

PGP-UK Genomics Report for ukB5E082

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 | 4997799 |
| Variants filtered out | 0 |
| Novel / existing variants | 511304 (10.3) / 4473937 (89.7) |
| Overlapped genes | 56713 |
| Overlapped transcripts | 67499 |
| Overlapped regulatory features | 166946 |

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.

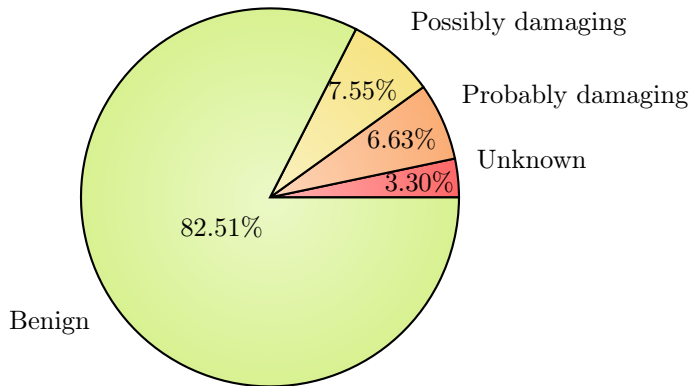


Figure 2: PolyPhen Summary

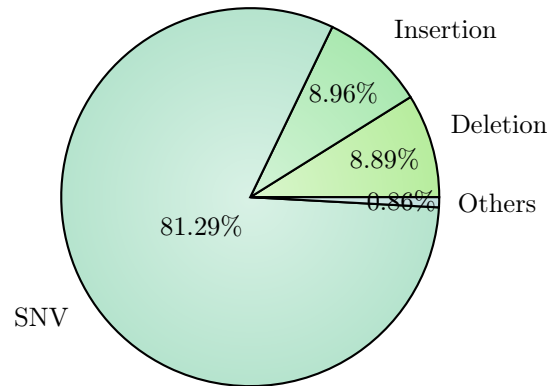


Figure 3: Variant Class

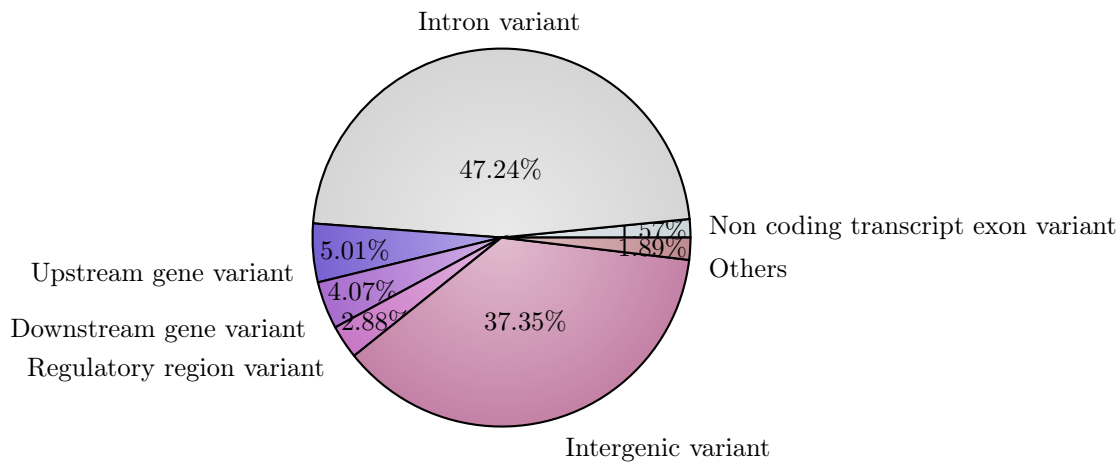


Figure 4: Consequence type

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).

