

PGP-UK Genomics Report for ukAE0694

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.

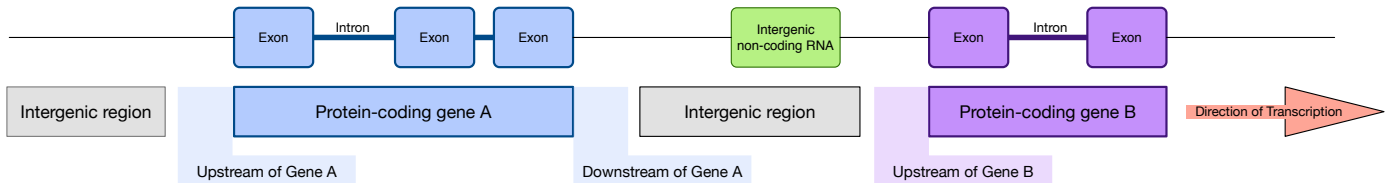


Figure 1: Diagram of gene structure indicating locations of potential variants

| Feature | Count |
|--------------------------------|--------------------------------|
| Lines of input read | 5006995 |
| Variants filtered out | 0 |
| Novel / existing variants | 521213 (10.4) / 4473631 (89.6) |
| Overlapped genes | 56656 |
| Overlapped transcripts | 67429 |
| Overlapped regulatory features | 166756 |

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.

