# PGP-UK Genomics Report for uk496F0B

## 1 Summary

This is the genome report was produced using collaborative research tools, including SNPedia and GetEvidence. This section shows an overview of all the small variants which were found in the genome for this individual, when compared with a reference genome. These variants are summarised in Table 1 and the pie-charts in Figures 2, 3 and 4.

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. The diagram in Figure 1 is a simplification of the usual gene structure. "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.

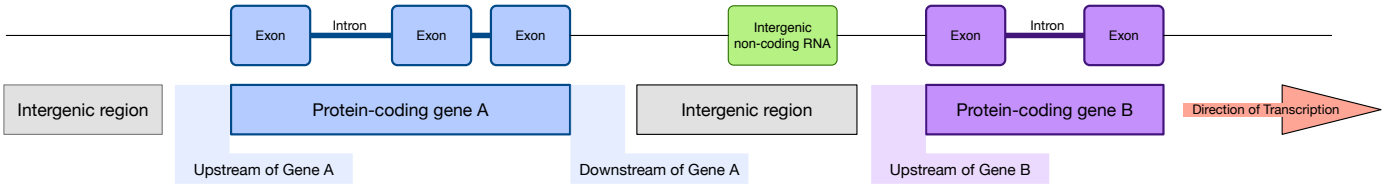


Figure 1: Diagram of gene structure indicating locations of potential variants

| Feature | Count |
|---|---|
| Lines of input read | 4964935 |
| Variants filtered out | 0 |
| Novel / existing variants | 496460 (10.0) / 4456347 (90.0) |
| Overlapped genes | 56732 |
| Overlapped transcripts | 67527 |
| Overlapped regulatory features | 166509 |

Table 1: Variant calling summary

There are several different types of genomic variants. The most common change is when one single building block of the DNA (called a nucleotide) is changed, called a single nucleotide variants (SNV). 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.

