might not be translated into the final protein. Not all transcripts are for protein-coding genes, with many containing non-coding RNAs that can be overlapping other genes, in introns or in intergenic regions. The diagram in Figure 1 is a simplification of the usual gene structure.

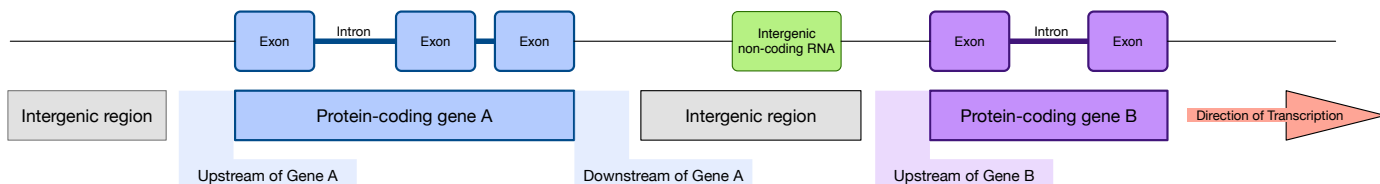


Figure 1: Diagram of gene structure indicating locations of potential variants

Feature	Count
Lines of input read	4277274
Variants remaining after filtering	4248448
Novel / existing variants	119803 (2.8%) / 4128645 (97.2%)
Overlapped genes	54789
Overlapped transcripts	64677
Overlapped regulatory features	216623

Table 1: Variant calling summary

There are several different types of genomic variants. The most common are single nucleotide variants (SNV) that correspond to the change of a single nucleotide in the DNA. Other variant types include insertions, where the DNA in the individual is longer than the reference sequence due to the insertion of one or more nucleotides; and deletions, where a few nucleotides are missing compared to the reference sequence.

Some of these changes will have no effect on the protein, while some changes may alter the protein function to varying degrees. The PolyPhen analysis software attempts to quantify the effect each mutation will have on the protein function. This ranges from "benign" where no change to the protein function is expected, to "probably damaging" where it is predicted that the mutation will affect protein function. It is nevertheless important to note that what is "damaging" for the protein is not necessarily damaging for the individual.