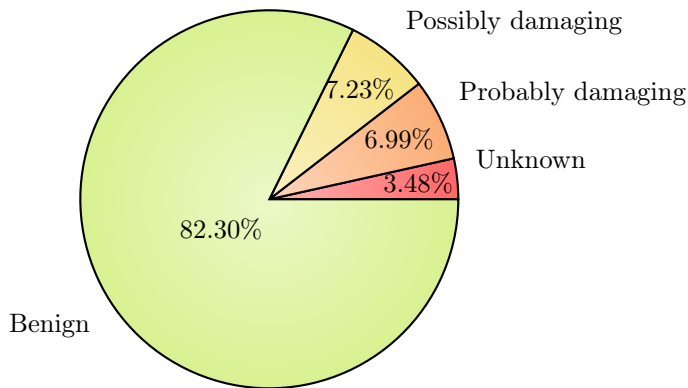


Figure 2: PolyPhen Summary

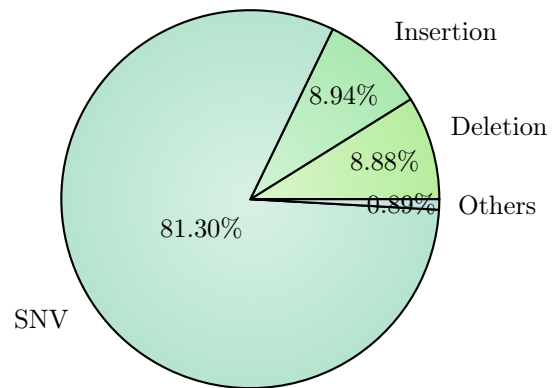


Figure 3: Variant Class

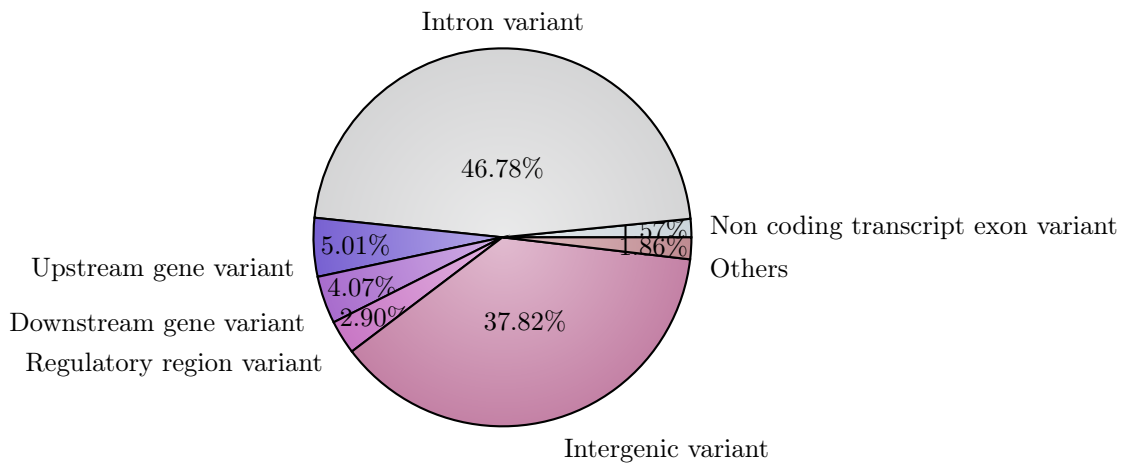


Figure 4: Consequence type

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).