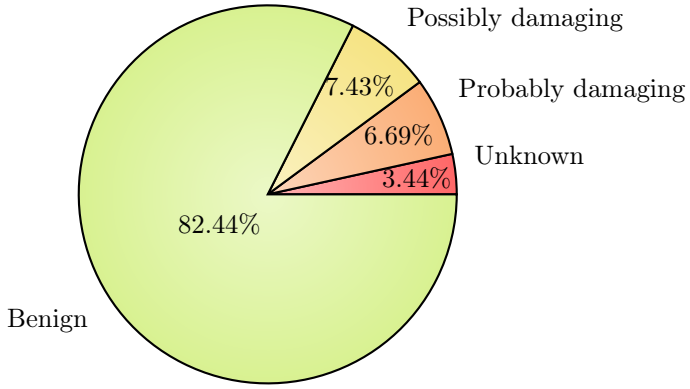


Figure 2: PolyPhen Summary

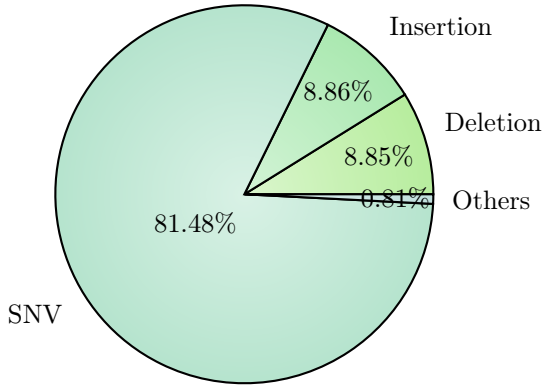


Figure 3: Variant Class

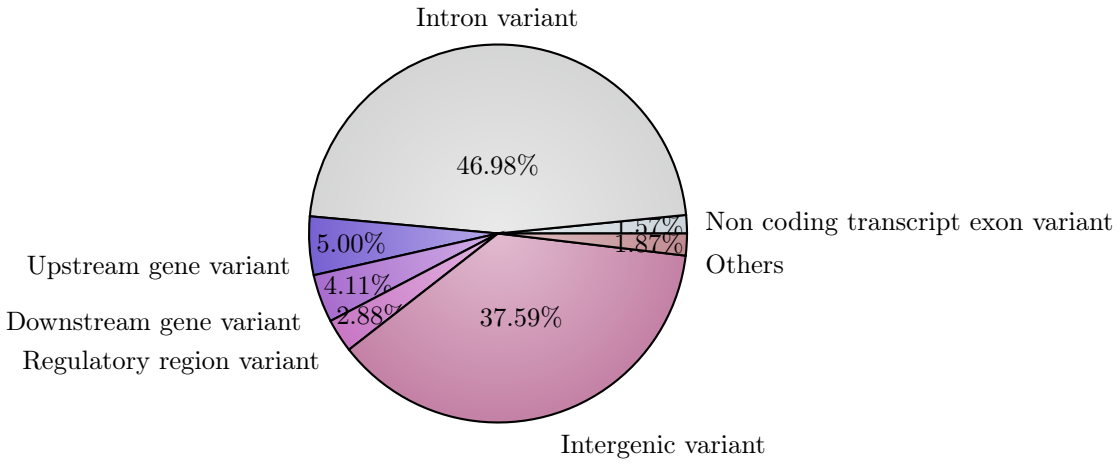


Figure 4: Consequence type

## 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.

Based on the populations defined in the 1000 genomes project (1kGP), the ancestry composition for this individual is inferred to be 100.0 percent European [British in England and Scotland].

Please note that this analysis is limited by the populations available in the 1kGP data. If there are European subpopulations reported, and the ancestry of the participant does not correspond to any of the 1kGP populations, the closest 1kGP sampled subpopulation will be shown (even though it might be different from the participant's actual ancestry).

