

PGP-UK Genomics Report for uk072667

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 | 4946542 |
| Variants filtered out | 0 |
| Novel / existing variants | 483218 (9.8) / 4452676 (90.2) |
| Overlapped genes | 56691 |
| Overlapped transcripts | 67517 |
| Overlapped regulatory features | 166612 |

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.

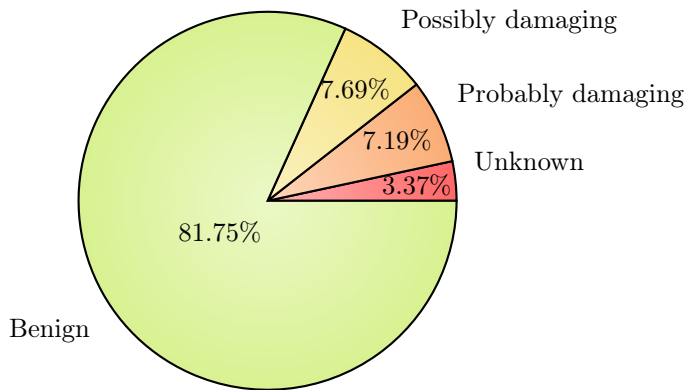


Figure 2: PolyPhen Summary

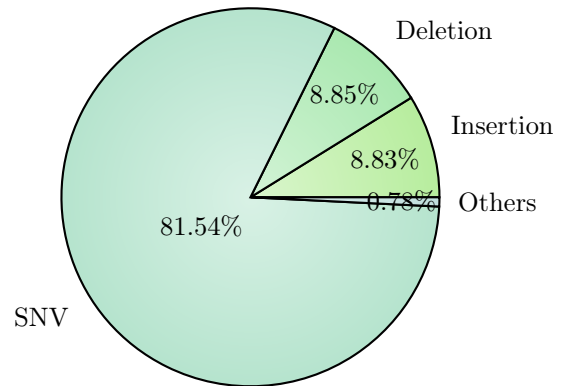


Figure 3: Variant Class

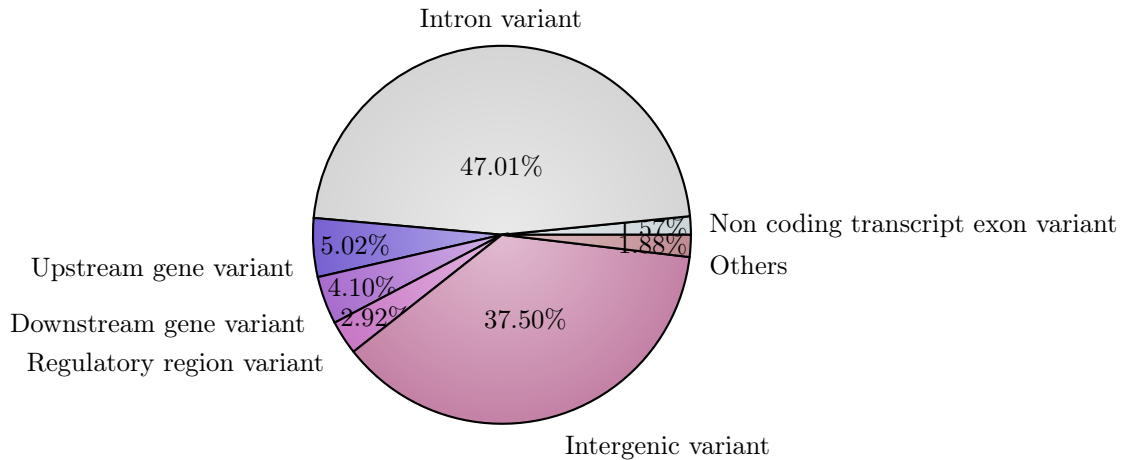


Figure 4: Consequence type

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).