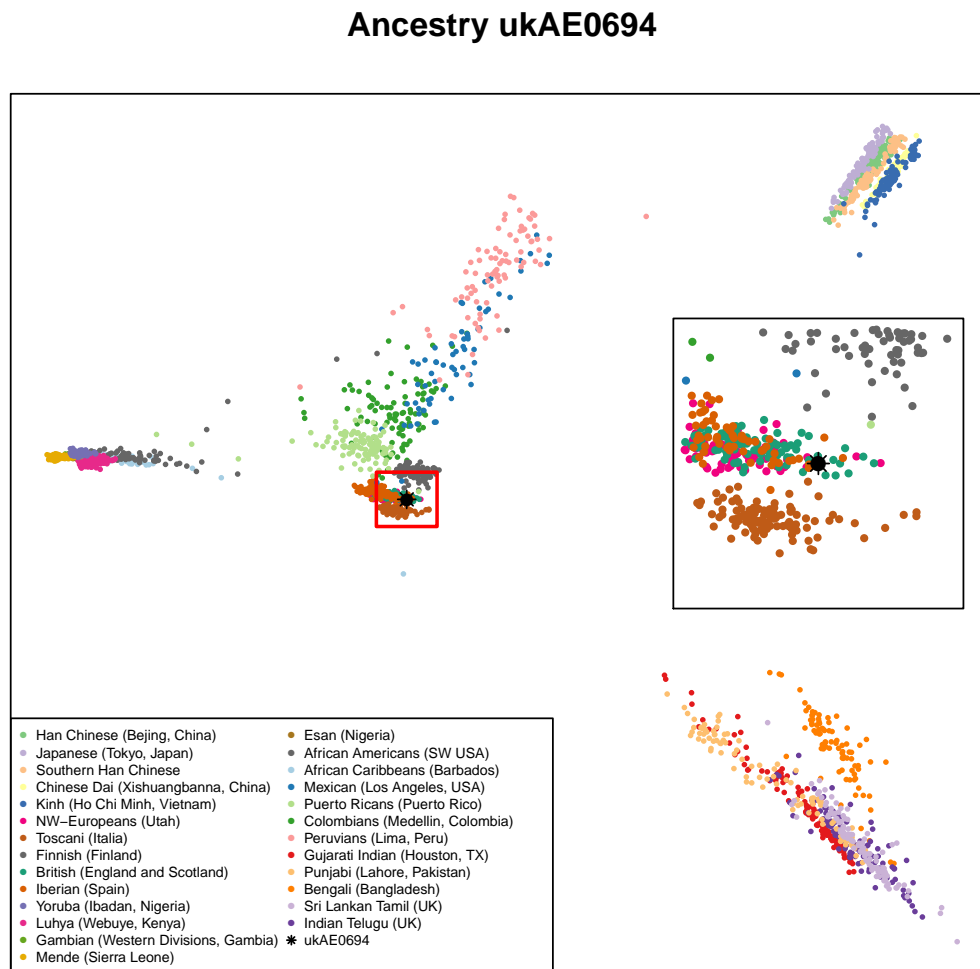


Figure 5: Ancestry Principal Component Analysis

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 | 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 | rs11045585 | (A;A) | 24% chance (lower than average) of docetaxel-in... | | Link | |
| 2 | rs1544410 | (G;G) | Decreased risk of low bone mineral density diso... | | Link | |
| 2 | rs17070145 | (C;T) | Increased memory performance | | | Link |
| 2 | rs1800972 | (G;G) | Reduced risk for Crohn's disease; reduced risk ... | Link | | |
| 2 | rs1864163 | (A;G) | Associated with higher HDL cholesterol | | Link | |
| 2 | rs2243250 | (C;T) | 0.6x decreased risk for myocardial infarction i... | | | |
| 2 | rs2292813 | (C;T) | Decreased risk of autism | | | |
| 2 | rs2542052 | (C;C) | Better odds of living to 100 | | | |
| 2 | rs261332 | (A;G) | Associated with higher HDL cholesterol | | | |
| 2 | rs3736309 | (G;G) | 0.44x decreased risk for chronic obstructive pu... | | | |
| 2 | rs3750817 | (C;T) | 0.78x reduced risk for breast cancer | | | |
| 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 | rs763110 | (C;T) | ~0.80x reduced cancer risk | | | 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... | | | |
| 2 | rs9642880 | (G;G) | Slightly lower risk of Bladder Cancer. | | Link | |
| 1.8 | rs1800588 | (C;T) | Higher HDL-C levels | Link | Link | |
| 1.8 | rs266729 | (C;G) | 0.73x decreased risk for colorectal cancer | | Link | |
| 1.8 | rs3814113 | (C;T) | 0.8x decreased risk for ovarian cancer | | Link | |
| 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 | rs2007153 | (A;A) | Decreased risk of schizophrenia in limited stud... | | | |

| Mag. | Identifier | Genotype | Summary | ExAC | GetEvidence | ClinVar |
|------|------------|----------|--|------|-------------|---------|
| 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;T) | Associated with higher HDL cholesterol | | Link | |
| 1.5 | rs729302 | (A;C) | 0.89x decreased risk of developing rheumatoid a... | | | |
| 1.4 | rs1165205 | (A;T) | 0.85x decreased gout risk | | Link | |
| 1.4 | rs6700125 | (C;C) | 0.7x decreased risk for ALS | | | |
| 1.4 | rs9402571 | (G;T) | Slightly decreased risk for type-2 diabetes | | | |
| 1.3 | rs9306160 | (T;T) | 0.75x (reduced) risk for metastasis in LN-/ER+ ... | Link | Link | |
| 1.2 | rs11172113 | (C;C) | 0.8x lower risk for migraines | | | |
| 1.2 | rs4320932 | (A;G) | 0.87x decreased risk for ovarian cancer | | | |
| 1.2 | rs6048 | (G;G) | Slightly lower risk (10-20%) of deep vein throm... | Link | Link | 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 | rs182549 | (T;T) | Can digest milk. | | | Link |
| 1 | rs2952768 | (C;T) | Slightly less drug dependence: decreased effect... | | | Link |
| 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 | 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 | 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 | rs74315403 | (G;G) | PrP codon 178 (D) - non pathogenic variant | | | Link |
| 0 | rs7495174 | (A;A) | Blue/gray eyes more likely | | Link | |