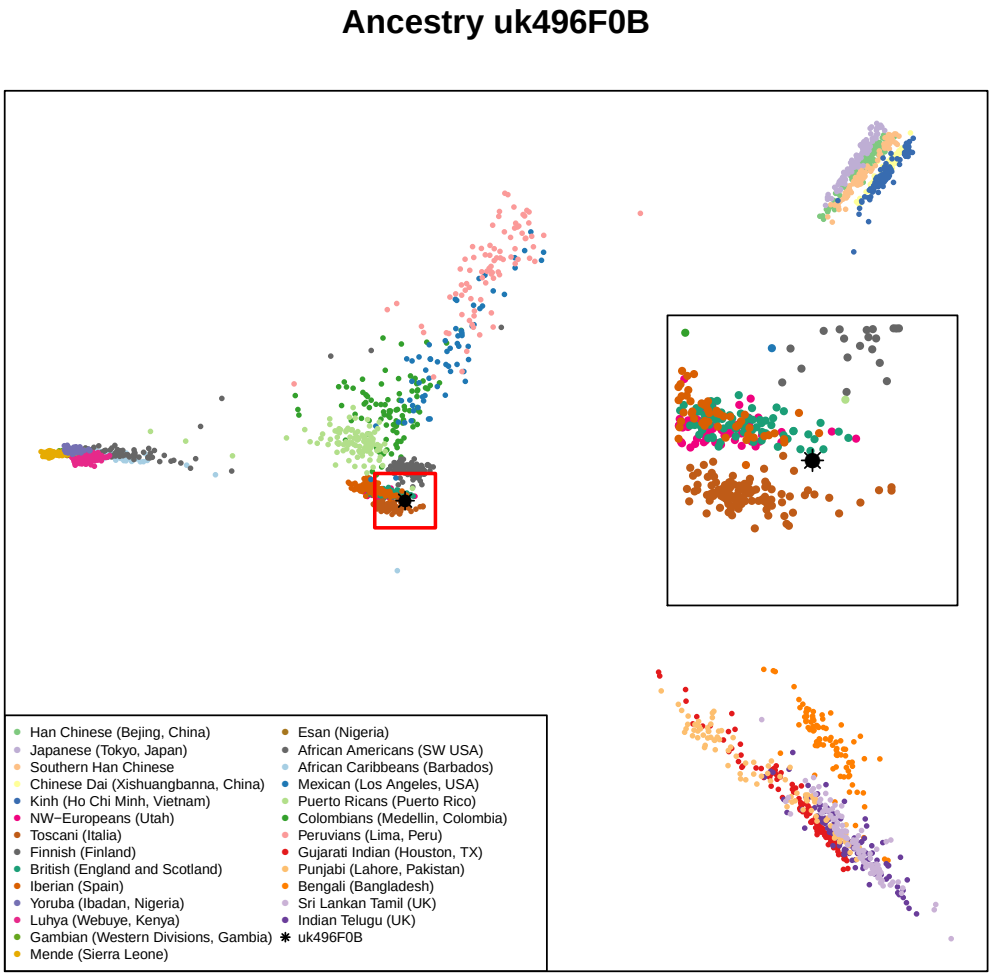


Figure 5: Ancestry Principal Component Analysis

# 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.

## 3.1 Possibly Beneficial Traits

| Mag. | Identifier | Genotype | Summary | ExAC | GetEvidence | ClinVar |
|---|---|---|---|---|---|---|
| 3 | rs7294919 | (C;T) | Moderately enhanced hippocampal volume | | | |
| 2.4 | rs2802288 | (A;A) | Longer lifespan | | | |
| 2.2 | rs2511989 | (A;A) | 0.44x decreased age-related macular degeneratio... | | Link | |
| 2.1 | rs3775291 | (A;G) | 0.71x decreased risk for dry age related macula... | Link | Link | Link |
| 2.1 | rs6505162 | (A;A) | 0.43x decreased risk for esophageal cancer | Link | | |
| 2 | rs1012053 | (A;C) | 0.625x reduced risk of Bipolar Disorder. | | Link | |
| 2 | rs11045585 | (A;A) | 24% chance (lower than average) of docetaxel-in... | | Link | |
| 2 | rs1136410 | (C;T) | 0.80x reduced risk for glioblastoma | Link | Link | |
| 2 | rs12979860 | (C;C) | ~80% of such hepatitis C patients respond to tr... | | Link | Link |
| 2 | rs1544410 | (G;G) | Decreased risk of low bone mineral density diso... | | Link | |
| 2 | rs17070145 | (C;T) | Increased memory performance | | | Link |
| 2 | rs1864163 | (G;G) | Associated with higher HDL cholesterol | | Link | |
| 2 | rs261332 | (A;G) | Associated with higher HDL cholesterol | | | |
| 2 | rs2707466 | (A;A) | Stronger bones | Link | Link | |
| 2 | rs2764264 | (C;C) | Greater odds of living to 95 | | | |
| 2 | rs2908004 | (T;T) | Stronger bones | Link | Link | |
| 2 | rs3738579 | (C;T) | 0.5x decreased risk for cervical cancer: HNSCC:... | | | |
| 2 | rs3764261 | (G;T) | Associated with higher HDL cholesterol | | Link | Link |
| 2 | rs3782179 | (C;T) | 3x lower odds of testicular cancer risk for men... | | | |
| 2 | rs3819331 | (T;T) | Lower risk of autism | Link | | |
| 2 | rs3914132 | (C;T) | Lower otosclerosis risk | | Link | |
| 2 | rs4149268 | (G;G) | Associated with higher HDL cholesterol | | Link | |
| 2 | rs6807362 | (G;G) | Decreased autism risk | Link | Link | |
| 2 | rs7216389 | (C;C) | 0.69x lower risk of Childhood Asthma. | | Link | |
| 2 | rs801114 | (T;T) | 0.78x decreased Basal Cell Carcinoma risk. | | Link | |
| 2 | rs8070723 | (A;G) | 0.18x reduced risk of developing progressive su... | | | |
| 1.8 | rs1128535 | (A;G) | 0.77x risk for Crohn's disease | | | |

