# Genomics Report for PGP-UK8/uk174659

## 1 Summary

This is the genome report for participant PGP-UK8/uk174659 . 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.

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.

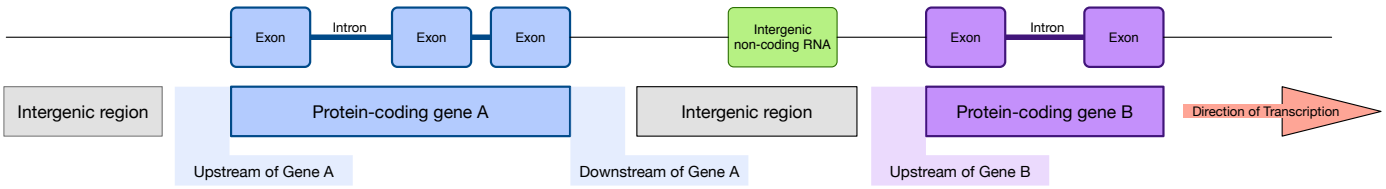


Figure 1: Diagram of gene structure indicating locations of potential variants

| Feature | Count |
|---|---|
| Lines of input read | 4207534 |
| Variants remaining after filtering | 4174671 |
| Novel / existing variants | 113893 (2.7%) / 4060778 (97.3%) |
| Overlapped genes | 55194 |
| Overlapped transcripts | 65014 |
| Overlapped regulatory features | 213504 |

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.

