# PGP-UK Genomics Report for uk79373B

## 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.

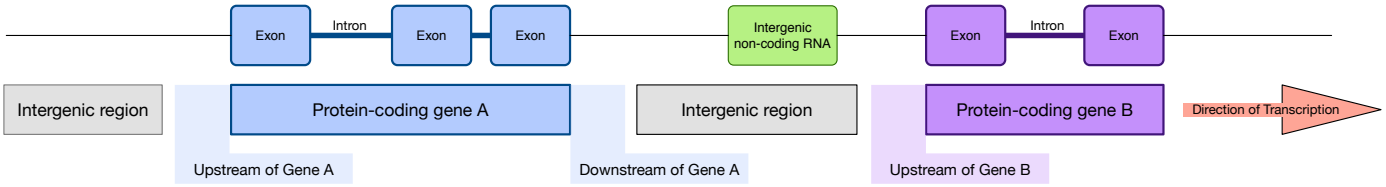


Figure 1: Diagram of gene structure indicating locations of potential variants

| Feature | Count |
|---|---|
| Lines of input read | 4956903 |
| Variants filtered out | 0 |
| Novel / existing variants | 500244 (10.1) / 4443860 (89.9) |
| Overlapped genes | 56736 |
| Overlapped transcripts | 67518 |
| Overlapped regulatory features | 166411 |

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.