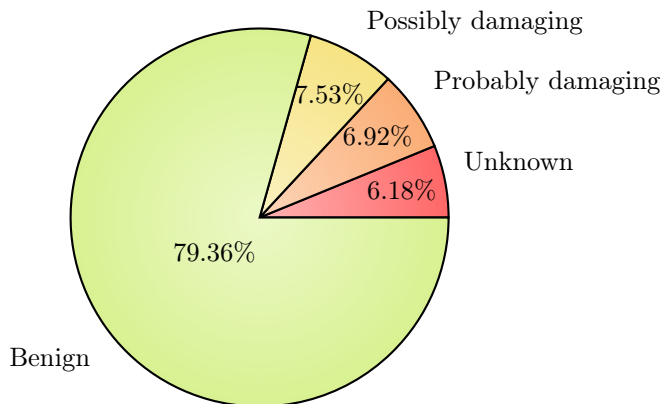


Figure 2: PolyPhen Summary

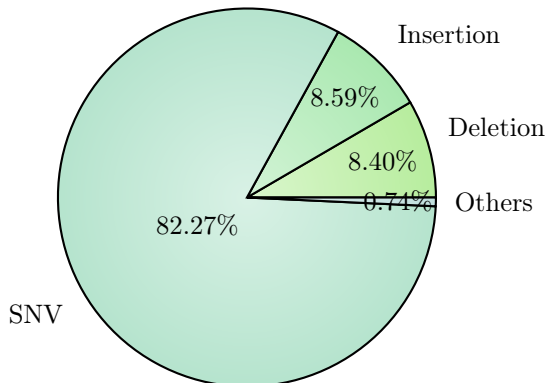


Figure 3: Variant Class

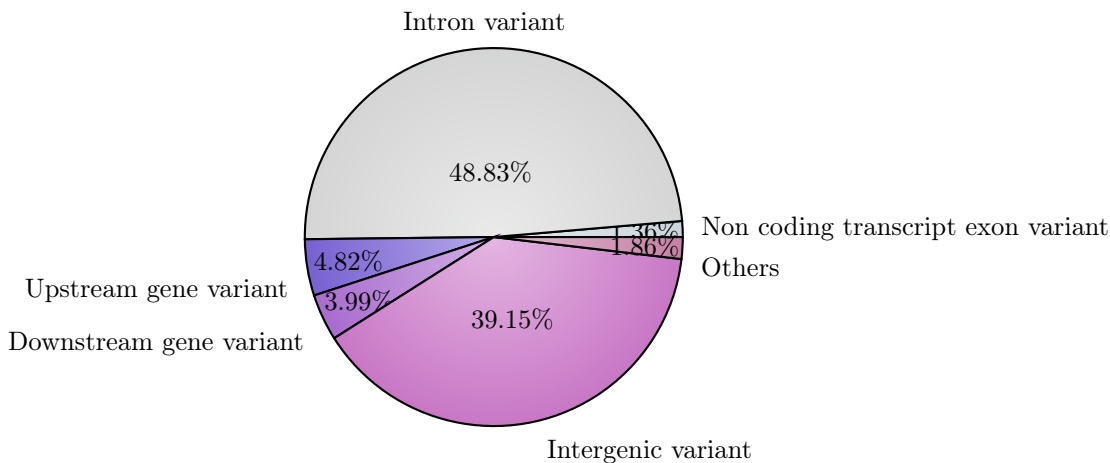


Figure 4: Consequence type

## 2 Ancestry

This plot shows the distribution of the genomes of different populations. Data from several studies which used whole genome sequencing was used to see the relationships between the genomes of the populations. It shows how closely related certain populations are genetically: Groups which cluster closely are more genetically similar than groups which are further apart. The black star symbol shows where this PGP-UK participant sits in relation to other populations, indicating their ancestry and their most closely related populations according to genetic sequence.

