

PGP-UK Genomics Report for uk0AD39D

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 | 4973383 |
| Variants filtered out | 0 |
| Novel / existing variants | 487884 (9.8) / 4474345 (90.2) |
| Overlapped genes | 56769 |
| Overlapped transcripts | 67628 |
| Overlapped regulatory features | 167186 |

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.

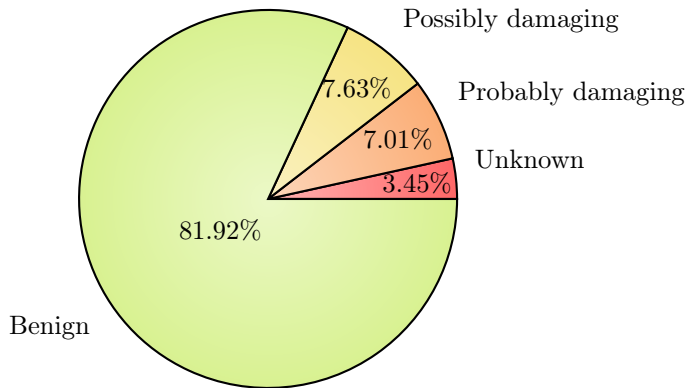


Figure 2: PolyPhen Summary

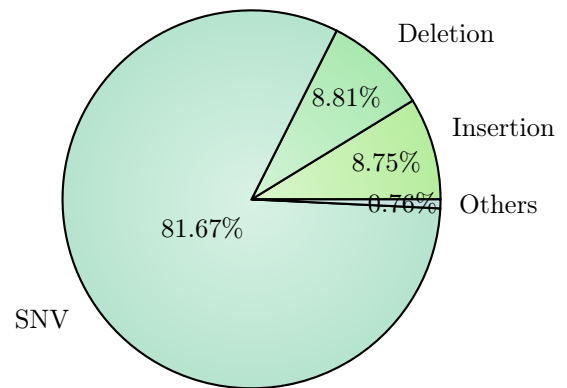


Figure 3: Variant Class

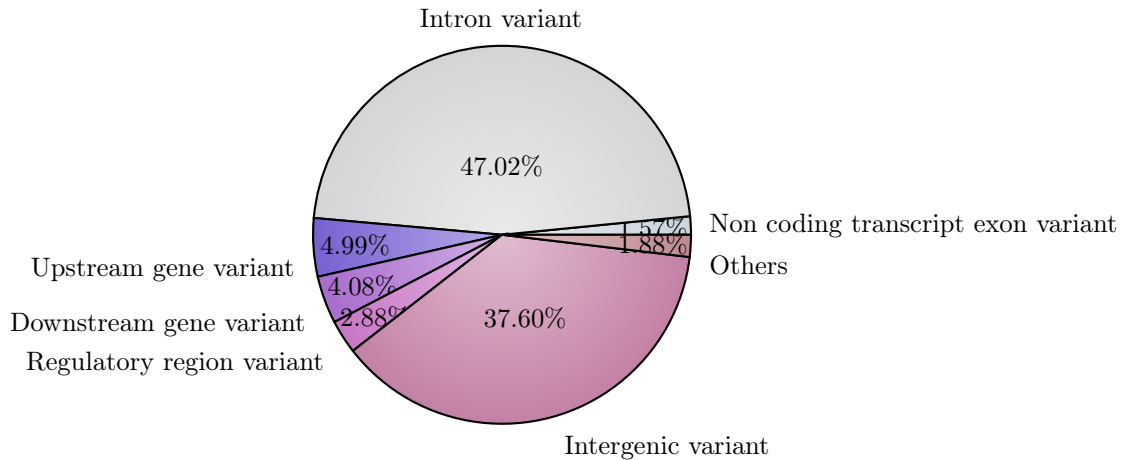


Figure 4: Consequence type

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).

