# Genomics Report for PGP-UK7/uk1097F9

## 1 Summary

This is the genome report for participant PGP-UK7/uk1097F9 . 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.

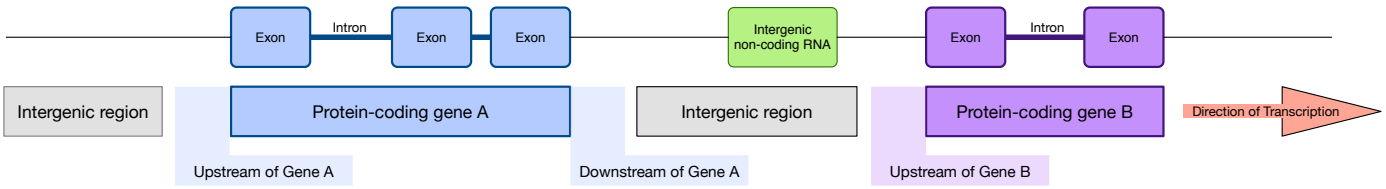


Figure 1: Diagram of gene structure indicating locations of potential variants

| Feature | Count |
|---|---|
| Lines of input read | 4164214 |
| Variants remaining after filtering | 4136609 |
| Novel / existing variants | 112341 (2.7%) / 4024268 (97.3%) |
| Overlapped genes | 54668 |
| Overlapped transcripts | 64432 |
| Overlapped regulatory features | 211697 |

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.

