

PGP-UK Genomics Report for uk55955C

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.

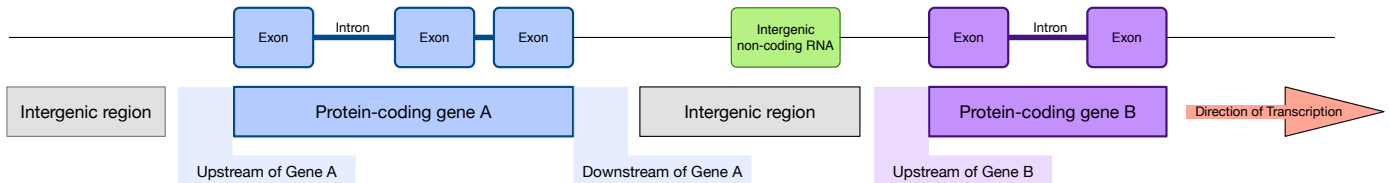


Figure 1: Diagram of gene structure indicating locations of potential variants

| Feature | Count |
|--------------------------------|-------------------------------|
| Lines of input read | 4954427 |
| Variants filtered out | 0 |
| Novel / existing variants | 485538 (9.8) / 4457216 (90.2) |
| Overlapped genes | 56761 |
| Overlapped transcripts | 67583 |
| Overlapped regulatory features | 167295 |

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.