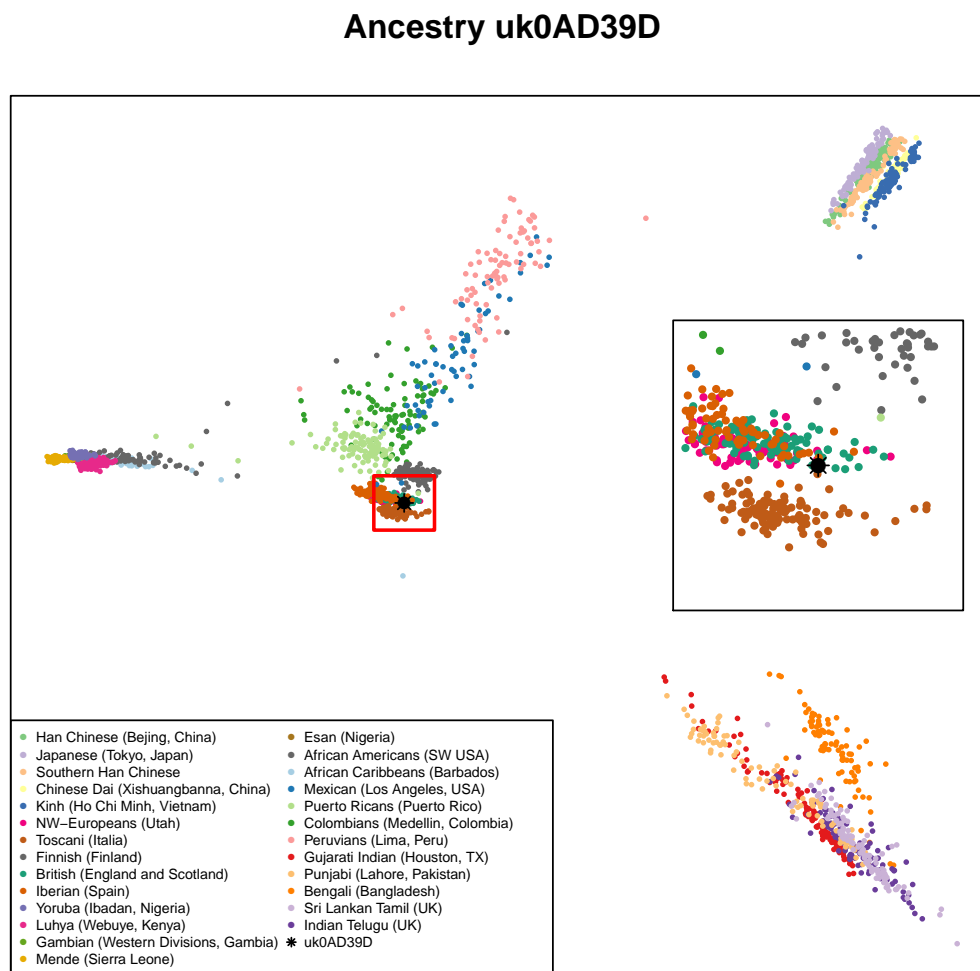


Figure 5: Ancestry Principal Component Analysis

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 |
|------|------------|----------|--|----------------------|----------------------|----------------------|
| 2.8 | rs6046 | (T;T) | Better blood pressure: lower risk of myocardial... | Link | Link | Link |
| 2.5 | rs3764261 | (T;T) | Associated with higher HDL cholesterol. HDL | | Link | Link |
| 2.4 | rs2802288 | (A;A) | Longer lifespan | | | |
| 2.1 | rs2511989 | (A;G) | 0.63x decreased age-related macular degeneratio... | | Link | |
| 2 | rs1012053 | (A;C) | 0.625x reduced risk of Bipolar Disorder. | | Link | |
| 2 | rs1026732 | (A;A) | <0.70x risk for restless legs | | Link | |
| 2 | rs10468017 | (C;T) | Associated with higher HDL cholesterol | | 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 | rs11635424 | (A;A) | <0.70x risk for restless legs | | Link | |
| 2 | rs12593813 | (A;A) | <0.71x risk for restless legs | | Link | |
| 2 | rs1864163 | (G;G) | Associated with higher HDL cholesterol | | Link | |
| 2 | rs2241423 | (A;A) | 0.79 decreased risk for obesity | | | |
| 2 | rs2243250 | (C;T) | 0.6x decreased risk for myocardial infarction i... | | | |
| 2 | rs2542052 | (C;C) | Better odds of living to 100 | | | |
| 2 | rs2764264 | (C;C) | Greater odds of living to 95 | | | |
| 2 | rs3738579 | (C;T) | 0.5x decreased risk for cervical cancer: HNSCC:... | | | |
| 2 | rs3750817 | (C;T) | 0.78x reduced risk for breast cancer | | | |
| 2 | rs6505162 | (A;C) | 0.58x decreased risk for esophageal cancer | Link | | |
| 2 | rs763110 | (C;T) | ~0.80x reduced cancer risk | | | Link |
| 2 | rs801114 | (T;T) | 0.78x decreased Basal Cell Carcinoma risk. | | Link | |
| 1.8 | rs3814113 | (C;C) | 0.8x decreased risk for ovarian cancer | | Link | |
| 1.6 | rs1061170 | (T;T) | Lower risk for AMD: generally longer live than ... | Link | Link | Link |
| 1.6 | rs10801935 | (C;C) | 0.3x decreased risk of breast cancer | | | |
| 1.5 | rs11212617 | (A;C) | Somewhat increased likelihood of treatment succ... | | | Link |
| 1.5 | rs1165205 | (A;A) | 0.85x decreased gout risk | | Link | |
| 1.5 | rs309375 | (G;G) | Smaller mosquito bites | | | |