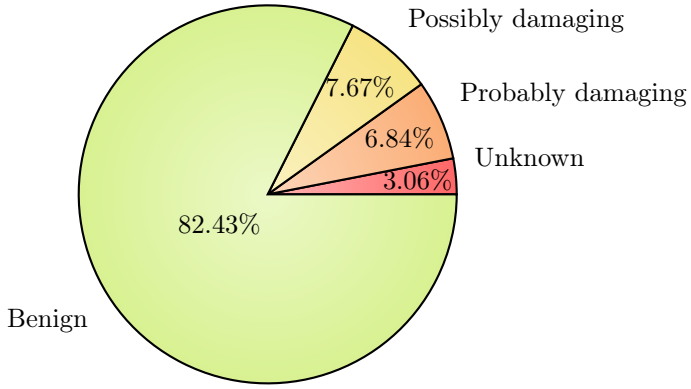


Figure 2: PolyPhen Summary

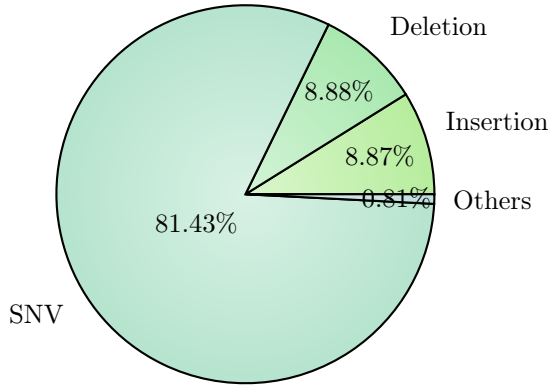


Figure 3: Variant Class

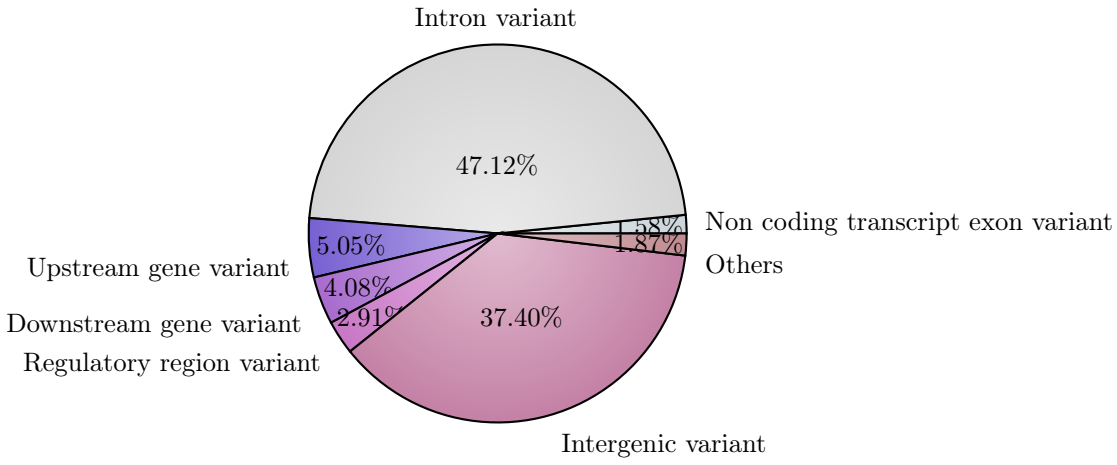


Figure 4: Consequence type

## 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).

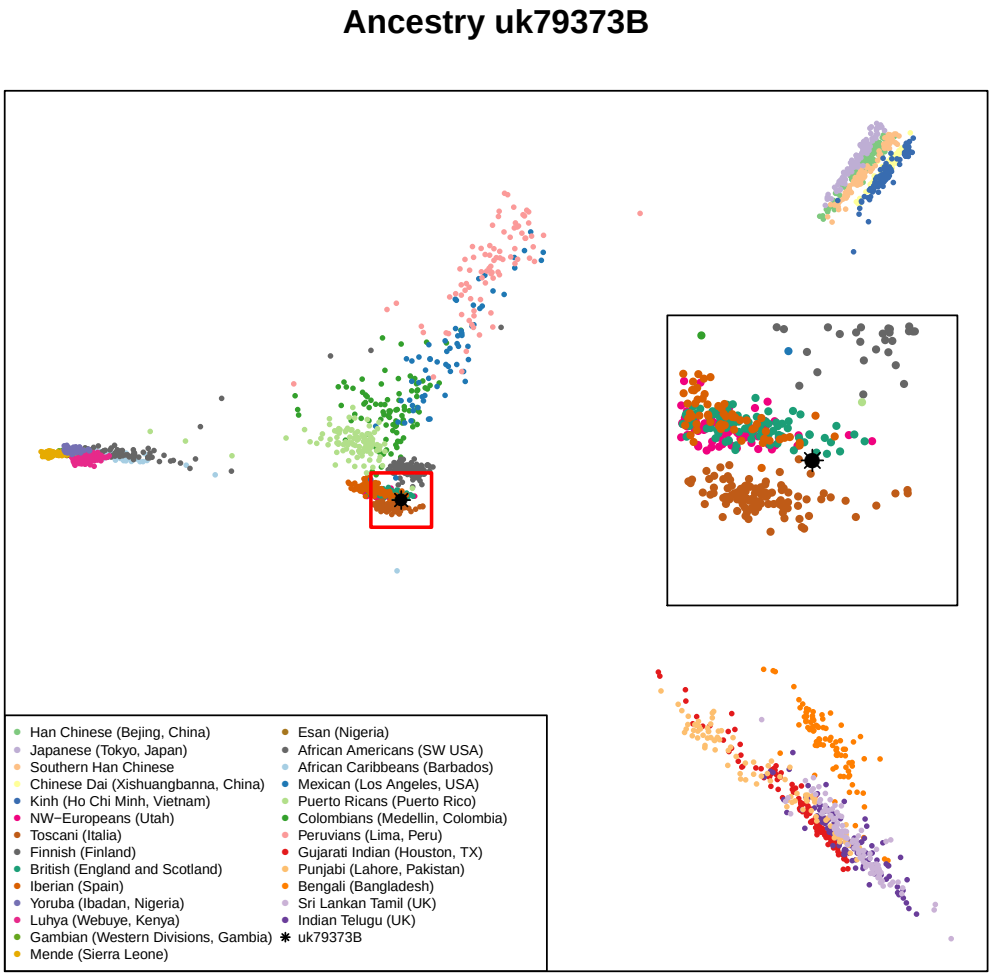


Figure 5: Ancestry Principal Component Analysis

# 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 |
|---|---|---|---|---|---|---|
| 3 | rs925391 | (T;T) | Unlikely to go bald | | | |
| 2.1 | rs2511989 | (A;G) | 0.63x decreased age-related macular degeneratio... | | Link | |
| 2 | rs1012053 | (A;C) | 0.625x reduced risk of Bipolar Disorder. | | Link | |
| 2 | rs10503669 | (A;C) | Associated with higher HDL cholesterol | | 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 | rs2073963 | (T;T) | Reduced risk of baldness | | | |
| 2 | rs2235015 | (G;T) | Somewhat more likely to respond to certain anti... | Link | Link | |
| 2 | rs2243250 | (C;T) | 0.6x decreased risk for myocardial infarction i... | | | |
| 2 | rs261332 | (A;A) | Associated with higher HDL cholesterol | | | |