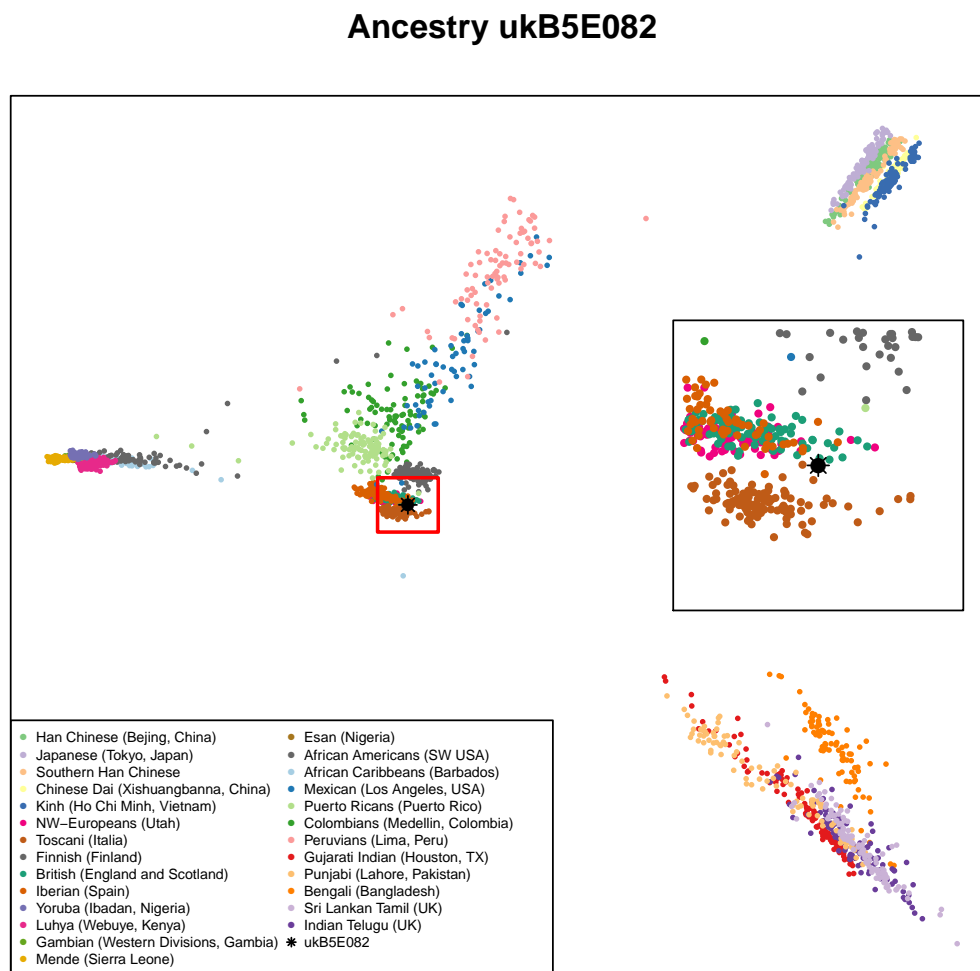


Figure 5: Ancestry Principal Component Analysis

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 |
|------|------------|----------|--|----------------------|----------------------|----------------------|
| 2.1 | rs2511989 | (A;G) | 0.63x decreased age-related macular degeneratio... | | Link | |
| 2.1 | rs547154 | (A;C) | 0.47x decreased risk for AMD | | | Link |
| 2 | rs10468017 | (C;T) | Associated with higher HDL cholesterol | | Link | |
| 2 | rs1056836 | (G;G) | 0.3x decreased risk for prostate cancer | Link | Link | Link |
| 2 | rs11045585 | (A;A) | 24% chance (lower than average) of docetaxel-in... | | Link | |
| 2 | rs1160312 | (G;G) | Reduced risk of Baldness. | | Link | |
| 2 | rs12979860 | (C;C) | ~80% of such hepatitis C patients respond to tr... | | Link | Link |
| 2 | rs17070145 | (C;T) | Increased memory performance | | | Link |
| 2 | rs1799884 | (G;G) | Mothers have typical Birth-Weight babies. Sligh... | | | |
| 2 | rs1864163 | (G;G) | Associated with higher HDL cholesterol | | Link | |
| 2 | rs2243250 | (C;T) | 0.6x decreased risk for myocardial infarction i... | | | |
| 2 | rs261332 | (A;G) | Associated with higher HDL cholesterol | | | |
| 2 | rs3736309 | (A;G) | 0.44x decreased risk for chronic obstructive pu... | | | |
| 2 | rs3738579 | (C;T) | 0.5x decreased risk for cervical cancer: HNSCC:... | | | |
| 2 | rs3750817 | (C;T) | 0.78x reduced risk for breast cancer | | | |
| 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 | rs6505162 | (A;C) | 0.58x decreased risk for esophageal cancer | Link | | |
| 2 | rs6511720 | (G;T) | Slightly lower odds of developing CHD. | | Link | Link |
| 2 | rs6855911 | (A;G) | 0.62x decreased risk for gout | | Link | |
| 2 | rs7216389 | (C;C) | 0.69x lower risk of Childhood Asthma. | | Link | |
| 2 | rs763110 | (C;T) | ~0.80x reduced cancer risk | | | Link |
| 2 | rs7776725 | (T;T) | Stronger bones | | Link | |
| 2 | rs801114 | (T;T) | 0.78x decreased Basal Cell Carcinoma risk. | | Link | |
| 2 | rs9272346 | (A;G) | 0.3x risk type-1 diabetes | | Link | |
| 2 | rs9642880 | (G;G) | Slightly lower risk of Bladder Cancer. | | Link | |