| Mag. | Identifier | Genotype | Summary | ExAC | GetEvidence | ClinVar |
|---|---|---|---|---|---|---|
| 1.8 | rs1746048 | (C;T) | 0.94 decreased risk for coronary heart disease | | Link | |
| 1.8 | rs1800588 | (C;T) | Higher HDL-C levels | Link | Link | |
| 1.8 | rs4714156 | (C;C) | <0.61x risk for restless legs | | | |
| 1.5 | rs1063192 | (C;C) | 0.71x reduced risk of myocardial infarction | | | |
| 1.5 | rs11212617 | (A;C) | Somewhat increased likelihood of treatment succ... | | | 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;C) | Associated with higher HDL (good) cholesterol. | | | |
| 1.5 | rs4939883 | (C;C) | Associated with higher HDL cholesterol | | Link | |
| 1.5 | rs610932 | (A;A) | A allele associated with reduced risk of Alzhei... | | | |
| 1.4 | rs1165205 | (A;T) | 0.85x decreased gout risk | | Link | |
| 1.4 | rs2294008 | (C;C) | Lower risk of gastric and bladder cancer | Link | Link | |
| 1.4 | rs6495446 | (C;T) | 0.8x reduced risk for chronic kidney disease | | | |
| 1.3 | rs9306160 | (T;T) | 0.75x (reduced) risk for metastasis in LN-/ER+ ... | Link | Link | |
| 1.2 | rs11246226 | (A;C) | Decreased risk of schizophrenia in limited stud... | | Link | |
| 1.2 | rs4320932 | (A;G) | 0.87x decreased risk for ovarian cancer | | | |
| 1.2 | rs4686484 | (G;G) | Slightly decreased risk for celiac disease | | | |
| 1.1 | rs13333226 | (A;G) | Slightly lower risk for hypertension | | | Link |
| 1.1 | rs2293347 | (G;G) | Among NSCLC patients: better Gefitinib response... | Link | | Link |
| 1.1 | rs4988235 | (T;T) | Can digest milk | | | Link |
| 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 | (T;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 | rs7850258 | (A;G) | Typical odds of developing primary hypothyroidi... | | | |
| 1 | rs800292 | (C;T) | 1% decreased risk of macular degeneration | Link | Link | Link |
| 0.1 | rs1726866 | (C;C) | Can taste bitter | 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 | rs12252 | (T;T) | More resistant to influenza | Link | | Link |
| 0 | rs16990018 | (A;A) | PrP Codon 171 Asn - Non-pathogenic variant | Link | | Link |
| 0 | rs17244841 | (A;A) | More responsive to statin treatment | | 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 | rs242941 | (G;G) | Better response to inhaled corticosteroid in pa... | | Link | |
| 0 | rs28933385 | (G;G) | Prion protein Codon 200 (E) - Non pathogenic va... | | | Link |
| 0 | rs5065 | (A;A) | 1.12x risk on diuretic; if hypertensive: better... | Link | Link | Link |
| 0 | rs6259 | (G;G) | Best inverse correlation between tea-drinking: ... | Link | Link | |
| 0 | rs74315403 | (G;G) | PrP codon 178 (D) - non pathogenic variant | | | Link |