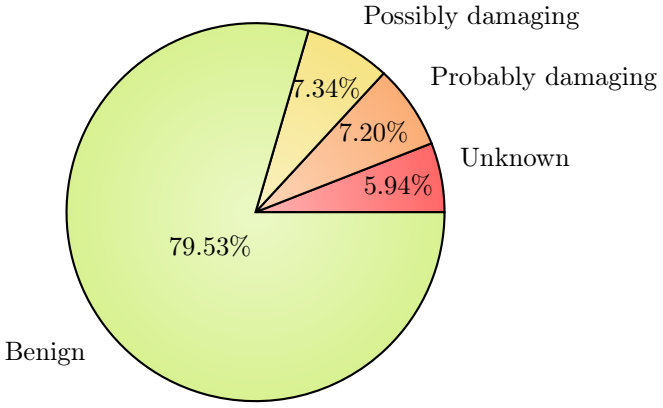


Figure 2: PolyPhen Summary

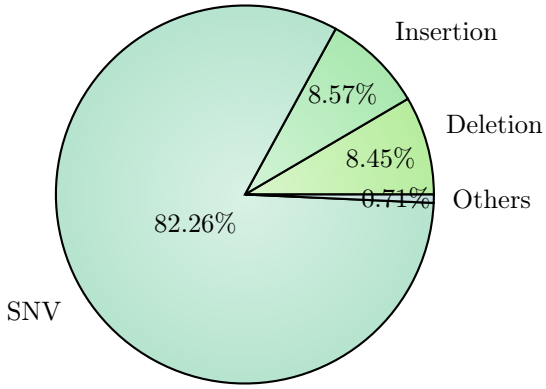


Figure 3: Variant Class

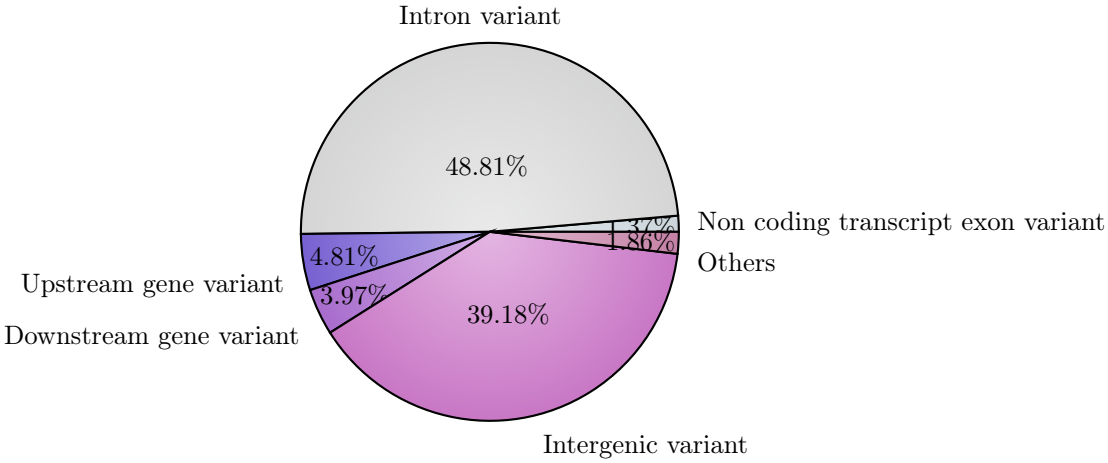


Figure 4: Consequence type

## 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.

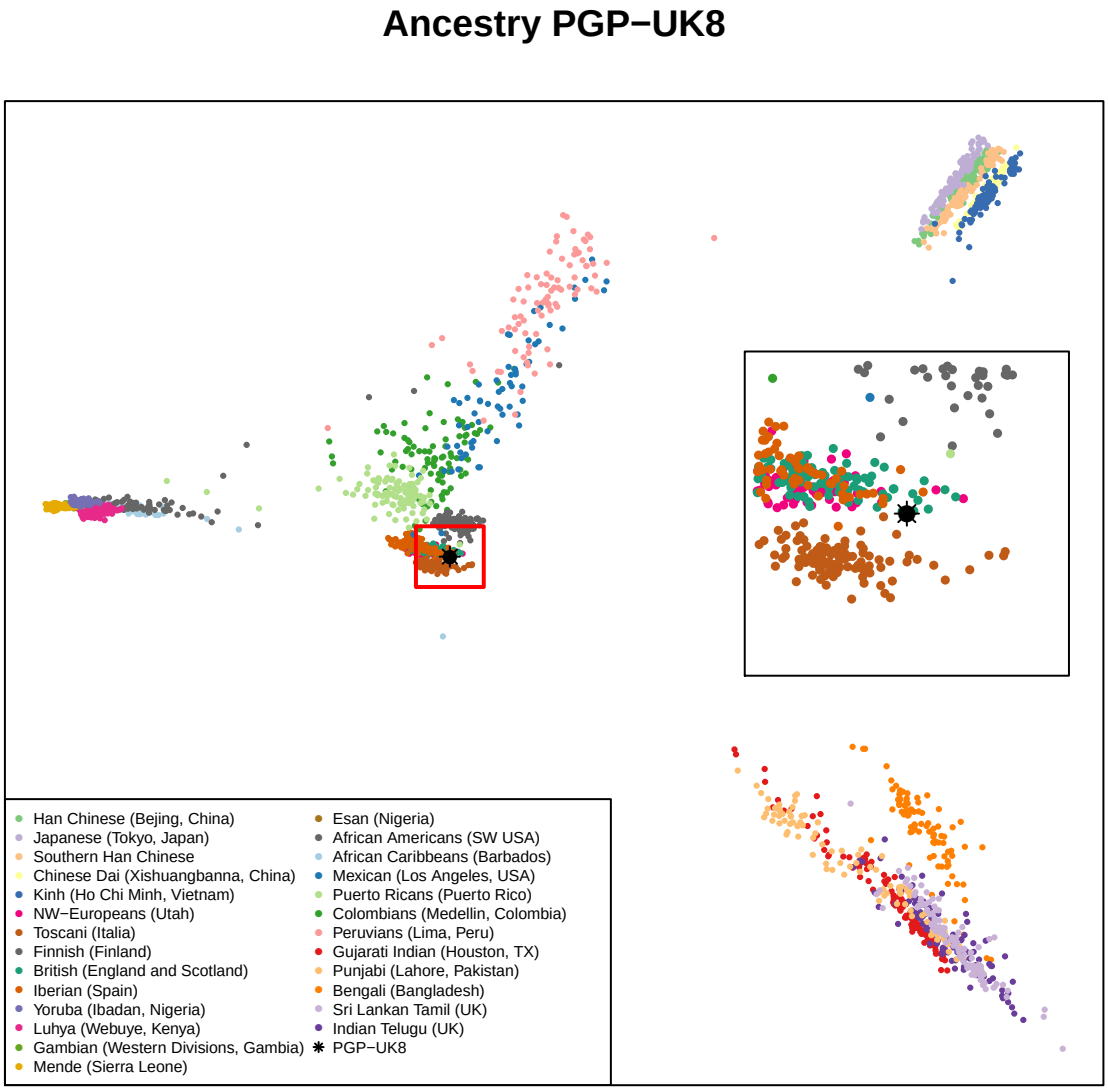


Figure 5: Ancestry Principal Component Analysis

# 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 |
|---|---|---|---|---|---|---|
| 2.2 | rs2511989 | (A;A) | 0.44x decreased age-related macular degeneratio... | | Link | |
| 2.1 | rs3775291 | (A;G) | 0.71x decreased risk for dry age related macula... | Link | Link | Link |
| 2.1 | rs738409 | (G;G) | Most common genotype; slightly less damage from... | Link | Link | |
| 2.1 | rs806380 | (G;G) | Uncommon. lowest odds of cannabis dependence | | | |
| 2 | rs10468017 | (T;T) | Associated with higher HDL cholesterol | | Link | |
| 2 | rs11045585 | (A;A) | 24% chance (lower than average) of docetaxel-in... | | Link | |
| 2 | rs12979860 | (C;C) | ~80% of such hepatitis C patients respond to tr... | | Link | |
| 2 | rs1544410 | (G;G) | Decreased risk of low bone mineral density diso... | | Link | |
| 2 | rs17070145 | (C;T) | Increased memory performance | | | Link |
| 2 | rs1799884 | (G;G) | Mothers have typical Birth-Weight babies. Sligh... | | | |
| 2 | rs1864163 | (A;G) | Associated with higher HDL cholesterol | | Link | |
| 2 | rs3738579 | (C;T) | 0.5x decreased risk for cervical cancer: HNSCC:... | | | |
| 2 | rs3819331 | (T;T) | Lower risk of autism | Link | | |
| 2 | rs4149268 | (G;G) | Associated with higher HDL cholesterol | | Link | |
| 2 | rs505922 | (T;T) | Blood type O | | Link | |
| 2 | rs6855911 | (G;G) | Rare: but 0.62x decreased risk for gout | | Link | |
| 2 | rs7216389 | (C;C) | 0.69x lower risk of Childhood Asthma. | | Link | |
