

PGP-UK Genomics Report for ukE53A44

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 | 5066350 |
| Variants filtered out | 0 |
| Novel / existing variants | 515286 (10.2) / 4538444 (89.8) |
| Overlapped genes | 56821 |
| Overlapped transcripts | 67677 |
| Overlapped regulatory features | 168466 |

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.

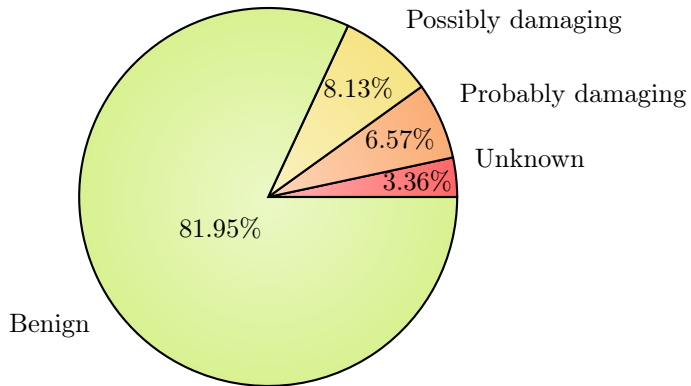


Figure 2: PolyPhen Summary

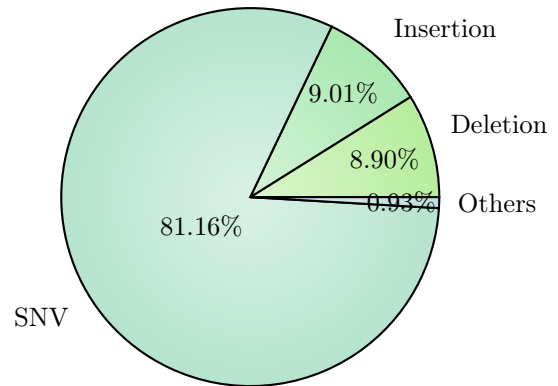


Figure 3: Variant Class

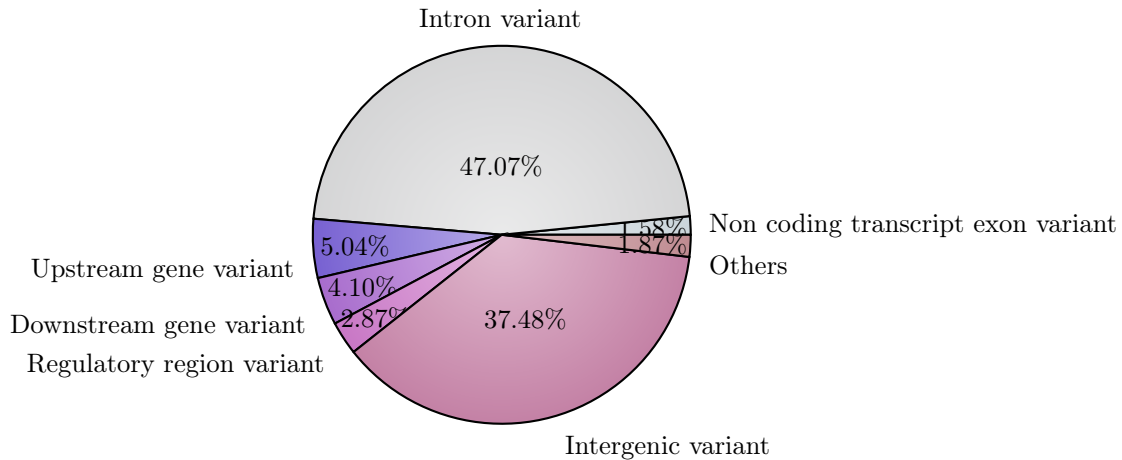


Figure 4: Consequence type

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).

