

PGP-UK Genomics Report for uk48CE67

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 | 4950583 |
| Variants filtered out | 0 |
| Novel / existing variants | 498878 (10.1) / 4438094 (89.9) |
| Overlapped genes | 56750 |
| Overlapped transcripts | 67522 |
| Overlapped regulatory features | 166216 |

Table 1: Variant calling summary

There are several different types of genomic variants. The most common change is when one single building block of the DNA (called a nucleotide) is changed, called a single nucleotide variants (SNV). Other variant types include insertions, where the DNA in the individual is longer than the reference sequence due to the insertion of one or more nucleotides; and deletions, where a few nucleotides are missing compared to the reference sequence.

Some of these changes will have no effect on the protein, while some changes may alter the protein function to varying degrees. The PolyPhen analysis software attempts to quantify the effect each mutation will have on the protein function. This ranges from "benign" where no change to the protein function is expected, to "probably damaging" where it is predicted that the mutation will affect protein function. It is nevertheless important to note that what is "damaging" for the protein is not necessarily damaging for the individual.

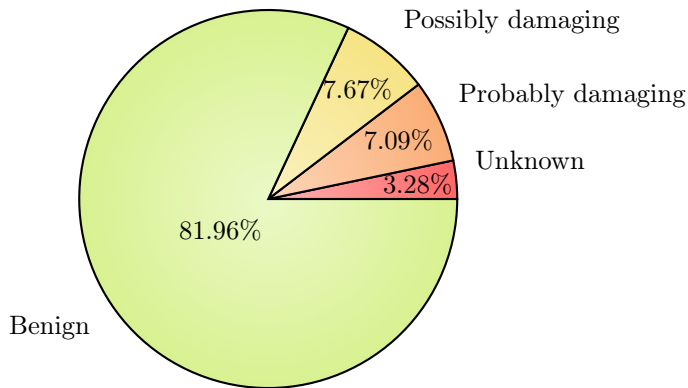


Figure 2: PolyPhen Summary

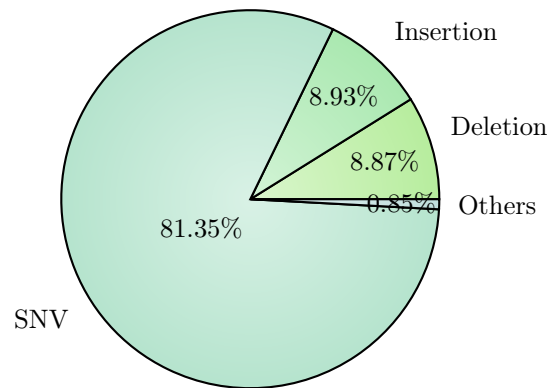


Figure 3: Variant Class

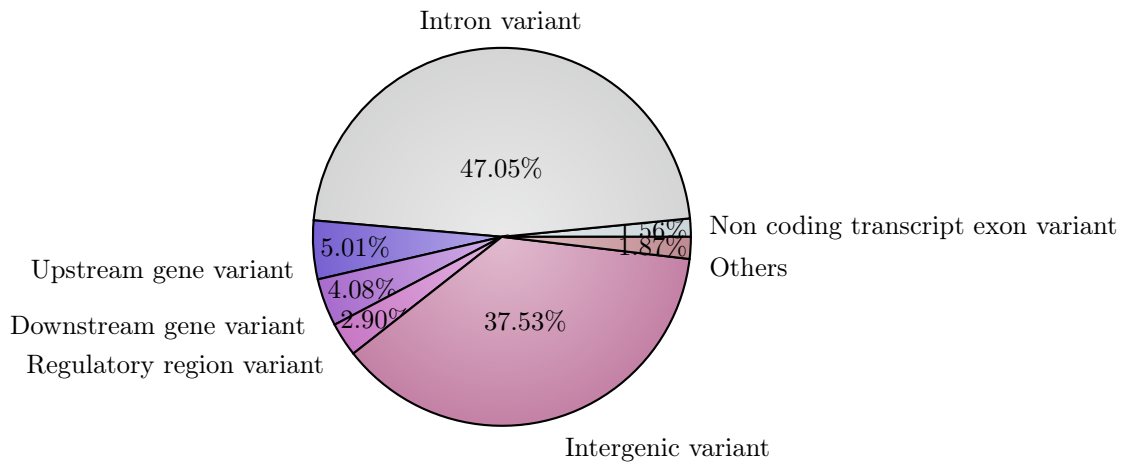


Figure 4: Consequence type

2 Ancestry

This plot shows the distribution of the genomes of different populations. Data from several studies which used whole genome sequencing was used to see the relationships between the genomes of the populations. It shows how closely related certain populations are genetically: Groups which cluster closely are more genetically similar than groups which are further apart. The black star symbol shows where this PGP-UK participant sits in relation to other populations, indicating their ancestry and their most closely related populations according to genetic sequence.

Based on the populations defined in the 1000 genomes project (1kGP), the ancestry composition for this individual is inferred to be 100.0 percent European [British in England and Scotland].

Please note that this analysis is limited by the populations available in the 1kGP data. If there are European subpopulations reported, and the ancestry of the participant does not correspond to any of the 1kGP populations, the closest 1kGP sampled subpopulation will be shown (even though it might be different from the participant's actual ancestry).