| 2 | rs763110 | (C;T) | ~0.80x reduced cancer risk | | | Link |
| 2 | rs7776725 | (T;T) | Stronger bones | | Link | |
| 2 | rs8070723 | (A;G) | 0.18x reduced risk of developing progressive su... | | | |
| 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 | rs1165205 | (A;A) | 0.85x decreased gout risk | | Link | |
| 1.5 | rs16991615 | (A;G) | Slight increase (11 months) in avg age at menop... | Link | Link | |
| 1.5 | rs2007153 | (A;A) | Decreased risk of schizophrenia in limited stud... | | | |
| 1.5 | rs3851179 | (A;G) | 0.85x decreased risk for Alzheimer's disease | | Link | |
| 1.5 | rs4149274 | (C;C) | Associated with higher HDL (good) cholesterol. | | | |
| 1.5 | rs464049 | (C;C) | Decreased risk of schizophrenia in limited stud... | | | |

| Mag. | Identifier | Genotype | Summary | ExAC | GetEvidence | ClinVar |
|---|---|---|---|---|---|---|
| 1.5 | rs4939883 | (C;T) | Associated with higher HDL cholesterol | | Link | |
| 1.5 | rs9939609 | (T;T) | Lower risk of obesity and Type-2 diabetes | | Link | |
| 1.4 | rs2294008 | (C;C) | Lower risk of cancer | Link | Link | |
| 1.2 | rs13333226 | (G;G) | ~15-20% lower risk for hypertension or cardiova... | | | Link |
| 1.2 | rs4320932 | (A;G) | 0.87x decreased risk for ovarian cancer | | | |
| 1.2 | rs6048 | (G;G) | Slightly lower risk (10-20%) of deep vein throm... | Link | Link | Link |
| 1.1 | rs11172113 | (C;T) | 0.9x lower risk for migraines | | | |
| 1.1 | rs4988235 | (T;T) | Can digest milk | | | Link |
| 1 | rs182549 | (T;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;G) | Typical odds of developing primary hypothyroidi... | | | |
| 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 | rs1056836 | (G;G) | 0.3x decreased risk for prostate cancer | 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 | 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... | | | |