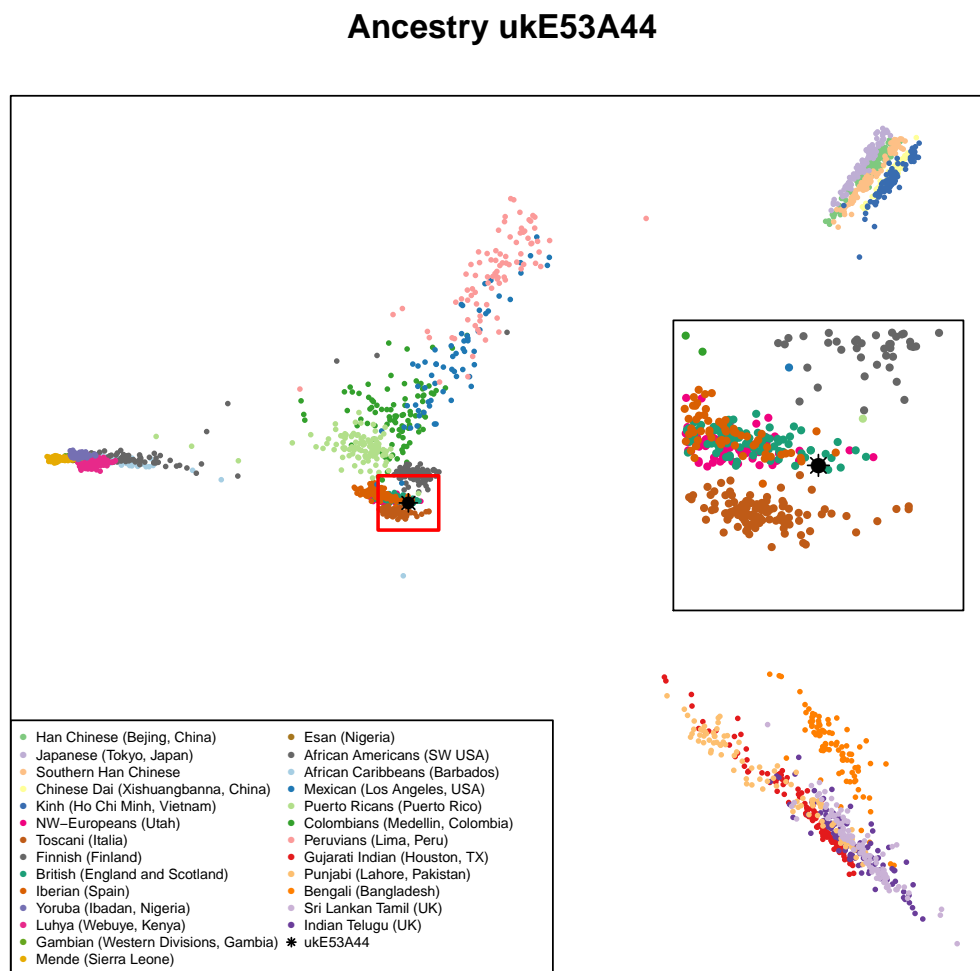


Figure 5: Ancestry Principal Component Analysis

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 |
|------|------------|----------|--|----------------------|----------------------|----------------------|
| 2.2 | rs2511989 | (A;A) | 0.44x decreased age-related macular degeneratio... | | Link | |
| 2 | rs10468017 | (T;T) | Associated with higher HDL cholesterol | | Link | |
| 2 | rs10504861 | (A;G) | Reduced risk of migraine without aura | | | |
| 2 | rs11045585 | (A;A) | 24% chance (lower than average) of docetaxel-in... | | Link | |
| 2 | rs1864163 | (A;G) | Associated with higher HDL cholesterol | | Link | |
| 2 | rs2235015 | (G;T) | Somewhat more likely to respond to certain anti... | Link | Link | |
| 2 | rs2243250 | (C;T) | 0.6x decreased risk for myocardial infarction i... | | | |
| 2 | rs3750817 | (C;T) | 0.78x reduced risk for breast cancer | | | |
| 2 | rs3764261 | (G;T) | Associated with higher HDL cholesterol | | Link | Link |
| 2 | rs3782179 | (C;T) | 3x lower odds of testicular cancer risk for men... | | | |
| 2 | rs4073582 | (A;A) | Lower risk for gout | Link | | |
| 2 | rs4149268 | (A;G) | Associated with higher HDL cholesterol | | 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 | rs800292 | (T;T) | 5% decreased risk of macular degeneration | Link | Link | 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 | rs925391 | (C;T) | Lower odds of going bald | | | |
| 2 | rs9272346 | (A;G) | 0.3x risk type-1 diabetes | | Link | |
| 2 | rs9525638 | (C;C) | Stronger bones | | | |
| 2 | rs9642880 | (G;G) | Slightly lower risk of Bladder Cancer. | | Link | |
| 1.8 | rs1128535 | (A;G) | 0.77x risk for Crohn's disease | | | |
| 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 | |
| 1.8 | rs4714156 | (C;C) | <0.61x risk for restless legs | | | |
| 1.8 | rs6897932 | (C;T) | 0.91x decreased risk for multiple sclerosis | Link | Link | Link |