3.2 Possibly Harmful Traits

| Mag. | Identifier | Genotype | Summary | ExAC | GetEvidence | ClinVar |
|------|------------|----------|--|------|-------------|---------|
| 3.8 | rs5186 | (C;C) | 7.3x increased risk of hypertension | Link | Link | Link |
| 3.1 | rs10830963 | (G;G) | Increased type-2 diabetes risk; higher gestatio... | | 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 | rs3903239 | (C;C) | Higher frequency of atrial fibrillation | | | |
| 3 | rs55705857 | (A;G) | 6x increased risk of glioma of IDH1/IDH2 subtyp... | | | |
| 3.0 | rs1142345 | (A;G) | TPMT*3C . impaired drug metabolism | Link | Link | Link |
| 2.5 | rs10490924 | (G;T) | 2.7x risk for age related macular degeneration | Link | Link | Link |
| 2.5 | rs1121980 | (C;T) | 1.67x risk for obesity | | Link | |
| 2.5 | rs1421085 | (C;T) | ~1.3x increased obesity risk | | Link | Link |
| 2.5 | rs16969968 | (A;G) | Slightly higher risk for nicotine dependence: l... | Link | Link | Link |
| 2.5 | rs187238 | (G;G) | Hypertension increases risk 3.75x for sudden ca... | | | |
| 2.5 | rs2254958 | (C;C) | 1.61x increased risk for Alzheimer's | | | |
| 2.5 | rs3780374 | (A;G) | Substantially increased odds of developing V617... | | | |
| 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 | rs7574865 | (G;T) | 1.3x risk of rheumatoid arthritis; 1.55x risk o... | | Link | Link |
| 2.5 | rs8034191 | (C;T) | 1.27x lung cancer risk | | Link | |
| 2.5 | rs9934438 | (A;A) | Coumadin resistance | | Link | Link |
| 2.4 | rs1143679 | (A;G) | 1.78x increased risk for SLE | Link | Link | |
| 2.4 | rs7966230 | (G;G) | Slightly lower levels of plasma VWF | | | |
| 2.3 | rs37973 | (G;G) | Among asthmatics: 2.3x more likely to show less... | | | Link |
| 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 | rs1329428 | (G;G) | 2x increased risk for macular degeneration | | | |
| 2.1 | rs1360780 | (T;T) | 1.3x increased risk for depression | | Link | |
| 2.1 | rs2294008 | (T;T) | Increased risk of gastric and bladder cancer | Link | Link | |
| 2.1 | rs2494732 | (C;C) | Greater odds of cannabis-associated psychosis | Link | Link | |
| 2.1 | rs380390 | (C;C) | Increased risk for ARMD | | Link | |
| 2.1 | rs4430796 | (A;A) | 1.38x increased risk for prostate cancer | | Link | |
| 2.1 | rs5751876 | (T;T) | Significantly higher anxiety levels after moder... | Link | | |
| 2.1 | rs6457617 | (T;T) | 5.2x risk of rheumatoid arthritis | | Link | |
| 2.1 | rs646776 | (A;A) | 1.2x risk of coronary artery disease | | Link | |
| 2.1 | rs6742078 | (T;T) | +16% bilirubin levels increased risk of gallsto... | | Link | Link |
| 2.1 | rs7837688 | (G;T) | 1.7x increased risk for prostate cancer | | | |
| 2.1 | rs944289 | (C;T) | 1.3x increased thyroid cancer risk | | Link | |
| 2 | rs10086908 | (C;T) | 1.7x increased risk for prostate cancer | | | |
| 2 | rs10090154 | (C;T) | 1.4x increased risk for prostate cancer | | | |
| 2 | rs10248420 | (A;A) | 7x less likely to respond to certain antidepres... | | Link | |
| 2 | rs1050152 | (C;T) | 2.1x increased risk of Crohn's disease | Link | Link | Link |
| 2 | rs10513789 | (G;T) | Increased risk of Parkinson's disease | | | |
| 2 | rs1051730 | (C;T) | 1.3x increased risk of lung cancer | Link | Link | Link |
| 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 | rs11190870 | (C;T) | Possibly increased risk of scoliosis | | | |
| 2 | rs11229030 | (C;C) | Higher odds of Crohn's disease | | | |
| 2 | rs1160312 | (A;G) | 1.6x increased risk of Male Pattern Baldness. | | Link | |
| 2 | rs11650354 | (T;T) | 8x risk for allergic asthma | Link | | |
| 2 | rs11983225 | (T;T) | 7x less likely to respond to certain antidepres... | | Link | |
| 2 | rs12696304 | (C;G) | Prone to aging faster: at least in European pop... | | | |
| 2 | rs13376333 | (T;T) | ~2x higher risk of atrial fibrillation | | Link | |

