## Genomics Report for PGP-UK10/uk481F67

## 1 Summary

This is the genome report for participant PGP-UK10/uk481F67. 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 | 4932231 |
| Variants remaining after filtering | 4900960 |
| Novel / existing variants | $165833(3.4 \%) / 4735127(96.6 \%)$ |
| Overlapped genes | 55493 |
| Overlapped transcripts | 65993 |
| Overlapped regulatory features | 242649 |

Table 1: Variant calling summary

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

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


Figure 2: PolyPhen Summary


Figure 3: Variant Class


Figure 4: Consequence type

## 2 Ancestry

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

## Ancestry PGP-UK10



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 |
| :---: | :---: | :---: | :---: | :---: | :---: | :---: |
| 3 | rs925391 | (T;T) | Unlikely to go bald |  |  |  |
| 2.5 | rs3782179 | (C;C) | 9x lower odds of testicular cancer |  |  |  |
| 2.4 | rs2802288 | (A;A) | Longer lifespan |  |  |  |
| 2.1 | rs2511989 | (A;G) | 0.63x decreased age-related macular degeneratio... |  | Link |  |
| 2.1 | rs6505162 | ( $\mathrm{A} ; \mathrm{A}$ ) | 0.43 x decreased risk for esophageal cancer | Link |  |  |
| 2.1 | rs995030 | ( $\mathrm{A} ; \mathrm{A}$ ) | Reduced risk of testicular cancer |  | Link |  |
| 2 | rs10504861 | (A;G) | Reduced risk of migraine without aura |  |  |  |
| 2 | rs11045585 | (A;A) | $24 \%$ chance (lower than average) of docetaxel-in... |  | Link |  |
| 2 | rs1160312 | (G;G) | Reduced risk of Baldness. |  | Link |  |
| 2 | rs11635424 | ( $\mathrm{A} ; \mathrm{A}$ ) | $<0.70 \mathrm{x}$ risk for restless legs |  | Link |  |
| 2 | rs12593813 | ( $\mathrm{A} ; \mathrm{A}$ ) | $<0.71 \mathrm{x}$ risk for restless legs |  | Link |  |
| 2 | rs13706 | ( $\mathrm{A} ; \mathrm{A}$ ) | Decreased risk for certain cancers | Link | Link | Link |
| 2 | rs1544410 | (G;G) | Decreased risk of low bone mineral density diso... |  | Link |  |
| 2 | rs16890979 | (T;T) | Lower systolic blood pressure: serum uric acid ... | Link | Link |  |
| 2 | rs1799884 | (G;G) | Mothers have typical Birth-Weight babies. Sligh... |  |  |  |
| 2 | rs1864163 | (A;G) | Associated with higher HDL cholesterol |  | Link |  |
| 2 | rs2235015 | (G;T) | Somewhat more likely to respond to certain anti... | Link | Link |  |
| 2 | rs2243250 | (T;T) | 0.33x decreased risk for myocardial infarction ... |  |  |  |
| 2 | rs2707466 | ( $\mathrm{A} ; \mathrm{A}$ ) | Stronger bones | Link | Link |  |
| 2 | rs2764264 | (C;C) | Greater odds of living to 95 |  |  |  |
| 2 | rs2908004 | (T;T) | Stronger bones | Link | Link |  |
| 2 | rs3178250 | (C;C) | Lower otosclerosis risk |  |  |  |
| 2 | rs3764261 | (G;T) | Associated with higher HDL cholesterol |  | Link | Link |
| 2 | rs3819331 | (T;T) | Lower risk of autism | Link |  |  |
| 2 | rs3914132 | (C;T) | Lower otosclerosis risk |  | Link |  |
| 2 | rs4149268 | (A;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 |  |