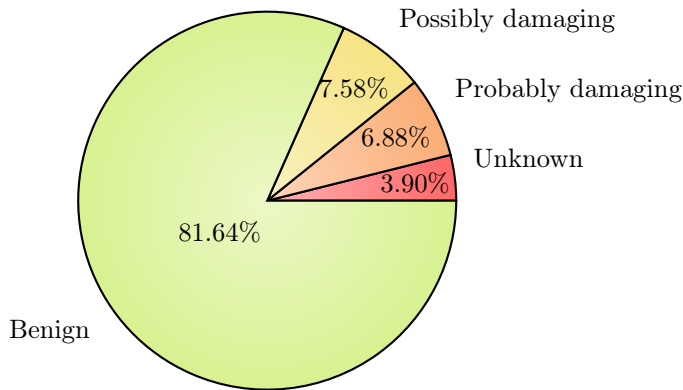


Figure 2: PolyPhen Summary

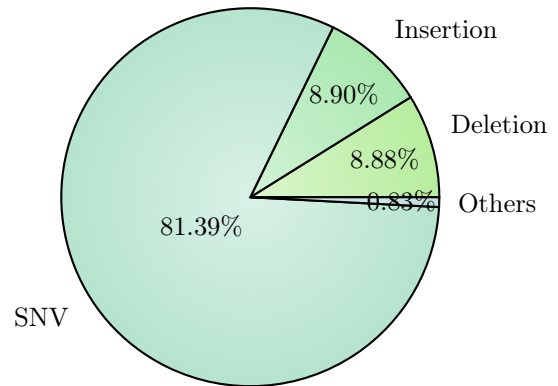


Figure 3: Variant Class

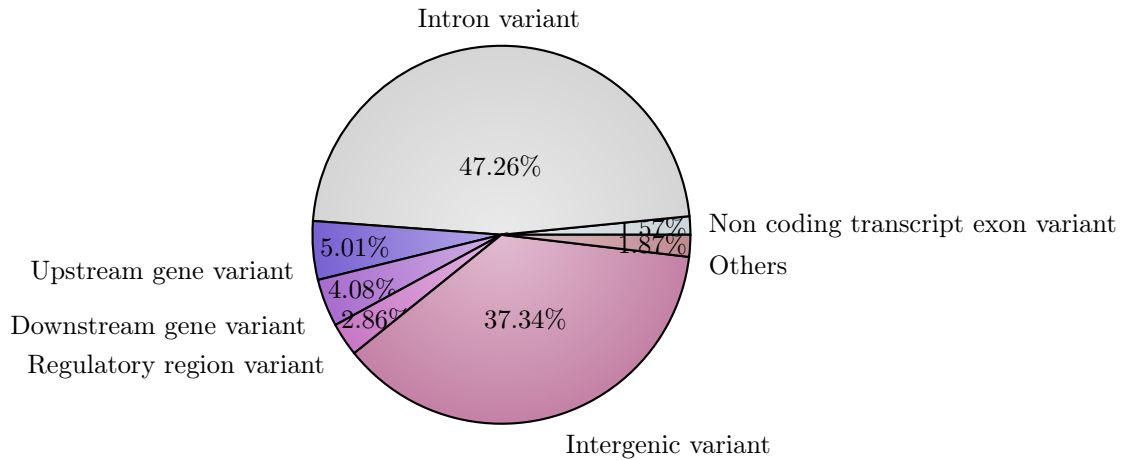


Figure 4: Consequence type

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).