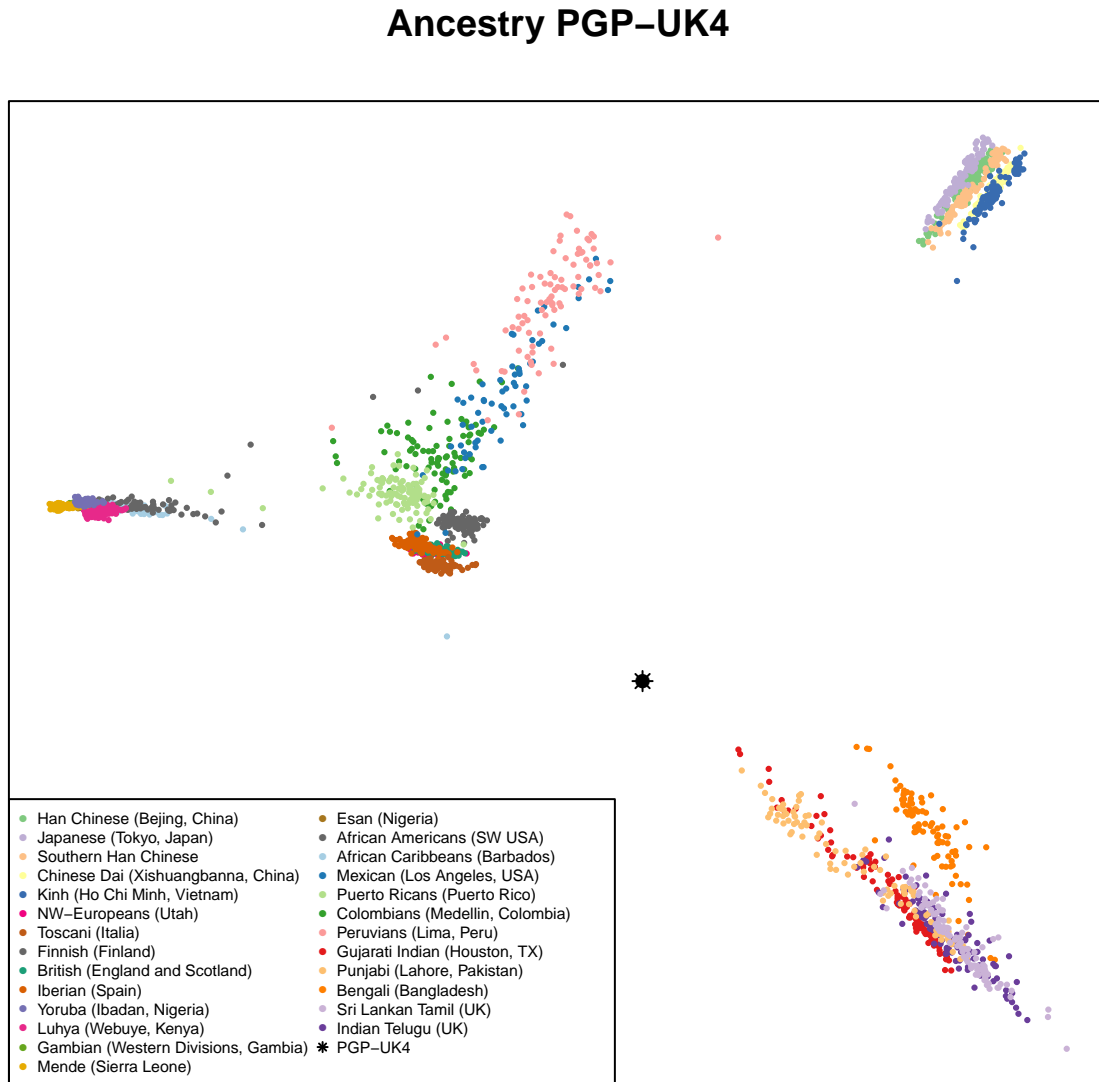


Figure 5: Ancestry Principal Component Analysis

### 3 Traits (based on SNPedia information)

Existing research has associated many variants with phenotypic traits, some of which can be perceived as beneficial while others appear to have a harmful effect. Some traits are complex and can be affected by several variants. It is likely that some of these would confer a higher risk while others a lower risk of trait manifestation. These can not be combined linearly to produce an actual risk of disease.

It is important to note that in most cases genomic data is probabilistic, not deterministic- i.e. having a genetic predisposition for a disease is not a diagnosis; rather, it shows an increased likelihood of developing that disease. Also, one person can have both potentially beneficial and harmful variants in the same gene, or associated with the same disease.

Some variants can also affect certain populations more, or will only affect a particular gender. For example, a variant for higher risk of endometriosis in the sequence of a male will not directly affect that person, but can be passed on to descendants.

While many traits are the result of a unique variant, many are the combination of several variants throughout the genome. In SNPedia, these are called *genosets*. These can integrate some of the information already present in the single variant tables, or be the combination of variants that have no phenotypic effect on their own, but contribute to a trait when together.

The variants in the following tables are sorted by magnitude. This is an subjective measure defined in SNPedia to highlight the perceived importance of the genotype described. At the moment this scale goes from 0 to 10. You can read more about it by visiting their explanatory [webpage](#).

As our knowledge grows, the interpretation of the effect of certain variants might change. Clicking on the links in the genome report tables will take you to websites containing more information about each variant.

- Possibly Beneficial Traits

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
2.1	rs2511989	(A;G)	0.63x decreased age-related macular degeneratio...		<a href="#">Link</a>	
2	rs10468017	(C;T)	Associated with higher HDL cholesterol		<a href="#">Link</a>	
2	rs10504861	(A;G)	Reduced risk of migraine without aura			
2	rs11045585	(A;A)	24% chance (lower than average) of docetaxel-in...		<a href="#">Link</a>	
2	rs1544410	(G;G)	Decreased risk of low bone mineral density diso...		<a href="#">Link</a>	
2	rs1799884	(G;G)	Mothers have typical Birth-Weight babies. Sligh...			
2	rs1864163	(G;G)	Associated with higher HDL cholesterol		<a href="#">Link</a>	
2	rs2073963	(T;T)	Reduced risk of baldness			
2	rs3738579	(C;T)	0.5x decreased risk for cervical cancer: HNSCC:...			
2	rs4143094	(G;G)	No increased risk of colorectal cancer correlat...			
2	rs4149268	(G;G)	Associated with higher HDL cholesterol		<a href="#">Link</a>	
2	rs6807362	(G;G)	Decreased autism risk	<a href="#">Link</a>	<a href="#">Link</a>	
2	rs6855911	(A;G)	0.62x decreased risk for gout		<a href="#">Link</a>	
2	rs763110	(C;T)	~0.80x reduced cancer risk			<a href="#">Link</a>
2	rs7776725	(T;T)	Stronger bones		<a href="#">Link</a>	
2	rs801114	(T;T)	0.78x decreased Basal Cell Carcinoma risk.		<a href="#">Link</a>	
2	rs925391	(C;T)	Lower odds of going bald			
1.5	rs1050631	(C;C)	Mean Survival Time of 32 months for esophageal ...	<a href="#">Link</a>		
1.5	rs11136000	(T;T)	0.84x decreased risk for Alzheimer's disease		<a href="#">Link</a>	
1.5	rs3790844	(C;T)	Slightly reduced risk (0.77x) for pancreatic ca...			
1.5	rs3851179	(A;G)	0.85x decreased risk for Alzheimer's disease		<a href="#">Link</a>	
1.5	rs4149274	(C;C)	Associated with higher HDL (good) cholesterol.			
1.5	rs4939883	(C;C)	Associated with higher HDL cholesterol		<a href="#">Link</a>	
1.5	rs610932	(A;A)	A allele associated with reduced risk of Alzhei...			
1.5	rs6427528	(A;G)	For rheumatoid arthritis patients: better respo...			
1.5	rs729302	(A;C)	0.89x decreased risk of developing rheumatoid a...			
1.3	rs2361502	(C;C)	Possible higher levels of serum bilirubin and l...			
1.2	rs4320932	(A;G)	0.87x decreased risk for ovarian cancer			