| Mag. | Identifier | Genotype | Summary | ExAC | GetEvidence | ClinVar |
| :---: | :---: | :---: | :---: | :---: | :---: | :---: |
| 2 | rs763110 | (T;T) | 0.80x reduced cancer risk |  |  | Link |
| 2 | rs7776725 | ( $\mathrm{T} ; \mathrm{T}$ ) | Stronger bones |  | Link |  |
| 1.6 | rs10801935 | (C;C) | 0.3x decreased risk of breast cancer |  |  |  |
| 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 | rs11136000 | (C;T) | 0.84x decreased risk for Alzheimer's disease |  | Link |  |
| 1.5 | rs2007153 | (A;A) | Decreased risk of schizophrenia in limited stud... |  |  |  |
| 1.5 | rs28362263 | (A;G) | Modest reduction in LDL-C and coronary risk | Link | 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 | rs3851179 | (A;G) | 0.85x decreased risk for Alzheimer's disease |  | Link |  |
| 1.5 | rs4149274 | (C;T) | Associated with higher HDL (good) cholesterol |  |  |  |
| 1.5 | rs4489954 | (G;T) | 0.69x risk risk of developing restless legs syn... |  | Link |  |
| 1.5 | rs4939883 | (C;T) | Associated with higher HDL cholesterol |  | 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... |  |  |  |
| 1.5 | rs9939609 | (T;T) | Lower risk of obesity and Type-2 diabetes |  | Link |  |
| 1.3 | rs10166942 | (C;C) | 0.7x lower risk for migraines |  |  |  |
| 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 | rs13333226 | (A;G) | Slightly lower risk for hypertension |  |  | Link |
| 1.1 | rs16891982 | (C;C) | Generally non-European: but if European: 7x mor... | Link | Link | Link |
| 1.1 | rs2293347 | (G;G) | Among NSCLC patients: better Gefitinib response... | Link |  |  |
| 1.1 | rs7568369 | (G;T) | 0.90x reduced risk of obesity |  |  |  |
| 1 | rs10248420 | (G;G) | 7x more likely to respond to certain antidepres... |  | Link |  |
| 1 | rs11983225 | (C;T) | 7x more likely to respond to certain antidepres... |  | Link |  |
| 1 | rs182549 | (C;T) | Can digest milk. |  |  | Link |
| 1 | rs2235040 | (A;G) | 7x more likely to respond to certain antidepres... | Link | Link |  |
| 1 | rs2235067 | (A;G) | 7x more likely to respond to certain antidepres... |  |  |  |
| 1 | rs2546890 | (G;G) | Lower risk of multiple sclerosis |  |  |  |
| 1 | rs4148739 | (A;G) | 7x more likely to respond to certain antidepres... |  | Link |  |
| 1 | rs4939827 | (C;T) | 0.86 x decreased risk for colorectal cancer |  | Link |  |
| 1 | rs800292 | (C;T) | $1 \%$ decreased risk of macular degeneration | Link | Link | Link |
| 1.0 | rs2283123 | (C;T) | Decreased risk of schizophrenia in limited stud... |  |  |  |
| 1.0 | rs6583817 | (C;T) | $\sim_{0} 0.80 \mathrm{x}$ (lower) risk for late onset Alzheimer's ... |  |  |  |
| 0.5 | rs36094464 | (A;T) | Most likely benign: though reported years ago t... | Link | Link | Link |
| 0.1 | rs891512 | (G;G) | Lower blood pressure than those with an A allel... | Link |  |  |
| 0 | rs1047781 | (A;A) | ABH blood group "Secretor" status if Japanese | Link | Link | Link |
| 0 | rs1056836 | (G;G) | 0.3 x decreased risk for prostate cancer | Link | Link | Link |
| 0 | rs10897346 | (C;C) | If depressed: 2.6 x more likely to not respond t... |  |  |  |
| 0 | rs12252 | (T;T) | More resistant to influenza | Link |  | Link |
| 0 | rs16990018 | ( $\mathrm{A} ; \mathrm{A}$ ) | PrP Codon 171 Asn - Non-pathogenic variant | Link |  | 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 | 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 | rs8028689 | ( $\mathrm{T} ; \mathrm{T}$ ) | 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 | rs185919705 | (C;T) | Corneal dystrophy: fuchs endothelial: 8 | Link |  | Link |
| 4 | rs7158782 | (G;G) | 4x higher risk of adverse side-effect if taking... |  |  |  |
| 3 | rs13266634 | (C;C) | Increased risk for type-2 diabetes | Link | Link | Link |
| 3 | rs3738579 | ( $\mathrm{T} ; \mathrm{T}$ ) | 1.5x-2x increased risk for cervical cancer: H... |  |  |  |