| Mag. | Identifier | Genotype | Summary | ExAC | GetEvidence | ClinVar |
|------|------------|----------|--|------|-------------|---------|
| 1.8 | rs1128535 | (A;G) | 0.77x risk for Crohn's disease | | | |
| 1.8 | rs1800588 | (C;T) | Higher HDL-C levels | Link | Link | |
| 1.8 | rs187238 | (C;G) | Hypertension not a risk factor for sudden cardi... | | | |
| 1.8 | rs4714156 | (C;C) | <0.61x risk for restless legs | | | |
| 1.6 | rs1061170 | (T;T) | Lower risk for AMD: generally longer live than ... | Link | Link | Link |
| 1.6 | rs3775948 | (C;C) | Slightly lower risk for gout | | | |
| 1.5 | rs10513789 | (G;G) | Lower risk of Parkinson's disease | | | |
| 1.5 | rs11136000 | (C;T) | 0.84x decreased risk for Alzheimer's disease | | Link | |
| 1.5 | rs11212617 | (A;C) | Somewhat increased likelihood of treatment succ... | | | Link |
| 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 | rs464049 | (C;C) | Decreased risk of schizophrenia in limited stud... | | | |
| 1.5 | rs4939883 | (C;C) | Associated with higher HDL cholesterol | | Link | |
| 1.5 | rs5888 | (C;C) | Higher HDL cholesterol but lower risk for age-r... | Link | | |
| 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 | | | |
| 1.4 | rs6700125 | (C;C) | 0.7x decreased risk for ALS | | | |
| 1.25 | rs10088218 | (A;G) | 0.76x decreased risk for ovarian cancer | | | |
| 1.1 | rs10166942 | (C;T) | 0.85x lower risk for migraines | | | |
| 1.1 | rs11172113 | (C;T) | 0.9x lower risk for migraines | | | |
| 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 |
| 1.1 | rs7568369 | (G;T) | 0.90x reduced risk of obesity | | | |
| 1 | rs12510549 | (C;C) | Decreased risk for high uric acid levels and go... | | | |
| 1 | rs182549 | (T;T) | Can digest milk. | | | Link |
| 1 | rs2546890 | (G;G) | Lower risk of multiple sclerosis | | | |
| 1.0 | rs2283123 | (C;T) | Decreased risk of schizophrenia in limited stud... | | | |
| 1.0 | rs6583817 | (C;T) | ~0.80x (lower) risk for late onset Alzheimer's ... | | | |
| 0.1 | rs1726866 | (C;C) | Can taste bitter | Link | Link | 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 | rs16947 | (A;A) | Homozygous for CYP2D6 variants (non-CYP2D6*1) | Link | 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 | 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 | 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 | rs7305115 | (A;A) | Individuals showed a significantly lower risk o... | Link | Link | |
| 0 | rs74315403 | (G;G) | PrP codon 178 (D) - non pathogenic variant | | | Link |
| 0 | rs9394492 | (C;C) | <0.76x risk for restless legs | | | |
| 0 | rs9951307 | (A;G) | 0.10 decreased risk for brain edema after a str... | | | |

