# Genomics Report for PGP-UK9/uk85AA3B

#### 1 Summary

This is the genome report for participant PGP-UK9/uk85AA3B . 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 <u>personal and research purposes</u> 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. "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.

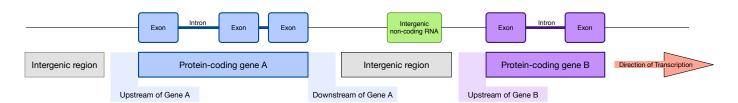


Figure 1: Diagram of gene structure indicating locations of potential variants

Feature	Count
Lines of input read	4202948
Variants remaining after filtering	4178750
Novel / existing variants	118816 (2.8%) / 4059934 (97.2%)
Overlapped genes	54735
Overlapped transcripts	64536
Overlapped regulatory features	213237

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.

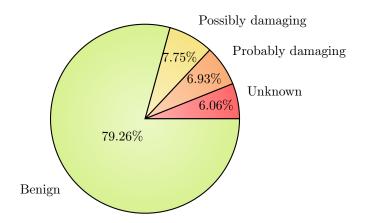


Figure 2: PolyPhen Summary

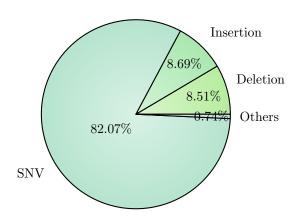


Figure 3: Variant Class

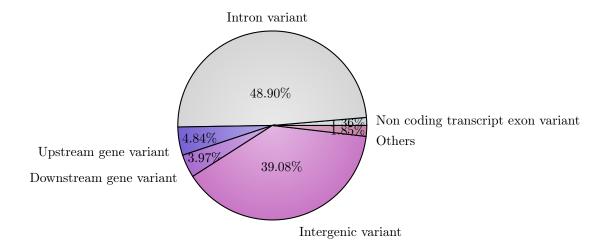


Figure 4: Consequence type

#### 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.

### **Ancestry PGP-UK9**

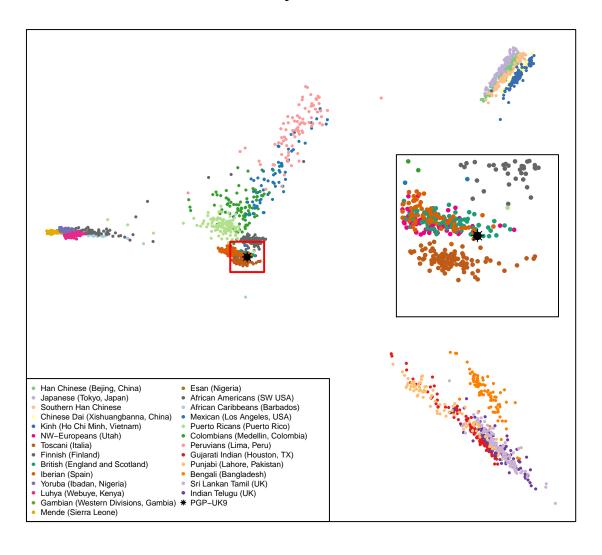


Figure 5: Ancestry Principal Component Analysis

### 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
2.4	rs2802288	(A;A)	Longer lifespan			
2.1	rs2511989	(A;G)	0.63x decreased age-related macular degeneratio		Link	
2.1	rs3775291	(A;G)	0.71x decreased risk for dry age related macula	Link	Link	Link
2	rs1012053	(A;C)	0.625x reduced risk of Bipolar Disorder.		Link	
2	rs10468017	(C;T)	Associated with higher HDL cholesterol		Link	
2	rs11045585	(A;A)	24% chance (lower than average) of docetaxel-in		Link	
2	rs17070145	(C;T)	Increased memory performance			Link
2	rs1864163	(G;G)	Associated with higher HDL cholesterol		Link	
2	rs2060793	(A;A)	Lower serum levels of vitamin D			
2	rs2243250	(C;T)	0.6x decreased risk for myocardial infarction i			
2	rs2542052	(C;C)	Better odds of living to 100			
2	rs261332	(A;G)	Associated with higher HDL cholesterol			
2	rs2764264	(C;C)	Greater odds of living to 95			
2	rs3738579	(C;T)	0.5x decreased risk for cervical cancer: HNSCC:			
2	rs3782179	(C;T)	3x lower odds of testicular cancer risk for men			
2	rs4307059	(C;C)	Reduced Autism risk		Link	
2	rs763110	(C;T)	~0.80x reduced cancer risk			Link
2	rs800292	(T;T)	5% decreased risk of macular degeneration	Link	Link	Link
1.5	rs1026732	(A;G)	0.70x risk for restless legs		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			
1.5	rs11136000	(C;T)	0.84x decreased risk for Alzheimer's disease		Link	
1.5	rs11465804	(G;T)	0.68x lower risk for spondylitis	Link	Link	
1.5	rs11635424	(A;G)	0.70x risk for restless legs		Link	
1.5	rs12593813	(A;G)	0.71x risk for restless legs		Link	
1.5	rs3784709	(C;T)	0.71x risk of developing restless legs syndrome		Link	
1.5	rs3790844	(C;T)	Slightly reduced risk (0.77x) for pancreatic ca			
1.5	rs4489954	(G;T)	0.69x risk risk of developing restless legs syn		Link	