| Mag. | Identifier | Genotype | Summary | ExAC | GetEvidence | ClinVar |
|------|------------|----------|--|------|-------------|---------|
| 2 | rs16944 | (A;A) | Increased risk for osteoarthritis | | Link | |
| 2 | rs17228212 | (C;C) | >1.26x increased risk for heart disease | | Link | |
| 2 | rs1734791 | (A;A) | 1.4x increased risk for lupus | | | |
| 2 | rs17576 | (A;G) | Higher risk for MI and lung cancer: and COPD in... | Link | Link | |
| 2 | 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 | rs1800896 | (A;G) | 1.6x increased prostate cancer risk | | | |
| 2 | rs2056116 | (G;G) | 1.41x risk for breast cancer | | | |
| 2 | rs2073963 | (G;T) | Increased risk of baldness | | | |
| 2 | rs2201841 | (T;T) | 2.4x increased risk for Graves' disease | | Link | |
| 2 | rs2230201 | (G;G) | >1.4x risk of lupus | Link | | |
| 2 | rs2235015 | (G;G) | Somewhat less likely to respond to certain anti... | Link | Link | |
| 2 | rs2235040 | (G;G) | 7x less likely to respond to certain antidepres... | Link | Link | |
| 2 | rs2235067 | (G;G) | 7x less likely to respond to certain antidepres... | | | |
| 2 | rs2274223 | (A;G) | 1.5x increased risk for stomach and esophageal ... | Link | Link | Link |
| 2 | rs2305480 | (C;T) | 3.5x increase in risk of asthma for Han Chinese... | 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 | rs25487 | (A;G) | 2x higher risk for skin cancer; possibly other ... | Link | Link | Link |
| 2 | rs2736990 | (C;C) | Increased risk of developing Parkinson's Diseas... | | Link | |
| 2 | rs326 | (A;A) | Lower HDL cholesterol | | Link | Link |
| 2 | rs351855 | (C;T) | 1.2x increased risk for prostate cancer | Link | Link | Link |
| 2 | rs3738579 | (C;C) | 0.6x decreased risk for cervical cancer: but 1... | | | |
| 2 | rs3738919 | (A;C) | 1.94x risk of developing rheumatoid arthritis | | | |
| 2 | rs3775948 | (G;G) | Slightly higher risk for gout | | | |
| 2 | rs4148739 | (A;A) | 7x less likely to respond to certain antidepres... | | Link | |
| 2 | rs4242382 | (A;G) | 1.7x increased risk for prostate cancer | | 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 | rs486907 | (A;A) | 2x increased prostate cancer risk | Link | Link | Link |
| 2 | rs493258 | (G;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 | rs6441286 | (G;T) | 1.54x chance of developing primary biliary cirr... | | Link | |
| 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;T) | 1.2x higher risk for hypertension | | | |
| 2 | rs699 | (C;T) | Increased risk of hypertension | Link | Link | Link |
| 2 | rs7442295 | (A;A) | ~4x higher risk for hyperuracemia | | Link | |
| 2 | 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 | 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 | rs9652490 | (A;A) | ~2x increased risk for Parkinson's disease: and... | | Link | |
| 2 | rs9954153 | (G;T) | ~2.5x higher risk for Fuchs' dystrophy: a corne... | | | |
| 2.0 | rs28997576 | (C;G) | Increased risk for breast cancer: at least in p... | Link | Link | Link |
| 2.0 | rs4911414 | (G;T) | 2-4x higher risk of sun sensitivity if part of ... | | Link | |
| 1.9 | rs7923837 | (A;G) | 1.6x risk for T2D | | | |
| 1.8 | rs1136287 | (C;T) | 1.5x increased risk of wet ARMD in a Taiwanese ... | Link | Link | |
| 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 | |
| 1.8 | rs4474514 | (A;G) | 3x increased testicular cancer risk for men | | Link | |
| 1.61 | rs4295627 | (G;G) | 1.85x higher risk for glioma development | | Link | |