| 3 | rs4244285 | (A;G) | Poorer metabolizer of several popular medicines... | Link | Link | Link |
| 2.9 | rs16901979 | ( $\mathrm{A} ; \mathrm{A}$ ) | 1.5x increased risk for prostate cancer |  | Link |  |
| 2.6 | rs13361189 | (C;C) | 2.6x increased risk for Crohn's disease |  | Link |  |
| 2.6 | rs4958847 | (A;A) | 2.6x increased risk for Crohn's disease |  |  |  |
| 2.5 | rs1121980 | (C;T) | 1.67 x risk for obesity |  | Link |  |
| 2.5 | rs12803066 | (A;G) | Increased risk of myopia |  |  |  |
| 2.5 | rs17595731 | (C;G) | ~5 fold higher risk for Fuchs' dystrophy: a cor... |  |  |  |
| 2.5 | rs2241880 | (C;C) | 2x-3x increased risk for Crohn's disease in Cau... | Link | Link | Link |
| 2.5 | rs339331 | ( $\mathrm{T} ; \mathrm{T}$ ) | Prostate cancer risk |  |  |  |
| 2.5 | rs3780374 | (A;G) | Substantially increased odds of developing V617... |  |  |  |
| 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 | rs664143 | (C;T) | Higher risk for number of cancers |  |  |  |
| 2.3 | rs1859962 | (G;G) | 1.28 x increased risk for prostate cancer |  | Link |  |
| 2.2 | rs2231137 | (G;G) | ${ }^{\sim} 1.5$-3x increased risk for ischemic stroke | Link | Link | Link |
| 2.1 | rs10090154 | (T;T) | 1.4 x increased risk for prostate cancer |  |  |  |
| 2.1 | rs10411210 | ( $\mathrm{T} ; \mathrm{T}$ ) | 1.15x increased risk of colorectal cancer |  | Link |  |
| 2.1 | rs1219648 | (G;G) | 1.64 x risk for breast cancer |  | Link |  |
| 2.1 | rs1329428 | (G;G) | 2 x increased risk for macular degeneration |  |  |  |
| 2.1 | rs17070145 | (C;C) | Reduced memory abilities |  |  | Link |
| 2.1 | rs2254958 | ( $\mathrm{C} ; \mathrm{T}$ ) | 1.24x increased risk for Alzheimer's |  |  |  |
| 2.1 | rs2294008 | (T;T) | Increased risk of gastric and bladder cancer | Link | Link |  |
| 2.1 | rs2383207 | (G;G) | Increased risk for heart disease |  |  |  |
| 2.1 | rs2420946 | (T;T) | 1.64 x risk for breast cancer |  |  |  |
| 2.1 | rs380390 | (C;C) | Increased risk for ARMD |  | Link |  |
| 2.1 | rs4402960 | (T;T) | 1.2x increased risk for type-2 diabetes |  | Link | Link |
| 2.1 | rs4430796 | ( $\mathrm{A} ; \mathrm{A}$ ) | 1.38 x increased risk for prostate cancer |  | Link |  |
| 2.1 | rs4444903 | (G;G) | 3.5x risk of hep-cancer in cirrhosis patients; ... |  |  |  |
| 2.1 | rs7837688 | (G;G) | 1.7 x increased risk for prostate cancer |  |  |  |
| 2.1 | rs9272346 | (A;G) | 5.5x risk type-1 diabetes |  | Link |  |
| 2 | rs10086908 | (C;T) | 1.7 x increased risk for prostate cancer |  |  |  |
| 2 | rs1024611 | (C;T) | Increased risk of exercise induced ischemia |  |  | Link |
| 2 | rs10811661 | (C;T) | 1.2x increased risk for type-2 diabetes |  | Link |  |
| 2 | rs10889677 | (C;C) | 1x increased risk for certain autoimmune diseas... |  | Link |  |
| 2 | rs10984447 | (A;A) | $>1.17 \mathrm{x}$ increased risk for multiple sclerosis |  | Link |  |
| 2 | rs11229030 | (C;C) | Higher odds of Crohn's disease |  |  |  |
| 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 | rs1375144 | (C;C) | 1.59x increased risk of developing bipolar diso... |  |  |  |
| 2 | rs16942 | (A;G) | Very slightly increased breast cancer risk | Link | Link | Link |
| 2 | rs17228212 | (C;T) | 1.26x increased risk for heart disease |  | Link |  |
| 2 | rs17435 | ( $\mathrm{T} ; \mathrm{T}$ ) | 1.4 x increased risk for lupus |  |  |  |
| 2 | rs1799732 | $(-; \mathrm{C})$ | 1.3x increased adenoma recurrence risk |  | Link |  |
| 2 | rs2073963 | (G;T) | Increased risk of baldness |  |  |  |
| 2 | rs2143340 | (C;T) | Increased risk of dyslexia and poor reading per... |  |  |  |
| 2 | rs2156921 | (A;G) | 1.29x increased risk for depression |  |  |  |
| 2 | rs2201841 | (T; T ) | 2.4x increased risk for Graves' disease |  | Link |  |
| 2 | rs2230201 | (G;G) | $>1.4 \mathrm{x}$ risk of lupus | Link |  |  |