| Mag. | Identifier | Genotype | Summary | ExAC | GetEvidence | ClinVar |
|------|------------|----------|--|------|-------------|---------|
| 1.6 | rs1061170 | (T;T) | Lower risk for AMD: generally longer live than ... | Link | Link | Link |
| 1.5 | rs10513789 | (G;G) | Lower risk of Parkinson's disease | | | |
| 1.5 | rs11136000 | (T;T) | 0.84x decreased risk for Alzheimer's disease | | Link | |
| 1.5 | rs11212617 | (A;C) | Somewhat increased likelihood of treatment succ... | | | Link |
| 1.5 | rs16991615 | (A;G) | Slight increase (11 months) in avg age at menop... | Link | Link | |
| 1.5 | rs34516635 | (A;G) | Possibly increased longevity for Ashkenazi Jewi... | Link | | Link |
| 1.5 | rs3851179 | (A;A) | 0.85x decreased risk for Alzheimer's disease | | Link | |
| 1.5 | rs4149274 | (C;T) | 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 | rs6700125 | (C;C) | 0.7x decreased risk for ALS | | | |
| 1.4 | rs9402571 | (G;T) | Slightly decreased risk for type-2 diabetes | | | |
| 1.2 | rs11246226 | (A;C) | Decreased risk of schizophrenia in limited stud... | | Link | |
| 1.2 | rs4320932 | (A;G) | 0.87x decreased risk for ovarian cancer | | | |
| 1.2 | rs9306160 | (C;T) | 0.75x (reduced) risk for metastasis in LN-/ER+ ... | Link | Link | |
| 1.1 | rs10166942 | (C;T) | 0.85x lower risk for migraines | | | |
| 1.1 | rs11172113 | (C;T) | 0.9x lower risk for migraines | | | |
| 1.1 | rs2293347 | (G;G) | Among NSCLC patients: better Gefitinib response... | Link | | Link |
| 1 | rs182549 | (C;T) | Can digest milk. | | | Link |
| 1 | rs2952768 | (C;T) | Slightly less drug dependence: decreased effect... | | | Link |
| 0.1 | rs1726866 | (C;C) | Can taste bitter | Link | Link | Link |
| 0.1 | rs891512 | (G;G) | Lower blood pressure than those with an A allel... | Link | | |
| 0 | rs1047781 | (A;A) | ABH blood group "Secretor" status if Japanese | Link | Link | Link |
| 0 | rs12252 | (T;T) | More resistant to influenza | Link | | Link |
| 0 | rs16990018 | (A;A) | PrP Codon 171 Asn - Non-pathogenic variant | Link | | Link |
| 0 | rs17244841 | (A;A) | More responsive to statin treatment | | Link | Link |
| 0 | rs1799782 | (C;C) | Lower risk for skin cancer | Link | Link | |
| 0 | rs1799945 | (C;C) | Not a H63D hemochromatosis carrier. | Link | Link | Link |
| 0 | rs1800562 | (G;G) | Not a C282Y hemochromatosis carrier. | Link | Link | Link |
| 0 | rs242941 | (G;G) | Better response to inhaled corticosteroid in pa... | | Link | |
| 0 | rs28933385 | (G;G) | Prion protein Codon 200 (E) - Non pathogenic va... | | | Link |
| 0 | 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 | |
| 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 | 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 | rs1805008 | (T;T) | ~7-10x higher likelihood of red hair; higher ri... | Link | | Link |
| 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 | rs3738579 | (T;T) | 1.5x - 2x increased risk for cervical cancer: H... | | | |
| 3 | rs3803662 | (T;T) | 1.6x increased risk for breast cancer | | Link | |