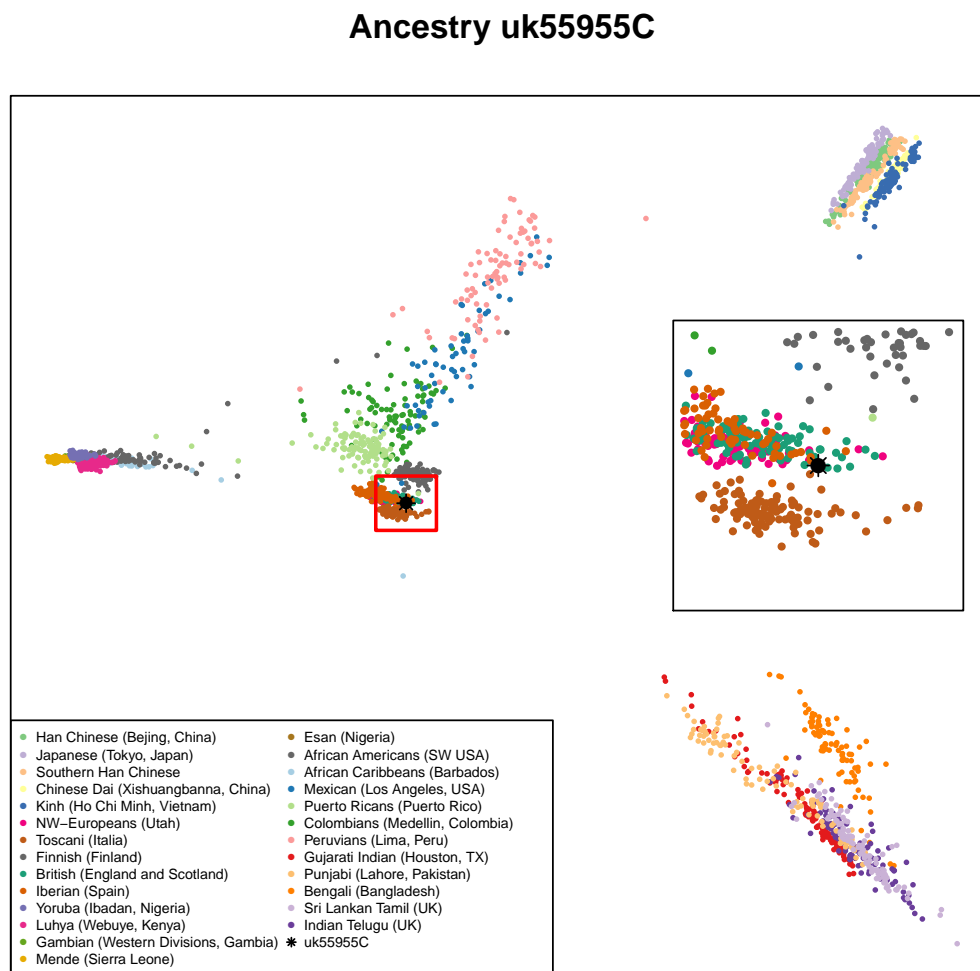


Figure 5: Ancestry Principal Component Analysis

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 |
|------|------------|----------|--|------|-------------|---------|
| 2.4 | rs3750817 | (T;T) | 0.64x reduced risk for breast cancer: and high... | | | |
| 2.4 | rs9272346 | (G;G) | 0.08x risk type-1 diabetes | | Link | |
| 2.1 | rs1136410 | (C;C) | 0.80x reduced risk for glioblastoma | Link | Link | |
| 2.1 | rs2511989 | (A;G) | 0.63x decreased age-related macular degeneratio... | | Link | |
| 2 | rs1026732 | (A;A) | <0.70x risk for restless legs | | Link | |
| 2 | rs10503669 | (A;C) | 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 | rs1128535 | (G;G) | Reduced risk (0.77x) for Crohn's disease | | | |
| 2 | rs1160312 | (G;G) | Reduced risk of Baldness. | | Link | |
| 2 | rs11635424 | (A;A) | <0.70x risk for restless legs | | Link | |
| 2 | rs12593813 | (A;A) | <0.71x risk for restless legs | | Link | |
| 2 | rs12678919 | (A;G) | Associated with higher HDL cholesterol | | Link | |
| 2 | rs12979860 | (C;C) | ~80% of such hepatitis C patients respond to tr... | | Link | Link |
| 2 | rs1799884 | (G;G) | Mothers have typical Birth-Weight babies. Sligh... | | | |
| 2 | rs1864163 | (A;G) | Associated with higher HDL cholesterol | | Link | |
| 2 | rs2241423 | (A;G) | 0.79 decreased risk for obesity | | | |
| 2 | rs3218536 | (A;A) | Lower risk for breast: ovarian cancer | Link | Link | |
| 2 | rs3738579 | (C;T) | 0.5x decreased risk for cervical cancer: HNSCC:... | | | |
| 2 | rs4149268 | (G;G) | Associated with higher HDL cholesterol | | Link | |
| 2 | rs6511720 | (G;T) | Slightly lower odds of developing CHD. | | Link | Link |
| 2 | rs7105934 | (A;G) | 0.69 times lower odds of developing renal cell ... | | | |
| 2 | rs763110 | (C;T) | ~0.80x reduced cancer risk | | | Link |
| 2 | rs7776725 | (T;T) | Stronger bones | | Link | |
| 2 | rs801114 | (T;T) | 0.78x decreased Basal Cell Carcinoma risk. | | Link | |
| 2 | rs9525638 | (C;C) | Stronger bones | | | |
| 1.8 | rs4714156 | (C;C) | <0.61x risk for restless legs | | | |