| 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 | rs2352028 | (T;T) | Increased risk of lung cancer in non-smokers an... |  | Link |  |
| 2 | rs2383206 | (A;G) | 1.4 x increased risk for heart disease |  |  |  |
| 2 | rs25487 | (A;G) | 2x higher risk for skin cancer; possibly other ... | Link | Link |  |
| 2 | rs2736990 | (C;C) | Increased risk of developing Parkinson's Diseas... |  | Link |  |
| 2 | rs3212227 | $(\mathrm{A} ; \mathrm{C})$ | Significantly increased risk of developing cerv... |  |  |  |
| 2 | rs358806 | (C;C) | 1.78x increased risk of developing Type-2 diabe... |  | Link |  |
| 2 | rs3738919 | ( $\mathrm{A} ; \mathrm{C}$ ) | 1.94x risk of developing rheumatoid arthritis |  |  |  |
| 2 | rs3790565 | (C;C) | Increased risk of developing primary biliary ci... |  |  |  |
| 2 | rs4242382 | (A;A) | 1.7x increased risk for prostate cancer |  | Link |  |
| 2 | rs449647 | (A;A) | Lower levels of ApoE |  |  |  |
| 2 | rs4538475 | (G;G) | Increased risk of developing Parkinson's Diseas... |  | Link |  |
| 2 | rs4633 | (C;T) | Higher risk for endometrial cancer | Link | Link |  |
| 2 | rs4792311 | (A;G) | Increased risk of prostate cancer | Link | Link | Link |
| 2 | rs4825476 | (G;G) | 1.9x higher risk of suicidal thoughts when taki... |  | Link |  |
| 2 | rs493258 | (A;G) | 1.15x risk of Age Related Macular Degeneration |  |  |  |
| 2 | rs4968451 | (A;C) | 1.61x increased risk for meningioma |  |  |  |
| 2 | rs520354 | (A;G) | Increased risk in men for biliary conditions |  |  |  |
| 2 | rs6441286 | (G;T) | 1.54 x chance of developing primary biliary cirr... |  | Link |  |
| 2 | rs6457617 | (C;T) | 2.3x risk of rheumatoid arthritis |  | Link |  |
| 2 | rs6498169 | (A;A) | $>1.14 \mathrm{x}$ risk of multiple sclerosis |  | Link |  |
| 2 | rs6532197 | (G;G) | Increased risk of developing Parkinson's Diseas... |  | 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;G) | 1.5x higher risk for hypertension |  |  |  |
| 2 | rs699 | (C;C) | Increased risk of hypertension | Link | Link | Link |
| 2 | rs7190458 | (A;G) | Slightly higher pancreatic cancer risk | Link |  |  |
| 2 | rs738409 | (C;G) | Increased liver fat: odds of alcoholic liver di... | Link | Link |  |
| 2 | rs744373 | (C;T) | 1.17x risk of Alzheimer's |  |  |  |
| 2 | rs763361 | (T; T) | Increased risk for multiple autoimmune diseases... | Link | 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 | rs7923837 | (G;G) | 3.2x risk for T2D |  |  |  |
| 2 | rs7961152 | (A;C) | 1.2 x higher risk for hypertension |  |  |  |
| 2 | rs9303277 | (T;T) | 1.46x Increased risk of developing primary bili... |  |  |  |
| 2 | rs9543325 | (C; C ) | Slightly higher pancreatic cancer risk |  |  |  |
| 2.0 | rs9642880 | (T;T) | 1.5x increased bladder cancer risk |  | Link |  |
| 1.7 | rs1447295 | (A;A) | 1.7x increased risk of prostate cancer |  | Link |  |
| 1.6 | rs11523871 | (A;C) | 1.6x increased breast cancer risk for women ove... | Link | Link |  |
| 1.6 | rs1537415 | (C;G) | 1.6x increased risk for periodontitis |  | Link |  |
| 1.6 | rs2059693 | (T;T) | 1.6x increased risk for testicular cancer |  |  |  |
| 1.6 | rs2981745 | (C;T) | 1.6x increased risk for breast cancer in female... |  |  |  |
| 1.6 | rs356219 | (G;G) | 1.6x increased risk for Parkinson's disease |  |  |  |
| 1.6 | rs3775948 | (C;G) | Slightly higher risk for gout |  |  |  |
| 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 | rs10883365 | (A;G) | 1.2 x increased risk for developing Crohn's dise... |  | Link |  |
| 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 | rs13149290 | (C;C) | Slightly increased risk of developing prostate ... |  |  |  |
| 1.5 | rs1360517 | (A;G) | Higher susceptibility for AIDS |  | Link |  |