| 2.6 | rs110419 | (A;A) | 2.6x increased risk for neuroblastoma | | | |
| 2.5 | rs10490924 | (G;T) | 2.7x risk for age related macular degeneration | Link | Link | Link |
| 2.5 | rs11190870 | (T;T) | Possibly even more increased risk of scoliosis | | | |
| 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 | rs16847548 | (C;C) | 2.6x increased risk for sudden cardiac death in... | | | |
| 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 | rs2004640 | (T;T) | 1.4x increased risk for SLE | | Link | Link |
| 2.5 | rs2254958 | (C;C) | 1.61x increased risk for Alzheimer's | | | |
| 2.5 | rs5219 | (T;T) | 2.5x increased risk for type-2 diabetes | Link | Link | Link |
| 2.5 | rs5888 | (C;T) | 3x higher risk for age-related macular degenera... | Link | | |
| 2.5 | rs663048 | (T;T) | 3x increased risk of developing lung cancer | Link | Link | |
| 2.5 | rs664143 | (C;T) | Higher risk for number of cancers | | | |
| 2.5 | rs795484 | (A;A) | Even more increased morphine dose requirement a... | | | |
| 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 | |
| 2.2 | rs944289 | (T;T) | 1.69x increased thyroid cancer risk | | Link | |
| 2.1 | rs1695 | (G;G) | 3.5x asthma risk in certain populations | Link | Link | Link |
| 2.1 | rs17563 | (C;C) | Risk for otosclerosis | Link | Link | Link |
| 2.1 | rs2383207 | (G;G) | Increased risk for heart disease | | | |
| 2.1 | rs2494732 | (C;C) | Greater odds of cannabis-associated psychosis | Link | 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 | rs1045642 | (C;T) | Slower metaboliser for some drugs | Link | Link | Link |
| 2 | rs10757272 | (T;T) | 1.54x increased risk for Coronary artery diseas... | | | |
| 2 | rs10811661 | (C;T) | 1.2x increased risk for type-2 diabetes | | Link | |
| 2 | rs10984447 | (A;A) | >1.17x increased risk for multiple sclerosis | | Link | |
| 2 | rs1160312 | (A;G) | 1.6x increased risk of Male Pattern Baldness. | | Link | |
| 2 | rs11983225 | (T;T) | 7x less likely to respond to certain antidepres... | | Link | |
| 2 | rs1219648 | (A;G) | 1.20x risk for breast cancer | | Link | |
| 2 | rs12431733 | (T;T) | Increased risk of developing Parkinson's Diseas... | | Link | |
| 2 | rs12696304 | (C;G) | Prone to aging faster: at least in European pop... | | | |
| 2 | rs13254738 | (C;C) | 1.18x prostate cancer risk | | Link | |
| 2 | rs1360780 | (C;T) | 1.3x increased risk for depression | | Link | |
| 2 | rs1585215 | (A;G) | 2x increased risk for Hodgkin lymphoma | | | |
| 2 | rs1691053 | (A;G) | Increased risk of developing prostate cancer | | | |
| 2 | rs17228212 | (C;T) | 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 | rs1800896 | (A;G) | 1.6x increased prostate cancer risk | | | |
| 2 | rs2073963 | (G;T) | Increased risk of baldness | | | |
| 2 | rs2201841 | (T;T) | 2.4x increased risk for Graves' disease | | Link | |
| 2 | rs2230199 | (C;G) | 1.6x+ risk of ARMD | Link | Link | Link |
| 2 | rs2230201 | (G;G) | >1.4x risk of lupus | Link | | |
| 2 | rs2235040 | (G;G) | 7x less likely to respond to certain antidepres... | Link | Link | |