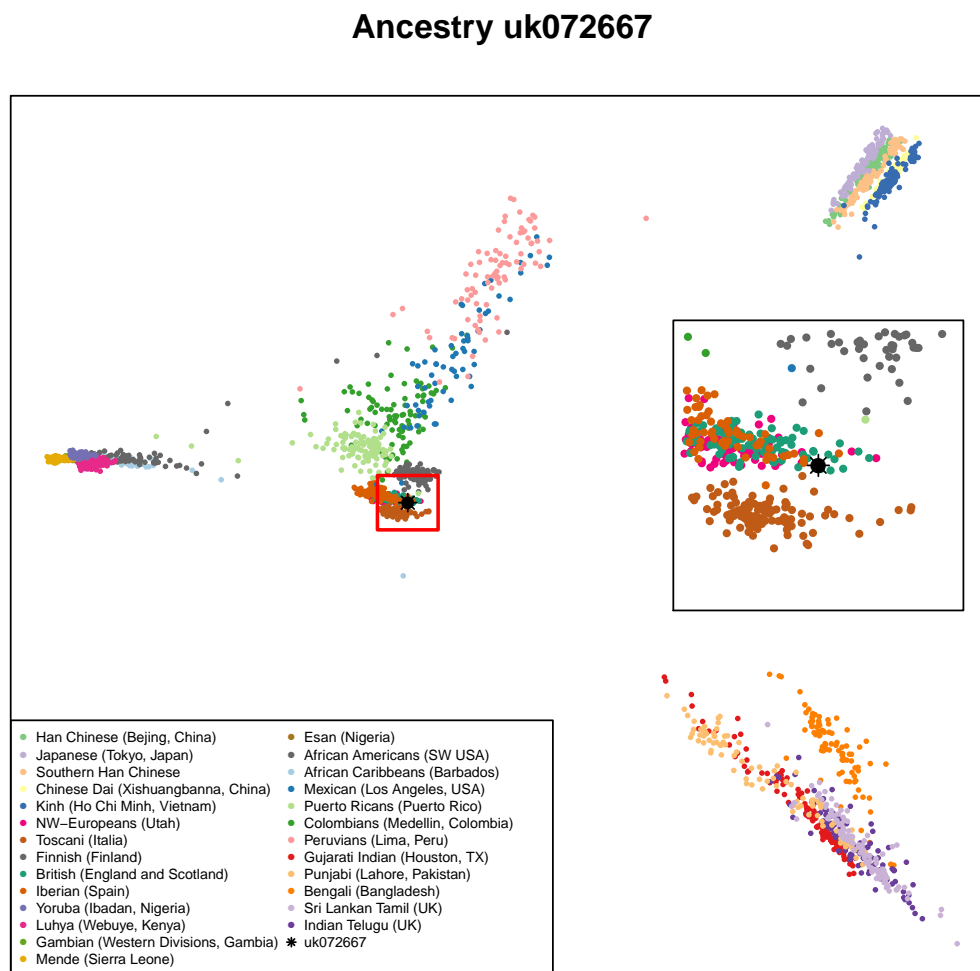


Figure 5: Ancestry Principal Component Analysis

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 |
| 2.1 | rs2511989 | (A;G) | 0.63x decreased age-related macular degeneratio... | | Link | |
| 2.1 | rs3775291 | (A;G) | 0.71x decreased risk for dry age related macula... | Link | Link | Link |
| 2 | rs10088218 | (A;A) | 0.5x decreased risk for ovarian cancer | | | |
| 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 | rs12979860 | (C;C) | ~80% of such hepatitis C patients respond to tr... | | Link | Link |
| 2 | rs17070145 | (C;T) | Increased memory performance | | | Link |
| 2 | rs1800012 | (T;T) | Possibly reduced risk of anterior cruciate liga... | | Link | Link |
| 2 | rs1864163 | (G;G) | Associated with higher HDL cholesterol | | Link | |
| 2 | rs2073963 | (T;T) | Reduced risk of baldness | | | |
| 2 | rs2235015 | (G;T) | Somewhat more likely to respond to certain anti... | Link | Link | |
| 2 | rs2241423 | (A;G) | 0.79 decreased risk for obesity | | | |
| 2 | rs2542052 | (C;C) | Better odds of living to 100 | | | |
| 2 | rs261332 | (A;G) | Associated with higher HDL cholesterol | | | |
| 2 | rs266729 | (G;G) | 0.73x decreased risk for colorectal cancer | | Link | |
| 2 | rs3738579 | (C;T) | 0.5x decreased risk for cervical cancer: HNSCC:... | | | |
| 2 | rs3764261 | (G;T) | Associated with higher HDL cholesterol | | Link | Link |
| 2 | rs3819331 | (T;T) | Lower risk of autism | Link | | |
| 2 | rs4149268 | (A;G) | Associated with higher HDL cholesterol | | Link | |
| 2 | rs6505162 | (A;C) | 0.58x decreased risk for esophageal cancer | Link | | |
| 2 | rs6855911 | (A;G) | 0.62x decreased risk for gout | | Link | |
| 2 | rs7776725 | (T;T) | Stronger bones | | Link | |
| 2 | rs9272346 | (A;G) | 0.3x risk type-1 diabetes | | Link | |
| 1.8 | rs1128535 | (A;G) | 0.77x risk for Crohn's disease | | | |
| 1.8 | rs1746048 | (C;T) | 0.94 decreased risk for coronary heart disease | | Link | |