- Possibly Harmful Traits

| Mag. | Identifier | Genotype | Summary | ExAC | GetEvidence | ClinVar |
|---|---|---|---|---|---|---|
| 3 | rs1021737 | (T;T) | Significantly higher plasma total homocysteine ... | Link | Link | 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... | | | |
| 3 | rs2981582 | (C;T) | 1.3x higher risk of ER+ breast cancer | | Link | |
| 3 | rs4244285 | (A;G) | Poorer metabolizer of several popular medicines... | Link | Link | Link |
| 3 | rs75193786 | (C;T) | Carrier of Phenylketonuria allele | Link | | Link |
| 3 | rs7754840 | (C;C) | 1.3x increased risk for type-2 diabetes | | Link | |
| 2.5 | rs10484554 | (C;T) | 2.8x increased risk for psoriasis | | Link | |
| 2.5 | rs10490924 | (G;T) | 2.7x risk for age related macular degeneration | Link | Link | Link |
| 2.5 | rs11190870 | (T;T) | Possibly even more increased risk of scoliosis | | | |
| 2.5 | rs16969968 | (A;G) | Slightly higher risk for nicotine dependence: l... | Link | Link | Link |
| 2.5 | rs2254958 | (C;C) | 1.61x increased risk for Alzheimer's | | | |
| 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 | 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... | | | |
| 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... | | Link | Link |
| 2.5 | rs8034191 | (C;T) | 1.27x lung cancer risk | | Link | |
| 2.5 | rs9934438 | (A;A) | Coumadin resistance | | Link | Link |
| 2.3 | rs3798220 | (C;T) | 2-3x higher risk for cardiovascular events: whi... | Link | Link | |
| 2.2 | rs2231137 | (G;G) | ~1.5-3x increased risk for ischemic stroke | Link | Link | Link |
| 2.2 | rs944289 | (T;T) | 2.6x increased thyroid cancer risk | | Link | |
| 2.1 | rs1050631 | (T;T) | Mean Survival Time of 17 months for esophageal ... | Link | | |
| 2.1 | rs10811661 | (T;T) | 1.2x increased risk for type-2 diabetes | | Link | |
| 2.1 | rs1329428 | (G;G) | 2x increased risk for macular degeneration | | | |
| 2.1 | rs17077540 | (A;G) | 1.6x major depressive disorder risk | | | |
| 2.1 | rs2187668 | (A;G) | Somewhat increased autoimmune disorder (lupus: ... | | | |
| 2.1 | rs380390 | (C;C) | Increased risk for ARMD | | Link | |
| 2.1 | rs5186 | (A;C) | ~1.4x increased risk of hypertension | Link | Link | Link |
| 2 | rs10086908 | (C;T) | 1.7x increased risk for prostate cancer | | | |
| 2 | rs10090154 | (C;T) | 1.4x 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 | rs10260404 | (C;C) | 1.60x risk of developing ALS | | Link | |
| 2 | rs1041981 | (A;A) | Higher myocardial infarction risk | Link | Link | Link |
| 2 | rs1051730 | (C;T) | 1.3x increased risk of lung cancer | Link | Link | Link |
| 2 | rs10889677 | (A;C) | 1.5x increased risk for certain autoimmune dise... | | Link | |
| 2 | rs10984447 | (A;A) | >1.17x increased risk for multiple sclerosis | | Link | |
| 2 | rs11229030 | (C;C) | Higher odds of Crohn's disease | | | |
| 2 | rs1160312 | (A;G) | 1.6x increased risk of Male Pattern Baldness. | | Link | |
| 2 | rs1169300 | (A;A) | ~2x increased lung cancer risk | | | |
| 2 | rs11983225 | (T;T) | 7x less likely to respond to certain antidepres... | | Link | |
| 2 | rs1219648 | (A;G) | 1.20x risk for breast cancer | | Link | |
| 2 | rs12567232 | (A;G) | Increased risk for Crohn's Disease | | Link | |
| 2 | rs1265181 | (C;G) | Increased risk for psoriasis | | Link | |
| 2 | rs1333048 | (A;C) | 1.3x increased coronary artery disease risk | | | |
| 2 | rs1360780 | (C;T) | 1.3x increased risk for depression | | Link | |
| 2 | rs1375144 | (C;C) | 1.59x increased risk of developing bipolar diso... | | | |
| 2 | rs17228212 | (C;T) | 1.26x increased risk for heart disease | | Link | |
| 2 | rs1734791 | (A;A) | 1.4x increased risk for lupus | | | |
| 2 | rs17576 | (G;G) | Higher risk for lung cancer: and COPD in smoker... | Link | Link | |

