# 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 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 | 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


Intergenic variant
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) | 0.71 x decreased risk for dry age related macula... | Link | Link | Link |
| 2 | rs1012053 | $(\mathrm{A} ; \mathrm{C})$ | 0.625x reduced risk of Bipolar Disorder. |  | Link |  |
| 2 | rs10468017 | ( $\mathrm{C} ; \mathrm{T}$ ) | Associated with higher HDL cholesterol |  | Link |  |
| 2 | rs11045585 | ( $\mathrm{A} ; \mathrm{A}$ ) | 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) | 3 x 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) | 0.71 x 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 | $(\mathrm{A} ; \mathrm{C})$ | 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: 2.6 x more likely to not respond t... |  |  |  |
| 0 | rs1126742 | (T;T) | Higher hypertension risk | Link | Link |  |
| 0 | rs12252 | ( $\mathrm{T} ; \mathrm{T}$ ) | 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 | ( $\mathrm{A} ; \mathrm{T}$ ) | $26 \%$ risk of bad reaction to anti-epileptic carb... |  |  | Link |
| 3 | rs1983132 | (C;T) | $2-3 \mathrm{x}$ higher prostate cancer risk if routinely... |  |  |  |
| 3 | rs2066844 | (C;T) | 3 x 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 | ( $\mathrm{C} ; \mathrm{T}$ ) | 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) | $\sim 1.3 \mathrm{x}$ increased obesity risk |  | Link | Link |
| 2.5 | rs1800255 | ( $\mathrm{A} ; \mathrm{A}$ ) | 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 | ( $\mathrm{T} ; \mathrm{T}$ ) | 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) | ${ }^{1} 1.5-3 \mathrm{x}$ 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 | ( $\mathrm{A} ; \mathrm{C}$ ) | 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 | ( $\mathrm{T} ; \mathrm{T}$ ) | 5.2x risk of rheumatoid arthritis |  | Link |  |
| 2.1 | rs646776 | ( $\mathrm{A} ; \mathrm{A}$ ) | 1.2 x risk of coronary artery disease |  | Link |  |
| 2.1 | rs7837688 | (G;G) | 1.7 x 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) | 1.3 x 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) | 1.62 x increased risk for developing Crohn's dis... |  | Link |  |
| 2 | rs10889677 | (C;C) | 1x increased risk for certain autoimmune diseas... |  | Link |  |
| 2 | rs10984447 | ( $\mathrm{A} ; \mathrm{A}$ ) | $>1.17 \mathrm{x}$ increased risk for multiple sclerosis |  | Link |  |
| 2 | rs1160312 | $(\mathrm{A} ; \mathrm{A})$ | 1.6x increased risk of Male Pattern Baldness. |  | Link |  |
| 2 | rs11983225 | (T;T) | 7x less likely to respond to certain antidepres... |  | Link |  |
| 2 | rs12431733 | ( $\mathrm{T} ; \mathrm{T}$ ) | Increased risk of developing Parkinson's Diseas... |  | Link |  |
| 2 | rs13254738 | ( $\mathrm{A} ; \mathrm{C}$ ) | 1.18x prostate cancer risk |  | Link |  |
| 2 | rs1333048 | (A;C) | 1.3x increased coronary artery disease risk |  |  |  |
| 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) | 1.4 x 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) | $>1.4 \mathrm{x}$ risk of lupus | Link |  |  |
| 2 | rs2235015 | (G;G) | Somewhat less likely to respond to certain anti... | Link | Link |  |
| 2 | rs2235040 | (G;G) | 7 x 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) | 1.4 x 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) | 1.3 x 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) | $\sim 4 \mathrm{x}$ higher risk for hyperuracemia |  | Link |  |
| 2 | rs744373 | (C;C) | 1.17x risk of Alzheimer's |  |  |  |
| 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 | ( $\mathrm{A} ; \mathrm{A}$ ) | $\sim^{\sim} 2 \mathrm{x}$ 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) | $2-4 \mathrm{x}$ 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: 1.5 x more likely to show less... |  |  | Link |
| 1.8 | rs4474514 | (A;G) | 3 x 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 ( $\sim 1.2 \mathrm{x}$ ) increase in endometriosis risk |  |  |  |
| 1.5 | rs10980705 | (C;T) | 2.3x increased risk for knee osteoarthritis |  |  |  |
| 1.5 | rs11171739 | (C;T) | 1.34 x 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) | $>1.36 \mathrm{x}$ 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) | 1.43 x 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) | $\sim 1.2 \mathrm{x}$ 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) | 1.4 x increased risk for type-2 diabetes |  | Link |  |
| 1.5 | rs4585 | ( $\mathrm{T} ; \mathrm{T}$ ) | 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) | 1.7 x 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) | ${ }^{\sim} 2 \mathrm{x}$ increased risk of adverse drug reactions fr... |  | Link |  |
| 1.5 | rs9642880 | (G;T) | 1.2 x increased bladder cancer risk |  | Link |  |
| 1.4 | rs1126497 | (C;T) | 1.4 x increased risk for breast cancer | Link | Link | Link |
| 1.4 | rs1801157 | (A;G) | 1.4x higher risk for breast cancer |  |  |  |
| 1.4 | rs3131296 | (G;G) | 1.4x increased risk for schizophrenia |  | Link |  |
| 1.4 | rs4959039 | (A;G) | 1.4x higher risk for multiple sclerosis |  |  |  |
| 1.34 | rs17465637 | (C;C) | 1.34x higher risk for myocardial infarction | Link | Link |  |
| 1.3 | rs1042713 | (A;G) | 1.3x increased risk that pediatric inhaler use ... | Link | Link | Link |
| 1.3 | rs1047286 | (C;T) | 1.3x increased risk for age-related macular deg... | Link | Link | Link |
| 1.3 | rs10947262 | (C;C) | 1.3 x increased risk for osteoarthritis |  |  |  |
| 1.3 | rs1434536 | (A;G) | 1.29x increased breast cancer risk |  |  |  |
| 1.3 | rs16847548 | (C;T) | 1.3 x increased risk for sudden cardiac death in... |  |  |  |
| 1.3 | rs2295490 | (A;G) | 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... |  |  |  |
| 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 |  |


| Mag. | 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 | ( $\mathrm{T} ; \mathrm{T}$ ) | 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 | ( $\mathrm{A} ; \mathrm{A}$ ) | 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: 1.17 x ... |  | Link |  |
| 1.2 | rs6897876 | (C;C) | Slight increase in testicular cancer risk for m... |  |  |  |
| 1.2 | rs8050136 | $(\mathrm{A} ; \mathrm{C})$ | 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 | ( $\mathrm{A} ; \mathrm{A}$ ) | Nicotine dependence |  |  |  |
| 1 | rs2546890 | ( $\mathrm{A} ; \mathrm{A}$ ) | Higher risk of multiple sclerosis |  |  |  |
| 1 | rs3194051 | (A;A) | $>1.1 \mathrm{x}$ 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 | ( $\mathrm{T} ; \mathrm{T}$ ) | 1.1x increased risk for schizophrenia |  | Link |  |
| 1 | rs987525 | $(\mathrm{A} ; \mathrm{C})$ | 2.5x increased risk for cleft lip |  | Link |  |
| 1.0 | rs11246226 | ( $\mathrm{A} ; \mathrm{A}$ ) | 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) | 1.24 x increased myocardial infarction risk: $1.2 \ldots$ |  | 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) | $<0.61$ x 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 $1 \ldots$ |
| 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).