| Mag. | Identifier | Genotype | Summary | ExAC | GetEvidence | ClinVar |
| :---: | :---: | :---: | :---: | :---: | :---: | :---: |
| 1.5 | rs140701 | (A;G) | Increased risk for anxiety disorders |  |  |  |
| 1.5 | rs16944 | (A;G) | Minorly increased risk of mental illness and os... |  | Link |  |
| 1.5 | rs1801020 | (C;T) | 1.31x increased risk of heart disease | Link |  | Link |
| 1.5 | rs2280714 | (A;G) | 1.4x increased risk of SLE |  |  |  |
| 1.5 | rs28694718 | (A;G) | 2 x higher risk for schizophrenia |  |  |  |
| 1.5 | rs309375 | ( $\mathrm{T} ; \mathrm{T}$ ) | Larger mosquito bites |  |  |  |
| 1.5 | rs356220 | ( $\mathrm{T} ; \mathrm{T}$ ) | Increased risk of Parkinson's Disease |  |  |  |
| 1.5 | rs3745516 | (A;G) | Slightly increased risk of developing primary b... |  |  |  |
| 1.5 | rs3825776 | (A;G) | 1.3x increased risk for ALS |  | Link |  |
| 1.5 | rs464049 | (T; $\mathrm{T}^{\text {) }}$ | Increased risk of schizophrenia in limited stud... |  |  |  |
| 1.5 | rs4656461 | (A;G) | 1.5x increased risk for open angle glaucoma |  |  |  |
| 1.5 | rs4785763 | (A;C) | 1.5x higher risk for melanoma |  | Link |  |
| 1.5 | rs4979462 | (C;T) | Slightly increased risk of developing primary b... |  |  |  |
| 1.5 | rs699473 | (C;T) | $\sim 1.5 \mathrm{x}$ increased brain tumor risk |  |  |  |
| 1.5 | rs7850258 | (G;G) | Slightly higher odds of developing primary hypo... |  |  |  |
| 1.5 | rs966221 | (C;C) | 1.5x increased stroke risk certain populations |  |  |  |
| 1.4 | rs1126497 | (C;T) | 1.4 x increased risk for breast cancer | Link | Link | Link |
| 1.4 | rs12770228 | (A;G) | 1.4x increased risk for meningioma |  |  |  |
| 1.4 | rs2046210 | (C;T) | 1.4x increased breast cancer risk |  | Link |  |
| 1.4 | rs3131296 | (G;G) | 1.4 x increased risk for schizophrenia |  | Link |  |
| 1.4 | rs6010620 | (G;G) | 1.4x higher risk for glioma development; but th... |  | Link |  |
| 1.3 | rs10947262 | (C;C) | 1.3x increased risk for osteoarthritis |  |  |  |
| 1.3 | rs110419 | (A;G) | 1.3x increased risk for neuroblastoma |  |  |  |
| 1.3 | rs2736100 | (G;T) | 1.3x higher risk for glioma development |  | Link |  |
| 1.3 | rs34330 | (C;T) | 1.3x higher risk for endometrial cancer (in Chi... |  |  |  |
| 1.3 | rs501120 | (A;G) | 1.3x increased risk for heart disease |  | Link |  |
| 1.25 | rs13387042 | (A;A) | 1.24 x increased risk for breast cancer |  | Link |  |
| 1.25 | rs748404 | ( $\mathrm{T} ; \mathrm{T}$ ) | Slightly increased risk (1.25) for lung cancer... |  | Link |  |
| 1.2 | rs10865331 | (A;G) | 1.2x higher risk for ankylosing spondylitis |  |  |  |
| 1.2 | rs11037909 | ( $\mathrm{T} ; \mathrm{T}$ ) | 1.47x type II diabetes risk | Link |  |  |
| 1.2 | rs1344706 | ( $\mathrm{T} ; \mathrm{T}$ ) | 1.2x increased risk for schizophrenia |  | Link |  |
| 1.2 | rs2072590 | (G;T) | 1.2 x increased risk for ovarian cancer |  |  |  |
| 1.2 | rs2076295 | (G;T) | One copy of the risk allele (G): slightly incre... |  |  |  |
| 1.2 | rs2651899 | (G;G) | 1.2 x higher risk for migraines |  |  |  |
| 1.2 | rs3740878 | (A;A) | 1.46x type II diabetes risk; common | Link |  |  |
| 1.2 | rs4496877 | ( $\mathrm{T} ; \mathrm{T}$ ) | For type-1 diabetics: 1.6x increased nephropath... |  |  |  |
| 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 |  |
| 1.17 | rs17465637 | (A;C) | 1.17 x higher risk for myocardial infarction | Link | Link |  |
| 1.1 | rs11110912 | (C;C) | 1.3x high blood pressure risk |  |  |  |
| 1.1 | rs11650354 | (C;T) | Possible risk for allergic asthma | Link |  |  |
| 1.1 | rs11650494 | (A;G) | Slightly higher prostate cancer risk |  |  |  |
| 1.1 | rs1799966 | (A;G) | Very slightly increased breast cancer risk | 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 | 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) | 1.2 x increased risk for chronic kidney disease |  |  |  |
| 1.1 | rs6897876 | (C;T) | Slight increase in testicular cancer risk for m... |  |  |  |
| 1.1 | rs7171755 | (A;A) | Very slight descrease in cortical thickness and... |  |  |  |
| 1.1 | rs7412 | (C;C) | More likely to gain weight if taking olanzapine... | Link | Link | Link |
| 1.1 | rs889312 | ( $\mathrm{A} ; \mathrm{C}$ ) | Very slightly higher risk for breast cancer |  | Link |  |
| 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 | rs10761659 | (A;G) | 1.2x risk of Crohn's disease |  | Link |  |