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
1.1	rs10166942	(C;T)	0.85x lower risk for migraines			
1.1	rs11172113	(C;T)	0.9x lower risk for migraines			
1.1	rs2293347	(G;G)	Among NSCLC patients: better Gefitinib response...	Link		
1.1	rs7568369	(T;T)	0.90x reduced risk of obesity			
1	rs11601907	(C;T)	Variant allele is designated benign in ClinVar	Link		Link
1	rs182549	(C;T)	Can digest milk.			Link
1	rs2351299	(G;T)	Possible reduced risk of Autism			
1	rs2952768	(C;T)	Slightly less drug dependence: decreased effect...			
1	rs4939827	(C;T)	0.86x decreased risk for colorectal cancer		Link	
1	rs7850258	(A;A)	Slightly lower odds of developing primary hypot...			
1	rs800292	(C;T)	1% decreased risk of macular degeneration	Link	Link	Link
0.1	rs891512	(G;G)	Lower blood pressure than those with an A allel...	Link		
0	rs1047781	(A;A)	ABH blood group "Secretor" status if Japanese	Link	Link	Link
0	rs10897346	(C;C)	If depressed: 2.6x more likely to not respond t...			
0	rs1126742	(T;T)	Higher hypertension risk	Link	Link	
0	rs12252	(T;T)	More resistant to influenza	Link		Link
0	rs12593929	(A;A)	Blue eye color more likely			
0	rs16990018	(A;A)	PrP Codon 171 Asn - Non-pathogenic variant	Link		Link
0	rs17244841	(A;A)	More responsive to statin treatment		Link	
0	rs1799782	(C;C)	Lower risk for skin cancer	Link	Link	
0	rs1799945	(C;C)	Not a H63D hemochromatosis carrier.	Link	Link	Link
0	rs1800562	(G;G)	Not a C282Y hemochromatosis carrier.	Link	Link	Link
0	rs2240203	(A;A)	Blue eye color more likely			
0	rs242941	(G;G)	Better response to inhaled corticosteroid in pa...		Link	
0	rs28933385	(G;G)	Prion protein Codon 200 (E) - Non pathogenic va...			Link
0	rs312481	(C;C)	Better response to certain calcium channel bloc...			
0	rs403016	(C;C)	2x risk for lupus		Link	
0	rs5746059	(A;A)	Slightly higher fat mass			
0	rs6259	(G;G)	Best inverse correlation between tea-drinking: ...	Link	Link	
0	rs74315403	(G;G)	PrP codon 178 (D) - non pathogenic variant			Link
0	rs7495174	(A;A)	Blue/gray eyes more likely		Link	
0	rs8028689	(T;T)	Blue eye color if part of blue eye color haplot...			
0	rs9394492	(C;C)	<0.76x risk for restless legs			
0	rs9951307	(A;G)	0.10 decreased risk for brain edema after a str...			