| Mag. | Identifier | Genotype | Summary | ExAC | GetEvidence | ClinVar |
|------|------------|----------|--|------|-------------|---------|
| 1.6 | rs11523871 | (A;C) | 1.6x increased breast cancer risk for women ove... | Link | Link | |
| 1.6 | rs2736100 | (G;G) | 1.6x higher risk for glioma development | | 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 | rs10260404 | (C;T) | 1.20x risk of developing ALS | | Link | |
| 1.5 | rs10492519 | (A;G) | Slightly increased risk of developing prostate ... | | | |
| 1.5 | rs10883365 | (A;G) | 1.2x increased risk for developing Crohn's dise... | | 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 | |
| 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 | rs12498742 | (A;A) | 1.25 increased risk for gout | | | |
| 1.5 | rs13149290 | (C;C) | Slightly increased risk of developing prostate ... | | | |
| 1.5 | rs1375144 | (C;C) | 1.59x increased risk of developing bipolar diso... | | | |
| 1.5 | rs1801020 | (C;T) | 1.31x increased risk of heart disease | Link | | Link |
| 1.5 | rs1801274 | (C;T) | Complex; generally greater risk for cancer prog... | Link | Link | Link |
| 1.5 | rs1975197 | (C;T) | 1.3x increased risk of developing restless legs... | | Link | |
| 1.5 | rs1994090 | (G;T) | Slightly increased risk of developing Parkinson... | | Link | |
| 1.5 | rs199533 | (C;T) | Slightly increased risk of developing Parkinson... | Link | | |
| 1.5 | rs2272127 | (C;C) | Associated with herpes and schizophrenia | | | |
| 1.5 | rs2697962 | (A;G) | Slightly increased risk of developing Parkinson... | | | |
| 1.5 | rs27388 | (A;G) | Slightly increased risk of developing schizophr... | | | |
| 1.5 | rs2881766 | (G;T) | Slightly increased risk for pregnancy-induced h... | | | |
| 1.5 | rs3212227 | (A;A) | 1.43x increased risk of developing psoriasis an... | | | |
| 1.5 | rs358806 | (A;C) | 0.86x increased risk of developing Type-2 diabe... | | Link | |
| 1.5 | rs3790565 | (C;T) | Slightly increased risk of developing primary b... | | | |
| 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 | rs464049 | (C;T) | Increased risk of schizophrenia in limited stud... | | | |
| 1.5 | rs4785763 | (A;C) | 1.5x higher risk for melanoma | | Link | |
| 1.5 | rs4845618 | (G;T) | 1.7x increased melanoma risk | | | |
| 1.5 | rs4982731 | (C;C) | Possible higher risk of childhood acute lymphob... | | | |
| 1.5 | rs5219 | (C;T) | 1.3x increased risk for type-2 diabetes | Link | Link | Link |
| 1.5 | rs5746059 | (A;G) | Slightly higher fat mass | | | |
| 1.5 | rs6498169 | (A;G) | 1.14x risk of multiple sclerosis | | Link | |
| 1.5 | rs6601764 | (C;T) | 1.16x increased risk of developing Crohn's dise... | | Link | |
| 1.5 | rs6710341 | (A;G) | Slightly increased risk of developing restless ... | | | |
| 1.5 | rs6896702 | (C;T) | Slightly increased risk of developing Parkinson... | | | |
| 1.5 | rs699473 | (C;T) | ~1.5x increased brain tumor risk | | | |
| 1.5 | rs7454108 | (C;T) | Single HLA-DQ8 haplotype | | | |
| 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 | rs9303277 | (C;T) | 1.46x Slightly increased risk of developing pri... | | | |
| 1.5 | rs9561778 | (G;T) | ~2x increased risk of adverse drug reactions fr... | | Link | |
| 1.4 | rs12770228 | (A;G) | 1.4x increased risk for meningioma | | | |
| 1.4 | rs1447295 | (A;C) | 1.4x increased risk of prostate cancer | | Link | |
| 1.4 | rs1800693 | (G;G) | Slight (1.4x) increase in risk for multiple scl... | Link | Link | Link |
| 1.4 | rs3131296 | (G;G) | 1.4x increased risk for schizophrenia | | Link | |
| 1.4 | rs4977756 | (G;G) | 1.93x higher risk for glioma development | | Link | |
| 1.4 | rs498872 | (T;T) | 1.4x higher risk for glioma development | | Link | |
| 1.34 | rs17465637 | (C;C) | 1.34x higher risk for myocardial infarction | Link | Link | |
| 1.3 | rs110419 | (A;G) | 1.3x increased risk for neuroblastoma | | | |