| Mag. | Identifier | Genotype | Summary | ExAC | GetEvidence | ClinVar |
|------|------------|----------|--|------|-------------|---------|
| 1.8 | rs1800588 | (C;T) | Higher HDL-C levels | Link | 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 |
| 1.6 | rs1061170 | (T;T) | Lower risk for AMD: generally longer live than ... | Link | Link | Link |
| 1.6 | rs3025786 | (C;T) | Slightly decreased Alzheimer's disease risk amo... | Link | | |
| 1.5 | rs1026732 | (A;G) | 0.70x risk for restless legs | | Link | |
| 1.5 | rs1063192 | (C;T) | 0.71x reduced risk of myocardial infarction | | | |
| 1.5 | rs11465804 | (G;T) | 0.68x lower risk for spondylitis | Link | Link | |
| 1.5 | rs11635424 | (A;G) | 0.70x risk for restless legs | | Link | |
| 1.5 | rs12593813 | (A;G) | 0.71x risk for restless legs | | Link | |
| 1.5 | rs3784709 | (C;T) | 0.71x risk of developing restless legs syndrome... | | Link | |
| 1.5 | rs3790844 | (C;T) | Slightly reduced risk (0.77x) for pancreatic ca... | | | |
| 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 | rs4939883 | (C;T) | Associated with higher HDL cholesterol | | Link | |
| 1.5 | rs5888 | (C;C) | Higher HDL cholesterol but lower risk for age-r... | Link | | |
| 1.5 | rs9939609 | (T;T) | Lower risk of obesity and Type-2 diabetes | | Link | |
| 1.4 | rs1165205 | (A;T) | 0.85x decreased gout risk | | Link | |
| 1.4 | rs6700125 | (C;C) | 0.7x decreased risk for ALS | | | |
| 1.2 | rs4320932 | (A;G) | 0.87x decreased risk for ovarian cancer | | | |
| 1.2 | rs6048 | (G;G) | Slightly lower risk (10-20%) of deep vein throm... | Link | Link | Link |
| 1.2 | 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 | rs13333226 | (A;G) | Slightly lower risk for hypertension | | | Link |
| 1 | rs10248420 | (A;G) | 7x more likely to respond to certain antidepres... | | Link | |
| 1 | rs11983225 | (C;T) | 7x more likely to respond to certain antidepres... | | Link | |
| 1 | rs182549 | (C;T) | Can digest milk. | | | Link |
| 1 | rs2235040 | (A;G) | 7x more likely to respond to certain antidepres... | Link | Link | |
| 1 | rs2235067 | (A;G) | 7x more likely to respond to certain antidepres... | | | |
| 1 | rs2494732 | (T;T) | Lower odds of psychosis | Link | Link | |
| 1 | rs2546890 | (G;G) | Lower risk of multiple sclerosis | | | |
| 1 | rs4148739 | (A;G) | 7x more likely to respond to certain antidepres... | | 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 | 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 | rs5065 | (A;A) | 1.12x risk on diuretic; if hypertensive: better... | Link | Link | Link |
| 0 | rs6259 | (G;G) | Best inverse correlation between tea-drinking: ... | Link | Link | |
| 0 | rs74315403 | (G;G) | PrP codon 178 (D) - non pathogenic variant | | | Link |
| 0 | rs7495174 | (A;A) | Blue/gray eyes more likely | | Link | |