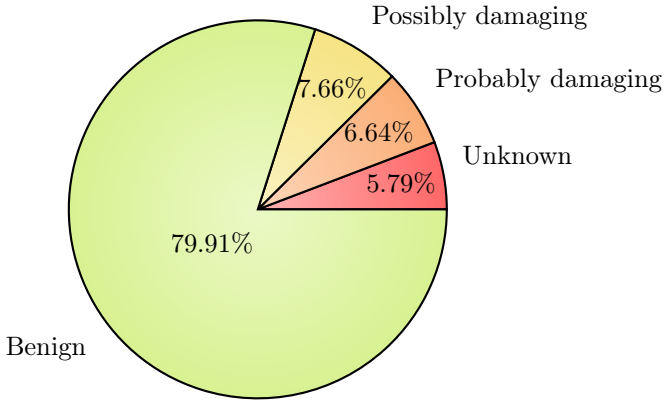


Figure 2: PolyPhen Summary

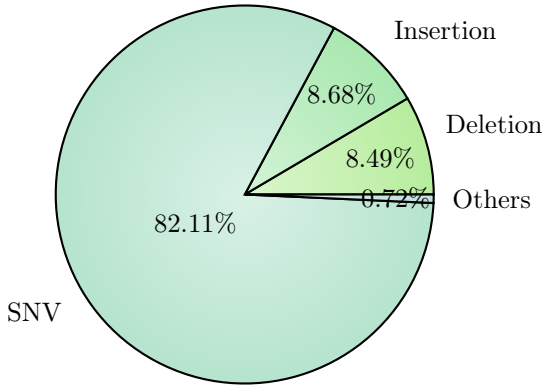


Figure 3: Variant Class

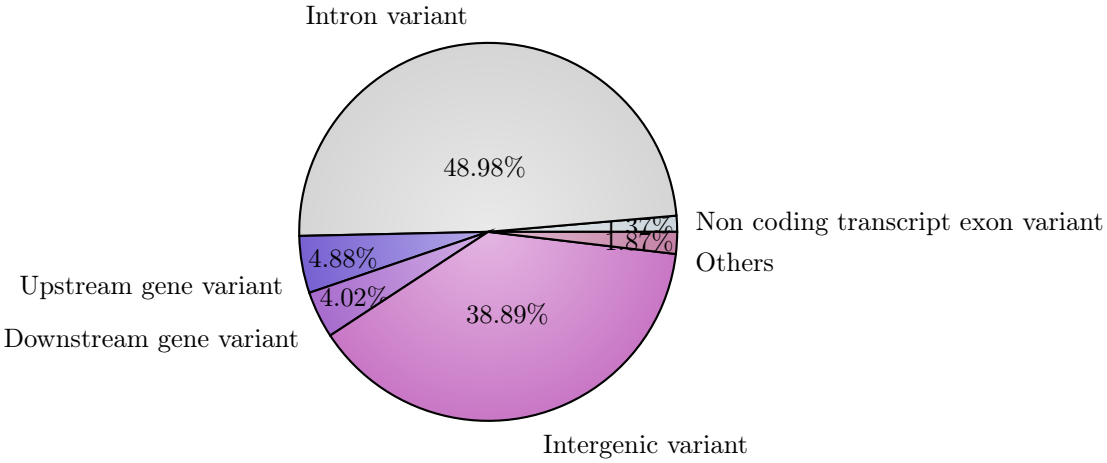


Figure 4: Consequence type

## 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.

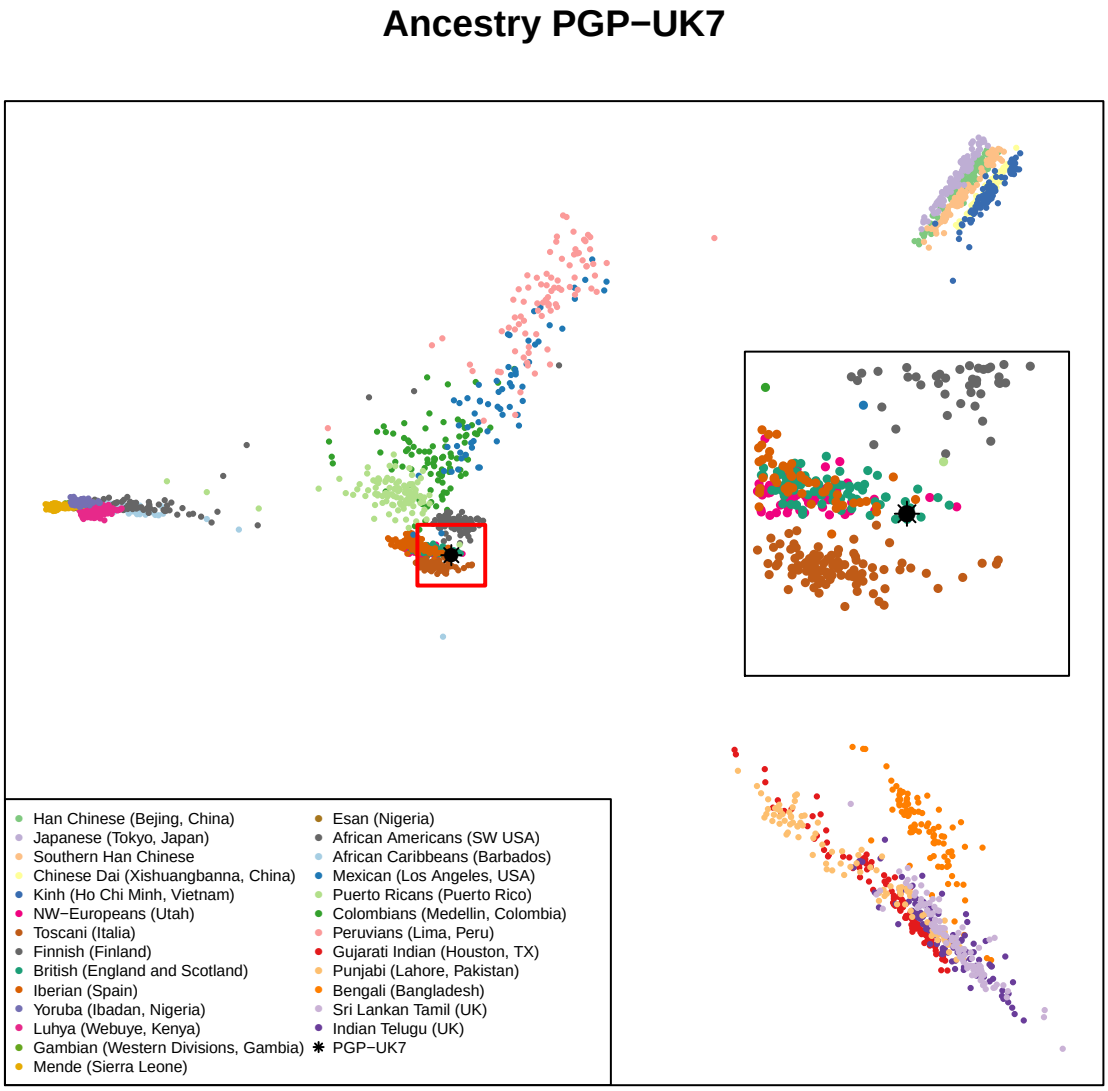


Figure 5: Ancestry Principal Component Analysis

# 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 |
|---|---|---|---|---|---|---|
| 2.5 | rs3764261 | (T;T) | Associated with higher HDL cholesterol. HDL | | Link | Link |
| 2.1 | rs2511989 | (A;G) | 0.63x decreased age-related macular degeneratio... | | Link | |
| 2.1 | rs6505162 | (A;A) | 0.43x decreased risk for esophageal cancer | Link | | |
| 2.1 | rs738409 | (G;G) | Most common genotype; slightly less damage from... | Link | 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 | rs1128535 | (G;G) | Reduced risk (0.77x) for Crohn's disease | | | |
| 2 | rs1544410 | (G;G) | Decreased risk of low bone mineral density diso... | | Link | |
| 2 | rs1799884 | (G;G) | Mothers have typical Birth-Weight babies. Sligh... | | | |