| Mag. | Identifier | Genotype | Summary | ExAC | GetEvidence | ClinVar |
|------|------------|----------|--|------|-------------|---------|
| 1.5 | rs4149274 | (C;T) | Associated with higher HDL (good) cholesterol | | | |
| 1.5 | rs4489954 | (G;T) | 0.69x risk risk of developing restless legs syn... | | Link | |
| 1.5 | 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 | | |
| 1.5 | rs729302 | (A;C) | 0.89x decreased risk of developing rheumatoid a... | | | |
| 1.4 | rs6495446 | (C;T) | 0.8x reduced risk for chronic kidney disease | | | |
| 1.4 | rs6700125 | (C;C) | 0.7x decreased risk for ALS | | | |
| 1.4 | rs9402571 | (G;T) | Slightly decreased risk for type-2 diabetes | | | |
| 1.3 | rs2361502 | (C;C) | Possible higher levels of serum bilirubin and l... | | | |
| 1.25 | rs10088218 | (A;G) | 0.76x decreased risk for ovarian cancer | | | |
| 1.2 | rs4320932 | (A;G) | 0.87x decreased risk for ovarian cancer | | | |
| 1.1 | rs13333226 | (A;G) | Slightly lower risk for hypertension | | | Link |
| 1.1 | rs7568369 | (G;T) | 0.90x reduced risk of obesity | | | |
| 1 | rs182549 | (C;T) | Can digest milk. | | | Link |
| 1 | rs7850258 | (A;G) | Typical odds of developing primary hypothyroidi... | | | |
| 1 | rs800292 | (C;T) | 1% decreased risk of macular degeneration | Link | Link | Link |
| 1.0 | rs11246226 | (C;C) | Decreased risk of schizophrenia in limited stud... | | 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 | rs1126809 | (A;G) | Slight increase in skin cancer risk | 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 | rs1799782 | (C;C) | Lower risk for skin cancer | Link | Link | |
| 0 | rs1800562 | (G;G) | Not a C282Y hemochromatosis carrier. | 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 | rs74315403 | (G;G) | PrP codon 178 (D) - non pathogenic variant | | | Link |
| 0 | rs7495174 | (A;A) | Blue/gray eyes more likely | | Link | |
| 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 |
|------|------------|----------|--|------|-------------|---------|
| 4 | rs1333049 | (C;C) | 1.9x increased risk for coronary artery disease... | | Link | |
| 3 | rs10897346 | (C;C) | If depressed: 2.6x more likely to not respond t... | | | |
| 3 | rs2306402 | (C;C) | 1.18x increased risk for late-onset Alzheimer's... | | | |
| 3 | rs2981582 | (C;T) | 1.3x higher risk of ER+ breast cancer | | Link | |
| 3 | rs7754840 | (C;C) | 1.3x increased risk for type-2 diabetes | | Link | |
| 2.5 | rs10484554 | (C;T) | 2.8x increased risk for psoriasis | | Link | |
| 2.5 | rs1057910 | (A;C) | CYP2C9*3 carrier; average 40% reduction in warf... | Link | Link | Link |
| 2.5 | rs13266634 | (C;T) | Increased risk for type-2 diabetes | Link | Link | Link |
| 2.5 | rs1421085 | (C;T) | ~1.3x increased obesity risk | | Link | Link |
| 2.5 | rs1799971 | (A;G) | Stronger cravings for alcohol. if alcoholic: na... | 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 | rs3738919 | (C;C) | 1.94x risk of developing rheumatoid arthritis | | | |
| 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 | rs795484 | (A;A) | Even more increased morphine dose requirement a... | | | |
| 2.4 | rs1143679 | (A;G) | 1.78x increased risk for SLE | Link | Link | |
| 2.4 | rs7966230 | (G;G) | Slightly lower levels of plasma VWF | | | |
| 2.2 | rs2004640 | (G;T) | 1.4x increased risk for SLE | | Link | Link |
| 2.2 | rs2231137 | (G;G) | ~1.5-3x increased risk for ischemic stroke | Link | Link | Link |
| 2.2 | rs2305089 | (T;T) | Higher risk for chordoma | Link | Link | |
| 2.1 | rs10811661 | (T;T) | 1.2x increased risk for type-2 diabetes | | Link | |
| 2.1 | rs17070145 | (C;C) | Reduced memory abilities | | | Link |
| 2.1 | rs2254958 | (C;T) | 1.24x increased risk for Alzheimer's | | | |
| 2.1 | rs2294008 | (T;T) | Increased risk of gastric and bladder cancer | Link | Link | |
| 2.1 | rs2383207 | (G;G) | Increased risk for heart disease | | | |
| 2.1 | rs4961 | (T;T) | 1.8x increased risk for high blood pressure | Link | Link | Link |
| 2.1 | rs944289 | (C;T) | 1.3x increased thyroid cancer risk | | Link | |
| 2 | rs10086908 | (C;T) | 1.7x increased risk for prostate cancer | | | |
| 2 | rs1024611 | (C;T) | Increased risk of exercise induced ischemia | | | Link |
| 2 | rs10248420 | (A;A) | 7x less likely to respond to certain antidepres... | | Link | |
| 2 | rs1045642 | (C;T) | Slower metaboliser for some drugs | Link | Link | Link |
| 2 | rs10488631 | (C;T) | 2x increased risk of developing SLE; 1.6x incre... | | Link | |
| 2 | rs10757272 | (T;T) | 1.54x increased risk for Coronary artery diseas... | | | |
| 2 | rs10984447 | (A;A) | >1.17x increased risk for multiple sclerosis | | Link | |
| 2 | rs10994336 | (C;T) | 1.45x increased odds of developing bipolar diso... | | Link | |
| 2 | rs11190870 | (C;T) | Possibly increased risk of scoliosis | | | |
| 2 | rs1160312 | (A;G) | 1.6x increased risk of Male Pattern Baldness. | | Link | |
| 2 | rs1169300 | (A;A) | ~2x increased lung cancer risk | | | |
| 2 | rs11983225 | (T;T) | 7x less likely to respond to certain antidepres... | | Link | |
| 2 | rs12037606 | (A;A) | 1.52x risk of developing Crohn's disease | | | |
| 2 | rs1219648 | (A;G) | 1.20x risk for breast cancer | | 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 | rs13254738 | (A;C) | 1.18x prostate cancer risk | | Link | |
| 2 | rs144848 | (G;G) | Very slightly increased breast cancer risk | Link | Link | Link |
| 2 | rs1537415 | (G;G) | 2x increased risk for periodontitis | | Link | |
| 2 | rs1691053 | (A;G) | Increased risk of developing prostate cancer | | | |
| 2 | rs16944 | (G;G) | Increased risk of mental disorders | | Link | |
| 2 | rs17001266 | (-;C) | 1.58x increased risk for schizophrenia in males... | | | |
| 2 | rs1734791 | (A;A) | 1.4x increased risk for lupus | | | |
| 2 | rs17435 | (A;T) | 1.4x increased risk for lupus | | | |
| 2 | rs17576 | (A;G) | Higher risk for MI and lung cancer: and COPD in... | Link | Link | |