3.2 Possibly Harmful Traits

| Mag. | Identifier | Genotype | Summary | ExAC | GetEvidence | ClinVar |
|------|------------|----------|--|------|-------------|---------|
| 3 | rs10897346 | (C;C) | If depressed: 2.6x more likely to not respond t... | | | |
| 3 | rs1801282 | (C;G) | Unconfirmed higher risk of cardiovascular disea... | Link | Link | Link |
| 3 | rs1983132 | (C;T) | 2 - 3x higher prostate cancer risk if routinely... | | | |
| 3 | rs2237717 | (T;T) | Reduced abilities related to neurocognition and... | | | |
| 3 | rs2306402 | (C;T) | 1.18x increased risk for late-onset Alzheimer's... | | | |
| 3 | rs2981582 | (C;T) | 1.3x higher risk of ER+ breast cancer | | Link | |
| 3 | rs3803662 | (T;T) | 1.6x increased risk for breast cancer | | Link | |
| 3 | rs7754840 | (C;G) | 1.3x increased risk for type-2 diabetes | | Link | |
| 2.9 | rs1061235 | (A;T) | 26% risk of bad reaction to anti-epileptic carb... | | | Link |
| 2.7 | rs10830963 | (C;G) | Increased type-2 diabetes risk; higher gestatio... | | 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 | rs16969968 | (A;G) | Slightly higher risk for nicotine dependence: l... | Link | Link | Link |
| 2.5 | rs187238 | (G;G) | Hypertension increases risk 3.75x for sudden ca... | | | |
| 2.5 | 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 | rs3780374 | (A;G) | Substantially increased odds of developing V617... | | | |
| 2.5 | rs613872 | (G;T) | ~5 fold higher risk for Fuchs' dystrophy: a cor... | | | |
| 2.5 | rs7574865 | (G;T) | 1.3x risk of rheumatoid arthritis; 1.55x risk o... | | Link | Link |
| 2.5 | rs8034191 | (C;T) | 1.27x lung cancer risk | | Link | |
| 2.5 | rs891512 | (A;G) | Higher blood pressure than G;G | Link | | |
| 2.4 | rs7966230 | (G;G) | Slightly lower levels of plasma VWF | | | |
| 2.3 | rs1859962 | (G;G) | 1.28x increased risk for prostate cancer | | Link | |
| 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 |
| 2.1 | rs10811661 | (T;T) | 1.2x increased risk for type-2 diabetes | | Link | |
| 2.1 | rs1585215 | (G;G) | 3.5x increased risk for Hodgkin lymphoma | | | |
| 2.1 | rs17077540 | (A;G) | 1.6x major depressive disorder risk | | | |
| 2.1 | rs17563 | (C;C) | Risk for otosclerosis | Link | Link | Link |
| 2.1 | rs2254958 | (C;T) | 1.24x increased risk for Alzheimer's | | | |
| 2.1 | rs5186 | (A;C) | ~1.4x increased risk of hypertension | Link | Link | Link |
| 2.1 | rs646776 | (A;A) | 1.2x risk of coronary artery disease | | Link | |
| 2.1 | rs795484 | (A;G) | Increased morphine dose requirement and postope... | | | |
| 2.1 | rs944289 | (C;T) | 1.3x increased thyroid cancer risk | | Link | |
| 2 | rs10086908 | (C;T) | 1.7x increased risk for prostate cancer | | | |
| 2 | rs10260404 | (C;C) | 1.60x risk of developing ALS | | Link | |
| 2 | rs10455872 | (A;G) | 1:51x increased Coronary Heart disease risk | | | 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 | rs1051730 | (C;T) | 1.3x increased risk of lung cancer | Link | Link | Link |
| 2 | rs10871777 | (A;G) | Adults likely to be 0.22 BMI units higher | | | |
| 2 | rs10980705 | (T;T) | 3.7x increased risk for knee osteoarthritis | | | |
| 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 | rs1219648 | (A;G) | 1.20x risk for breast cancer | | Link | |
| 2 | rs13254738 | (A;C) | 1.18x prostate cancer risk | | Link | |
| 2 | rs1333048 | (A;C) | 1.3x increased coronary artery disease risk | | | |
| 2 | rs1360780 | (C;T) | 1.3x increased risk for depression | | Link | |
| 2 | rs1361600 | (G;G) | ~2x increased risk for adult-onset asthma in Ja... | | | |
| 2 | rs144848 | (G;G) | Very slightly increased breast cancer risk | Link | Link | Link |
| 2 | rs1691053 | (A;G) | Increased risk of developing prostate cancer | | | |
| 2 | rs16942 | (A;G) | Very slightly increased breast cancer risk | Link | Link | Link |