| 2 | rs1864163 | (G;G) | Associated with higher HDL cholesterol | | Link | |
| 2 | rs2542052 | (C;C) | Better odds of living to 100 | | | |
| 2 | rs261332 | (A;G) | Associated with higher HDL cholesterol | | | |
| 2 | rs3914132 | (C;T) | Lower otosclerosis risk | | Link | |
| 2 | rs4143094 | (G;G) | No increased risk of colorectal cancer correlat... | | | |
| 2 | rs4149268 | (G;G) | Associated with higher HDL cholesterol | | Link | |
| 2 | rs4585 | (G;G) | Slightly higher (1.35x) odds of good metformin ... | | | |
| 2 | rs6807362 | (G;G) | Decreased autism risk | Link | Link | |
| 2 | rs7776725 | (T;T) | Stronger bones | | Link | |
| 2 | rs801114 | (T;T) | 0.78x decreased Basal Cell Carcinoma risk. | | Link | |
| 2 | rs9642880 | (G;G) | Slightly lower risk of Bladder Cancer. | | Link | |
| 1.9 | rs1015362 | (A;A) | Probably tans instead of freckles and sunburns.... | | 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 | rs11635424 | (A;G) | 0.70x risk for restless legs | | Link | |
| 1.5 | rs12593813 | (A;G) | 0.71x risk for restless legs | | Link | |
| 1.5 | rs4149274 | (C;C) | Associated with higher HDL (good) cholesterol. | | | |