| Mag. | Identifier | Genotype | Summary | ExAC | GetEvidence | ClinVar |
|------|------------|----------|--|------|-------------|---------|
| 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 | rs241448 | (C;T) | 1.51x increased risk for Alzheimer's | Link | | Link |
| 2 | rs2420946 | (C;T) | 1.20x risk for breast cancer | | | |
| 2 | rs25487 | (A;G) | 2x higher risk for skin cancer; possibly other ... | Link | Link | Link |
| 2 | rs2707466 | (G;G) | Weaker bones | Link | Link | |
| 2 | rs27388 | (A;A) | Increased risk of developing schizophrenia | | | |
| 2 | rs2908004 | (C;C) | Weaker bones | Link | Link | |
| 2 | rs3129934 | (C;T) | Increased risk of Multiple Sclerosis. | | Link | |
| 2 | rs326 | (A;A) | Lower HDL cholesterol | | Link | Link |
| 2 | rs3746444 | (C;T) | ~1.2x increased risk for cancer | Link | | |
| 2 | rs3775948 | (G;G) | Slightly higher risk for gout | | | |
| 2 | rs4027132 | (A;A) | 1.51x increased risk of developing bipolar diso... | | | |
| 2 | rs4148739 | (A;A) | 7x less likely to respond to certain antidepres... | | Link | |
| 2 | rs4633 | (C;T) | Higher risk for endometrial cancer | Link | Link | Link |
| 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 | rs6441286 | (G;T) | 1.54x chance of developing primary biliary cirr... | | Link | |
| 2 | rs6457617 | (C;T) | 2.3x risk of rheumatoid arthritis | | Link | |
| 2 | rs662799 | (A;G) | 1.4x higher early heart attack risk; less weigh... | | Link | |
| 2 | rs6896702 | (T;T) | Increased risk of developing Parkinson's Diseas... | | | |
| 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 | rs7794745 | (A;T) | Slightly increased risk for autism | | Link | Link |
| 2 | rs7923837 | (G;G) | 3.2x risk for T2D | | | |
| 2 | rs7961152 | (A;C) | 1.2x higher risk for hypertension | | | |
| 2 | rs854560 | (A;T) | Higher risk for heart disease: diabetic retinop... | Link | Link | Link |
| 2 | rs855913 | (G;T) | Reduced survival with ALS | | Link | |
| 2 | rs9303277 | (T;T) | 1.46x Increased risk of developing primary bili... | | | |
| 1.8 | rs1136287 | (C;T) | 1.5x increased risk of wet ARMD in a Taiwanese ... | Link | Link | |
| 1.8 | rs2278206 | (T;T) | 1.16x increased risk for asthma | Link | Link | |
| 1.8 | rs37973 | (A;G) | Among asthmatics: 1.5x more likely to show less... | | | Link |
| 1.8 | rs4474514 | (A;G) | 3x increased testicular cancer risk for men | | Link | |
| 1.7 | rs1042713 | (A;A) | 1.7x increased risk that pediatric inhaler use ... | Link | Link | Link |
| 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 | rs10859871 | (A;C) | Slight (~1.2x) increase in endometriosis risk | | | |
| 1.5 | rs10883365 | (A;G) | 1.2x increased risk for developing Crohn's dise... | | Link | |
| 1.5 | rs11171739 | (C;T) | 1.34x risk of developing Type-1 diabetes | | Link | |
| 1.5 | rs1169300 | (A;G) | ~1.5x increased lung cancer risk | | | |
| 1.5 | rs12498742 | (A;A) | 1.25 increased risk for gout | | | |
| 1.5 | rs13149290 | (C;C) | Slightly increased risk of developing prostate ... | | | |
| 1.5 | rs13181 | (G;T) | 1.12x increased risk for cutaneous melanoma | Link | Link | Link |
| 1.5 | rs13376333 | (C;T) | 1.5x higher risk of atrial fibrillation | | Link | |
| 1.5 | rs1360517 | (A;G) | Higher susceptibility for AIDS | | Link | |
| 1.5 | rs140701 | (A;A) | Increased risk for anxiety disorders | | | |
| 1.5 | rs144848 | (G;T) | Very slightly increased breast cancer risk | Link | Link | Link |
| 1.5 | rs16944 | (A;G) | Minorly increased risk of mental illness and os... | | Link | |
| 1.5 | rs1994090 | (G;T) | Slightly increased risk of developing Parkinson... | | Link | |