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
1.5	rs4939883	(C;T)	Associated with higher HDL cholesterol		Link	
1.5	rs5888	(C;C)	Higher HDL cholesterol but lower risk for age-r	Link		
1.5	rs6427528	(A;G)	For rheumatoid arthritis patients: better respo			
1.5	rs729302	(A;C)	0.89x decreased risk of developing rheumatoid a			
1.4	rs2294008	(C;C)	Lower risk of cancer	Link	Link	
1.25	rs10088218	(A;G)	0.76x decreased risk for ovarian cancer			
1.2	rs4686484	(G;G)	Slightly decreased risk for celiac disease			
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	rs2952768	(C;T)	Slightly less drug dependence: decreased effect			
1	rs4939827	(C;T)	0.86x decreased risk for colorectal cancer		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	rs28933385	(G;G)	Prion protein Codon 200 (E) - Non pathogenic va			Link
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	rs8028689	(T;T)	Blue eye color if part of blue eye color haplot			
0	rs9951307	(A;G)	0.10 decreased risk for brain edema after a str			

## • Possibly Harmful Traits

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
4	rs5186	(C;C)	7.3x increased risk of hypertension	Link	Link	Link
3.2	rs1061235	(A;T)	26% risk of bad reaction to anti-epileptic carb			Link
3	rs1983132	(C;T)	2 - 3x higher prostate cancer risk if routinely			
3	rs2066844	(C;T)	3x higher risk for Crohn's disease	Link	Link	Link
3	rs6920220	(A;G)	1.2x risk Rheumatoid Arthritis		Link	
3	rs7754840	(C;G)	1.3x increased risk for type-2 diabetes		Link	
2.8	rs3780374	(A;A)	Substantially increased odds of developing V617			
2.7	rs10830963	(C;G)	Increased type-2 diabetes risk; higher gestatio		Link	
2.5	rs11190870	(T;T)	Possibly even more increased risk of scoliosis			
2.5	rs1121980	(C;T)	1.67x risk for obesity		Link	
2.5	rs12803066	(A;G)	Increased risk of myopia			
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
2.5	rs1800255	(A;A)	Increased risk for pelvic organ prolapse	Link	Link	Link
2.5	rs2254958	(C;C)	1.61x increased risk for Alzheimer's			
2.5	rs2943634	(C;C)	Higher risk of ischemic stroke		Link	
2.5	rs339331	(T;T)	Prostate cancer risk			
2.5	rs3738919	(C;C)	1.94x risk of developing rheumatoid arthritis			
2.5	rs4143094	(G;T)	Slightly (17%) higher risk of colorectal cancer			
2.5	rs613872	(G;G)	~20-30x higher risk for Fuchs' dystrophy: a cor			
2.5	rs891512	(A;G)	Higher blood pressure than G;G	Link		
2.4	rs7966230	(G;G)	Slightly lower levels of plasma VWF			
2.2	rs2231137	(G;G)	~1.5-3x increased risk for ischemic stroke	Link	Link	Link
2.1	rs10427255	(C;C)	Highest odds of photic sneeze reflex			
2.1	rs17563	(C;C)	Risk for otosclerosis	Link	Link	Link
2.1	rs2231142	(A;C)	1.74x increased gout risk; gefinitib takers 4x	Link	Link	Link
2.1	rs5751876	(T;T)	Significantly higher anxiety levels after moder	Link		
2.1	rs6457617	(T;T)	5.2x risk of rheumatoid arthritis		Link	
2.1	rs646776	(A;A)	1.2x risk of coronary artery disease		Link	
2.1	rs7837688	(G;G)	1.7x increased risk for prostate cancer			
2.1	rs795484	(A;G)	Increased morphine dose requirement and postope			
2.1	rs9272346	(A;G)	5.5x risk type-1 diabetes		Link	
2.1	rs944289	(C;T)	1.3x increased thyroid cancer risk		Link	
2	rs10086908	(C;T)	1.7x increased risk for prostate cancer			
2	rs1024611	(C;T)	Increased risk of exercise induced ischemia			Link
2	rs10248420	(A;A)	7x less likely to respond to certain antidepres		Link	
2	rs1045642	(C;T)	Slower metaboliser for some drugs	Link	Link	
2	rs1050152	(C;T)	2.1x increased risk of Crohn's disease	Link	Link	Link
2	rs10811661	(C;T)	1.2x increased risk for type-2 diabetes		Link	
2	rs10883365	(G;G)	1.62x increased risk for developing Crohn's dis		Link	
2	rs10889677	(C;C)	1x increased risk for certain autoimmune diseas		Link	
2	rs10984447	(A;A)	>1.17x increased risk for multiple sclerosis		Link	
2	rs1160312	(A;A)	1.6x increased risk of Male Pattern Baldness.		Link	
2	rs11983225	(T;T)	7x less likely to respond to certain antidepres		Link	
2	rs12431733	(T;T)	Increased risk of developing Parkinson's Diseas		Link	
2	rs13254738	(A;C)	1.18x prostate cancer risk		Link	
2	rs1333048	(A;C)	1.3x increased coronary artery disease risk		T 1	
2	rs1360780	(C;T)	1.3x increased risk for depression		Link	
2	rs16944	(G;G)	Increased risk of mental disorders		Link	
2	rs1734791	(A;A)	1.4x increased risk for lupus			
2	rs2073963	(G;T)	Increased risk of baldness			
2	rs2156921	(A;G)	1.29x increased risk for depression			
2	rs2201841	(T;T)	2.4x increased risk for Graves' disease		Link	