• Possibly Harmful Traits

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
3.2	rs2981582	(T;T)	1.7x higher risk of ER+ breast cancer		<a href="#">Link</a>	
3.1	rs1421085	(C;C)	~1.7x increased obesity risk		<a href="#">Link</a>	<a href="#">Link</a>
3	rs1121980	(T;T)	2.76x risk for obesity		<a href="#">Link</a>	
3	rs13266634	(C;C)	Increased risk for type-2 diabetes	<a href="#">Link</a>	<a href="#">Link</a>	<a href="#">Link</a>
3	rs2145418	(G;G)	9.2x increased thyroid cancer risk			
3	rs7754840	(C;C)	1.3x increased risk for type-2 diabetes		<a href="#">Link</a>	
2.5	rs1799971	(A;G)	Stronger cravings for alcohol. if alcoholic: na...	<a href="#">Link</a>	<a href="#">Link</a>	<a href="#">Link</a>
2.5	rs2241880	(C;C)	2x-3x increased risk for Crohn's disease in Cau...	<a href="#">Link</a>	<a href="#">Link</a>	<a href="#">Link</a>
2.5	rs339331	(T;T)	Prostate cancer risk			
2.5	rs3780374	(A;G)	Substantially increased odds of developing V617...			
2.5	rs5888	(C;T)	3x higher risk for age-related macular degenera...	<a href="#">Link</a>		
2.5	rs664143	(C;T)	Higher risk for number of cancers			
2.5	rs7574865	(G;T)	1.3x risk of rheumatoid arthritis; 1.55x risk o...		<a href="#">Link</a>	<a href="#">Link</a>
2.3	rs7966230	(C;G)	Slightly lower levels of plasma VWF			
2.1	rs10811661	(T;T)	1.2x increased risk for type-2 diabetes		<a href="#">Link</a>	
2.1	rs1219648	(G;G)	1.64x risk for breast cancer		<a href="#">Link</a>	
2.1	rs1585215	(G;G)	3.5x increased risk for Hodgkin lymphoma			
2.1	rs17563	(C;C)	Risk for otosclerosis	<a href="#">Link</a>	<a href="#">Link</a>	<a href="#">Link</a>
2.1	rs2231137	(A;G)	~1.5-3x increased risk for ischemic stroke	<a href="#">Link</a>	<a href="#">Link</a>	<a href="#">Link</a>
2.1	rs2254958	(C;T)	1.24x increased risk for Alzheimer's			
2.1	rs2294008	(T;T)	Increased risk of gastric and bladder cancer	<a href="#">Link</a>	<a href="#">Link</a>	
2.1	rs2420946	(T;T)	1.64x risk for breast cancer			
2.1	rs6457617	(T;T)	5.2x risk of rheumatoid arthritis		<a href="#">Link</a>	
2.1	rs7837688	(G;G)	1.7x increased risk for prostate cancer			
2.1	rs9272346	(A;G)	5.5x risk type-1 diabetes		<a href="#">Link</a>	
2.1	rs944289	(C;T)	1.3x increased thyroid cancer risk		<a href="#">Link</a>	
2	rs10096097	(G;G)	Increased Anorexia Nervosa risk			
2	rs1024611	(C;T)	Increased risk of exercise induced ischemia			<a href="#">Link</a>
2	rs10248420	(A;A)	7x less likely to respond to certain antidepres...		<a href="#">Link</a>	
2	rs1045642	(C;T)	Slower metaboliser for some drugs	<a href="#">Link</a>	<a href="#">Link</a>	
2	rs10883365	(G;G)	1.62x increased risk for developing Crohn's dis...		<a href="#">Link</a>	
2	rs10889677	(A;C)	1.5x increased risk for certain autoimmune dise...		<a href="#">Link</a>	
2	rs10984447	(A;G)	1.17x increased risk for multiple sclerosis		<a href="#">Link</a>	
2	rs11190870	(C;T)	Possibly increased risk of scoliosis			
2	rs1136287	(C;T)	1.5x increased risk of wet ARMD	<a href="#">Link</a>	<a href="#">Link</a>	
2	rs1160312	(A;G)	1.6x increased risk of Male Pattern Baldness.		<a href="#">Link</a>	
2	rs11983225	(T;T)	7x less likely to respond to certain antidepres...		<a href="#">Link</a>	
2	rs12431733	(T;T)	Increased risk of developing Parkinson's Diseas...		<a href="#">Link</a>	
2	rs12469063	(G;G)	Increased risk of developing restless legs synd...			
2	rs12567232	(A;G)	Increased risk for Crohn's Disease		<a href="#">Link</a>	
2	rs13254738	(A;C)	1.18x prostate cancer risk		<a href="#">Link</a>	
2	rs1333048	(A;C)	1.3x increased coronary artery disease risk			
2	rs16942	(G;G)	Very slightly increased breast cancer risk	<a href="#">Link</a>	<a href="#">Link</a>	<a href="#">Link</a>
2	rs16944	(G;G)	Increased risk of mental disorders		<a href="#">Link</a>	
2	rs17228212	(C;T)	1.26x increased risk for heart disease		<a href="#">Link</a>	
2	rs1734791	(A;T)	1.4x increased risk for lupus			
2	rs17435	(T;T)	1.4x increased risk for lupus			
2	rs17576	(A;G)	Higher risk for MI and lung cancer: and COPD in...	<a href="#">Link</a>	<a href="#">Link</a>	
2	rs1799966	(G;G)	Very slightly increased risk for breast cancer	<a href="#">Link</a>	<a href="#">Link</a>	<a href="#">Link</a>
2	rs1800896	(A;A)	1.8x increased prostate cancer risk			
2	rs1867277	(A;A)	2x increased risk for thyroid cancer			
2	rs1994090	(G;G)	Increased risk of developing Parkinson's Diseas...		<a href="#">Link</a>	
2	rs2075650	(A;G)	2x higher Alzheimer's risk	<a href="#">Link</a>	<a href="#">Link</a>	