| 2 | rs3736309 | (A;G) | 0.44x decreased risk for chronic obstructive pu... | | | |
| 2 | rs3738579 | (C;T) | 0.5x decreased risk for cervical cancer: HNSCC:... | | | |
| 2 | rs3764261 | (G;T) | Associated with higher HDL cholesterol | | Link | Link |
| 2 | rs3782179 | (C;T) | 3x lower odds of testicular cancer risk for men... | | | |
| 2 | rs3819331 | (T;T) | Lower risk of autism | Link | | |
| 2 | rs3914132 | (C;T) | Lower otosclerosis risk | | Link | |
| 2 | rs4149268 | (A;G) | Associated with higher HDL cholesterol | | Link | |
| 2 | rs4307059 | (C;C) | Reduced Autism risk | | Link | |
| 2 | rs6495446 | (T;T) | 0.64x reduced risk for chronic kidney disease | | | |
| 2 | rs6505162 | (A;C) | 0.58x decreased risk for esophageal cancer | Link | | |
| 2 | rs6807362 | (G;G) | Decreased autism risk | Link | Link | |
| 2 | rs7776725 | (T;T) | Stronger bones | | Link | |
| 2 | rs801114 | (T;T) | 0.78x decreased Basal Cell Carcinoma risk. | | Link | |
| 1.8 | rs1128535 | (A;G) | 0.77x risk for Crohn's disease | | | |
| 1.8 | rs1800588 | (T;T) | Higher HDL-C levels | Link | Link | |