| Mag. | Identifier | Genotype | Summary | ExAC | GetEvidence | ClinVar |
|------|------------|----------|--|------|-------------|---------|
| 1.5 | rs199533 | (C;T) | Slightly increased risk of developing Parkinson... | Link | | |
| 1.5 | rs2007153 | (G;G) | Increased risk of schizophrenia in limited stud... | | | |
| 1.5 | rs2272127 | (C;C) | Associated with herpes and schizophrenia | | | |
| 1.5 | rs2280714 | (A;A) | 1.4x increased risk of SLE | | | |
| 1.5 | rs2464196 | (C;T) | ~1.5x increased lung cancer risk | Link | Link | Link |
| 1.5 | rs2736990 | (C;T) | Slightly increased risk of developing Parkinson... | | Link | |
| 1.5 | rs2881766 | (G;T) | Slightly increased risk for pregnancy-induced h... | | | |
| 1.5 | rs309375 | (T;T) | Larger mosquito bites | | | |
| 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 | rs3745516 | (A;G) | Slightly increased risk of developing primary b... | | | |
| 1.5 | rs3790565 | (C;T) | Slightly increased risk of developing primary b... | | | |
| 1.5 | rs3814570 | (T;T) | 1.3x increased risk for Crohn's disease with il... | | | |
| 1.5 | rs3825776 | (A;G) | 1.3x increased risk for ALS | | Link | |
| 1.5 | rs4506565 | (A;T) | 1.4x increased risk for type-2 diabetes | | Link | |
| 1.5 | rs464049 | (T;T) | Increased risk of schizophrenia in limited stud... | | | |
| 1.5 | rs4785763 | (A;A) | 2x higher risk for melanoma | | Link | |
| 1.5 | rs5746059 | (A;A) | 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 | rs699473 | (C;C) | ~1.5x increased brain tumor risk | | | |
| 1.5 | rs7341475 | (G;G) | 1.58x increased schizophrenia risk for women | | Link | |
| 1.5 | rs7454108 | (C;T) | Single HLA-DQ8 haplotype | | | |
| 1.5 | rs7536563 | (A;G) | 1.12x risk of multiple sclerosis | | Link | |
| 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 | rs9561778 | (G;T) | ~2x increased risk of adverse drug reactions fr... | | Link | |
| 1.5 | rs9652490 | (A;G) | Slightly increased risk of developing Parkinson... | | Link | |
| 1.4 | rs1126497 | (C;T) | 1.4x increased risk for breast cancer | Link | Link | Link |
| 1.4 | rs12770228 | (A;G) | 1.4x increased risk for meningioma | | | |
| 1.4 | rs1800693 | (G;G) | Slight (1.4x) increase in risk for multiple scl... | Link | Link | Link |
| 1.4 | rs2252586 | (A;A) | 1.4x higher risk for glioma development | | | |
| 1.4 | rs3131296 | (G;G) | 1.4x increased risk for schizophrenia | | Link | |
| 1.4 | rs4959039 | (A;G) | 1.4x higher risk for multiple sclerosis | | | |
| 1.4 | rs498872 | (T;T) | 1.4x higher risk for glioma development | | Link | |
| 1.4 | rs6010620 | (G;G) | 1.4x higher risk for glioma development; but th... | | Link | |
| 1.3 | rs1047286 | (C;T) | 1.3x increased risk for age-related macular deg... | Link | Link | Link |
| 1.3 | rs10947262 | (C;C) | 1.3x increased risk for osteoarthritis | | | |
| 1.3 | 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 | 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 | |
| 1.25 | rs13387042 | (A;A) | 1.24x increased risk for breast cancer | | Link | |
| 1.2 | rs11037909 | (T;T) | 1.47x type II diabetes risk | Link | | |
| 1.2 | rs143383 | (C;T) | 1.1x increased risk for osteoarthritis | | Link | Link |
| 1.2 | rs2056116 | (A;G) | 1.18x risk for breast cancer | | | |
| 1.2 | rs2076295 | (G;T) | One copy of the risk allele (G): slightly incre... | | | |
| 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 | rs393152 | (A;G) | Slight increased risk of both PD and AD | Link | Link | |
| 1.2 | rs419788 | (A;G) | 2.0x risk for lupus | Link | | |
| 1.2 | rs4324715 | (C;C) | >1.5x increased testicular cancer risk for men | | | |