3.2 Possibly Harmful Traits

| Mag. | Identifier | Genotype | Summary | ExAC | GetEvidence | ClinVar |
|------|------------|----------|--|------|-------------|---------|
| 3.5 | rs6920220 | (A;A) | 1.7x risk of Rheumatoid Arthritis | | Link | |
| 3 | rs10897346 | (C;C) | If depressed: 2.6x more likely to not respond t... | | | |
| 3 | rs13266634 | (C;C) | Increased risk for type-2 diabetes | Link | Link | Link |
| 3 | rs2306402 | (C;T) | 1.18x increased risk for late-onset Alzheimer's... | | | |
| 3 | rs7754840 | (C;G) | 1.3x increased risk for type-2 diabetes | | Link | |
| 2.7 | rs10830963 | (C;G) | Increased type-2 diabetes risk; higher gestatio... | | Link | |
| 2.5 | rs1121980 | (C;T) | 1.67x risk for obesity | | Link | |
| 2.5 | rs12803066 | (A;G) | Increased risk of myopia | | | |
| 2.5 | rs1421085 | (C;T) | ~1.3x increased obesity risk | | Link | Link |
| 2.5 | rs2004640 | (T;T) | 1.4x increased risk for SLE | | Link | Link |
| 2.5 | rs2241880 | (C;C) | 2x-3x increased risk for Crohn's disease in Cau... | Link | Link | Link |
| 2.5 | rs2254958 | (C;C) | 1.61x increased risk for Alzheimer's | | | |
| 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 | rs664143 | (C;T) | Higher risk for number of cancers | | | |
| 2.5 | rs891512 | (A;G) | Higher blood pressure than G;G | Link | | |
| 2.4 | rs2274223 | (G;G) | 1.9x increased risk for stomach and esophageal ... | Link | Link | Link |
| 2.4 | rs7966230 | (G;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 | rs10811661 | (T;T) | 1.2x increased risk for type-2 diabetes | | Link | |
| 2.1 | rs2494732 | (C;C) | Greater odds of cannabis-associated psychosis | Link | Link | |
| 2.1 | rs4430796 | (A;A) | 1.38x increased risk for prostate cancer | | Link | |
| 2.1 | rs4693596 | (C;C) | 2x odds of myopathy if taking statins | | | |
| 2.1 | rs5186 | (A;C) | ~1.4x increased risk of hypertension | Link | Link | 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 | rs10086908 | (C;T) | 1.7x increased risk for prostate cancer | | | |
| 2 | rs10248420 | (A;A) | 7x less likely to respond to certain antidepres... | | Link | |
| 2 | rs10260404 | (C;C) | 1.60x risk of developing ALS | | Link | |
| 2 | rs1045642 | (C;T) | Slower metaboliser for some drugs | Link | Link | Link |
| 2 | rs1050152 | (C;T) | 2.1x increased risk of Crohn's disease | Link | Link | Link |
| 2 | rs10871777 | (A;G) | Adults likely to be 0.22 BMI units higher | | | |
| 2 | rs10980705 | (T;T) | 3.7x increased risk for knee osteoarthritis | | | |
| 2 | rs10984447 | (A;G) | 1.17x increased risk for multiple sclerosis | | Link | |
| 2 | rs11190870 | (C;T) | Possibly increased risk of scoliosis | | | |
| 2 | rs11983225 | (T;T) | 7x less likely to respond to certain antidepres... | | Link | |
| 2 | rs12567232 | (A;G) | Increased risk for Crohn's Disease | | Link | |
| 2 | rs12770228 | (A;A) | 2x increased risk for meningioma | | | |
| 2 | rs1333048 | (A;C) | 1.3x increased coronary artery disease risk | | | |
| 2 | rs1544410 | (A;A) | Increased risk of low bone mineral density diso... | | Link | |
| 2 | rs16944 | (G;G) | Increased risk of mental disorders | | Link | |
| 2 | rs17228212 | (C;T) | 1.26x increased risk for heart disease | | Link | |
| 2 | rs1734791 | (A;A) | 1.4x increased risk for lupus | | | |
| 2 | rs17696736 | (A;G) | 1.34x risk of type-1 diabetes | | Link | |
| 2 | rs17782313 | (C;T) | Adults likely to be 0.22 BMI units higher | | Link | Link |
| 2 | rs1799732 | (-;C) | 1.3x increased adenoma recurrence risk | | Link | |
| 2 | rs1800888 | (C;T) | Increased risk of coronary artery disease | Link | Link | 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 | rs2230201 | (G;G) | >1.4x risk of lupus | Link | | |