| Mag. | Identifier | Genotype | Summary | ExAC | GetEvidence | ClinVar |
|---|---|---|---|---|---|---|
| 1.5 | rs4489954 | (G;T) | 0.69x risk risk of developing restless legs syn... | | Link | |
| 1.5 | rs4939883 | (C;C) | Associated with higher HDL cholesterol | | Link | |
| 1.5 | rs729302 | (A;C) | 0.89x decreased risk of developing rheumatoid a... | | | |
| 1.5 | rs9939609 | (T;T) | Lower risk of obesity and Type-2 diabetes | | Link | |
| 1.2 | rs11246226 | (A;C) | Decreased risk of schizophrenia in limited stud... | | Link | |
| 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 | rs4988235 | (T;T) | Can digest milk | | | Link |
| 1 | rs182549 | (T;T) | Can digest milk. | | | Link |
| 1 | rs2494732 | (T;T) | Lower odds of psychosis | Link | Link | |
| 1 | rs2546890 | (G;G) | Lower risk of multiple sclerosis | | | |
| 1 | rs2952768 | (C;T) | Slightly less drug dependence: decreased effect... | | | |
| 1 | rs4939827 | (C;T) | 0.86x decreased risk for colorectal cancer | | Link | |
| 1.0 | rs6583817 | (C;T) | ~0.80x (lower) risk for late onset Alzheimer's ... | | | |
| 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 | rs1126809 | (A;G) | Slight increase in skin cancer risk | Link | 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 | rs2306402 | (C;C) | 1.18x increased risk for LOAD | | | |
| 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 | | | |

- Possibly Harmful Traits

| Mag. | Identifier | Genotype | Summary | ExAC | GetEvidence | ClinVar |
|---|---|---|---|---|---|---|
| 3 | rs2237717 | (T;T) | Reduced abilities related to neurocognition and... | | | |
| 3 | rs3738579 | (T;T) | 1.5x - 2x increased risk for cervical cancer: H... | | | |
| 3 | rs7754840 | (C;G) | 1.3x increased risk for type-2 diabetes | | Link | |
| 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 | rs13266634 | (C;T) | Increased risk for type-2 diabetes | Link | Link | Link |
| 2.5 | rs16969968 | (A;G) | Slightly higher risk for nicotine dependence: l... | Link | Link | Link |
| 2.5 | rs1800629 | (A;A) | Complex; generally higher risk for certain dise... | Link | Link | |
| 2.5 | rs2943634 | (C;C) | Higher risk of ischemic stroke | | Link | |
| 2.5 | rs339331 | (T;T) | Prostate cancer risk | | | |
| 2.5 | rs664143 | (T;T) | Higher risk for number of cancers | | | |
| 2.5 | rs8034191 | (C;T) | 1.27x lung cancer risk | | Link | |
| 2.5 | rs9934438 | (A;A) | Coumadin resistance | | Link | Link |
| 2.3 | rs7966230 | (C;G) | Slightly lower levels of plasma VWF | | | |
| 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 | rs1064395 | (A;A) | Having the A allele at this SNP makes your risk... | | | |
| 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 | rs2254958 | (C;T) | 1.24x increased risk for Alzheimer's | | | |
| 2.1 | rs380390 | (C;C) | Increased risk for ARMD | | Link | |
| 2.1 | rs4430796 | (A;A) | 1.38x increased risk for prostate cancer | | 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 | rs7837688 | (G;G) | 1.7x increased risk for prostate cancer | | | |
| 2.1 | rs795484 | (A;G) | Increased morphine dose requirement and postope... | | | |
| 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 | rs1041981 | (A;A) | Higher myocardial infarction risk | Link | Link | Link |
| 2 | rs1050152 | (C;T) | 2.1x increased risk of Crohn's disease | Link | Link | Link |
| 2 | rs1051730 | (C;T) | 1.3x increased risk of lung cancer | Link | Link | Link |
| 2 | rs10889677 | (C;C) | 1x increased risk for certain autoimmune diseas... | | Link | |
| 2 | rs10984447 | (A;G) | 1.17x increased risk for multiple sclerosis | | Link | |
| 2 | rs1136287 | (C;T) | 1.5x increased risk of wet ARMD | Link | 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 | rs1223271 | (A;A) | Increased risk of developing Parkinson's Diseas... | | Link | |
| 2 | rs1265181 | (C;G) | Increased risk for psoriasis | | Link | |
| 2 | rs1333048 | (A;C) | 1.3x increased coronary artery disease risk | | | |
| 2 | rs16942 | (A;G) | Very slightly increased breast cancer risk | Link | Link | Link |
| 2 | rs1734791 | (A;A) | 1.4x increased risk for lupus | | | |
| 2 | rs17576 | (A;G) | Higher risk for MI and lung cancer: and COPD in... | Link | Link | |
| 2 | rs1800896 | (A;A) | 1.8x increased prostate cancer risk | | | |
| 2 | rs187238 | (G;G) | Hypertension increases risk 3.75x for sudden ca... | | | |
| 2 | rs2073963 | (G;T) | Increased risk of baldness | | | |
| 2 | rs2075650 | (A;G) | 2x higher Alzheimer's risk | Link | Link | |
| 2 | rs2156921 | (A;G) | 1.29x increased risk for depression | | | |
| 2 | rs2201841 | (T;T) | 2.4x increased risk for Graves' disease | | Link | |
| 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... | | | |