| Mag. | Identifier | Genotype | Summary | ExAC | GetEvidence | ClinVar |
|------|------------|----------|--|------|-------------|---------|
| 1.5 | rs1063192 | (C;T) | 0.71x reduced risk of myocardial infarction | | | |
| 1.5 | rs11212617 | (A;C) | Somewhat increased likelihood of treatment succ... | | | Link |
| 1.5 | rs309375 | (G;G) | Smaller mosquito bites | | | |
| 1.5 | rs3790844 | (C;T) | Slightly reduced risk (0.77x) for pancreatic ca... | | | |
| 1.5 | rs3851179 | (A;G) | 0.85x decreased risk for Alzheimer's disease | | Link | |
| 1.5 | rs4149274 | (C;C) | Associated with higher HDL (good) cholesterol. | | | |
| 1.5 | rs4939883 | (C;C) | Associated with higher HDL cholesterol | | Link | |
| 1.4 | 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 | rs2361502 | (C;C) | Possible higher levels of serum bilirubin and l... | | | |
| 1.1 | rs11172113 | (C;T) | 0.9x lower risk for migraines | | | |
| 1.1 | rs13333226 | (A;G) | Slightly lower risk for hypertension | | | Link |
| 1.1 | rs2293347 | (G;G) | Among NSCLC patients: better Gefitinib response... | Link | | Link |
| 1.1 | rs7568369 | (G;T) | 0.90x reduced risk of obesity | | | |
| 1 | rs182549 | (C;T) | Can digest milk. | | | Link |
| 1 | rs2351299 | (G;T) | Possible reduced risk of Autism | | | |
| 1 | rs2494732 | (T;T) | Lower odds of psychosis | Link | Link | |
| 1 | rs7850258 | (A;G) | Typical odds of developing primary hypothyroidi... | | | |
| 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 | 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 | rs5065 | (A;A) | 1.12x risk on diuretic; if hypertensive: better... | Link | Link | Link |
| 0 | rs6259 | (G;G) | Best inverse correlation between tea-drinking: ... | Link | Link | |
| 0 | rs7305115 | (A;A) | Individuals showed a significantly lower risk o... | Link | Link | |
| 0 | rs74315403 | (G;G) | PrP codon 178 (D) - non pathogenic variant | | | Link |
| 0 | rs7495174 | (A;A) | Blue/gray eyes more likely | | Link | |
| 0 | rs9394492 | (C;C) | <0.76x risk for restless legs | | | |
| 0 | rs9951307 | (A;G) | 0.10 decreased risk for brain edema after a str... | | | |