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
2	rs2156921	(A;G)	1.29x increased risk for depression			
2	rs2201841	(C;T)	1.5x increased risk for Crohn's disease; 2x inc...		Link	
2	rs2235015	(G;G)	Somewhat less likely to respond to certain anti...	Link	Link	
2	rs2235040	(G;G)	7x less likely to respond to certain antidepres...	Link	Link	
2	rs2235067	(G;G)	7x less likely to respond to certain antidepres...			
2	rs2274223	(A;G)	1.5x increased risk for stomach and esophageal ...	Link	Link	
2	rs2286812	(T;T)	~4x higher risk for Fuchs' dystrophy: a corneal...			
2	rs2383206	(A;G)	1.4x increased risk for heart disease			
2	rs2383207	(A;G)	Increased risk for heart disease			
2	rs241448	(C;T)	1.51x increased risk for Alzheimer's	Link		Link
2	rs25487	(G;G)	2x higher risk for skin cancer; possibly other ...	Link	Link	
2	rs2707466	(G;G)	Weaker bones	Link	Link	
2	rs2736100	(T;T)	Higher risk of Interstitial lung disease: and t...		Link	
2	rs2908004	(C;C)	Weaker bones	Link	Link	
2	rs3184504	(C;T)	Increased risk for celiac disease	Link	Link	
2	rs358806	(C;C)	1.78x increased risk of developing Type-2 diabe...		Link	
2	rs3738919	(A;C)	1.94x risk of developing rheumatoid arthritis			
2	rs3775948	(G;G)	Slightly higher risk for gout			
2	rs4129148	(C;G)	3x risk of schizophrenia.		Link	
2	rs4148739	(A;A)	7x less likely to respond to certain antidepres...		Link	
2	rs4402960	(G;T)	1.2x increased risk for type-2 diabetes		Link	Link
2	rs4420638	(A;G)	~3x increased Alzheimer's risk; 1.4x increased ...		Link	Link
2	rs4444903	(A;G)	3.5x risk of hep-cancer in cirrhosis patients; ...			
2	rs4633	(C;T)	Higher risk for endometrial cancer	Link	Link	
2	rs493258	(G;G)	1.15x risk of Age Related Macular Degeneration			
2	rs4961	(G;T)	1.8x increased risk for high blood pressure	Link	Link	Link
2	rs520354	(A;G)	Increased risk in men for biliary conditions			
2	rs629242	(C;T)	Somewhat higher risk for prostate cancer			
2	rs6441286	(G;T)	1.54x chance of developing primary biliary cirr...		Link	
2	rs6498169	(A;A)	>1.14x risk of multiple sclerosis		Link	
2	rs657152	(T;T)	Multiple risks: see details		Link	
2	rs6601764	(C;C)	1.52x increased risk of developing Crohn's dise...		Link	
2	rs6897932	(C;C)	1.5x increased risk for multiple sclerosis	Link	Link	Link
2	rs6908425	(C;C)	1.95x increased risk of developing Crohn's dise...		Link	
2	rs6997709	(G;T)	1.2x higher risk for hypertension			
2	rs699	(C;T)	Increased risk of hypertension	Link	Link	Link
2	rs7216389	(T;T)	1.5x increased risk for Childhood Asthma.		Link	
2	rs738409	(C;G)	Increased liver fat: odds of alcoholic liver di...	Link	Link	
2	rs7442295	(A;A)	~4x higher risk for hyperuracemia		Link	
2	rs744373	(C;T)	1.17x risk of Alzheimer's			
2	rs7794745	(A;T)	Slightly increased risk for autism		Link	Link
2	rs7961152	(A;C)	1.2x higher risk for hypertension			
2	rs828907	(T;T)	Increased risk of bladder cancer and 2x risk of...			
2	rs9652490	(A;A)	~2x increased risk for Parkinson's disease: and...		Link	
2	rs965513	(A;A)	3.5x increased thyroid cancer risk		Link	
2	rs9954153	(G;T)	~2.5x higher risk for Fuchs' dystrophy: a corne...			
2.0	rs2305795	(A;A)	1.64x higher risk of narcolepsy compared to (G;...			Link
2.0	rs9642880	(T;T)	1.5x increased bladder cancer risk		Link	
1.8	rs37973	(A;G)	Among asthmatics: 1.5x more likely to show less...			Link
1.6	rs11523871	(A;C)	1.6x increased breast cancer risk for women ove...	Link	Link	
1.6	rs2981745	(C;T)	1.6x increased risk for breast cancer in female...			
1.6	rs3764880	(A;A)	1.2 - 1.8x increased tuberculosis risk	Link	Link	
1.5	rs10260404	(C;T)	1.20x risk of developing ALS		Link	
1.5	rs10757272	(C;T)	1.30x increased risk for Coronary artery diseas...			
1.5	rs10859871	(A;C)	Slight (~1.2x) increase in endometriosis risk			