| Mag. | Identifier | Genotype | Summary | ExAC | GetEvidence | ClinVar |
|------|------------|----------|--|------|-------------|---------|
| 2 | rs17001266 | (-;C) | 1.58x increased risk for schizophrenia in males... | | | |
| 2 | rs17228212 | (C;C) | >1.26x increased risk for heart disease | | Link | |
| 2 | rs17435 | (T;T) | 1.4x increased risk for lupus | | | |
| 2 | rs17576 | (A;G) | Higher risk for MI and lung cancer: and COPD in... | Link | Link | |
| 2 | rs17696736 | (A;G) | 1.34x risk of type-1 diabetes | | Link | |
| 2 | rs17782313 | (C;T) | Adults likely to be 0.22 BMI units higher | | Link | Link |
| 2 | rs1800629 | (A;G) | Complex; generally higher risk for certain dise... | Link | Link | Link |
| 2 | rs1800896 | (A;G) | 1.6x increased prostate cancer risk | | | |
| 2 | rs1801160 | (A;G) | Possible 5-fluorouracil toxicity | Link | Link | Link |
| 2 | rs2056116 | (G;G) | 1.41x risk for breast cancer | | | |
| 2 | rs2201841 | (T;T) | 2.4x increased risk for Graves' disease | | Link | |
| 2 | rs2230201 | (G;G) | >1.4x risk of lupus | Link | | |
| 2 | rs2305480 | (C;T) | 3.5x increase in risk of asthma for Han Chinese... | Link | Link | |
| 2 | rs2383206 | (A;G) | 1.4x increased risk for heart disease | | | |
| 2 | rs2383207 | (A;G) | Increased risk for heart disease | | | |
| 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 | (G;G) | 2x higher risk for skin cancer; possibly other ... | Link | Link | Link |
| 2 | rs2736100 | (T;T) | Higher risk of Interstitial lung disease: and t... | | Link | |
| 2 | rs3129934 | (C;T) | Increased risk of Multiple Sclerosis. | | Link | |
| 2 | rs3212227 | (A;C) | Significantly increased risk of developing cerv... | | | |
| 2 | rs3775948 | (G;G) | Slightly higher risk for gout | | | |
| 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 | (C;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 | rs4961 | (G;T) | 1.8x increased risk for high blood pressure | Link | Link | Link |
| 2 | rs520354 | (A;A) | Increased risk in men for biliary conditions | | | |
| 2 | rs5759167 | (T;T) | Higher prostate cancer risk | | Link | |
| 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 | rs6498169 | (A;A) | >1.14x risk of multiple sclerosis | | Link | |
| 2 | rs663048 | (G;T) | Significantly increased risk of developing lung... | Link | 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;G) | 1.5x higher risk for hypertension | | | |
| 2 | rs7442295 | (A;A) | ~4x higher risk for hyperuracemia | | Link | |
| 2 | rs744373 | (C;C) | 1.17x risk of Alzheimer's | | | |
| 2 | rs7807268 | (C;C) | 1.4x risk for Crohn's disease | | Link | |
| 2 | rs7923837 | (G;G) | 3.2x risk for T2D | | | |
| 2 | rs7961152 | (A;C) | 1.2x higher risk for hypertension | | | |
| 2 | rs828907 | (T;T) | Increased risk of bladder cancer and 2x risk of... | | | |
| 2 | rs854560 | (A;A) | Higher risk for heart disease: diabetic retinop... | Link | Link | Link |
| 2 | rs855913 | (G;T) | Reduced survival with ALS | | Link | |
| 2 | rs9525638 | (T;T) | Weaker bones | | | |
| 2 | rs9543325 | (C;C) | 1.37x Slightly higher pancreatic cancer risk | | | |
| 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 | rs9954153 | (G;T) | ~2.5x higher risk for Fuchs' dystrophy: a corne... | | | |
| 2.0 | rs1044396 | (C;C) | Increased risk of Nicotine dependence among mal... | Link | Link | Link |
| 2.0 | rs2156921 | (G;G) | 1.29x increased risk for depression | | | |
| 1.8 | rs143383 | (T;T) | 1.3x increased risk for osteoarthritis | | Link | Link |
| 1.8 | rs2278206 | (T;T) | 1.16x increased risk for asthma | Link | Link | |
| 1.6 | rs11523871 | (A;C) | 1.6x increased breast cancer risk for women ove... | Link | Link | |