3.2 Possibly Harmful Traits

| Mag. | Identifier | Genotype | Summary | ExAC | GetEvidence | ClinVar |
|------|------------|----------|--|------|-------------|---------|
| 3.1 | rs1421085 | (C;C) | ~1.7x increased obesity risk | | Link | Link |
| 3 | rs10897346 | (C;C) | If depressed: 2.6x more likely to not respond t... | | | |
| 3 | rs1121980 | (T;T) | 2.76x risk for obesity | | Link | |
| 3 | rs13266634 | (C;C) | Increased risk for type-2 diabetes | Link | Link | Link |
| 3 | rs16969968 | (A;A) | Higher risk for nicotine dependence; lower risk... | Link | Link | Link |
| 3 | rs1800460 | (A;G) | (TPMT*3B) impaired drug metabolism | Link | Link | Link |
| 3 | rs1801282 | (C;G) | Unconfirmed higher risk of cardiovascular disea... | Link | Link | Link |
| 3 | rs2306402 | (C;T) | 1.18x increased risk for late-onset Alzheimer's... | | | |
| 3 | rs3803662 | (T;T) | 1.6x increased risk for breast cancer | | Link | |
| 3 | rs55705857 | (A;G) | 6x increased risk of glioma of IDH1/IDH2 subtyp... | | | |
| 3 | rs7754840 | (C;G) | 1.3x increased risk for type-2 diabetes | | Link | |
| 3.0 | rs1142345 | (A;G) | TPMT*3C . impaired drug metabolism | Link | Link | Link |
| 2.9 | rs16901979 | (A;C) | 1.5x increased risk for prostate cancer | | Link | |
| 2.7 | rs10830963 | (C;G) | Increased type-2 diabetes risk; higher gestatio... | | Link | |
| 2.6 | rs8034191 | (C;C) | 1.80x lung cancer risk; decreased response to a... | | Link | |
| 2.5 | rs1051730 | (T;T) | 1.8x increased risk of lung cancer; reduced res... | 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 | rs2943634 | (C;C) | Slightly higher risk of ischemic stroke | | Link | |
| 2.5 | rs339331 | (T;T) | Prostate cancer risk | | | |
| 2.5 | rs3738919 | (C;C) | 1.94x risk of developing rheumatoid arthritis | | | |
| 2.5 | rs5888 | (C;T) | 3x higher risk for age-related macular degenera... | 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 |
| 2.4 | rs1143679 | (A;G) | 1.78x increased risk for SLE | Link | Link | |
| 2.3 | rs7966230 | (C;G) | Slightly lower levels of plasma VWF | | | |
| 2.2 | rs1024611 | (C;C) | Increased risk of exercise induced ischemia: In... | | | 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 | rs1329428 | (G;G) | 2x increased risk for macular degeneration | | | |
| 2.1 | rs1360780 | (T;T) | 1.3x increased risk for depression | | Link | |
| 2.1 | rs17070145 | (C;C) | Reduced memory abilities | | | Link |
| 2.1 | rs2231137 | (A;G) | ~1.5-3x increased risk for ischemic stroke | Link | Link | Link |
| 2.1 | rs2254958 | (C;T) | 1.24x increased risk for Alzheimer's | | | |
| 2.1 | rs2270641 | (G;G) | 3.7x higher risk for schizophrenia | Link | Link | |
| 2.1 | rs2294008 | (T;T) | Increased risk of gastric and bladder cancer | Link | Link | |
| 2.1 | rs4149056 | (C;T) | Reduced breakdown of some drugs; 5x increased m... | Link | Link | Link |
| 2.1 | rs4363657 | (C;T) | 4.5x increased myopathy risk for statin users | | Link | |
| 2.1 | rs4402960 | (T;T) | 1.2x increased risk for type-2 diabetes: 1.5x r... | | Link | Link |
| 2.1 | rs4430796 | (A;A) | 1.38x increased risk for prostate cancer | | Link | |
| 2.1 | rs4444903 | (G;G) | 3.5x risk of hep-cancer in cirrhosis patients; ... | | | Link |
| 2.1 | rs795484 | (A;G) | Increased morphine dose requirement and postope... | | | |
| 2.1 | rs944289 | (C;T) | 1.3x increased thyroid cancer risk | | Link | |
| 2 | rs10248420 | (A;A) | 7x less likely to respond to certain antidepres... | | Link | |
| 2 | rs10260404 | (C;C) | 1.60x risk of developing ALS | | Link | |
| 2 | rs1042838 | (G;T) | 1.28x risk for endometrial ovarian cancer; over... | Link | Link | |
| 2 | rs1045642 | (C;T) | Slower metaboliser for some drugs | Link | Link | Link |
| 2 | rs1050152 | (C;T) | 2.1x increased risk of Crohn's disease | Link | Link | Link |
| 2 | rs10937823 | (C;T) | Some association with bipolar disorder | | | |
| 2 | rs10984447 | (A;A) | >1.17x increased risk for multiple sclerosis | | Link | |
| 2 | rs11190870 | (C;T) | Possibly increased risk of scoliosis | | | |
| 2 | rs1169300 | (A;A) | ~2x increased lung cancer risk | | | |
| 2 | rs11983225 | (T;T) | 7x less likely to respond to certain antidepres... | | Link | |