| Mag. | Identifier | Genotype | Summary | ExAC | GetEvidence | ClinVar |
|------|------------|----------|--|------|-------------|---------|
| 2 | rs2235015 | (G;G) | Somewhat less likely to respond to certain anti... | Link | Link | |
| 2 | rs2235040 | (G;G) | 7x less likely to respond to certain antidepres... | Link | Link | |
| 2 | rs2235067 | (G;G) | 7x less likely to respond to certain antidepres... | | | |
| 2 | rs2305480 | (T;T) | If 4 years old or younger: ~3x increased asthma... | Link | Link | |
| 2 | rs2305795 | (A;G) | 1.28x higher risk of narcolepsy compared to (G;... | | | Link |
| 2 | rs2383206 | (A;G) | 1.4x increased risk for heart disease | | | |
| 2 | rs2383207 | (A;G) | Increased risk for heart disease | | | |
| 2 | rs25487 | (G;G) | 2x higher risk for skin cancer; possibly other ... | Link | Link | Link |
| 2 | rs268 | (A;G) | 3X increased risk for venous thromboembolism | Link | Link | Link |
| 2 | rs2707466 | (G;G) | Weaker bones | Link | Link | |
| 2 | rs2908004 | (C;C) | Weaker bones | Link | Link | |
| 2 | rs3025039 | (C;T) | 2.6x increased risk for ARMD in a Taiwanese pop... | | | |
| 2 | rs326 | (A;A) | Lower HDL cholesterol | | Link | Link |
| 2 | rs358806 | (C;C) | 1.78x increased risk of developing Type-2 diabe... | | Link | |
| 2 | rs3738919 | (A;C) | 1.94x risk of developing rheumatoid arthritis | | | |
| 2 | 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; ... | | | Link |
| 2 | rs4633 | (C;T) | Higher risk for endometrial cancer | Link | Link | Link |
| 2 | rs493258 | (A;G) | 1.15x risk of Age Related Macular Degeneration | | | |
| 2 | rs4961 | (G;T) | 1.8x increased risk for high blood pressure | Link | Link | Link |
| 2 | rs4968451 | (A;C) | 1.61x increased risk for meningioma | | | |
| 2 | rs5174 | (A;G) | 1.3x increased risk for heart disease | Link | Link | Link |
| 2 | rs520354 | (A;G) | Increased risk in men for biliary conditions | | | |
| 2 | rs5759167 | (T;T) | Higher prostate cancer risk | | Link | |
| 2 | rs6232 | (A;G) | Higher risk of obesity and insulin sensitivity | Link | Link | Link |
| 2 | rs6441286 | (G;T) | 1.54x chance of developing primary biliary cirr... | | Link | |
| 2 | rs6498169 | (A;A) | >1.14x risk of multiple sclerosis | | Link | |
| 2 | rs663048 | (G;T) | Significantly increased risk of developing lung... | 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;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 | rs744373 | (C;T) | 1.17x risk of Alzheimer's | | | |
| 2 | rs7807268 | (C;G) | 1.3x risk for Crohn's disease | | Link | |
| 2 | rs800292 | (C;C) | 5% higher risk of Age related macular degenerat... | Link | Link | Link |
| 2 | rs828907 | (G;T) | Slightly increased risk of bladder cancer and 2... | | | |
| 2 | rs854560 | (A;A) | Higher risk for heart disease: diabetic retinop... | Link | Link | Link |
| 2 | rs9303277 | (T;T) | 1.46x Increased risk of developing primary bili... | | | |
| 1.9 | rs7923837 | (A;G) | 1.6x risk for T2D | | | |
| 1.8 | rs143383 | (T;T) | 1.3x increased risk for osteoarthritis | | Link | Link |
| 1.8 | rs2278206 | (T;T) | 1.16x increased risk for asthma | Link | 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 | rs33980500 | (C;T) | 1.6x increase in risk for psoriatic arthritis | Link | Link | Link |
| 1.6 | rs3764880 | (A;A) | 1.2 - 1.8x increased tuberculosis risk | Link | Link | |
| 1.5 | rs10492519 | (A;G) | Slightly increased risk of developing prostate ... | | | |
| 1.5 | rs10757272 | (C;T) | 1.30x increased risk for Coronary artery diseas... | | | |
| 1.5 | rs10883365 | (A;G) | 1.2x increased risk for developing Crohn's dise... | | Link | |
| 1.5 | rs10895068 | (A;A) | 2.5x increased odds of breast cancer among horm... | | | |
| 1.5 | rs11171739 | (C;T) | 1.34x risk of developing Type-1 diabetes | | Link | |
| 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 | |