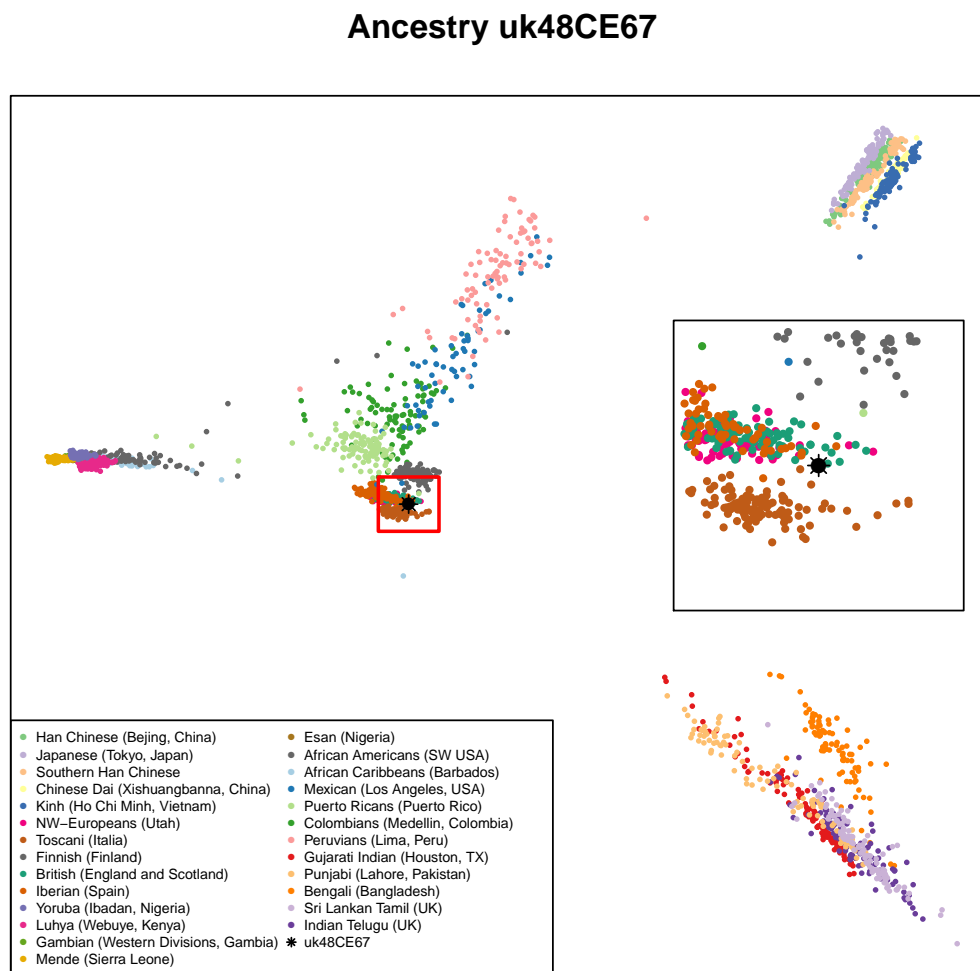


Figure 5: Ancestry Principal Component Analysis

3 Traits (based on SNPedia information)

Existing research has associated many variants with phenotypic traits, some of which can be perceived as beneficial while others appear to have a harmful effect. Some traits are complex and can be affected by several variants. It is likely that some of these would confer a higher risk while others a lower risk of trait manifestation. These can not be combined linearly to produce an actual risk of disease.

It is important to note that in most cases genomic data is probabilistic, not deterministic- i.e. having a genetic predisposition for a disease is not a diagnosis; rather, it shows an increased likelihood of developing that disease. Also, one person can have both potentially beneficial and harmful variants in the same gene, or associated with the same disease.

Some variants can also affect certain populations more, or will only affect a particular gender. For example, a variant for higher risk of endometriosis in the sequence of a male will not directly affect that person, but can be passed on to descendants.

While many traits are the result of a unique variant, many are the combination of several variants throughout the genome. In SNPedia, these are called *genosets*. These can integrate some of the information already present in the single variant tables, or be the combination of variants that have no phenotypic effect on their own, but contribute to a trait when together.

The variants in the following tables are sorted by magnitude. This is an subjective measure defined in SNPedia to highlight the perceived importance of the genotype described. At the moment this scale goes from 0 to 10. You can read more about it by visiting their explanatory [webpage](#).

As our knowledge grows, the interpretation of the effect of certain variants might change. Clicking on the links in the genome report tables will take you to websites containing more information about each variant.