| Mag. | Identifier | Genotype | Summary | ExAC | GetEvidence | ClinVar |
|------|------------|----------|--|------|-------------|---------|
| 2 | rs12567232 | (A;G) | Increased risk for Crohn's Disease | | Link | |
| 2 | rs12770228 | (A;A) | 2x increased risk for meningioma | | | |
| 2 | rs1361600 | (G;G) | ~2x increased risk for adult-onset asthma in Ja... | | | |
| 2 | rs1585215 | (A;G) | 2x increased risk for Hodgkin lymphoma | | | |
| 2 | rs16942 | (A;G) | Very slightly increased breast cancer risk | Link | Link | Link |
| 2 | rs16944 | (G;G) | Increased risk of mental disorders | | Link | |
| 2 | rs1734791 | (A;A) | 1.4x increased risk for lupus | | | |
| 2 | rs17576 | (G;G) | Higher risk for lung cancer: and COPD in smoker... | Link | Link | |
| 2 | rs1800896 | (A;A) | 1.8x increased prostate cancer risk | | | |
| 2 | rs2073963 | (G;T) | Increased risk of baldness | | | |
| 2 | rs2143340 | (C;T) | Increased risk of dyslexia and poor reading per... | | | |
| 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 | rs2274223 | (A;G) | 1.5x increased risk for stomach and esophageal ... | Link | 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 | 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 | rs2707466 | (G;G) | Weaker bones | Link | Link | |
| 2 | rs2736990 | (C;C) | Increased risk of developing Parkinson's Diseas... | | Link | |
| 2 | rs2908004 | (C;C) | Weaker bones | Link | Link | |
| 2 | rs3197999 | (T;T) | 1.2x risk of Crohn's | Link | Link | |
| 2 | rs3212227 | (C;C) | Significantly increased risk of developing cerv... | | | |
| 2 | rs358806 | (C;C) | 1.78x increased risk of developing Type-2 diabe... | | Link | |
| 2 | rs3746444 | (C;T) | ~1.2x increased risk for cancer | Link | | |
| 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 | rs449647 | (A;A) | Lower levels of ApoE | | | |
| 2 | rs4792311 | (A;A) | Increased risk of prostate cancer | Link | Link | Link |
| 2 | rs493258 | (A;G) | 1.15x risk of Age Related Macular Degeneration | | | |
| 2 | rs4968451 | (A;C) | 1.61x increased risk for meningioma | | | |
| 2 | rs5174 | (A;A) | 1.3x increased risk for heart disease | Link | Link | Link |
| 2 | rs520354 | (A;G) | Increased risk in men for biliary conditions | | | |
| 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 | rs6897932 | (C;C) | 1.08x increased risk for multiple sclerosis | Link | Link | Link |
| 2 | rs6997709 | (G;G) | 1.5x higher risk for hypertension | | | |
| 2 | rs7216389 | (T;T) | 1.5x increased risk for Childhood Asthma. | | Link | |
| 2 | rs7442295 | (A;A) | ~4x higher risk for hyperuracemia | | Link | |
| 2 | rs744373 | (C;C) | 1.17x risk of Alzheimer's | | | |
| 2 | rs7536563 | (A;A) | >1.12x risk of multiple sclerosis | | Link | |
| 2 | rs763361 | (T;T) | Increased risk for multiple autoimmune diseases... | Link | Link | |
| 2 | rs7639618 | (C;T) | 1.45x increased osteoarthritis risk | Link | | |
| 2 | rs7794745 | (A;T) | Slightly increased risk for autism | | Link | Link |
| 2 | rs7807268 | (C;G) | 1.3x risk for Crohn's disease | | Link | |
| 2 | rs7961152 | (A;A) | 1.5x higher risk for hypertension | | | |
| 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 | rs965513 | (A;G) | 1.77x increased thyroid cancer risk | | Link | |
| 1.9 | rs7923837 | (A;G) | 1.6x risk for T2D | | | |