| Mag. | Identifier | Genotype | Summary | ExAC | GetEvidence | ClinVar |
|---|---|---|---|---|---|---|
| 1.8 | rs187238 | (C;G) | Hypertension not a risk factor for sudden cardi... | | | |
| 1.8 | rs4714156 | (C;C) | <0.61x risk for restless legs | | | |
| 1.5 | rs1026732 | (A;G) | 0.70x risk for restless legs | | Link | |
| 1.5 | rs11136000 | (T;T) | 0.84x decreased risk for Alzheimer's disease | | 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 | rs4149274 | (C;T) | Associated with higher HDL (good) cholesterol | | | |
| 1.5 | rs4489954 | (G;T) | 0.69x risk risk of developing restless legs syn... | | Link | |
| 1.5 | rs464049 | (C;C) | Decreased risk of schizophrenia in limited stud... | | | |
| 1.5 | rs4939883 | (C;C) | Associated with higher HDL cholesterol | | Link | |
| 1.5 | rs5888 | (C;C) | Higher HDL cholesterol but lower risk for age-r... | Link | | |
| 1.5 | rs5968255 | (C;C) | Slower AIDS progression (8 years) | | | |
| 1.5 | rs729302 | (A;C) | 0.89x decreased risk of developing rheumatoid a... | | | |
| 1.2 | rs9306160 | (C;T) | 0.75x (reduced) risk for metastasis in LN-/ER+ ... | Link | Link | |
| 1.1 | rs11172113 | (C;T) | 0.9x lower risk for migraines | | | |
| 1.1 | rs2293347 | (G;G) | Among NSCLC patients: better Gefitinib response... | Link | | Link |
| 1.1 | rs4988235 | (T;T) | Can digest milk | | | Link |
| 1 | rs182549 | (T;T) | Can digest milk. | | | Link |
| 1 | rs2952768 | (C;T) | Slightly less drug dependence: decreased effect... | | | Link |
| 1 | rs7850258 | (A;G) | Typical odds of developing primary hypothyroidi... | | | |
| 1.0 | rs11246226 | (C;C) | Decreased risk of schizophrenia in limited stud... | | Link | |
| 0.1 | rs891512 | (G;G) | Lower blood pressure than those with an A allel... | Link | | |
| 0 | rs1047781 | (A;A) | ABH blood group "Secretor" status if Japanese | Link | Link | Link |
| 0 | rs1126809 | (A;G) | Slight increase in skin cancer risk | Link | Link | Link |
| 0 | rs12252 | (T;T) | More resistant to influenza | Link | | Link |
| 0 | rs16947 | (A;A) | Homozygous for CYP2D6 variants (non-CYP2D6*1) | Link | Link | Link |
| 0 | rs16990018 | (A;A) | PrP Codon 171 Asn - Non-pathogenic variant | Link | | Link |
| 0 | rs17244841 | (A;A) | More responsive to statin treatment | | Link | Link |
| 0 | rs1799782 | (C;C) | Lower risk for skin cancer | Link | Link | |
| 0 | rs1800562 | (G;G) | Not a C282Y hemochromatosis carrier. | Link | Link | 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 | |
| 0 | rs9951307 | (A;G) | 0.10 decreased risk for brain edema after a str... | | | |