| Mag. | Identifier | Genotype | Summary | ExAC | GetEvidence | ClinVar |
|------|------------|----------|--|------|-------------|---------|
| 1.6 | rs1537415 | (C;G) | 1.6x increased risk for periodontitis | | Link | |
| 1.6 | rs2981745 | (C;T) | 1.6x increased risk for breast cancer in female... | | | |
| 1.6 | rs33980500 | (C;T) | 1.6x increase in risk for psoriatic arthritis | Link | Link | Link |
| 1.6 | rs3764880 | (A;A) | 1.2 - 1.8x increased tuberculosis risk | Link | Link | |
| 1.5 | rs10492519 | (A;G) | Slightly increased risk of developing prostate ... | | | |
| 1.5 | rs10757272 | (C;T) | 1.30x increased risk for Coronary artery diseas... | | | |
| 1.5 | 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 | rs12431733 | (C;T) | Slightly increased risk of developing Parkinson... | | Link | |
| 1.5 | rs12469063 | (A;G) | Slightly increased risk of developing restless ... | | | |
| 1.5 | rs12498742 | (A;A) | 1.25 increased risk for gout | | | |
| 1.5 | rs13149290 | (C;T) | Slightly increased risk of developing prostate ... | | | |
| 1.5 | rs13181 | (G;T) | 1.12x increased risk for cutaneous melanoma | Link | Link | Link |
| 1.5 | rs140701 | (A;G) | Increased risk for anxiety disorders | | | |
| 1.5 | rs16944 | (A;G) | Minorly increased risk of mental illness and os... | | Link | |
| 1.5 | rs1801274 | (C;T) | Complex; generally greater risk for cancer prog... | Link | Link | Link |
| 1.5 | rs1975197 | (C;T) | 1.3x increased risk of developing restless legs... | | Link | |
| 1.5 | rs2007153 | (G;G) | Increased risk of schizophrenia in limited stud... | | | |
| 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 | rs2286812 | (C;T) | ~2x higher risk for Fuchs' dystrophy: a corneal... | | | |
| 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 | rs3087243 | (G;G) | Increased risk for autoimmune diseases | | Link | |
| 1.5 | rs309375 | (T;T) | Larger mosquito bites | | | |
| 1.5 | rs358806 | (A;C) | 0.86x increased risk of developing Type-2 diabe... | | Link | |
| 1.5 | rs3814570 | (C;T) | 1.3x increased risk for Crohn's disease with il... | | | |
| 1.5 | rs393152 | (A;A) | Increased risk of both PD and AD | Link | Link | |
| 1.5 | rs4027132 | (A;G) | 1.39x increased risk of developing bipolar diso... | | | |
| 1.5 | rs4506565 | (A;T) | 1.4x increased risk for type-2 diabetes | | Link | |
| 1.5 | rs4538475 | (A;G) | Slightly increased risk of developing Parkinson... | | Link | |
| 1.5 | rs4585 | (T;T) | Slightly poorer (0.75x) response to metformin i... | | | |
| 1.5 | rs464049 | (C;T) | Increased risk of schizophrenia in limited stud... | | | |
| 1.5 | rs486907 | (A;G) | 1.5x increased prostate cancer risk | 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 | rs6435862 | (G;T) | 1.7x higher risk of aggressive neuroblastoma | | Link | |
| 1.5 | rs6601764 | (C;T) | 1.16x 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 | 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 | rs9303277 | (C;T) | 1.46x Slightly increased risk of developing pri... | | | |
| 1.5 | rs9642880 | (G;T) | 1.2x increased bladder cancer risk | | 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 | |