3.1 Possibly Beneficial Traits

| Mag. | Identifier | Genotype | Summary | ExAC | GetEvidence | ClinVar |
|------|------------|----------|--|----------------------|----------------------|----------------------|
| 2.1 | rs5471154 | (A;C) | 0.47x decreased risk for AMD | | | Link |
| 2.1 | rs6897932 | (T;T) | 0.70x decreased risk for multiple sclerosis | Link | Link | 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 | rs12678919 | (A;G) | Associated with higher HDL cholesterol | | Link | |
| 2 | rs174537 | (T;T) | Lower LDL-C and total cholesterol | | | |
| 2 | rs2542052 | (C;C) | Better odds of living to 100 | | | |
| 2 | rs3750817 | (C;T) | 0.78x reduced risk for breast cancer | | | |
| 2 | rs3782179 | (C;T) | 3x lower odds of testicular cancer risk for men... | | | |
| 2 | rs3819331 | (T;T) | Lower risk of autism | Link | | |
| 2 | rs4149268 | (A;G) | Associated with higher HDL cholesterol | | Link | |
| 2 | rs6505162 | (A;C) | 0.58x decreased risk for esophageal cancer | Link | | |
| 2 | rs6511720 | (G;T) | Slightly lower odds of developing CHD. | | Link | 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 | rs187238 | (C;G) | Hypertension not a risk factor for sudden cardi... | | | |
| 1.8 | rs4714156 | (C;C) | <0.61x risk for restless legs | | | |
| 1.8 | rs9402571 | (G;G) | 0.85x decreased risk for type-2 diabetes | | | |
| 1.6 | rs1061170 | (T;T) | Lower risk for AMD: generally longer live than ... | Link | Link | Link |
| 1.6 | rs10801935 | (C;C) | 0.3x decreased risk of breast cancer | | | |
| 1.5 | rs1063192 | (C;T) | 0.71x reduced risk of myocardial infarction | | | |
| 1.5 | rs11136000 | (C;T) | 0.84x decreased risk for Alzheimer's disease | | Link | |
| 1.5 | rs11212617 | (A;C) | Somewhat increased likelihood of treatment succ... | | | Link |
| 1.5 | rs1165205 | (A;A) | 0.85x decreased gout risk | | Link | |