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
2	rs2230199	(C;G)	1.6x+ risk of ARMD	Link	Link	Link
2	rs2230201	(G;G)	>1.4x risk of lupus	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	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	(A;G)	2x higher risk for skin cancer; possibly other	Link	Link	
2	rs3025039	(C;T)	2.6x increased risk for ARMD			
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	rs3775948	(G;G)	Slightly higher risk for gout			
2	rs3793784	(C;G)	1.5x risk for ARMD		Link	Link
2	rs4148739	(A;A)	7x less likely to respond to certain antidepres		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	rs669	(G;G)	3.8x or higher increased risk for Alzheimers	Link	Link	Link
2	rs6807362	(C;C)	Increased autism risk	Link	Link	
2	rs6896702	(T;T)	Increased risk of developing Parkinson's Diseas			
2	rs6897932	(C;T)	1.3x increased risk for multiple sclerosis	Link	Link	Link
2	rs6997709	(G;G)	1.5x higher risk for hypertension			
2	rs699	(C;T)	Increased risk of hypertension	Link	Link	Link
2	rs700651	(G;G)	~1.56x increased risk of aneurysm		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)	<sup>~</sup> 4x higher risk for hyperuracemia	231111	Link	
2	rs744373	(C;C)	1.17x risk of Alzheimer's		231111	
2	rs763361	(T;T)	Increased risk for multiple autoimmune diseases	Link	Link	
2	rs7639618	(T;T)	1.45x increased osteoarthritis risk	Link		
2	rs7774434	(C;C)	Increased risk of developing primary biliary ci			
2	rs7794745	(T;T)	Slightly increased risk for autism		Link	Link
2	rs7807268	(C;C)	1.4x risk for Crohn's disease		Link	
2	rs7961152	(A;C)	1.2x higher risk for hypertension			
2	rs828907	(G;T)	Slightly increased risk of bladder cancer and 2			
2	rs9525638	(T;T)	Weaker bones			
2	rs9652490	(A;A)	~2x increased risk for Parkinson's disease: and		Link	
2	rs9954153	(G;G)	~5x higher risk for Fuchs' dystrophy: a corneal			
2.0	rs2305795	(A;A)	1.64x higher risk of narcolepsy compared to (G;			Link
2.0	rs4911414	(G;T)	2-4x higher risk of sun sensitivity if part of		Link	
1.9	rs7923837	(A;G)	1.6x risk for T2D			
1.8	rs37973	(A;G)	Among asthmatics: 1.5x more likely to show less			Link
1.8	rs4474514	(A;G)	3x increased testicular cancer risk for men		Link	
1.6	rs1537415	(C;G)	1.6x increased risk for periodontitis		Link	
1.6	rs2736100	(G;G)	1.6x higher risk for glioma development		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			
1.5	rs10980705	(C;T)	2.3x increased risk for knee osteoarthritis			
1.5	rs11171739	(C;T)	1.34x risk of developing Type-1 diabetes		Link	
1.5	rs12037606	(A;G)	1.22x risk of developing Crohn's disease			
1.5	rs12469063	(A;G)	Slightly increased risk of developing restless			
1.5	rs12498742	(A;A)	1.25 increased risk for gout			
1.5	rs13149290	(C;C)	Slightly increased risk of developing prostate			
1.5	rs1375144	(C;T)	1.32x increased risk of developing bipolar diso			