## 3.2 Possibly Harmful Traits

| Mag. | Identifier | Genotype | Summary | ExAC | GetEvidence | ClinVar |
|---|---|---|---|---|---|---|
| 4 | rs1333049 | (C;C) | 1.9x increased risk for coronary artery disease... | | Link | |
| 3 | rs10897346 | (C;C) | If depressed: 2.6x more likely to not respond t... | | | |
| 3 | rs16969968 | (A;A) | Higher risk for nicotine dependence: lower risk... | Link | Link | Link |
| 3 | rs1805008 | (T;T) | ~7-10x higher likelihood of red hair; higher ri... | Link | | Link |
| 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 | rs3903239 | (C;C) | Higher frequency of atrial fibrillation | | | |
| 3 | rs6920220 | (A;G) | 1.2x risk Rheumatoid Arthritis | | Link | |
| 3 | rs7754840 | (C;C) | 1.3x increased risk for type-2 diabetes | | Link | |
| 2.9 | rs16901979 | (A;A) | 1.5x increased risk for prostate cancer | | 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 | | | |
| 2.6 | rs8034191 | (C;C) | 1.80x lung cancer risk; decreased response to a... | | Link | |
| 2.5 | rs10490924 | (G;T) | 2.7x risk for age related macular degeneration | Link | Link | Link |
| 2.5 | rs1051730 | (T;T) | 1.8x increased risk of lung cancer; reduced res... | Link | Link | Link |
| 2.5 | rs1121980 | (C;T) | 1.67x risk for obesity | | Link | |
| 2.5 | rs13266634 | (C;T) | Increased risk for type-2 diabetes | Link | Link | Link |
| 2.5 | rs1421085 | (C;T) | ~1.3x increased obesity risk | | Link | Link |
| 2.5 | rs17696736 | (G;G) | 1.94x risk of type-1 diabetes | | Link | |
| 2.5 | rs2943634 | (C;C) | Slightly higher risk of ischemic stroke | | Link | |
| 2.5 | rs339331 | (T;T) | Prostate cancer risk | | | |
| 2.5 | rs7574865 | (G;T) | 1.3x risk of rheumatoid arthritis; 1.55x risk o... | | Link | Link |
| 2.3 | rs1859962 | (G;G) | 1.28x increased risk for prostate cancer | | 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 |
| 2.2 | rs944289 | (T;T) | 1.69x increased thyroid cancer risk | | Link | |
| 2.1 | rs10427255 | (C;C) | Highest odds of photic sneeze reflex | | | |
| 2.1 | rs10811661 | (T;T) | 1.2x increased risk for type-2 diabetes | | Link | |
| 2.1 | rs1219648 | (G;G) | 1.64x risk for breast cancer | | Link | |
| 2.1 | rs1329428 | (G;G) | 2x increased risk for macular degeneration | | | |
| 2.1 | rs17070145 | (C;C) | Reduced memory abilities | | | Link |
| 2.1 | rs17077540 | (A;G) | 1.6x major depressive disorder risk | | | |
| 2.1 | rs17563 | (C;C) | Risk for otosclerosis | Link | Link | Link |
| 2.1 | rs2231142 | (A;C) | 1.74x increased gout risk; gefinitib takers 4x ... | Link | Link | Link |
| 2.1 | rs2254958 | (C;T) | 1.24x increased risk for Alzheimer's | | | |
| 2.1 | rs2294008 | (T;T) | Increased risk of gastric and bladder cancer | Link | Link | |
| 2.1 | rs2383207 | (G;G) | Increased risk for heart disease | | | |
| 2.1 | rs7837688 | (G;T) | 1.7x increased risk for prostate cancer | | | |
| 2 | rs10086908 | (C;T) | 1.7x increased risk for prostate cancer | | | |
| 2 | rs10090154 | (C;T) | 1.4x increased risk for prostate cancer | | | |
| 2 | rs1024611 | (C;T) | Increased risk of exercise induced ischemia | | | Link |
| 2 | rs10248420 | (A;A) | 7x less likely to respond to certain antidepres... | | Link | |
| 2 | 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 | rs10757272 | (T;T) | 1.54x increased risk for Coronary artery diseas... | | | |
| 2 | rs10984447 | (A;A) | >1.17x increased risk for multiple sclerosis | | Link | |
| 2 | rs11045585 | (A;G) | 63% chance (higher than average) of docetaxel-i... | | Link | |
| 2 | rs11190870 | (C;T) | Possibly increased risk of scoliosis | | | |
| 2 | rs1160312 | (A;A) | 1.6x increased risk of Male Pattern Baldness. | | Link | |
| 2 | rs11983225 | (T;T) | 7x less likely to respond to certain antidepres... | | Link | |
| 2 | rs12037606 | (A;A) | 1.52x risk of developing Crohn's disease | | | |