| Mag. | Identifier | Genotype | Summary | ExAC | GetEvidence | ClinVar |
|------|------------|----------|--|------|-------------|---------|
| 1.5 | rs16991615 | (A;G) | Slight increase (11 months) in avg age at menop... | Link | Link | |
| 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 | |
| 1.5 | rs5888 | (C;C) | Higher HDL cholesterol but lower risk for age-r... | Link | | |
| 1.3 | rs9306160 | (T;T) | 0.75x (reduced) risk for metastasis in LN-/ER+ ... | Link | Link | |
| 1.2 | rs11172113 | (C;C) | 0.8x lower risk for migraines | | | |
| 1.2 | rs4320932 | (A;G) | 0.87x decreased risk for ovarian cancer | | | |
| 1.2 | rs6048 | (G;G) | Slightly lower risk (10-20%) of deep vein throm... | Link | Link | Link |
| 1.1 | rs2293347 | (G;G) | Among NSCLC patients: better Gefitinib response... | Link | | Link |
| 1.1 | rs4988235 | (T;T) | Can digest milk | | | Link |
| 1 | rs182549 | (T;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 | rs2952768 | (C;T) | Slightly less drug dependence: decreased effect... | | | Link |
| 1 | rs800292 | (C;T) | 1% decreased risk of macular degeneration | Link | Link | Link |
| 1.0 | rs2283123 | (C;T) | Decreased risk of schizophrenia in limited stud... | | | |
| 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 | rs10427255 | (T;T) | Lowest odds of photic sneeze reflex | | | |
| 0 | rs1047781 | (A;A) | ABH blood group "Secretor" status if Japanese | Link | Link | Link |
| 0 | rs16990018 | (A;A) | PrP Codon 171 Asn - Non-pathogenic variant | Link | | Link |
| 0 | rs1799782 | (C;C) | Lower risk for skin cancer | Link | Link | |
| 0 | rs1799945 | (C;C) | Not a H63D hemochromatosis carrier. | Link | Link | Link |
| 0 | rs1800562 | (G;G) | Not a C282Y hemochromatosis carrier. | Link | Link | Link |
| 0 | rs28933385 | (G;G) | Prion protein Codon 200 (E) - Non pathogenic va... | | | Link |
| 0 | rs312481 | (C;C) | Better response to certain calcium channel bloc... | | | |
| 0 | rs5065 | (A;A) | 1.12x risk on diuretic; if hypertensive: better... | Link | Link | Link |
| 0 | 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 |
|------|------------|----------|--|------|-------------|---------|
| 4 | rs1333049 | (C;C) | 1.9x increased risk for coronary artery disease... | | Link | |
| 3.5 | rs875858 | (C;T) | Docetaxel sensitive? | | | |
| 3 | rs10897346 | (C;C) | If depressed: 2.6x more likely to not respond t... | | | |
| 3 | rs13266634 | (C;C) | Increased risk for type-2 diabetes | Link | Link | Link |
| 3 | rs2306402 | (C;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 | rs3892097 | (A;A) | CYP2D6 poor metabolizer; many associations rela... | Link | Link | Link |
| 3 | rs3903239 | (C;C) | Higher frequency of atrial fibrillation | | | |
| 3 | rs4244285 | (A;G) | Poorer metabolizer of several popular medicines... | Link | Link | Link |
| 2.7 | rs10830963 | (C;G) | Increased type-2 diabetes risk; higher gestatio... | | Link | |
| 2.6 | rs110419 | (A;A) | 2.6x increased risk for neuroblastoma | | | |