| Mag. | Identifier | Genotype | Summary | ExAC | GetEvidence | ClinVar |
|------|------------|----------|--|------|-------------|---------|
| 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 |
| 1.6 | rs1537415 | (C;G) | 1.6x increased risk for periodontitis | | Link | |
| 1.6 | rs2059693 | (T;T) | 1.6x increased risk for testicular cancer | | | |
| 1.6 | rs2736100 | (G;G) | 1.6x higher risk for glioma development | | Link | |
| 1.6 | rs33980500 | (C;T) | 1.6x increase in risk for psoriatic arthritis | Link | Link | Link |
| 1.6 | rs3764880 | (A;A) | 1.2 - 1.8x increased tuberculosis risk | Link | Link | |
| 1.6 | rs3775948 | (C;G) | Slightly higher risk for gout | | | |
| 1.5 | rs10859871 | (A;C) | Slight (~1.2x) increase in endometriosis risk | | | |
| 1.5 | rs11171739 | (C;T) | 1.34x risk of developing Type-1 diabetes | | Link | |
| 1.5 | rs12037606 | (A;G) | 1.22x risk of developing Crohn's disease | | | |
| 1.5 | rs12210050 | (C;T) | Slightly higher risk for basal cell carcinoma | | Link | |
| 1.5 | rs1223271 | (A;G) | 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 | rs13181 | (G;T) | 1.12x increased risk for cutaneous melanoma | Link | Link | Link |
| 1.5 | rs13376333 | (C;T) | 1.5x higher risk of atrial fibrillation | | Link | |
| 1.5 | rs140701 | (A;G) | Increased risk for anxiety disorders | | | |
| 1.5 | rs1801274 | (T;T) | Complex; generally greater risk for cancer prog... | Link | Link | Link |
| 1.5 | rs1867277 | (A;G) | 1.5x increased risk for thyroid cancer | | | |
| 1.5 | rs1975197 | (C;T) | 1.3x increased risk of developing restless legs... | | Link | |
| 1.5 | rs2240340 | (A;G) | Slightly increased (1.5x) risk for RA | Link | | |
| 1.5 | rs2280714 | (A;A) | 1.4x increased risk of SLE | | | |
| 1.5 | rs2697962 | (A;G) | Slightly increased risk of developing Parkinson... | | | |
| 1.5 | rs2881766 | (T;T) | Slightly increased risk for pregnancy-induced h... | | | |
| 1.5 | rs393152 | (A;A) | Increased risk of both PD and AD | Link | Link | |
| 1.5 | rs401681 | (C;C) | ~1.2x increased risk for several types of cance... | | Link | |
| 1.5 | rs4464148 | (C;T) | 1.10x increased risk for colorectal cancer | | | |
| 1.5 | rs464049 | (C;T) | Increased risk of schizophrenia in limited stud... | | | |
| 1.5 | rs4656461 | (A;G) | 1.5x increased risk for open angle glaucoma | | | |
| 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;G) | Slightly higher fat mass | | | |
| 1.5 | rs619203 | (C;G) | Increases susceptibility to Myocardial Infarcti... | Link | Link | |
| 1.5 | rs642961 | (A;G) | 1.68x increased risk of cleft lip | | Link | |
| 1.5 | rs6498169 | (A;G) | 1.14x risk of multiple sclerosis | | Link | |
| 1.5 | rs6601764 | (C;T) | 1.16x increased risk of developing Crohn's dise... | | Link | |
| 1.5 | rs6896702 | (C;T) | Slightly increased risk of developing Parkinson... | | | |
| 1.5 | rs6908425 | (C;T) | 1.63x increased risk of developing Crohn's dise... | | Link | |
| 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 | 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 | rs9642880 | (G;T) | 1.2x increased bladder cancer risk | | Link | |
| 1.5 | rs9652490 | (A;G) | Slightly increased risk of developing Parkinson... | | Link | |
| 1.5 | rs995030 | (G;G) | Non-protective against testicular cancer | | Link | |
| 1.4 | rs1126497 | (C;T) | 1.4x increased risk for breast cancer | Link | Link | Link |
| 1.4 | rs2046210 | (C;T) | 1.4x increased breast cancer risk | | Link | Link |
| 1.4 | rs3131296 | (G;G) | 1.4x increased risk for schizophrenia | | Link | |
| 1.4 | rs4959039 | (A;G) | 1.4x higher risk for multiple sclerosis | | | |
| 1.4 | rs6010620 | (G;G) | 1.4x higher risk for glioma development; but th... | | Link | |
| 1.4 | rs8050136 | (A;A) | 1.4x increased risk for T2D in some populations... | | Link | |
| 1.34 | rs17465637 | (C;C) | 1.34x higher risk for myocardial infarction | Link | Link | |
| 1.3 | rs1042713 | (A;G) | 1.3x increased risk that pediatric inhaler use ... | Link | Link | Link |