| Mag. | Identifier | Genotype | Summary | ExAC | GetEvidence | ClinVar |
|---|---|---|---|---|---|---|
| 2 | rs12469063 | (G;G) | Increased risk of developing restless legs synd... | | | |
| 2 | rs12567232 | (A;G) | Increased risk for Crohn's Disease | | Link | |
| 2 | rs1265181 | (C;G) | Increased risk for psoriasis | | Link | |
| 2 | rs1360780 | (C;T) | 1.3x increased risk for depression | | Link | |
| 2 | rs16942 | (G;G) | Very slightly increased breast cancer risk | Link | Link | Link |
| 2 | rs16944 | (G;G) | Increased risk of mental disorders | | Link | |
| 2 | rs17001266 | (-;C) | 1.58x increased risk for schizophrenia in males... | | | |
| 2 | rs17228212 | (C;T) | 1.26x increased risk for heart disease | | Link | |
| 2 | rs1734791 | (A;A) | 1.4x increased risk for lupus | | | |
| 2 | rs1800629 | (A;G) | Complex; generally higher risk for certain dise... | Link | Link | Link |
| 2 | rs2156921 | (A;G) | 1.29x increased risk for depression | | | |
| 2 | rs2201841 | (C;T) | 1.5x increased risk for Crohn's disease; 2x inc... | | Link | |
| 2 | rs2230199 | (C;G) | 1.6x+ risk of ARMD | Link | Link | Link |
| 2 | rs2230201 | (G;G) | >1.4x risk of lupus | Link | | |
| 2 | 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 | 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 | rs27388 | (A;A) | Increased risk of developing schizophrenia | | | |
| 2 | rs3025039 | (C;T) | 2.6x increased risk for ARMD in a Taiwanese pop... | | | |
| 2 | rs351855 | (C;T) | 1.2x increased risk for prostate cancer | Link | Link | Link |
| 2 | rs358806 | (C;C) | 1.78x increased risk of developing Type-2 diabe... | | Link | |
| 2 | rs3738919 | (A;C) | 1.94x risk of developing rheumatoid arthritis | | | |
| 2 | rs3775948 | (G;G) | Slightly higher risk for gout | | | |
| 2 | rs4027132 | (A;A) | 1.51x increased risk of developing bipolar diso... | | | |
| 2 | rs4148739 | (A;A) | 7x less likely to respond to certain antidepres... | | Link | |
| 2 | rs4242382 | (A;G) | 1.7x increased risk for prostate cancer | | Link | |
| 2 | rs4444903 | (A;G) | 3.5x risk of hep-cancer in cirrhosis patients; ... | | | Link |
| 2 | rs4464148 | (C;C) | 1.35x increased risk for colorectal cancer | | | |
| 2 | rs4633 | (T;T) | Higher risk for endometrial 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 | rs4968451 | (A;C) | 1.61x increased risk for meningioma | | | |
| 2 | rs5174 | (A;G) | 1.3x increased risk for heart disease | Link | Link | Link |
| 2 | rs520354 | (A;G) | Increased risk in men for biliary conditions | | | |
| 2 | rs629242 | (C;T) | Somewhat higher risk for prostate cancer | | | |
| 2 | rs638405 | (G;G) | 2x increased ALZ risk in ApoE4 carriers | Link | | |
| 2 | rs6441286 | (G;T) | 1.54x chance of developing primary biliary cirr... | | Link | |
| 2 | rs6457617 | (C;T) | 2.3x risk of rheumatoid arthritis | | 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 | rs6897932 | (C;C) | 1.08x increased risk for multiple sclerosis | Link | Link | Link |
| 2 | rs6908425 | (C;C) | 1.95x increased risk of developing Crohn's dise... | | Link | |
| 2 | rs6997709 | (G;T) | 1.2x higher risk for hypertension | | | |
| 2 | rs699 | (C;T) | Increased risk of hypertension | Link | Link | Link |
| 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;T) | 1.17x risk of Alzheimer's | | | |
| 2 | rs7794745 | (T;T) | Slightly increased risk for autism | | Link | Link |
| 2 | rs7961152 | (A;C) | 1.2x higher risk for hypertension | | | |
| 2 | rs800292 | (C;C) | 5% higher risk of Age related macular degenerat... | Link | Link | Link |
| 2 | rs828907 | (T;T) | Increased risk of bladder cancer and 2x risk of... | | | |