| Mag. | Identifier | Genotype | Summary | ExAC | GetEvidence | ClinVar |
|---|---|---|---|---|---|---|
| 2 | rs2274223 | (A;G) | 1.5x increased risk for stomach and esophageal ... | Link | Link | |
| 2 | rs2305795 | (A;G) | 1.28x higher risk of narcolepsy compared to (G;... | | | Link |
| 2 | rs2383206 | (A;G) | 1.4x increased risk for heart disease | | | |
| 2 | rs2383207 | (A;G) | Increased risk for heart disease | | | |
| 2 | rs25487 | (G;G) | 2x higher risk for skin cancer; possibly other ... | Link | Link | |
| 2 | rs2707466 | (G;G) | Weaker bones | Link | Link | |
| 2 | rs27388 | (A;A) | Increased risk of developing schizophrenia | | | |
| 2 | rs2908004 | (C;C) | Weaker bones | Link | Link | |
| 2 | rs3025039 | (C;T) | 2.6x increased risk for ARMD | | | |
| 2 | rs3212227 | (A;C) | Significantly increased risk of developing cerv... | | | |
| 2 | rs326 | (A;A) | Lower HDL cholesterol | | 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 | 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 | (T;T) | Higher risk for endometrial cancer | Link | Link | |
| 2 | rs4792311 | (A;G) | Increased risk of prostate cancer | Link | Link | Link |
| 2 | rs493258 | (A;G) | 1.15x risk of Age Related Macular Degeneration | | | |
| 2 | rs4968451 | (A;C) | 1.61x increased risk for meningioma | | | |
| 2 | rs5174 | (A;G) | 1.3x increased risk for heart disease | Link | Link | Link |
| 2 | rs520354 | (A;G) | Increased risk in men for biliary conditions | | | |
| 2 | rs6603272 | (G;G) | >2.74x increased risk of developing schizophren... | | | |
| 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 | rs6908425 | (C;C) | 1.95x increased risk of developing Crohn's dise... | | Link | |
| 2 | rs6997709 | (G;G) | 1.5x higher risk for hypertension | | | |
| 2 | rs699 | (C;T) | Increased risk of hypertension | Link | Link | Link |
| 2 | rs7442295 | (A;A) | ~4x higher risk for hyperuracemia | | Link | |
| 2 | rs7794745 | (A;T) | Slightly increased risk for autism | | Link | Link |
| 2 | rs7807268 | (C;C) | 1.4x risk for Crohn's disease | | 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 | (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 | |
| 2.0 | rs4911414 | (T;T) | 2-4x higher risk of sun sensitivity if part of ... | | Link | |
| 1.8 | rs37973 | (A;G) | Among asthmatics: 1.5x more likely to show less... | | | Link |
| 1.6 | rs1537415 | (C;G) | 1.6x increased risk for periodontitis | | Link | |
| 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 | rs10492519 | (A;G) | Slightly increased risk of developing prostate ... | | | |
| 1.5 | rs10757272 | (C;T) | 1.30x increased risk for Coronary artery diseas... | | | |
| 1.5 | rs10883365 | (A;G) | 1.2x increased risk for developing Crohn's dise... | | Link | |
| 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 | rs1154155 | (G;T) | 1.94x increased risk for narcolepsy | | Link | |
| 1.5 | rs1169300 | (A;G) | ~1.5x increased lung cancer risk | | | |
| 1.5 | rs12037606 | (A;G) | 1.22x risk of developing Crohn's disease | | | |
| 1.5 | rs12431733 | (C;T) | Slightly increased risk of developing Parkinson... | | Link | |
| 1.5 | rs12498742 | (A;A) | 1.25 increased risk for gout | | | |
| 1.5 | rs13149290 | (C;C) | Slightly increased risk of developing prostate ... | | | |