| 2.5 | rs1121980 | (C;T) | 1.67x risk for obesity | | Link | |
| 2.5 | rs1421085 | (C;T) | ~1.3x increased obesity risk | | Link | Link |
| 2.5 | rs16969968 | (A;G) | Slightly higher risk for nicotine dependence: l... | Link | Link | Link |
| 2.5 | rs1799971 | (A;G) | Stronger cravings for alcohol. if alcoholic: na... | Link | Link | Link |
| 2.5 | rs2254958 | (C;C) | 1.61x increased risk for Alzheimer's | | | |
| 2.5 | rs2943634 | (C;C) | Slightly higher risk of ischemic stroke | | Link | |
| 2.5 | rs3780374 | (A;G) | Substantially increased odds of developing V617... | | | |
| 2.5 | rs664143 | (C;T) | Higher risk for number of cancers | | | |
| 2.5 | rs7574865 | (G;T) | 1.3x risk of rheumatoid arthritis; 1.55x risk o... | | Link | Link |
| 2.5 | rs8034191 | (C;T) | 1.27x lung cancer risk | | Link | |
| 2.3 | rs7966230 | (C;G) | Slightly lower levels of plasma VWF | | | |
| 2.2 | rs2004640 | (G;T) | 1.4x increased risk for SLE | | Link | Link |
| 2.2 | rs2231137 | (G;G) | ~1.5-3x increased risk for ischemic stroke | Link | Link | Link |
| 2.1 | rs1050152 | (T;T) | 2.1x increased risk of Crohn's disease | Link | Link | Link |
| 2.1 | rs17070145 | (C;C) | Reduced memory abilities | | | Link |
| 2.1 | rs2270641 | (G;G) | 3.7x higher risk for schizophrenia | Link | Link | |
| 2.1 | rs2383207 | (G;G) | Increased risk for heart disease | | | |
| 2.1 | rs4444903 | (G;G) | 3.5x risk of hep-cancer in cirrhosis patients; ... | | | Link |
| 2.1 | rs5186 | (A;C) | ~1.4x increased risk of hypertension | Link | Link | Link |
| 2.1 | rs6457617 | (T;T) | 5.2x risk of rheumatoid arthritis | | Link | |
| 2.1 | rs646776 | (A;A) | 1.2x risk of coronary artery disease | | Link | |
| 2 | rs10086908 | (C;C) | 1.7x increased risk for prostate cancer | | | |
| 2 | rs10096097 | (G;G) | Increased Anorexia Nervosa risk | | | |
| 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 | rs10306114 | (A;G) | Higher risk of bleeding during coronary angiogr... | | | Link |
| 2 | rs10488631 | (C;T) | 2x increased risk of developing SLE; 1.6x incre... | | Link | |
| 2 | rs10513789 | (G;T) | Increased risk of Parkinson's disease | | | |
| 2 | rs1051730 | (C;T) | 1.3x increased risk of lung cancer | Link | Link | Link |
| 2 | 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 | rs10871777 | (A;G) | Adults likely to be 0.22 BMI units higher | | | |
| 2 | rs10984447 | (A;A) | >1.17x increased risk for multiple sclerosis | | Link | |
| 2 | rs11190870 | (C;T) | Possibly increased risk of scoliosis | | | |
| 2 | rs11229030 | (C;C) | Higher odds of Crohn's disease | | | |
| 2 | rs1160312 | (A;A) | 1.6x increased risk of Male Pattern Baldness. | | Link | |
| 2 | rs1169300 | (A;A) | ~2x increased lung cancer risk | | | |
| 2 | rs11983225 | (T;T) | 7x less likely to respond to certain antidepres... | | Link | |
| 2 | rs1219648 | (A;G) | 1.20x risk for breast cancer | | Link | |
| 2 | rs12567232 | (A;G) | Increased risk for Crohn's Disease | | Link | |
| 2 | rs12770228 | (A;A) | 2x increased risk for meningioma | | | |