| Mag. | Identifier | Genotype | Summary | ExAC | GetEvidence | ClinVar |
| :--- | :--- | :--- | :--- | :--- | :--- | :--- |
| 1 | rs1143674 | (A;G) | 1.3x increased autism risk | Link |  |  |
| 1 | rs1804197 | (A;C) | Increased risk of familial colorectal cancer an... |  |  | Link |
| 1 | rs2228000 | (T;T) | Statistically significant: but slight: increase... | Link | Link | Link |
| 1 | rs3194051 | (A;G) | $1.12 x$ risk of type-1 diabetes | Link | Link | Link |
| 1 | rs6932590 | (C;T) | $1.1 x$ increased risk for schizophrenia |  | Link |  |
| 1 | rs798766 | (T;T) | Increased susceptibility urinary bladder cancer... |  |  |  |
| 1 | rs987525 | (A;C) | 2.5x increased risk for cleft lip |  | Link |  |
| 0.1 | rs2070744 | (C;C) | Increased prostate cancer risk |  | Link | Link |
| 0.1 | rs601338 | (G;G) | Susceptible to Norovirus infections | Link | Link | Link |
| 0 | rs1333040 | (C;T) | $1.24 x$ increased myocardial infarction risk: $1.2 \ldots$ |  | Link |  |
| 0 | rs3761418 | (A;A) | $1.3 x$ increased risk for depression |  |  |  |
| 0 | rs6277 | (C;C) | $1.6 x$ higher schizophrenia risk | Link | Link | Link |

- Genosets (Multi-variant Phenotypes)

| Magnitude | Identifier | Summary |
| :--- | :--- | :--- |
| 4 | gs144 | Male |
| 3 | gs273 | Lowest risk (13\% of white women) of Atrial Fibr... |
| 2.5 | gs157 | More stimulated by coffee |
| 2.5 | gs281 | Part of the 88\% of the population claimed not t... |
| 2.2 | gs280 | Light hair color for europeans |
| 2 | gs101 | Probably able to digest milk |
| 2 | gs136 | Normal: but this haplotype in 8q24 increases ri... |
| 2 | gs140 | NAT2 slow metabolizer |
| 2 | gs154 | NAT2 Slow metabolizer |
| 2 | gs173 | CYP2D6*10 |
| 2 | gs221 | Autoimmune disorder risk in Europeans |
| 2 | $g s 246$ | APOE3/APOE3 |
| 1.5 | gs247 | Parkinson's Disease Risk |
| 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).