| Mag. | Identifier | Genotype | Summary | ExAC | GetEvidence | ClinVar |
|---|---|---|---|---|---|---|
| 1.5 | rs1360517 | (A;G) | Higher susceptibility for AIDS | | Link | |
| 1.5 | rs16944 | (A;G) | Minorly increased risk of mental illness and os... | | Link | |
| 1.5 | rs2241880 | (C;T) | 1.4x increased risk for Crohn's disease in Cauc... | Link | Link | Link |
| 1.5 | rs2272127 | (C;C) | Associated with herpes and schizophrenia | | | |
| 1.5 | rs2464196 | (C;T) | ~1.5x increased lung cancer risk | Link | Link | Link |
| 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 | rs28694718 | (A;G) | 2x higher risk for schizophrenia | | | |
| 1.5 | rs2881766 | (T;T) | Slightly increased risk for pregnancy-induced h... | | | |
| 1.5 | rs3087243 | (A;G) | Increased risk for auto-immune diseases | | Link | |
| 1.5 | rs3790565 | (C;T) | Slightly increased risk of developing primary b... | | | |
| 1.5 | rs3825776 | (A;G) | 1.3x increased risk for ALS | | Link | |
| 1.5 | rs393152 | (A;A) | Increased risk of both PD and AD | Link | 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 | rs464049 | (T;T) | Increased risk of schizophrenia in limited stud... | | | |
| 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 | 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 | rs7341475 | (G;G) | 1.58x increased schizophrenia risk for women | | Link | |
| 1.5 | rs7850258 | (G;G) | Slightly higher odds of developing primary hypo... | | | |
| 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 | rs1545843 | (A;A) | 1.4x increased risk for depression (for those u... | | | |
| 1.4 | rs2046210 | (C;T) | 1.4x increased breast cancer risk | | 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 | | | |
| 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 | rs2736100 | (G;T) | 1.3x higher risk for glioma development | | Link | |
| 1.3 | rs356219 | (A;G) | 1.3x increased risk for Parkinson's disease | | | |
| 1.2 | rs10865331 | (A;G) | 1.2x higher risk for ankylosing spondylitis | | | |
| 1.2 | rs11037909 | (T;T) | 1.47x type II diabetes risk | Link | | |
| 1.2 | rs143383 | (C;T) | 1.1x increased risk for osteoarthritis | | Link | Link |
| 1.2 | rs2072590 | (G;T) | 1.2x increased risk for ovarian cancer | | | |
| 1.2 | rs2076295 | (G;T) | One copy of the risk allele (G): slightly incre... | | | |
| 1.2 | rs2665390 | (C;T) | 1.2x increased risk for ovarian cancer | | | |
| 1.2 | rs2814707 | (A;G) | 1.2x increased risk for ALS | | Link | |
| 1.2 | rs3131296 | (A;G) | 1.2x increased risk for schizophrenia | | 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 | rs419788 | (A;G) | 2.0x risk for lupus | Link | | |
| 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 | rs17465637 | (A;C) | 1.17x higher risk for myocardial infarction | Link | Link | |
| 1.15 | rs748404 | (C;T) | Very slightly increased risk (1.15) for lung ca... | | Link | |
| 1.1 | rs11110912 | (C;G) | 1.3x high blood pressure risk | | | |
| 1.1 | rs1344706 | (G;T) | 1.1x increased risk for schizophrenia | | Link | |
| 1.1 | rs1799966 | (A;G) | Very slightly increased breast cancer risk | Link | Link | Link |
| 1.1 | rs34516635 | (G;G) | Less longevity for Ashkenazi Jewish women. | Link | | Link |