| Mag. | Identifier | Genotype | Summary | ExAC | GetEvidence | ClinVar |
|------|------------|----------|--|------|-------------|---------|
| 2 | rs17696736 | (A;G) | 1.34x risk of type-1 diabetes | | Link | |
| 2 | rs1800629 | (A;G) | Complex; generally higher risk for certain dise... | Link | Link | 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 | 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 | 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 | (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 | rs241448 | (C;T) | 1.51x increased risk for Alzheimer's | Link | | Link |
| 2 | rs2420946 | (C;T) | 1.20x risk for breast cancer | | | |
| 2 | rs2464196 | (T;T) | ~2x increased lung cancer risk | Link | Link | Link |
| 2 | rs25487 | (A;G) | 2x higher risk for skin cancer; possibly other ... | Link | Link | Link |
| 2 | rs2572886 | (A;G) | 1.4x increased risk of HIV infection | | | |
| 2 | rs2736100 | (T;T) | Higher risk of Interstitial lung disease: and t... | | 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 | rs358806 | (C;C) | 1.78x increased risk of developing Type-2 diabe... | | Link | |
| 2 | rs3745516 | (A;A) | Increased risk of developing primary biliary ci... | | | |
| 2 | rs3775948 | (G;G) | Slightly higher risk for gout | | | |
| 2 | rs3793784 | (C;G) | 1.5x risk for ARMD | | Link | Link |
| 2 | rs3825776 | (G;G) | >1.3x increased risk for ALS | | Link | |
| 2 | rs4148739 | (A;A) | 7x less likely to respond to certain antidepres... | | Link | |
| 2 | rs4402960 | (G;T) | 1.2x increased risk for type-2 diabetes: ~1x ri... | | Link | Link |
| 2 | rs4420638 | (A;G) | ~3x increased Alzheimer's risk; 1.4x increased ... | | Link | Link |
| 2 | rs4444903 | (A;G) | 3.5x risk of hep-cancer in cirrhosis patients; ... | | | Link |
| 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 | rs5174 | (A;G) | 1.3x increased risk for heart disease | Link | Link | Link |
| 2 | rs629242 | (C;T) | Somewhat higher risk for prostate cancer | | | |
| 2 | rs638405 | (G;G) | 2x increased ALZ risk in ApoE4 carriers | Link | | |
| 2 | rs6441286 | (G;T) | 1.54x chance of developing primary biliary cirr... | | Link | |
| 2 | rs6601764 | (C;C) | 1.52x increased risk of developing Crohn's dise... | | 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;T) | 1.2x higher risk for hypertension | | | |
| 2 | rs699 | (C;C) | Increased risk of hypertension | Link | Link | Link |
| 2 | rs7442295 | (A;A) | ~4x higher risk for hyperuracemia | | Link | |
| 2 | rs763361 | (T;T) | Increased risk for multiple autoimmune diseases... | Link | Link | |
| 2 | rs7794745 | (A;T) | Slightly increased risk for autism | | Link | Link |
| 2 | rs7807268 | (C;C) | 1.4x risk for Crohn's disease | | Link | |
| 2 | rs7961152 | (A;C) | 1.2x higher risk for hypertension | | | |
| 2 | rs828907 | (G;T) | Slightly increased risk of bladder cancer and 2... | | | |
| 2 | 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 | rs965513 | (A;G) | 1.77x increased thyroid cancer risk | | Link | |
| 2.0 | rs1434536 | (A;A) | 1.94x increased breast cancer risk | | | |