| Mag. | Identifier | Genotype | Summary | ExAC | GetEvidence | ClinVar |
|---|---|---|---|---|---|---|
| 2 | rs1800896 | (A;G) | 1.6x increased prostate cancer risk | | | |
| 2 | rs2070600 | (A;A) | 1.5x increased risk for gastric cancer | Link | Link | |
| 2 | rs2073963 | (G;T) | Increased risk of baldness | | | |
| 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 | rs2305480 | (T;T) | If 4 years old or younger: ~3x increased asthma... | Link | Link | |
| 2 | rs2383206 | (A;G) | 1.4x increased risk for heart disease | | | |
| 2 | rs2383207 | (A;G) | Increased risk for heart disease | | | |
| 2 | rs2420946 | (C;T) | 1.20x risk for breast cancer | | | |
| 2 | rs2464196 | (T;T) | ~2x increased lung cancer risk | Link | Link | Link |
| 2 | rs25487 | (A;G) | 2x higher risk for skin cancer; possibly other ... | Link | Link | |
| 2 | rs2707466 | (G;G) | Weaker bones | Link | 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 | rs3746444 | (C;T) | ~1.2x increased risk for cancer | Link | | |
| 2 | rs3775948 | (G;G) | Slightly higher risk for gout | | | |
| 2 | rs3793784 | (C;G) | 1.5x risk for ARMD | | Link | Link |
| 2 | rs4129148 | (C;G) | 3x risk of schizophrenia. | | Link | |
| 2 | rs4148739 | (A;A) | 7x less likely to respond to certain antidepres... | | Link | |
| 2 | rs4242382 | (A;G) | 1.7x increased risk for prostate cancer | | Link | |
| 2 | rs4444903 | (A;G) | 3.5x risk of hep-cancer in cirrhosis patients; ... | | | |
| 2 | rs520354 | (A;G) | Increased risk in men for biliary conditions | | | |
| 2 | rs629242 | (C;T) | Somewhat higher risk for prostate cancer | | | |
| 2 | rs6457617 | (C;T) | 2.3x risk of rheumatoid arthritis | | Link | |
| 2 | rs663048 | (G;T) | Significantly increased risk of developing lung... | Link | 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 | rs7923837 | (G;G) | 3.2x risk for T2D | | | |
| 2 | rs7961152 | (A;C) | 1.2x higher risk for hypertension | | | |
| 2 | rs800292 | (C;C) | 5% higher risk of Age related macular degenerat... | Link | Link | Link |
| 2 | rs828907 | (T;T) | Increased risk of bladder cancer and 2x risk of... | | | |
| 2 | rs855913 | (G;T) | Reduced survival with ALS | | Link | |
| 2 | rs9303277 | (T;T) | 1.46x Increased risk of developing primary bili... | | | |
| 2 | rs9652490 | (A;A) | ~2x increased risk for Parkinson's disease: and... | | Link | |
| 2 | rs965513 | (A;G) | 1.7x increased thyroid cancer risk | | Link | |
| 2.0 | rs1044396 | (C;C) | Increased risk of Nicotine dependence among mal... | Link | Link | Link |
| 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 | |
| 2.0 | rs9642880 | (T;T) | 1.5x increased bladder cancer risk | | Link | |
| 1.7 | rs8055236 | (G;T) | 1.9x risk for heart disease | | Link | |
| 1.6 | rs3764880 | (A;A) | 1.2 - 1.8x increased tuberculosis risk | Link | Link | |
| 1.6 | rs4959039 | (G;G) | 1.6x higher risk for multiple sclerosis | | | |
| 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 | rs10883365 | (A;G) | 1.2x increased risk for developing Crohn's dise... | | Link | |
| 1.5 | rs11171739 | (C;T) | 1.34x risk of developing Type-1 diabetes | | Link | |
| 1.5 | rs12210050 | (C;T) | Slightly higher risk for basal cell carcinoma | | Link | |
| 1.5 | rs12469063 | (A;G) | Slightly increased risk of developing restless ... | | | |
| 1.5 | rs13149290 | (C;C) | Slightly increased risk of developing prostate ... | | | |
| 1.5 | rs13376333 | (C;T) | 1.5x higher risk of atrial fibrillation | | Link | |