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
1.5	rs10980705	(C;T)	2.3x increased risk for knee osteoarthritis			
1.5	rs12037606	(A;G)	1.22x risk of developing Crohn's disease			
1.5	rs12498742	(A;A)	1.25 increased risk for gout			
1.5	rs13149290	(C;C)	Slightly increased risk of developing prostate ...			
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			
1.5	rs144848	(G;T)	Very slightly increased breast cancer risk	Link	Link	Link
1.5	rs17221417	(C;G)	1.3x higher risk for Crohn's disease		Link	
1.5	rs17756311	(A;A)	Possible higher risk of childhood acute lymphob...			
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	rs2007153	(G;G)	Increased risk of schizophrenia in limited stud...			
1.5	rs2280714	(A;G)	1.4x increased risk of SLE			
1.5	rs2697962	(A;G)	Slightly increased risk of developing Parkinson...			
1.5	rs2736990	(C;T)	Slightly increased risk of developing Parkinson...		Link	
1.5	rs27388	(A;G)	Slightly increased risk of developing schizophr...			
1.5	rs2881766	(G;T)	Slightly increased risk for pregnancy-induced h...			
1.5	rs3087243	(A;G)	Increased risk for auto-immune diseases		Link	
1.5	rs3212227	(A;A)	1.43x increased risk of developing psoriasis an...			
1.5	rs3814570	(C;T)	1.3x increased risk for Crohn's disease with il...			
1.5	rs393152	(A;A)	Increased risk of both PD and AD	Link	Link	
1.5	rs401681	(C;C)	~1.2x increased risk for several types of cance...		Link	
1.5	rs4027132	(A;G)	1.39x increased risk of developing bipolar diso...			
1.5	rs4506565	(A;T)	1.4x increased risk for type-2 diabetes		Link	
1.5	rs464049	(C;T)	Increased risk of schizophrenia in limited stud...			
1.5	rs4656461	(A;G)	1.5x increased risk for open angle glaucoma			
1.5	rs4785763	(A;C)	1.5x higher risk for melanoma		Link	
1.5	rs486907	(A;G)	1.5x increased prostate cancer risk	Link	Link	Link
1.5	rs4979462	(C;T)	Slightly increased risk of developing primary b...			
1.5	rs619203	(C;G)	Increases susceptibility to Myocardial Infarcti...	Link	Link	
1.5	rs6435862	(G;T)	1.7x higher risk of aggressive neuroblastoma		Link	
1.5	rs6532197	(A;G)	Slightly increased risk of developing Parkinson...		Link	
1.5	rs6896702	(C;T)	Slightly increased risk of developing Parkinson...			
1.5	rs699473	(C;C)	~1.5x increased brain tumor risk			
1.5	rs7341475	(G;G)	1.58x increased schizophrenia risk for women		Link	
1.5	rs7774434	(C;T)	Slightly increased risk of developing primary b...			
1.5	rs807701	(C;T)	Slightly increased dyslexia risk			
1.5	rs872071	(G;G)	~1.5x increased risk for chronic lymphocytic le...		Link	
1.5	rs9303277	(C;T)	1.46x Slightly increased risk of developing pri...			
1.5	rs995030	(G;G)	Non-protective against testicular cancer		Link	
1.4	rs1126497	(C;T)	1.4x increased risk for breast cancer	Link	Link	Link
1.4	rs2046210	(C;T)	1.4x increased breast cancer risk		Link	
1.4	rs2230201	(A;G)	1.4x risk of lupus	Link		
1.4	rs3131296	(G;G)	1.4x increased risk for schizophrenia		Link	
1.4	rs4959039	(A;G)	1.4x higher risk for multiple sclerosis			
1.4	rs6010620	(G;G)	1.4x higher risk for glioma development; but th...		Link	
1.4	rs8050136	(A;A)	1.4x increased risk for T2D in some populations...		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	rs10947262	(C;C)	1.3x increased risk for osteoarthritis			
1.3	rs13361189	(C;T)	1.3x increased risk for Crohn's disease		Link	
1.3	rs1434536	(A;G)	1.29x increased breast cancer risk			
1.3	rs16847548	(C;T)	1.3x increased risk for sudden cardiac death in...			
1.3	rs2059693	(C;T)	1.3x increased risk for testicular cancer			
1.3	rs34330	(C;T)	1.3x higher risk for endometrial cancer (in Chi...			



Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
1.3	rs356219	(A;G)	1.3x increased risk for Parkinson's disease			
1.3	rs4295627	(G;T)	1.3x higher risk for glioma development		Link	
1.3	rs4958847	(A;G)	1.3x increased risk for Crohn's disease			
1.3	rs501120	(A;G)	1.3x increased risk for heart disease		Link	
1.25	rs748404	(T;T)	Slightly increased risk (1.25) for lung cancer...		Link	
1.2	rs10865331	(A;G)	1.2x higher risk for ankylosing spondylitis			
1.2	rs143383	(C;T)	1.1x increased risk for osteoarthritis		Link	Link
1.2	rs1800693	(A;G)	Slight (1.2x) increase in risk for multiple scl...	Link	Link	Link
1.2	rs2056116	(A;G)	1.18x risk for breast cancer			
1.2	rs2076295	(G;T)	One copy of the risk allele (G): slightly incre...			
1.2	rs2814707	(A;G)	1.2x increased risk for ALS		Link	
1.2	rs3849942	(A;G)	1.2x increased risk for ALS		Link	
1.2	rs419788	(A;G)	2.0x risk for lupus	Link		
1.2	rs4795067	(A;G)	Slight increase in risk for psoriatic arthritis...			
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	
1.1	rs11110912	(C;C)	1.3x high blood pressure risk			
1.1	rs13387042	(A;G)	1.12x increased risk for breast cancer		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			
1.1	rs4977574	(A;G)	Some studies - but not others - report a slight...		Link	
1.1	rs6897876	(C;T)	Slight increase in testicular cancer risk for m...			
1.1	rs7171755	(A;G)	Very slight decrease in cortical thickness and ...			
1.1	rs7412	(C;T)	More likely to gain weight if taking olanzapine...	Link	Link	Link
1.1	rs889312	(A;C)	Very slightly higher risk for breast cancer		Link	
1.07	rs2291834	(C;C)	Very slightly higher risk for myocardial infarc...			
1	rs10761659	(A;G)	1.2x risk of Crohn's disease		Link	
1	rs12752888	(C;C)	Faster progression of mild cognitive impairment...			
1	rs2546890	(A;G)	Higher risk of multiple sclerosis			
1	rs3194051	(A;G)	1.12x risk of type-1 diabetes	Link	Link	Link
1	rs6932590	(T;T)	1.1x increased risk for schizophrenia		Link	
1	rs6974491	(A;G)	Higher risk of coeliac and/or inflammatory bowe...			
1	rs761100	(G;G)	Higher risk for dyslexia			
1.0	rs11246226	(A;A)	Increased risk of schizophrenia in limited stud...		Link	
0.1	rs601338	(G;G)	Susceptible to Norovirus infections	Link	Link	Link
0	rs1333040	(C;T)	1.24x increased myocardial infarction risk: 1.2...		Link	
0	rs4293393	(T;T)	1.25x Increased Risk of CKD for T allele in ...			
0	rs440446	(G;G)	Increased risk in men for biliary conditions	Link		
0	rs4714156	(C;C)	<0.61x risk for restless legs			
0	rs6314	(C;C)	Higher risk for RA	Link	Link	
0	rs7787082	(G;G)	7x less likely to respond to certain antidepres...		Link	
0	rs9298506	(A;A)	Higher Risk Aneurysm		Link	

- Genosets (Multi-variant Phenotypes)

Magnitude	Identifier	Summary
4	gs145	Female
3	gs273	Lowest risk (13% of white women) of Atrial Fibr...
2.5	gs155	CYP3A5 non-expressor
2.5	gs157	More stimulated by coffee
2.5	gs189	Probably APOE E2/E4: but maybe E1/E3. E1 is the...
2.5	gs281	Part of the 88% of the population claimed not t...
2.5	gs285	You will lose 2.5x as much weight on a low fat ...
2.4	gs297	Lower heart attack risk than average
2.3	gs255	Homozygous eye color haplotype #1
2.1	gs223	One copy of GCH1 variant associated with lower ...
2	gs101	Probably able to digest milk
2	gs103	Restless legs syndrome risk
2	gs140	NAT2 slow metabolizer
2	gs154	NAT2 Slow metabolizer
2	gs181	CYP2D6*2
2	gs188	One copy of APOE4 is possible: but not certain
1.5	gs247	Parkinson's Disease Risk
1.2	gs184	Able to taste bitterness.
1	gs163	CYP2D6*2A
0.1	gs233	Normal pain sensitivity

## 4 Report Metadata

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

Table 5: Analysis Pipeline Versions

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