| Mag. | Identifier | Genotype | Summary | ExAC | GetEvidence | ClinVar |
|------|------------|----------|--|------|-------------|---------|
| 2.0 | rs9642880 | (T;T) | 1.5x increased bladder cancer risk | | Link | |
| 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 | rs37973 | (A;G) | Among asthmatics: 1.5x more likely to show less... | | | Link |
| 1.7 | rs2024513 | (A;A) | 1.7x higher risk for schizophrenia (among Han C... | | | |
| 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 | 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 | 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;C) | Slightly increased risk of developing prostate ... | | | |
| 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 | rs17221417 | (C;G) | 1.3x higher risk for Crohn's disease | | Link | |
| 1.5 | rs1801274 | (C;T) | Complex; generally greater risk for cancer prog... | Link | Link | Link |
| 1.5 | rs2007153 | (G;G) | Increased risk of schizophrenia in limited stud... | | | |
| 1.5 | rs2076295 | (G;G) | Slightly increased risk for pulmonary fibrosis ... | | | |
| 1.5 | rs2177369 | (C;C) | 1.5x increased risk for Alzheimer's disease | | | |
| 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 | rs2280714 | (A;G) | 1.4x increased risk of SLE | | | |
| 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 | (T;T) | Slightly increased risk for pregnancy-induced h... | | | |
| 1.5 | rs3087243 | (G;G) | Increased risk for autoimmune diseases | | Link | |
| 1.5 | rs3212227 | (A;A) | 1.43x increased risk of developing psoriasis an... | | | |
| 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 | rs4464148 | (C;T) | 1.10x increased risk for colorectal cancer | | | |
| 1.5 | rs4785763 | (A;C) | 1.5x higher risk for melanoma | | Link | |
| 1.5 | rs486907 | (A;G) | 1.5x increased prostate cancer risk | Link | Link | Link |
| 1.5 | rs5219 | (C;T) | 1.3x increased risk for type-2 diabetes | Link | Link | Link |
| 1.5 | rs5746059 | (A;A) | Slightly higher fat mass | | | |
| 1.5 | rs619203 | (C;G) | Increases susceptibility to Myocardial Infarcti... | Link | Link | |
| 1.5 | rs7341475 | (G;G) | 1.58x increased schizophrenia risk for women | | Link | |
| 1.5 | rs7536563 | (A;G) | 1.12x risk of multiple sclerosis | | Link | |
| 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 | rs872071 | (A;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 | |
| 1.5 | rs995030 | (G;G) | Non-protective against testicular cancer | | Link | |
| 1.4 | rs1126497 | (T;T) | 1.4x increased risk for breast cancer | Link | Link | Link |
| 1.4 | rs12770228 | (A;G) | 1.4x increased risk for meningioma | | | |
| 1.4 | rs3184504 | (C;T) | Slightly increased risk for celiac disease | Link | Link | |
| 1.4 | rs4959039 | (A;G) | 1.4x higher risk for multiple sclerosis | | | |
| 1.4 | rs6010620 | (G;G) | 1.4x higher risk for glioma development; but th... | | Link | |
| 1.34 | rs17465637 | (C;C) | 1.34x higher risk for myocardial infarction | 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 | | | |