| Mag. | Identifier | Genotype | Summary | ExAC | GetEvidence | ClinVar |
|------|------------|----------|--|------|-------------|---------|
| 1.3 | rs1434536 | (A;G) | 1.29x increased breast cancer risk | | | |
| 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 | rs356219 | (A;G) | 1.3x increased risk for Parkinson's disease | | | |
| 1.3 | rs4958847 | (A;G) | 1.3x increased risk for Crohn's disease | | | |
| 1.25 | rs748404 | (T;T) | Slightly increased risk (1.25) for lung cancer... | | Link | |
| 1.2 | rs11037909 | (T;T) | 1.47x type II diabetes risk | Link | | |
| 1.2 | rs11842874 | (A;G) | +17% increased risk for osteoarthritis | | | |
| 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 | rs3176336 | (T;T) | Slightly higher (1.25x) higher risk for breast ... | | | |
| 1.2 | rs35677470 | (A;G) | 2x higher risk for scleroderma | Link | Link | |
| 1.2 | rs3740878 | (A;A) | 1.46x type II diabetes risk; common | Link | | Link |
| 1.2 | rs393152 | (A;G) | Slight increased risk of both PD and AD | Link | Link | |
| 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 | |
| 1.17 | rs3802842 | (A;C) | 1.17x increased risk of colorectal cancer | | Link | |
| 1.1 | rs11110912 | (C;G) | 1.3x high blood pressure risk | | | |
| 1.1 | rs11650494 | (A;G) | Slightly higher prostate cancer risk | | | |
| 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 | 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 | 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;T) | More likely to gain weight if taking olanzapine... | Link | Link | Link |
| 1.1 | rs889312 | (C;C) | Very slightly higher risk for breast cancer | | Link | |
| 1.1 | rs925391 | (C;C) | More likely to go bald; common | | | |
| 1.09 | rs12050604 | (A;C) | Very slightly increased risk for lung cancer | | | |
| 1.07 | rs2291834 | (C;C) | Very slightly higher risk for myocardial infarc... | | | |
| 1 | rs10504861 | (G;G) | Major allele: normal risk of migraine | | | |
| 1 | rs10761659 | (A;G) | 1.2x risk of Crohn's disease | | Link | |
| 1 | rs1143674 | (A;G) | 1.3x increased autism risk | Link | | |
| 1 | rs2282679 | (A;C) | Somewhat lower vitamin D levels | | | |
| 1 | rs2546890 | (A;G) | Higher risk of multiple sclerosis | | | |
| 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 | |
| 1 | rs6974491 | (A;G) | Higher risk of coeliac and/or inflammatory bowe... | | | |
| 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 | rs1042173 | (T;T) | Among alcoholics: likely to be heavier drinkers... | | | |
| 0 | rs1128503 | (T;T) | Likely to require more methadone during heroin ... | Link | Link | Link |
| 0 | rs1611115 | (T;T) | Somewhat more associated with impulsiveness and... | | | Link |
| 0 | rs3813929 | (C;C) | Possible weight gain if taking olanzapine | | Link | Link |
| 0 | rs4293393 | (T;T) | 1.25x Increased Risk of CKD for T allele in ... | | | |
| 0 | rs6277 | (C;C) | 1.6x higher schizophrenia risk | Link | Link | Link |
| 0 | rs7787082 | (G;G) | 7x less likely to respond to certain antidepres... | | Link | |

3.3 Genosets (Multi-variant Phenotypes)

| Magnitude | Identifier | Summary |
|-----------|------------|--|
| 3.1 | gs191 | Problem metabolizing NSAIDs |
| 3 | gs127 | Intermediate warfarin metabolizer |
| 3 | gs241 | Lighter green: brown or hazel eye color |
| 2.8 | gs295 | Lower heart attack risk than average |
| 2.5 | gs134 | Age related macular degeneration |
| 2.5 | gs155 | CYP3A5 non-expressor |
| 2.5 | gs161 | CYP2C9 Intermediate Metabolizers |
| 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 ... |
| 2.2 | gs280 | Light hair color for europeans |
| 2 | gs101 | Probably able to digest milk |
| 2 | gs104 | Restless legs syndrome risk |
| 2 | gs110 | Higher allergic asthma risk |
| 2 | gs154 | NAT2 Slow metabolizer |
| 2 | gs181 | CYP2D6*2 |
| 2 | gs249 | Parkinson's Disease Risk |
| 2 | gs269 | APOE E2/E3 |
| 1.5 | gs220 | HLA-B*1502? |
| 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 ERS1176629 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/ERS1176629>

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.