| Mag. | Identifier | Genotype | Summary | ExAC | GetEvidence | ClinVar |
|------|------------|----------|--|------|-------------|---------|
| 2 | rs1585215 | (A;G) | 2x increased risk for Hodgkin lymphoma | | | |
| 2 | rs16942 | (A;G) | Very slightly increased breast cancer risk | Link | Link | Link |
| 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 | 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 | rs1800896 | (A;G) | 1.6x increased prostate cancer risk | | | |
| 2 | rs1994090 | (G;G) | Increased risk of developing Parkinson's Diseas... | | Link | |
| 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 | 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 | rs241448 | (C;T) | 1.51x increased risk for Alzheimer's | Link | | Link |
| 2 | rs2420946 | (C;T) | 1.20x risk for breast cancer | | | |
| 2 | rs2464196 | (T;T) | ~2x increased lung cancer risk | Link | Link | Link |
| 2 | rs25487 | (G;G) | 2x higher risk for skin cancer; possibly other ... | Link | Link | Link |
| 2 | rs2707466 | (G;G) | Weaker bones | Link | Link | |
| 2 | rs2908004 | (C;C) | Weaker bones | Link | Link | |
| 2 | rs3212227 | (A;C) | Significantly increased risk of developing cerv... | | | |
| 2 | rs358806 | (C;C) | 1.78x increased risk of developing Type-2 diabe... | | Link | |
| 2 | rs3738919 | (A;C) | 1.94x risk of developing rheumatoid arthritis | | | |
| 2 | rs3746444 | (C;T) | ~1.2x increased risk for cancer | Link | | |
| 2 | rs3775948 | (G;G) | Slightly higher risk for gout | | | |
| 2 | rs3842787 | (C;T) | Higher risk of bleeding during coronary angiogr... | Link | Link | |
| 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 | rs4792311 | (A;G) | Increased risk of prostate cancer | Link | Link | Link |
| 2 | rs4825476 | (G;G) | 1.9x higher risk of suicidal thoughts when taki... | | Link | |
| 2 | rs493258 | (G;G) | 1.15x risk of Age Related Macular Degeneration | | | |
| 2 | rs5174 | (A;G) | 1.3x increased risk for heart disease | Link | Link | Link |
| 2 | rs5759167 | (T;T) | Higher prostate cancer risk | | Link | |
| 2 | rs6441286 | (G;T) | 1.54x chance of developing primary biliary cirr... | | Link | |
| 2 | rs6807362 | (C;C) | Increased autism risk | Link | Link | |
| 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 | rs699 | (C;T) | Increased risk of hypertension | Link | Link | Link |
| 2 | rs7216389 | (T;T) | 1.5x increased risk for Childhood Asthma. | | Link | |
| 2 | rs744373 | (C;T) | 1.17x risk of Alzheimer's | | | |
| 2 | rs7639618 | (T;T) | 1.45x increased osteoarthritis risk | Link | | |
| 2 | rs7774434 | (C;C) | Increased risk of developing primary biliary ci... | | | |
| 2 | rs7923837 | (G;G) | 3.2x risk for T2D | | | |
| 2 | rs7961152 | (A;C) | 1.2x higher risk for hypertension | | | |
| 2 | rs828907 | (G;T) | Slightly increased risk of bladder cancer and 2... | | | |
| 2 | rs854560 | (A;A) | Higher risk for heart disease: diabetic retinop... | Link | Link | Link |
| 2 | rs9652490 | (A;A) | ~2x increased risk for Parkinson's disease: and... | | Link | |
| 2.0 | rs2305795 | (A;A) | 1.64x higher risk of narcolepsy compared to (G;... | | | Link |
| 2.0 | rs4911414 | (G;T) | 2-4x higher risk of sun sensitivity if part of ... | | Link | |
| 1.8 | rs1136287 | (C;T) | 1.5x increased risk of wet ARMD in a Taiwanese ... | 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 | |
| 1.8 | rs6700125 | (C;T) | 1.2x increased risk for ALS | | | |