| Mag. | Identifier | Genotype | Summary | ExAC | GetEvidence | ClinVar |
|------|------------|----------|--|------|-------------|---------|
| 1.3 | rs1042713 | (A;G) | 1.3x increased risk that pediatric inhaler use ... | 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 | rs1260326 | (C;T) | Slightly higher risk for gout | Link | Link | Link |
| 1.3 | rs1375144 | (C;T) | 1.32x increased risk of developing bipolar diso... | | | |
| 1.3 | rs1434536 | (A;G) | 1.29x increased breast cancer risk | | | |
| 1.3 | rs16847548 | (C;T) | 1.3x increased risk for sudden cardiac death in... | | | |
| 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 | rs501120 | (A;G) | 1.3x increased risk for heart disease | | Link | |
| 1.25 | rs748404 | (T;T) | Slightly increased risk (1.25) for lung cancer... | | 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 | rs12050604 | (A;A) | Slightly increased risk for lung cancer | | | |
| 1.2 | rs2076295 | (G;T) | One copy of the risk allele (G): slightly incre... | | | |
| 1.2 | rs2252586 | (A;G) | 1.2x higher risk for glioma development | | | |
| 1.2 | rs3131296 | (A;G) | 1.2x increased risk for schizophrenia | | 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 | rs419788 | (A;G) | 2.0x risk for lupus | Link | | |
| 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 | |
| 1.2 | rs9858542 | (A;G) | 1.1x risk Crohn's Disease | Link | Link | |
| 1.1 | rs11110912 | (C;C) | 1.3x high blood pressure risk | | | |
| 1.1 | rs11650494 | (A;G) | Slightly higher prostate cancer risk | | | |
| 1.1 | rs13387042 | (A;G) | 1.12x increased risk for breast cancer | | Link | |
| 1.1 | rs1344706 | (G;T) | 1.1x increased risk for schizophrenia | | Link | |
| 1.1 | rs2651899 | (A;G) | 1.1x higher risk for migraines | | | |
| 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 | rs6800901 | (T;T) | 1.3x multiple myeloma risk | | | |
| 1.1 | rs688034 | (C;T) | 1.1x risk higher risk for coronary artery disea... | | Link | |
| 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 | 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 | rs12752888 | (C;C) | Faster progression of mild cognitive impairment... | | | |
| 1 | rs2273697 | (A;G) | Adverse reaction more likely to carbamazepine i... | Link | Link | Link |
| 1 | rs3194051 | (A;A) | >1.1x risk of type-1 diabetes | Link | Link | Link |
| 1 | rs6166 | (G;G) | Females slightly more likely to be sterile | Link | Link | Link |
| 1 | rs6932590 | (C;T) | 1.1x increased risk for schizophrenia | | Link | |
| 1 | rs761100 | (G;G) | Higher risk for dyslexia | | | |
| 1 | rs987525 | (A;C) | 2.5x increased risk for cleft lip | | Link | |
| 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 | rs1495965 | (A;A) | 1.2x higher risk for spondylitis | | | |
| 0 | rs3761418 | (A;A) | 1.3x increased risk for depression | | | |
| 0 | rs3813929 | (C;C) | Possible weight gain if taking olanzapine | | Link | Link |
| 0 | rs4712653 | (T;T) | 2x increased risk for neuroblastoma | | | |
| 0 | rs6314 | (C;C) | Higher risk for RA | Link | Link | |
| 0 | rs7652331 | (T;T) | Somewhat higher risk for prostate cancer | | | |

3.3 Genosets (Multi-variant Phenotypes)

| Magnitude | Identifier | Summary |
|-----------|------------|--|
| 3.3 | gs162 | CYP2C9 Poor Metabolizers |
| 3.1 | gs191 | Problem metabolizing NSAIDs |
| 3 | gs241 | Lighter green: brown or hazel eye color |
| 2.6 | gs296 | Lower heart attack risk than average |
| 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 | gs173 | CYP2D6*10 |
| 2 | gs246 | APOE3/APOE3 |
| 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 ERS1176641 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/ERS1176641>

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.