| Mag. | Identifier | Genotype | Summary | ExAC | GetEvidence | ClinVar |
|------|------------|----------|--|------|-------------|---------|
| 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 | rs110419 | (A;G) | 1.3x increased risk for neuroblastoma | | | |
| 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 | rs34330 | (C;T) | 1.3x higher risk for endometrial cancer (in Chi... | | | |
| 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 | |
| 1.3 | rs9858542 | (A;A) | 1.8x risk of Crohn's disease | Link | Link | |
| 1.2 | rs10865331 | (A;G) | 1.2x higher risk for ankylosing spondylitis | | | |
| 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 | rs1800693 | (A;G) | Slight (1.2x) increase in risk for multiple scl... | Link | Link | Link |
| 1.2 | rs2056116 | (A;G) | 1.18x risk for breast cancer | | | |
| 1.2 | rs2072590 | (G;T) | 1.2x increased risk for ovarian cancer | | | |
| 1.2 | rs2814707 | (A;G) | 1.2x increased risk for ALS | | Link | |
| 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 | rs4686484 | (A;A) | Slightly increased risk for celiac disease | | | |
| 1.2 | rs4795067 | (A;G) | Slight increase in risk for psoriatic arthritis... | | | |
| 1.2 | rs4977756 | (A;G) | 1.39x higher risk for glioma development | | Link | |
| 1.1 | rs1344706 | (G;T) | 1.1x increased risk for schizophrenia | | 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 | 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 | rs3818361 | (C;T) | 1.15x increased risk for late-onset Alzheimer's... | | | |
| 1.1 | rs4324715 | (C;T) | 1.5x increased testicular cancer risk for men | | | |
| 1.1 | rs6897876 | (C;T) | Slight increase in testicular cancer risk for m... | | | |
| 1.1 | rs7171755 | (A;G) | Very slight decrease in cortical thickness and ... | | | |
| 1.1 | rs7412 | (C;C) | More likely to gain weight if taking olanzapine... | Link | Link | Link |
| 1.1 | rs925391 | (C;C) | More likely to go bald; common | | | |
| 1.07 | rs2291834 | (C;C) | Very slightly higher risk for myocardial infarc... | | | |
| 1 | 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;A) | >1.1x risk of type-1 diabetes | Link | Link | Link |
| 1 | rs6932590 | (T;T) | 1.1x increased risk for schizophrenia | | Link | |
| 1.0 | rs11246226 | (A;A) | Increased risk of schizophrenia in limited stud... | | Link | |
| 0 | rs1061646 | (C;C) | 1.16x increased risk for breast cancer | Link | | Link |
| 0 | rs1128503 | (T;T) | Likely to require more methadone during heroin ... | Link | Link | Link |
| 0 | rs3761418 | (A;A) | 1.3x increased risk for depression | | | |
| 0 | rs3813929 | (C;C) | Possible weight gain if taking olanzapine | | Link | Link |
| 0 | rs7787082 | (G;G) | 7x less likely to respond to certain antidepres... | | Link | |

3.3 Genosets (Multi-variant Phenotypes)

| Magnitude | Identifier | Summary |
|-----------|------------|--|
| 3 | gs241 | Lighter green: brown or hazel eye color |
| 2.5 | gs102 | ALS risk |
| 2.5 | gs155 | CYP3A5 non-expressor |
| 2.5 | gs281 | Part of the 88% of the population claimed not t... |
| 2.5 | gs285 | You will lose 2.5x as much weight on a low fat ... |
| 2 | gs101 | Probably able to digest milk |
| 2 | gs154 | NAT2 Slow metabolizer |
| 2 | gs246 | APOE3/APOE3 |
| 1.5 | gs220 | HLA-B*1502? |
| 1.5 | gs247 | Parkinson's Disease Risk |
| 1.2 | gs184 | Able to taste bitterness. |
| 1 | gs182 | CYP2D6*39 |
| 0 | gs158 | CYP1A2 normal metabolizer |

4 Raw Data

The raw data used to create this report has been assigned the identifier ERS1176603 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/ERS1176603>

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.