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
1.5	rs140701	(A;A)	Increased risk for anxiety disorders			
1.5	rs1571801	(A;A)	>1.36x risk for prostate cancer			
1.5	rs17221417	(C;G)	1.3x higher risk for Crohn's disease		Link	
1.5	rs1801274	(C;T)	Complex; generally greater risk for cancer prog	Link	Link	Link
1.5	rs1975197	(C;T)	1.3x increased risk of developing restless legs		Link	
1.5	rs1994090	(G;T)	Slightly increased risk of developing Parkinson		Link	
1.5	rs2076295	(G;G)	Slightly increased risk for pulmonary fibrosis			
1.5	rs2280714	(A;A)	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	rs28694718	(A;G)	2x higher risk for schizophrenia			
1.5	rs2881766	(G;T)	Slightly increased risk for pregnancy-induced h			
1.5	rs3212227	(A;A)	1.43x increased risk of developing psoriasis an			
1.5	rs3764880	(A;G)	Possible 1.2 - 1.8x increased tuberculosis susc	Link	Link	
1.5	rs3790565	(C;T)	Slightly increased risk of developing primary b			
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;T)	~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	rs419788	(A;A)	2.3x risk for lupus	Link		
1.5	rs4464148	(C;T)	1.10x increased risk for colorectal cancer			
1.5	rs4506565	(A;T)	1.4x increased risk for type-2 diabetes		Link	
1.5	rs4585	(T;T)	Slightly poorer (0.75x) response to metformin i			
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	rs4845618	(G;T)	1.7x increased melanoma risk			
1.5	rs486907	(A;G)	1.5x increased prostate cancer risk	Link	Link	Link
1.5	rs5219	(C;T)	1.3x increased risk for type-2 diabetes	Link	Link	Link
1.5	rs619203	(C;G)	Increases susceptibility to Myocardial Infarcti	Link	Link	
1.5	rs642961	(A;G)	1.68x increased risk of cleft lip		Link	
1.5	rs6498169	(A;G)	1.14x risk of multiple sclerosis		Link	
1.5	rs6532197	(A;G)	Slightly increased risk of developing Parkinson		Link	
1.5	rs6710341	(A;G)	Slightly increased risk of developing restless			
1.5	rs6908425	(C;T)	1.63x increased risk of developing Crohn's dise		Link	
1.5	rs7341475	(G;G)	1.58x increased schizophrenia risk for women		Link	
1.5	rs7454108	(C;T)	Single HLA-DQ8 haplotype			
1.5	rs7850258	(G;G)	Slightly higher odds of developing primary hypo			
1.5	rs872071	(A;G)	~1.5x increased risk for chronic lymphocytic le		Link	
1.5	rs9561778	(G;T)	~2x increased risk of adverse drug reactions fr		Link	
1.5	rs9642880	(G;T)	1.2x increased bladder cancer risk	T. 1	Link	T 1
1.4	rs1126497	(C;T)	1.4x increased risk for breast cancer	Link	Link	Link
1.4	rs1801157	(A;G)	1.4x higher risk for breast cancer		T . 1	
1.4	rs3131296	(G;G)	1.4x increased risk for schizophrenia		Link	
1.4	rs4959039	(A;G)	1.4x higher risk for multiple sclerosis	T · 1	T :1.	
1.34	rs17465637	(C;C)	1.34x higher risk for myocardial infarction	Link	Link	T · 1
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			
1.3	rs1434536 rs16847548	(A;G)	1.29x increased breast cancer risk 1.3x increased risk for sudden cardiac death in			
1.3	rs2295490	(C;T) (A;G)	1.3x increased risk for sudden cardiac death in  1.32x increased risk of early-onset type-2 diab	Link	Link	
1.3	rs34330	(C;T)	1.3x higher risk for endometrial cancer (in Chi	LIIIK	THIK	
1.3	rs356219	$(C, \Gamma)$ $(A;G)$	1.3x increased risk for Parkinson's disease			
1.3	rs4295627	(G;T)	1.3x higher risk for glioma development		Link	
1.0	154230021	(0,1)	1.01 mgner risk for ghoma development		LIIIK	