## 3.2 Possibly Harmful Traits

| Mag. | Identifier | Genotype | Summary | ExAC | GetEvidence | ClinVar |
|---|---|---|---|---|---|---|
| 4 | rs1333049 | (C;C) | 1.9x increased risk for coronary artery disease... | | Link | |
| 3.2 | rs2981582 | (T;T) | 1.7x higher risk of ER+ breast cancer | | Link | |
| 3.1 | rs1421085 | (C;C) | ~1.7x increased obesity risk | | Link | Link |
| 3 | rs10897346 | (C;C) | If depressed: 2.6x more likely to not respond t... | | | |
| 3 | rs1121980 | (T;T) | 2.76x risk for obesity | | Link | |
| 3 | rs2306402 | (C;C) | 1.18x increased risk for late-onset Alzheimer's... | | | |
| 3 | rs4244285 | (A;G) | Poorer metabolizer of several popular medicines... | Link | Link | Link |
| 3 | rs7754840 | (C;G) | 1.3x increased risk for type-2 diabetes | | Link | |
| 2.5 | rs10490924 | (G;T) | 2.7x risk for age related macular degeneration | Link | Link | Link |
| 2.5 | rs13266634 | (C;T) | Increased risk for type-2 diabetes | Link | Link | Link |
| 2.5 | rs17595731 | (C;G) | ~5 fold higher risk for Fuchs' dystrophy: a cor... | | | |
| 2.5 | rs1799971 | (A;G) | Stronger cravings for alcohol. if alcoholic: na... | Link | Link | Link |
| 2.5 | rs1800255 | (A;A) | Increased risk for pelvic organ prolapse | Link | Link | Link |
| 2.5 | rs187238 | (G;G) | Hypertension increases risk 3.75x for sudden ca... | | | |
| 2.5 | rs2943634 | (C;C) | Slightly higher risk of ischemic stroke | | Link | |
| 2.5 | rs3780374 | (A;G) | Substantially increased odds of developing V617... | | | |
| 2.5 | rs5888 | (C;T) | 3x higher risk for age-related macular degenera... | Link | | |
| 2.5 | rs660895 | (G;G) | 6x higher risk of rheumatoid arthritis | | Link | |
| 2.5 | rs664143 | (C;T) | Higher risk for number of cancers | | | |
| 2.5 | rs9934438 | (A;A) | Coumadin resistance | | Link | Link |
| 2.3 | rs1859962 | (G;G) | 1.28x increased risk for prostate cancer | | 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 |
| 2.1 | rs10427255 | (C;C) | Highest odds of photic sneeze reflex | | | |
| 2.1 | rs10811661 | (T;T) | 1.2x increased risk for type-2 diabetes | | Link | |
| 2.1 | rs11887534 | (C;G) | 2x increased risk for gallstones | Link | Link | Link |
| 2.1 | rs1219648 | (G;G) | 1.64x risk for breast cancer | | Link | |
| 2.1 | rs1695 | (G;G) | 3.5x asthma risk in certain populations | Link | Link | Link |
| 2.1 | rs2383207 | (G;G) | Increased risk for heart disease | | | |
| 2.1 | rs2420946 | (T;T) | 1.64x risk for breast cancer | | | |
| 2.1 | rs4430796 | (A;A) | 1.38x increased risk for prostate cancer | | Link | |
| 2.1 | rs5186 | (A;C) | ~1.4x increased risk of hypertension | Link | Link | Link |
| 2.1 | rs6457617 | (T;T) | 5.2x risk of rheumatoid arthritis | | Link | |
| 2.1 | rs646776 | (A;A) | 1.2x risk of coronary artery disease | | Link | |
| 2.1 | rs795484 | (A;G) | Increased morphine dose requirement and postope... | | | |
| 2.1 | rs944289 | (C;T) | 1.3x increased thyroid cancer risk | | Link | |
| 2 | rs10260404 | (C;C) | 1.60x risk of developing ALS | | Link | |
| 2 | rs10492519 | (G;G) | Increased risk of developing prostate cancer | | | |
| 2 | rs10757272 | (T;T) | 1.54x increased risk for Coronary artery diseas... | | | |
| 2 | rs10871777 | (A;G) | Adults likely to be 0.22 BMI units higher | | | |
| 2 | rs10984447 | (A;G) | 1.17x increased risk for multiple sclerosis | | Link | |
| 2 | rs11229030 | (C;C) | Higher odds of Crohn's disease | | | |
| 2 | rs1160312 | (A;A) | 1.6x increased risk of Male Pattern Baldness. | | Link | |
| 2 | rs12431733 | (T;T) | Increased risk of developing Parkinson's Diseas... | | Link | |
| 2 | rs12567232 | (A;G) | Increased risk for Crohn's Disease | | Link | |
| 2 | rs12696304 | (C;G) | Prone to aging faster: at least in European pop... | | | |
| 2 | rs1360780 | (C;T) | 1.3x increased risk for depression | | Link | |
| 2 | rs1361600 | (G;G) | ~2x increased risk for adult-onset asthma in Ja... | | | |
| 2 | rs16942 | (A;G) | Very slightly increased breast cancer risk | Link | Link | Link |
| 2 | rs17001266 | (-;C) | 1.58x increased risk for schizophrenia in males... | | | |
| 2 | rs17228212 | (C;T) | 1.26x increased risk for heart disease | | Link | |
| 2 | rs1734791 | (A;A) | 1.4x increased risk for lupus | | | |
| 2 | rs17782313 | (C;T) | Adults likely to be 0.22 BMI units higher | | Link | Link |