| Mag. | Identifier | Genotype | Summary | ExAC | GetEvidence | ClinVar |
|---|---|---|---|---|---|---|
| 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 | rs6495446 | (C;T) | 1.2x increased risk for chronic kidney disease | | | |
| 1.1 | rs688034 | (C;T) | 1.1x risk higher risk for coronary artery disea... | | Link | |
| 1.1 | rs6897876 | (C;T) | Slight increase in testicular cancer risk for m... | | | |
| 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 | | | |
| 1.05 | rs2291834 | (C;T) | Very slightly higher risk for myocardial infarc... | | | |
| 1 | rs10504861 | (G;G) | Major allele: normal risk of migraine | | | |
| 1 | rs1143674 | (A;G) | 1.3x increased autism risk | Link | | |
| 1 | rs3194051 | (A;G) | 1.12x risk of type-1 diabetes | Link | Link | Link |
| 1 | rs6932590 | (C;T) | 1.1x increased risk for schizophrenia | | Link | |
| 1 | rs987525 | (A;C) | 2.5x increased risk for cleft lip | | Link | |
| 0.1 | rs601338 | (A;G) | Susceptible to Norovirus infections | Link | Link | Link |
| 0 | rs1004819 | (C;C) | 1.5x risk of Crohn's disease | | Link | |
| 0 | rs1495965 | (A;A) | 1.2x higher risk for spondylitis | | | |
| 0 | rs1800860 | (A;A) | 10% smaller kidneys as newborns | Link | | 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 | 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 | gs144 | Male |
| 3.1 | gs122 | 7x risk of baldness |
| 3.1 | gs237 | Blue eyes are much more likely |
| 3 | gs273 | Lowest risk (13% of white women) of Atrial Fibr... |
| 2.5 | gs134 | Age related macular degeneration |
| 2.5 | gs155 | CYP3A5 non-expressor |
| 2.5 | gs259 | Homozygous for eye color haplotype #3 |
| 2.5 | gs281 | Part of the 88% of the population claimed not t... |
| 2.3 | gs255 | Homozygous eye color haplotype #1 |
| 2.2 | gs280 | Light hair color for europeans |
| 2 | gs101 | Probably able to digest milk |
| 2 | gs129 | Unable to classify your ABO blood type |
| 2 | gs156 | NAT2 Rapid metabolizer. |
| 2 | gs159 | CYP1A2 fast metabolizer |
| 2 | gs179 | CYP2D6*41 |
| 2 | gs181 | CYP2D6*2 |
| 2 | gs187 | HLA-B*5801 homozygosity is possible. too common... |
| 2 | gs188 | One copy of APOE4 is possible: but not certain |
| 2 | gs213 | Haplogroup R (Y-DNA) |
| 1.5 | gs1001 | Mitochondrial Haplogroup H |
| 1.5 | gs139 | NAT2 intermediate metabolizer |
| 1.5 | gs186 | HLA-B*5801 heterozygosity is possible: unfortun... |
| 1.5 | gs226 | Novelty seeking behaviour potential |
| 1.5 | gs247 | Parkinson's Disease Risk |
| 1.2 | gs1039 | Mitochondrial Haplogroup H5 |
| 1.2 | gs1040 | Mitochondrial Haplogroup H5a |
| 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).