| Mag. | Identifier | Genotype | Summary | ExAC | GetEvidence | ClinVar |
|------|------------|----------|--|------|-------------|---------|
| 1.3 | rs1260326 | (C;T) | Slightly higher risk for gout | Link | Link | Link |
| 1.3 | rs1746048 | (C;C) | 1.03 increased risk for coronary heart disease | | Link | |
| 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 | | | |
| 1.25 | 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 | rs2072590 | (G;T) | 1.2x increased risk for ovarian cancer | | | |
| 1.2 | rs2814707 | (A;G) | 1.2x increased risk for ALS | | Link | |
| 1.2 | rs3176336 | (T;T) | Slightly higher (1.25x) higher risk for breast ... | | | |
| 1.2 | rs3740878 | (A;A) | 1.46x type II diabetes risk; common | Link | | Link |
| 1.2 | rs3849942 | (A;G) | 1.2x increased risk for ALS | | Link | |
| 1.2 | rs419788 | (A;G) | 2.0x risk for lupus | Link | | |
| 1.2 | rs4324715 | (C;C) | >1.5x increased testicular cancer risk for men | | | |
| 1.2 | rs4686484 | (A;A) | Slightly increased risk for celiac disease | | | |
| 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 | |
| 1.1 | rs1344706 | (G;T) | 1.1x increased risk for schizophrenia | | 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 | 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 | | | |
| 1.09 | 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 | rs2435357 | (A;A) | Slightly higher (2x?) risk for Hirschsprung dis... | | | Link |
| 1 | rs2546890 | (A;G) | Higher risk of multiple sclerosis | | | |
| 1 | rs3194051 | (A;G) | 1.12x risk of type-1 diabetes | Link | Link | Link |
| 1 | rs6974491 | (A;G) | Higher risk of coeliac and/or inflammatory bowe... | | | |
| 1 | rs761100 | (G;G) | Higher risk for dyslexia | | | |
| 0.1 | rs601338 | (A;G) | Susceptible to Norovirus infections | Link | Link | Link |
| 0 | rs1061646 | (C;C) | 1.16x increased risk for breast cancer | Link | | 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 |
|-----------|------------|--|
| 3.1 | gs191 | Problem metabolizing NSAIDs |
| 3 | gs241 | Lighter green: brown or hazel eye color |
| 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 | gs298 | Increased surveillance for colorectal cancer re... |
| 2.4 | gs297 | Lower heart attack risk than average |
| 2 | gs101 | Probably able to digest milk |
| 2 | gs154 | NAT2 Slow metabolizer |
| 2 | gs188 | One copy of APOE4 is possible: but not certain |
| 1.5 | gs186 | HLA-B*5801 heterozygosity is possible: unfortun... |
| 1.5 | gs247 | Parkinson's Disease Risk |
| 1.2 | gs184 | Able to taste bitterness. |
| 0 | gs158 | CYP1A2 normal metabolizer |

4 Raw Data

The raw data used to create this report has been assigned the identifier ERS1176559 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/ERS1176559>

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.