| Mag. | Identifier | Genotype | Summary | ExAC | GetEvidence | ClinVar |
|---|---|---|---|---|---|---|
| 2 | rs1799732 | (-;C) | 1.3x increased adenoma recurrence risk | | Link | |
| 2 | rs1800896 | (A;G) | 1.6x increased prostate cancer risk | | | |
| 2 | rs2073963 | (G;T) | Increased risk of baldness | | | |
| 2 | rs2156921 | (A;G) | 1.29x increased risk for depression | | | |
| 2 | rs2201841 | (C;T) | 1.5x increased risk for Crohn's disease; 2x inc... | | 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 | rs2305480 | (T;T) | If 4 years old or younger: ~3x increased asthma... | Link | Link | |
| 2 | rs25487 | (A;G) | 2x higher risk for skin cancer; possibly other ... | Link | Link | Link |
| 2 | rs2736100 | (T;T) | Higher risk of Interstitial lung disease: and t... | | Link | |
| 2 | rs27388 | (A;A) | Increased risk of developing schizophrenia | | | |
| 2 | rs351855 | (C;T) | 1.2x increased risk for prostate cancer | Link | 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 | rs4633 | (T;T) | Higher risk for endometrial cancer | Link | 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 | rs520354 | (A;G) | Increased risk in men for biliary conditions | | | |
| 2 | rs6435862 | (G;G) | 2.8x higher risk of aggressive neuroblastoma | | Link | |
| 2 | rs6700125 | (T;T) | 1.76x increased risk for ALS | | | |
| 2 | rs6897932 | (C;C) | 1.08x 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 | rs7190458 | (A;G) | Slightly higher pancreatic cancer risk | Link | | |
| 2 | rs7442295 | (A;A) | ~4x higher risk for hyperuracemia | | Link | |
| 2 | rs7639618 | (C;T) | 1.45x increased osteoarthritis risk | Link | | |
| 2 | rs7774434 | (C;C) | Increased risk of developing primary biliary ci... | | | |
| 2 | rs7776725 | (C;C) | Weaker bones | | 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;A) | 1.5x higher risk for hypertension | | | |
| 2 | rs854560 | (A;T) | Higher risk for heart disease: diabetic retinop... | Link | Link | Link |
| 2 | rs855913 | (G;T) | Reduced survival with ALS | | Link | |
| 2 | rs9303277 | (T;T) | 1.46x Increased risk of developing primary bili... | | | |
| 2 | rs9525638 | (T;T) | Weaker bones | | | |
| 2 | rs965513 | (A;G) | 1.77x increased thyroid cancer risk | | Link | |
| 2 | rs9954153 | (G;T) | ~2.5x higher risk for Fuchs' dystrophy: a corne... | | | |
| 2.0 | rs1434536 | (A;A) | 1.94x increased breast cancer risk | | | |
| 2.0 | rs2305795 | (A;A) | 1.64x higher risk of narcolepsy compared to (G;... | | | Link |
| 2.0 | rs9642880 | (T;T) | 1.5x increased bladder cancer risk | | Link | |
| 1.8 | rs1136287 | (C;T) | 1.5x increased risk of wet ARMD in a Taiwanese ... | Link | Link | |
| 1.8 | rs2278206 | (T;T) | 1.16x increased risk for asthma | Link | Link | |
| 1.8 | rs4474514 | (A;G) | 3x increased testicular cancer risk for men | | Link | |
| 1.8 | rs733618 | (A;G) | 1.87x risk for myasthenia gravis | | | |
| 1.7 | rs8055236 | (G;T) | 1.9x risk for heart disease | | 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 | rs2981745 | (C;T) | 1.6x increased risk for breast cancer in female... | | | |
| 1.6 | rs3764880 | (A;A) | 1.2 - 1.8x increased tuberculosis risk | Link | Link | |
| 1.5 | rs10464059 | (A;G) | Slightly increased risk of developing Parkinson... | | | |
| 1.5 | rs10883365 | (A;G) | 1.2x increased risk for developing Crohn's dise... | | Link | |