| Mag. | Identifier | Genotype | Summary | ExAC | GetEvidence | ClinVar |
|------|------------|----------|--|------|-------------|---------|
| 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 | |
| 1.15 | rs748404 | (C;T) | Very slightly increased risk (1.15) for lung ca... | | Link | |
| 1.1 | rs11110912 | (C;G) | 1.3x high blood pressure risk | | | |
| 1.1 | rs1344706 | (G;T) | 1.1x increased risk for schizophrenia | | Link | |
| 1.1 | rs1800450 | (A;G) | Carrier of mannose binding deficiency but of lo... | Link | Link | Link |
| 1.1 | rs2295190 | (G;T) | Slightly increased risk for ovarian cancer in w... | Link | Link | Link |
| 1.1 | rs249954 | (C;T) | Potentially increased risk of Breast Cancer | | | Link |
| 1.1 | rs2651899 | (A;G) | 1.1x higher risk for migraines | | | |
| 1.1 | rs7412 | (C;T) | More likely to gain weight if taking olanzapine... | Link | Link | Link |
| 1.05 | rs2291834 | (C;T) | Very slightly higher risk for myocardial infarc... | | | |
| 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... | | | |
| 0.1 | rs601338 | (A;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 | rs1495965 | (A;A) | 1.2x higher risk for spondylitis | | | |
| 0 | rs2296336 | (C;C) | 2.9x risk of type-1 diabetes | | | |
| 0 | rs3761418 | (A;A) | 1.3x increased risk for depression | | | |
| 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 | 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 |
|-----------|------------|--|
| 3.2 | gs238 | Red hair |
| 3 | gs241 | Lighter green: brown or hazel eye color |
| 2.5 | gs282 | You are part of the 12% of the population who c... |
| 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 | gs171 | CYP2D6*9 |
| 2 | gs181 | CYP2D6*2 |
| 2 | gs269 | APOE E2/E3 |
| 1.5 | gs186 | HLA-B*5801 heterozygosity is possible: unfortun... |
| 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 ERS1176573 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/ERS1176573>

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.