| Mag. | Identifier | Genotype | Summary | ExAC | GetEvidence | ClinVar |
|---|---|---|---|---|---|---|
| 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 | rs165599 | (G;G) | May indicate increased susceptibility to schizo... | | Link | |
| 1.5 | rs16944 | (A;G) | Minorly increased risk of mental illness and os... | | Link | |
| 1.5 | rs17221417 | (C;G) | 1.3x higher risk for Crohn's disease | | Link | |
| 1.5 | rs1799950 | (A;G) | Very slightly increased breast cancer risk | Link | Link | Link |
| 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 | | | |
| 1.5 | rs1975197 | (C;T) | 1.3x increased risk of developing restless legs... | | Link | |
| 1.5 | rs199533 | (C;T) | Slightly increased risk of developing Parkinson... | Link | | |
| 1.5 | rs2272127 | (C;C) | Associated with herpes and schizophrenia | | | |
| 1.5 | rs2280714 | (A;A) | 1.4x increased risk of SLE | | | |
| 1.5 | rs2282679 | (C;C) | Lower vitamin D levels | | | |
| 1.5 | rs2697962 | (A;G) | Slightly increased risk of developing Parkinson... | | | |
| 1.5 | rs27388 | (A;G) | Slightly increased risk of developing schizophr... | | | |
| 1.5 | rs28694718 | (A;A) | >2x higher risk for schizophrenia | | | |
| 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 | rs309375 | (T;T) | Larger mosquito bites | | | |
| 1.5 | rs3212227 | (A;A) | 1.43x increased risk of developing psoriasis an... | | | |
| 1.5 | rs3745516 | (A;G) | Slightly increased risk of developing primary b... | | | |
| 1.5 | rs3814570 | (C;T) | 1.3x increased risk for Crohn's disease with il... | | | |
| 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 | rs4464148 | (C;T) | 1.10x increased risk for colorectal cancer | | | |
| 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 | 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 | rs6435862 | (G;T) | 1.7x higher risk of aggressive neuroblastoma | | Link | |
| 1.5 | rs6498169 | (A;G) | 1.14x risk of multiple sclerosis | | Link | |
| 1.5 | rs6601764 | (C;T) | 1.16x increased risk of developing Crohn's dise... | | Link | |
| 1.5 | rs6710341 | (A;G) | Slightly increased risk of developing restless ... | | | |
| 1.5 | rs6896702 | (C;T) | Slightly increased risk of developing Parkinson... | | | |
| 1.5 | rs699473 | (C;T) | ~1.5x increased brain tumor risk | | | |
| 1.5 | rs7341475 | (G;G) | 1.58x increased schizophrenia risk for women | | Link | |
| 1.5 | rs7536563 | (A;G) | 1.12x risk of multiple sclerosis | | Link | |
| 1.5 | rs763035 | (C;T) | 1.2x increased risk for rosacea | | | |
| 1.5 | rs872071 | (G;G) | ~1.5x increased risk for chronic lymphocytic le... | | Link | |
| 1.5 | rs995030 | (G;G) | Non-protective against testicular cancer | | Link | |
| 1.4 | rs1126497 | (T;T) | 1.4x increased risk for breast cancer | Link | Link | Link |
| 1.4 | rs1447295 | (A;C) | 1.4x increased risk of prostate cancer | | Link | |
| 1.4 | rs1801157 | (A;G) | 1.4x higher risk for breast cancer | | | |
| 1.4 | rs2230201 | (A;G) | 1.4x risk of lupus | Link | | |
| 1.4 | rs3131296 | (G;G) | 1.4x increased risk for schizophrenia | | 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 | rs1260326 | (C;T) | Slightly higher risk for gout | Link | Link | 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 | rs2736100 | (G;T) | 1.3x higher risk for glioma development | | Link | |
| 1.3 | rs501120 | (A;G) | 1.3x increased risk for heart disease | | Link | |
| 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 | |