| Mag. | Identifier | Genotype | Summary | ExAC | GetEvidence | ClinVar |
|---|---|---|---|---|---|---|
| 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 | rs12210050 | (C;T) | Slightly higher risk for basal cell carcinoma | | Link | |
| 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;T) | Slightly increased risk of developing prostate ... | | | |
| 1.5 | rs13181 | (G;T) | 1.12x increased risk for cutaneous melanoma | Link | Link | Link |
| 1.5 | rs13376333 | (C;T) | 1.5x higher risk of atrial fibrillation | | Link | |
| 1.5 | rs1360517 | (A;G) | Higher susceptibility for AIDS | | Link | |
| 1.5 | rs1375144 | (C;C) | 1.59x increased risk of developing bipolar diso... | | | |
| 1.5 | rs140701 | (A;G) | Increased risk for anxiety disorders | | | |
| 1.5 | rs144848 | (G;T) | Very slightly increased breast cancer risk | Link | Link | Link |
| 1.5 | rs16944 | (A;G) | Minorly increased risk of mental illness and os... | | Link | |
| 1.5 | rs17115100 | (G;T) | Slightly increased risk of developing Parkinson... | Link | Link | |
| 1.5 | rs17221417 | (C;G) | 1.3x higher risk for Crohn's disease | | Link | |
| 1.5 | rs1801020 | (C;T) | 1.31x increased risk of heart disease | Link | | Link |
| 1.5 | rs1801274 | (T;T) | Complex; generally greater risk for cancer prog... | Link | Link | Link |
| 1.5 | rs199533 | (C;T) | Slightly increased risk of developing Parkinson... | Link | | |
| 1.5 | rs2007153 | (G;G) | Increased risk of schizophrenia in limited stud... | | | |
| 1.5 | rs2240340 | (A;G) | Slightly increased (1.5x) risk for RA | 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 | rs2881766 | (T;T) | Slightly increased risk for pregnancy-induced h... | | | |
| 1.5 | rs3087243 | (G;G) | Increased risk for autoimmune diseases | | Link | |
| 1.5 | rs309375 | (T;T) | Larger mosquito bites | | | |
| 1.5 | rs3212227 | (A;A) | 1.43x increased risk of developing psoriasis an... | | | |
| 1.5 | rs3790565 | (C;T) | Slightly increased risk of developing primary b... | | | |
| 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 | (C;T) | Increased risk of schizophrenia in limited stud... | | | |
| 1.5 | rs486907 | (A;G) | 1.5x increased prostate cancer risk | Link | Link | Link |
| 1.5 | rs5746059 | (A;A) | Slightly higher fat mass | | | |
| 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 | rs6896702 | (C;T) | Slightly increased risk of developing Parkinson... | | | |
| 1.5 | rs699473 | (C;C) | ~1.5x increased brain tumor risk | | | |
| 1.5 | rs7454108 | (C;C) | Homozygous for HLA-DQ8 haplotype | | | |
| 1.5 | rs9652490 | (A;G) | Slightly increased risk of developing Parkinson... | | Link | |
| 1.4 | rs1126497 | (C;T) | 1.4x increased risk for breast cancer | Link | Link | Link |
| 1.4 | rs2046210 | (C;T) | 1.4x increased breast cancer risk | | Link | Link |
| 1.4 | rs3131296 | (G;G) | 1.4x increased risk for schizophrenia | | Link | |
| 1.4 | rs4795067 | (G;G) | Slight increase in risk for psoriatic arthritis... | | | |
| 1.4 | rs4959039 | (A;G) | 1.4x higher risk for multiple sclerosis | | | |
| 1.4 | rs8050136 | (A;A) | 1.4x increased risk for T2D in some populations... | | Link | |
| 1.34 | rs17465637 | (C;C) | 1.34x higher risk for myocardial infarction | Link | Link | |
| 1.3 | rs1042713 | (A;G) | 1.3x increased risk that pediatric inhaler use ... | Link | Link | Link |
| 1.3 | 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 | rs1260326 | (C;T) | Slightly higher risk for gout | Link | Link | Link |