| Mag. | Identifier | Genotype | Summary | ExAC | GetEvidence | ClinVar |
|------|------------|----------|--|------|-------------|---------|
| 1.7 | rs2024513 | (A;A) | 1.7x higher risk for schizophrenia (among Han C... | | | |
| 1.6 | rs1537415 | (C;G) | 1.6x increased risk for periodontitis | | Link | |
| 1.6 | rs3764880 | (A;A) | 1.2 - 1.8x increased tuberculosis risk | Link | Link | |
| 1.6 | rs4959039 | (G;G) | 1.6x higher risk for multiple sclerosis | | | |
| 1.5 | rs10492519 | (A;G) | Slightly increased risk of developing prostate ... | | | |
| 1.5 | rs10980705 | (C;T) | 2.3x increased risk for knee osteoarthritis | | | |
| 1.5 | rs11171739 | (C;T) | 1.34x risk of developing Type-1 diabetes | | Link | |
| 1.5 | rs13149290 | (C;C) | Slightly increased risk of developing prostate ... | | | |
| 1.5 | rs140701 | (A;G) | 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 | rs1801020 | (C;T) | 1.31x increased risk of heart disease | Link | | Link |
| 1.5 | rs1801274 | (C;T) | Complex; generally greater risk for cancer prog... | Link | Link | Link |
| 1.5 | 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 | rs27388 | (A;G) | Slightly increased risk of developing schizophr... | | | |
| 1.5 | rs2881766 | (T;T) | Slightly increased risk for pregnancy-induced h... | | | |
| 1.5 | rs3087243 | (G;G) | Increased risk for autoimmune diseases | | Link | |
| 1.5 | rs3745516 | (A;G) | Slightly increased risk of developing primary b... | | | |
| 1.5 | rs3825776 | (A;G) | 1.3x increased risk for ALS | | Link | |
| 1.5 | rs393152 | (A;A) | Increased risk of both PD and AD | Link | Link | |
| 1.5 | rs401681 | (C;T) | ~1.2x increased risk for several types of cance... | | Link | |
| 1.5 | rs4027132 | (A;G) | 1.39x increased risk of developing bipolar diso... | | | |
| 1.5 | rs4464148 | (C;T) | 1.10x increased risk for colorectal cancer | | | |
| 1.5 | rs464049 | (C;T) | Increased risk of schizophrenia in limited stud... | | | |
| 1.5 | rs5746059 | (A;A) | Slightly higher fat mass | | | |
| 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 | 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 | rs699473 | (C;T) | ~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 | rs7850258 | (G;G) | Slightly higher odds of developing primary hypo... | | | |
| 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 | |
| 1.4 | rs1126497 | (T;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 | rs2230201 | (A;G) | 1.4x risk of lupus | Link | | |
| 1.4 | rs3131296 | (G;G) | 1.4x increased risk for schizophrenia | | Link | |
| 1.4 | rs3184504 | (C;T) | Slightly increased risk for celiac disease | Link | Link | |
| 1.4 | rs4795067 | (G;G) | Slight increase in risk for psoriatic arthritis... | | | |
| 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 | rs1042713 | (A;G) | 1.3x increased risk that pediatric inhaler use ... | Link | Link | Link |
| 1.3 | rs1047286 | (C;T) | 1.3x increased risk for age-related macular deg... | Link | Link | Link |
| 1.3 | rs10947262 | (C;C) | 1.3x increased risk for osteoarthritis | | | |
| 1.3 | rs1746048 | (C;C) | 1.03 increased risk for coronary heart disease | | Link | |
| 1.3 | rs2059693 | (C;T) | 1.3x increased risk for testicular cancer | | | |
| 1.3 | rs2295490 | (G;G) | 1.32x increased risk of early-onset type-2 diab... | Link | Link | |