| Mag. | Identifier | Genotype | Summary | ExAC | GetEvidence | ClinVar |
|------|------------|----------|--|------|-------------|---------|
| 1.5 | rs12431733 | (C;T) | Slightly increased risk of developing Parkinson... | | Link | |
| 1.5 | rs12469063 | (A;G) | Slightly increased risk of developing restless ... | | | |
| 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 | rs140701 | (A;G) | Increased risk for anxiety disorders | | | |
| 1.5 | rs144848 | (G;T) | Very slightly increased breast cancer risk | Link | Link | Link |
| 1.5 | rs1801274 | (T;T) | Complex; generally greater risk for cancer prog... | Link | Link | Link |
| 1.5 | rs2177369 | (C;C) | 1.5x increased risk for Alzheimer's disease | | | |
| 1.5 | rs2280714 | (A;A) | 1.4x increased risk of SLE | | | |
| 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 | 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 | rs3814570 | (T;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) | ~1.2x increased risk for several types of cance... | | Link | |
| 1.5 | rs4027132 | (A;G) | 1.39x increased risk of developing bipolar diso... | | | |
| 1.5 | rs4464148 | (C;T) | 1.10x increased risk for colorectal cancer | | | |
| 1.5 | rs4506565 | (A;T) | 1.4x increased risk for type-2 diabetes | | Link | |
| 1.5 | rs4785763 | (A;C) | 1.5x higher risk for melanoma | | Link | |
| 1.5 | rs4845618 | (G;T) | 1.7x increased melanoma risk | | | |
| 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 | rs6601764 | (C;T) | 1.16x increased risk of developing Crohn's dise... | | Link | |
| 1.5 | rs6710341 | (A;G) | Slightly increased risk of developing restless ... | | | |
| 1.5 | rs699473 | (C;C) | ~1.5x increased brain tumor risk | | | |
| 1.5 | rs763035 | (C;T) | 1.2x increased risk for rosacea | | | |
| 1.5 | rs7774434 | (C;T) | Slightly increased risk of developing primary b... | | | |
| 1.5 | rs7850258 | (G;G) | Slightly higher odds of developing primary hypo... | | | |
| 1.5 | rs807701 | (C;T) | Slightly increased dyslexia risk | | | |
| 1.5 | rs872071 | (G;G) | ~1.5x increased risk for chronic lymphocytic le... | | Link | |
| 1.5 | rs9652490 | (A;G) | Slightly increased risk of developing Parkinson... | | Link | |
| 1.5 | rs966221 | (C;C) | 1.5x increased stroke risk certain populations | | | |
| 1.5 | rs995030 | (G;G) | Non-protective against testicular cancer | | Link | |
| 1.4 | rs1126497 | (C;T) | 1.4x increased risk for breast cancer | Link | Link | Link |
| 1.4 | rs1545843 | (A;A) | 1.4x increased risk for depression (for those u... | | | |
| 1.4 | rs2046210 | (C;T) | 1.4x increased breast cancer risk | | Link | Link |
| 1.4 | rs3131296 | (G;G) | 1.4x increased risk for schizophrenia | | Link | |
| 1.4 | rs4959039 | (A;G) | 1.4x higher risk for multiple sclerosis | | | |
| 1.3 | rs110419 | (A;G) | 1.3x increased risk for neuroblastoma | | | |
| 1.3 | rs1260326 | (C;T) | Slightly higher risk for gout | Link | Link | Link |
| 1.3 | rs1375144 | (C;T) | 1.32x increased risk of developing bipolar diso... | | | |
| 1.3 | rs1746048 | (C;C) | 1.03 increased risk for coronary heart 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 | rs2736100 | (G;T) | 1.3x higher risk for glioma development: 2.1x r... | | Link | |
| 1.3 | rs356219 | (A;G) | 1.3x increased risk for Parkinson's disease | | | |
| 1.3 | rs4295627 | (G;T) | 1.36x higher risk for glioma development | | Link | |
| 1.2 | rs12050604 | (A;A) | Slightly increased risk for lung cancer | | | |
| 1.2 | rs1800693 | (A;G) | Slight (1.2x) increase in risk for multiple scl... | Link | Link | Link |