| Mag. | Identifier | Genotype | Summary | ExAC | GetEvidence | ClinVar |
|---|---|---|---|---|---|---|
| 1.3 | rs13361189 | (C;T) | 1.3x increased risk for Crohn's disease | | Link | |
| 1.3 | rs2024513 | (A;G) | 1.3x higher risk for schizophrenia (among Han C... | | | |
| 1.3 | rs2059693 | (C;T) | 1.3x increased risk for testicular cancer | | | |
| 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 | rs4958847 | (A;G) | 1.3x increased risk for Crohn's disease | | | |
| 1.3 | rs501120 | (A;G) | 1.3x increased risk for heart disease | | Link | |
| 1.25 | rs748404 | (T;T) | Slightly increased risk (1.25) for lung cancer... | | Link | |
| 1.2 | rs1800693 | (A;G) | Slight (1.2x) increase in risk for multiple scl... | Link | Link | Link |
| 1.2 | rs2076295 | (G;T) | One copy of the risk allele (G): slightly incre... | | | |
| 1.2 | rs2252586 | (A;G) | 1.2x higher risk for glioma development | | | |
| 1.2 | rs393152 | (A;G) | Slight increased risk of both PD and AD | Link | Link | |
| 1.2 | rs419788 | (A;G) | 2.0x risk for lupus | Link | | |
| 1.2 | rs449647 | (A;T) | Possibly lower levels of ApoE | | | |
| 1.2 | rs4977756 | (A;G) | 1.39x higher risk for glioma development | | Link | |
| 1.2 | rs498872 | (C;T) | 1.2x higher risk for glioma development | | Link | |
| 1.17 | rs3802842 | (A;C) | 1.17x increased risk of colorectal cancer | | Link | |
| 1.1 | rs11037909 | (C;T) | 1.27x type II diabetes risk | 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 | rs13387042 | (A;G) | 1.12x increased risk for breast cancer | | Link | |
| 1.1 | rs1344706 | (G;T) | 1.1x increased risk for schizophrenia | | Link | |
| 1.1 | rs2295190 | (G;T) | Slightly increased risk for ovarian cancer in w... | Link | Link | Link |
| 1.1 | rs249954 | (C;T) | Potentially increased risk of Breast Cancer | | | Link |
| 1.1 | rs2651899 | (A;G) | 1.1x higher risk for migraines | | | |
| 1.1 | rs34516635 | (G;G) | Less longevity for Ashkenazi Jewish women. | Link | | Link |
| 1.1 | rs3740878 | (A;G) | 1.26x type II diabetes risk | Link | | Link |
| 1.1 | rs3818361 | (C;T) | 1.15x increased risk for late-onset Alzheimer's... | | | |
| 1.1 | rs4324715 | (C;T) | 1.5x increased testicular cancer risk for men | | | |
| 1.1 | rs6800901 | (T;T) | 1.3x multiple myeloma risk | | | |
| 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 | 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 | rs925391 | (C;C) | More likely to go bald; common | | | |
| 1.07 | rs2291834 | (C;C) | Very slightly higher risk for myocardial infarc... | | | |
| 1 | rs10504861 | (G;G) | Major allele: normal risk of migraine | | | |
| 1 | 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 | (T;T) | 1.1x increased risk for schizophrenia | | Link | |
| 0.1 | rs601338 | (A;G) | Susceptible to Norovirus infections | Link | Link | Link |
| 0 | rs10761659 | (A;A) | 1.5x risk of Crohn's disease | | Link | |
| 0 | rs1495965 | (A;A) | 1.2x higher risk for spondylitis | | | |
| 0 | rs440446 | (G;G) | Increased risk in men for biliary conditions | Link | | |
| 0 | rs4712653 | (T;T) | 2x increased risk for neuroblastoma | | | |
| 0 | rs4795400 | (T;T) | If 4 years old or younger: ~2.5x increased asth... | | Link | |
| 0 | rs6314 | (C;C) | Higher risk for RA | Link | Link | |

### 3.3 Genosets (Multi-variant Phenotypes)

| Magnitude | Identifier | Summary |
|---|---|---|
| 3 | gs241 | Lighter green: brown or hazel eye color |
| 2.5 | gs102 | ALS risk |
| 2.5 | gs155 | CYP3A5 non-expressor |
| 2.5 | gs281 | Part of the 88% of the population claimed not t... |
| 2.4 | gs297 | Lower heart attack risk than average |
| 2 | gs101 | Probably able to digest milk |
| 2 | gs154 | NAT2 Slow metabolizer |
| 2 | gs167 | CYP2D6*6A |
| 2 | gs181 | CYP2D6*2 |
| 2 | gs239 | Reduced conversion of beta-carotene to retinol |
| 2 | gs246 | APOE3/APOE3 |
| 1.5 | gs247 | Parkinson's Disease Risk |
| 1.2 | gs184 | Able to taste bitterness. |
| 1 | gs163 | CYP2D6*2A |
| 0 | gs158 | CYP1A2 normal metabolizer |

## 4 Raw Data

The raw data used to create this report has been assigned the identifier ERS1176594 in the European Nucleotide Archive (ENA) hosted at the European Bioinformatics Institute (EBI).

These data will not be accessible unless the report is approved. This will happen by default one month after the report is issued, or if the report is approved for immediate release within the one month period. Participants can also withdraw from the study at any time in which case the report and the data will not be released and will be deleted.

If the data has already been released, it can be accessed at: http://www.ebi.ac.uk/ena/data/view/ERS1176594

## 5 Report Metadata

| Resource | Version | Website |
|---|---|---|
| Genome | GRCh38 | Link |
| BWA | 0.7.12 | Link |
| SAMtools | 1.3 | Link |
| GATK | 3.4-46 | Link |
| PLINK | v1.90b3.35 | Link |
| VEP | 88 | Link |
| SNPedia | 30-Jul-2017 | Link |
| ExAC | v0.3.1 | Link |
| GetEvidence | 16-Dec-2016 | Link |
| ClinVar | 16-Dec-2016 | Link |

Table 5: Analysis Pipeline Versions

Report generated on August 5, 2017.