| Mag. | Identifier | Genotype | Summary | ExAC | GetEvidence | ClinVar |
|------|------------|----------|--|------|-------------|---------|
| 1.3 | rs2736100 | (G;T) | 1.3x higher risk for glioma development: 2.1x r... | | Link | |
| 1.25 | rs748404 | (T;T) | Slightly increased risk (1.25) for lung cancer... | | Link | |
| 1.2 | rs1344706 | (T;T) | 1.2x increased risk for schizophrenia | | 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 | rs2076295 | (G;T) | One copy of the risk allele (G): slightly incre... | | | |
| 1.2 | rs2814707 | (A;G) | 1.2x increased risk for ALS | | Link | |
| 1.2 | rs3849942 | (A;G) | 1.2x increased risk for ALS | | Link | |
| 1.2 | rs419788 | (A;G) | 2.0x risk for lupus | Link | | |
| 1.2 | rs4977756 | (A;G) | 1.39x higher risk for glioma development | | Link | |
| 1.2 | rs6897876 | (C;C) | Slight increase in testicular cancer risk for m... | | | |
| 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 | |
| 1.1 | rs11037909 | (C;T) | 1.27x type II diabetes risk | Link | | |
| 1.1 | rs11110912 | (C;C) | 1.3x high blood pressure risk | | | |
| 1.1 | rs11650354 | (C;T) | Possible risk for allergic asthma | Link | | |
| 1.1 | rs1800450 | (A;G) | Carrier of mannose binding deficiency but of lo... | 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 | 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 | rs3740878 | (A;G) | 1.26x type II diabetes risk | Link | | Link |
| 1.1 | rs3818361 | (C;T) | 1.15x increased risk for late-onset Alzheimer's... | | | |
| 1.1 | rs7171755 | (A;G) | Very slight decrease in cortical thickness and ... | | | |
| 1.1 | rs7412 | (C;C) | More likely to gain weight if taking olanzapine... | Link | Link | Link |
| 1.1 | rs925391 | (C;C) | More likely to go bald; common | | | |
| 1.09 | rs12050604 | (A;C) | Very slightly increased risk for lung cancer | | | |
| 1.05 | rs2291834 | (C;T) | Very slightly higher risk for myocardial infarc... | | | |
| 1 | rs10761659 | (A;G) | 1.2x risk of Crohn's disease | | Link | |
| 1 | rs2273697 | (A;G) | Adverse reaction more likely to carbamazepine i... | Link | Link | Link |
| 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 | (C;T) | 1.1x increased risk for schizophrenia | | Link | |
| 1 | rs6974491 | (A;G) | Higher risk of coeliac and/or inflammatory bowe... | | | |
| 1 | rs761100 | (G;G) | Higher risk for dyslexia | | | |
| 1.0 | rs11246226 | (A;A) | Increased risk of schizophrenia in limited stud... | | Link | |
| 0 | rs1128503 | (T;T) | Likely to require more methadone during heroin ... | Link | Link | Link |
| 0 | rs4293393 | (T;T) | 1.25x Increased Risk of CKD for T allele in ... | | | |
| 0 | rs440446 | (G;G) | Increased risk in men for biliary conditions | Link | | |
| 0 | rs6314 | (C;C) | Higher risk for RA | Link | Link | |
| 0 | rs7787082 | (G;G) | 7x less likely to respond to certain antidepres... | | Link | |

3.3 Genosets (Multi-variant Phenotypes)

| Magnitude | Identifier | Summary |
|-----------|------------|--|
| 6 | gs216 | 2 copies of the APO- ϵ 4 <i>Allele</i> |
| 3.1 | gs191 | Problem metabolizing NSAIDs |
| 3 | gs127 | Intermediate warfarin metabolizer |
| 2.6 | gs296 | Lower heart attack risk than average |
| 2.5 | gs102 | ALS risk |
| 2.5 | gs155 | CYP3A5 non-expressor |
| 2.5 | gs161 | CYP2C9 Intermediate Metabolizers |
| 2.5 | gs281 | Part of the 88% of the population claimed not t... |
| 2.5 | gs298 | Increased surveillance for colorectal cancer re... |
| 2 | gs101 | Probably able to digest milk |
| 2 | gs154 | NAT2 Slow metabolizer |
| 2 | gs249 | Parkinson's Disease Risk |
| 1.2 | gs184 | Able to taste bitterness. |
| 0 | gs158 | CYP1A2 normal metabolizer |

4 Raw Data

The raw data used to create this report has been assigned the identifier ERS1176627 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/ERS1176627>

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.