| Mag. | Identifier | Genotype | Summary | ExAC | GetEvidence | ClinVar |
|------|------------|----------|--|------|-------------|---------|
| 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 | rs249954 | (T;T) | Potentially increased risk of Breast Cancer | | | Link |
| 1.2 | rs2814707 | (A;G) | 1.2x increased risk for ALS | | Link | |
| 1.2 | rs35677470 | (A;G) | 2x higher risk for scleroderma | Link | Link | |
| 1.2 | rs3849942 | (A;G) | 1.2x increased risk for ALS | | Link | |
| 1.2 | rs449647 | (A;T) | Possibly lower levels of ApoE | | | |
| 1.2 | rs4795067 | (A;G) | Slight increase in risk for psoriatic arthritis... | | | |
| 1.2 | rs8050136 | (A;C) | 1.2x increased risk for T2D in some populations... | | Link | |
| 1.2 | rs9858542 | (A;G) | 1.1x risk Crohn's Disease | Link | Link | |
| 1.17 | rs17465637 | (A;C) | 1.17x higher risk for myocardial infarction | Link | 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 | rs11650494 | (A;G) | Slightly higher prostate cancer risk | | | |
| 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 | rs2651899 | (A;G) | 1.1x higher risk for migraines | | | |
| 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 | rs3740878 | (A;G) | 1.26x type II diabetes risk | Link | | Link |
| 1.1 | rs4324715 | (C;T) | 1.5x increased testicular cancer risk for men | | | |
| 1.1 | rs6897876 | (C;T) | Slight increase in testicular cancer risk for m... | | | |
| 1.1 | rs7171755 | (A;A) | 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 | | | |
| 1.05 | rs2291834 | (C;T) | Very slightly higher risk for myocardial infarc... | | | |
| 1 | rs10504861 | (G;G) | Major allele: normal risk of migraine | | | |
| 1 | rs1143674 | (A;G) | 1.3x increased autism risk | Link | | |
| 1 | rs3194051 | (A;A) | >1.1x risk of type-1 diabetes | Link | Link | Link |
| 1 | rs5326 | (A;G) | Possible psychiatric risks | | | |
| 1 | rs6932590 | (T;T) | 1.1x increased risk for schizophrenia | | Link | |
| 1.0 | rs11246226 | (A;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 | rs1061646 | (C;C) | 1.16x increased risk for breast cancer | Link | | Link |
| 0 | rs3761418 | (A;A) | 1.3x increased risk for depression | | | |
| 0 | rs3813929 | (C;C) | Possible weight gain if taking olanzapine | | Link | Link |
| 0 | rs4795400 | (T;T) | If 4 years old or younger: ~2.5x increased asth... | | Link | |
| 0 | rs6314 | (C;C) | Higher risk for RA | Link | Link | |
| 0 | rs7787082 | (G;G) | 7x less likely to respond to certain antidepres... | | Link | |

3.3 Genosets (Multi-variant Phenotypes)

| Magnitude | Identifier | Summary |
|-----------|------------|--|
| 2.5 | gs102 | ALS risk |
| 2.5 | gs155 | CYP3A5 non-expressor |
| 2.5 | gs281 | Part of the 88% of the population claimed not t... |
| 2.5 | gs285 | You will lose 2.5x as much weight on a low fat ... |
| 2.4 | gs297 | Lower heart attack risk than average |
| 2 | gs101 | Probably able to digest milk |
| 2 | gs104 | Restless legs syndrome risk |
| 2 | gs154 | NAT2 Slow metabolizer |
| 2 | gs179 | CYP2D6*41 |
| 2 | gs246 | APOE3/APOE3 |
| 1.5 | gs185 | The beta blocker metoprolol is effective with 1... |
| 1.5 | gs220 | HLA-B*1502? |
| 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 ERS1176637 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/ERS1176637>

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 2, 2017.