	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
1.25	rs13387042	(A;A)	1.24x increased risk for breast cancer		Link	
1.25	rs748404	(T;T)	Slightly increased risk (1.25) for lung cancer		Link	
1.2	rs11037909	(T;T)	1.47x type II diabetes risk	Link		
1.2	rs2056116	(A;G)	1.18x risk for breast cancer			
1.2	rs2072590	(G;T)	1.2x increased risk for ovarian cancer			
1.2	rs2252586	(A;G)	1.2x higher risk for glioma development			
1.2	rs2814707	(A;G)	1.2x increased risk for ALS		Link	
1.2	rs3740878	(A;A)	1.46x type II diabetes risk; common	Link		
1.2	rs3849942	(A;G)	1.2x increased risk for ALS		Link	
1.2	rs4795067	(A;G)	Slight increase in risk for psoriatic arthritis			
1.2	rs4977756	(A;G)	1.2x higher risk for glioma development		Link	
1.2	rs498872	(C;T)	1.2x higher risk for glioma development		Link	
1.2	rs6010620	(A;G)	1.2x higher risk for glioma development: 1.17x		Link	
1.2	rs6897876	(C;C)	Slight increase in testicular cancer risk for m			
1.2	rs8050136	(A;C)	1.2x increased risk for T2D in some populations		Link	
1.2	rs9858542	(A;G)	1.1x risk Crohn's Disease	Link	Link	
1.1	rs11110912	(C;C)	1.3x high blood pressure risk			
1.1	rs11650494	(A;G)	Slightly higher prostate cancer risk			
1.1	rs1344706	(G;T)	1.1x increased risk for schizophrenia		Link	
1.1	rs1800450	(A;G)	Mannose binding deficiency but of low clinical	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	rs4977574	(A;G)	Some studies - but not others - report a slight		Link	
1.1	rs7171755	(A;G)	Very slight decrease in cortical thickness and			
1.1	rs7412	(C;C)	More likely to gain weight if taking olanzapine	Link	Link	Link
1.1	rs889312	(A;C)	Very slightly higher risk for breast cancer		Link	
1.1	rs925391	(C;C)	More likely to go bald; common			
1.09	rs12050604	(A;C)	Very slightly increased risk for lung cancer			
1.07	rs2291834	(C;C)	Very slightly higher risk for myocardial infarc			
1	rs10504861	(G;G)	Major allele: normal risk of migraine			
1	rs10761659	(A;G)	1.2x risk of Crohn's disease		Link	
1	rs12718541	(A;A)	Nicotine dependence			
1	rs2546890	(A;A)	Higher risk of multiple sclerosis			
1	rs3194051	(A;A)	>1.1x risk of type-1 diabetes	Link	Link	Link
1	rs4986761	(C;T)	Very slightly increased risk (1.05) for breast	Link	Link	Link
1	rs6932590	(T;T)	1.1x increased risk for schizophrenia		Link	
1	rs987525	(A;C)	2.5x increased risk for cleft lip		Link	
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	rs1004819	(C;C)	1.5x risk of Crohn's disease		Link	
0	rs1333040	(C;T)	1.24x increased myocardial infarction risk: 1.2		Link	
0	rs3761418	(A;A)	1.3x increased risk for depression			
0	rs4293393	(T;T)	1.25x Increased Risk of CKD for T allele in			
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	

#### • Genosets (Multi-variant Phenotypes)

Magnitude	Identifier	Summary
4	gs145	Female
3.5	gs126	Poor warfarin metabolizer
3.3	gs162	CYP2C9 Poor Metabolizers
3.1	gs191	Problem metabolizing NSAIDs
3	gs241	Lighter green: brown or hazel eye color
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	gs256	Blue eyes
2.5	gs259	Homozygous for eye color haplotype $\#3$
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
2.1	gs223	One copy of GCH1 variant associated with lower
2	gs104	Restless legs syndrome risk
2	gs129	Unable to classify your ABO blood type
2	gs156	NAT2 Rapid metabolizer.
2	gs159	CYP1A2 fast metabolizer
2	gs246	APOE3/APOE3
1.8	gs1002	Mitochondrial Haplogroup H1
1.5	gs1001	Mitochondrial Haplogroup H
1.5	gs139	NAT2 intermediate metabolizer
1.5	gs185	The beta blocker metoprolol is effective with 1
1.5	gs247	Parkinson's Disease Risk
1.2	gs184	Able to taste bitterness.
1	gs182	CYP2D6*39
0.1	gs233	Normal pain sensitivity

# 4 Report Metadata

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

Table 5: Analysis Pipeline Versions

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