| Mag. | Identifier | Genotype | Summary | ExAC | GetEvidence | ClinVar |
|---|---|---|---|---|---|---|
| 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 | rs2076295 | (G;T) | One copy of the risk allele (G): slightly incre... | | | |
| 1.2 | rs393152 | (A;G) | Slight increased risk of both PD and AD | Link | Link | |
| 1.2 | rs4324715 | (C;C) | >1.5x increased testicular cancer risk for men | | | |
| 1.2 | rs449647 | (A;T) | Possibly lower levels of ApoE | | | |
| 1.2 | rs4686484 | (A;A) | Slightly increased risk for celiac disease | | | |
| 1.2 | rs4977756 | (A;G) | 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 | rs9858542 | (A;G) | 1.1x risk Crohn's Disease | Link | Link | |
| 1.17 | rs3802842 | (A;C) | 1.17x increased risk of colorectal cancer | | Link | |
| 1.16 | rs2278206 | (C;C) | 1.16x increased risk for asthma | Link | Link | |
| 1.1 | rs11110912 | (C;C) | 1.3x high blood pressure risk | | | |
| 1.1 | rs1344706 | (G;T) | 1.1x increased risk for schizophrenia | | Link | |
| 1.1 | rs249954 | (C;T) | Slight if any increased risk of 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 | rs3818361 | (C;T) | 1.15x increased risk for late-onset Alzheimer's... | | | |
| 1.1 | rs4977574 | (A;G) | Some studies - but not others - report a slight... | | Link | |
| 1.1 | rs688034 | (C;T) | 1.1x risk higher risk for coronary artery disea... | | Link | |
| 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 | | | |
| 1.07 | 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 | rs1143674 | (A;A) | 1.3x increased autism risk | Link | | |
| 1 | rs12752888 | (C;C) | Faster progression of mild cognitive impairment... | | | |
| 1 | rs2546890 | (A;A) | Higher risk of multiple sclerosis | | | |
| 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 | (A;G) | Susceptible to Norovirus infections | Link | Link | Link |
| 0 | rs1333040 | (C;T) | 1.24x increased myocardial infarction risk: 1.2... | | Link | |
| 0 | rs1611115 | (T;T) | Somewhat more associated with impulsiveness and... | | | Link |
| 0 | rs2296336 | (C;C) | 2.9x risk of type-1 diabetes | | | |
| 0 | rs4712653 | (T;T) | 2x increased risk for neuroblastoma | | | |
| 0 | rs4795400 | (T;T) | If 4 years old or younger: ~2.5x increased asth... | | Link | |
| 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 | gs144 | Male |
| 3.1 | gs122 | 7x risk of baldness |
| 3.1 | gs237 | Blue eyes are much more likely |
| 3 | gs215 | R-L21 |
| 3 | gs273 | Lowest risk (13% of white women) of Atrial Fibr... |
| 3.0 | gs291 | Lower heart attack risk than average |
| 2.9 | gs192 | MTHFR polymorphisms affecting homocysteine |
| 2.5 | gs102 | ALS risk |
| 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 ... |
| 2.3 | gs255 | Homozygous eye color haplotype #1 |
| 2.2 | gs280 | Light hair color for europeans |
| 2 | gs101 | Probably able to digest milk |
| 2 | gs104 | Restless legs syndrome risk |
| 2 | gs159 | CYP1A2 fast metabolizer |
| 2 | gs179 | CYP2D6*41 |
| 2 | gs181 | CYP2D6*2 |
| 2 | gs213 | Haplogroup R (Y-DNA) |
| 2 | gs246 | APOE3/APOE3 |
| 1.5 | gs1105 | Mitochondrial Haplogroup U |
| 1.5 | gs186 | HLA-B*5801 heterozygosity is possible: unfortun... |
| 1.5 | gs247 | Parkinson's Disease Risk |
| 1.2 | gs184 | Able to taste bitterness. |
| 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).