| Mag. | Identifier | Genotype | Summary | ExAC | GetEvidence | ClinVar |
|---|---|---|---|---|---|---|
| 2 | rs854560 | (A;T) | Higher risk for heart disease: diabetic retinop... | Link | Link | Link |
| 2 | rs9525638 | (T;T) | Weaker bones | | | |
| 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 | rs4911414 | (G;T) | 2-4x higher risk of sun sensitivity if part of ... | | Link | |
| 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 | |
| 1.8 | 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 | | | |
| 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 | 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 | rs1169300 | (A;G) | ~1.5x increased lung cancer risk | | | |
| 1.5 | rs12431733 | (C;T) | Slightly increased risk of developing Parkinson... | | Link | |
| 1.5 | rs12498742 | (A;A) | 1.25 increased risk for gout | | | |
| 1.5 | rs13149290 | (C;T) | 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 | rs17115100 | (G;T) | Slightly increased risk of developing Parkinson... | Link | Link | |
| 1.5 | rs1801274 | (C;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 | rs2007153 | (G;G) | Increased risk of schizophrenia in limited stud... | | | |
| 1.5 | rs2076295 | (G;G) | Slightly increased risk for pulmonary fibrosis ... | | | |
| 1.5 | rs2177369 | (C;C) | 1.5x increased risk for Alzheimer's disease | | | |
| 1.5 | rs2240340 | (A;A) | Slightly increased (1.5x) risk for RA | Link | | |
| 1.5 | rs2272127 | (C;C) | Associated with herpes and schizophrenia | | | |
| 1.5 | rs2280714 | (A;A) | 1.4x increased risk of SLE | | | |
| 1.5 | rs2464196 | (C;T) | ~1.5x increased lung cancer risk | Link | Link | Link |
| 1.5 | rs2697962 | (A;G) | Slightly increased risk of developing Parkinson... | | | |
| 1.5 | rs2736990 | (C;T) | Slightly increased risk of developing Parkinson... | | Link | |
| 1.5 | rs2881766 | (T;T) | Slightly increased risk for pregnancy-induced h... | | | |
| 1.5 | rs3087243 | (A;G) | Increased risk for auto-immune diseases | | Link | |
| 1.5 | rs3212227 | (A;A) | 1.43x increased risk of developing psoriasis an... | | | |
| 1.5 | rs393152 | (A;A) | Increased risk of both PD and AD | Link | Link | |
| 1.5 | rs4506565 | (A;T) | 1.4x increased risk for type-2 diabetes | | Link | |
| 1.5 | rs4585 | (T;T) | Slightly poorer (0.75x) response to metformin i... | | | |
| 1.5 | rs4785763 | (A;A) | 2x higher risk for melanoma | | Link | |
| 1.5 | rs486907 | (A;G) | 1.5x increased prostate cancer risk | Link | Link | Link |
| 1.5 | rs5219 | (C;T) | 1.3x increased risk for type-2 diabetes | Link | Link | Link |
| 1.5 | rs5746059 | (A;A) | Slightly higher fat mass | | | |
| 1.5 | 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 | rs7341475 | (G;G) | 1.58x increased schizophrenia risk for women | | Link | |
| 1.5 | rs7454108 | (C;T) | Single HLA-DQ8 haplotype | | | |
| 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 | rs807701 | (C;T) | Slightly increased dyslexia risk | | | |
| 1.5 | rs872071 | (G;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 | (C;T) | 1.4x increased risk for breast cancer | Link | Link | Link |
| 1.4 | rs1447295 | (A;C) | 1.4x increased risk of prostate cancer | | Link | |
| 1.4 | rs1545843 | (A;A) | 1.4x increased risk for depression (for those u... | | | |

| Mag. | Identifier | Genotype | Summary | ExAC | GetEvidence | ClinVar |
|---|---|---|---|---|---|---|
| 1.4 | rs1801157 | (A;G) | 1.4x higher risk for breast cancer | | | |
| 1.4 | rs3849942 | (A;A) | 1.4x increased risk for ALS | | Link | |
| 1.4 | rs4795067 | (G;G) | Slight increase in risk for psoriatic arthritis... | | | |
| 1.4 | rs4959039 | (A;G) | 1.4x higher risk for multiple sclerosis | | | |
| 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 | rs1375144 | (C;T) | 1.32x increased risk of developing bipolar diso... | | | |
| 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 | rs2059693 | (C;T) | 1.3x increased risk for testicular cancer | | | |
| 1.3 | rs356219 | (A;G) | 1.3x increased risk for Parkinson's disease | | | |
| 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 | rs1344706 | (T;T) | 1.2x increased risk for schizophrenia | | Link | |
| 1.2 | rs2072590 | (G;T) | 1.2x increased risk for ovarian cancer | | | |
| 1.2 | rs2252586 | (A;G) | 1.2x higher risk for glioma development | | | |
| 1.2 | rs2651899 | (G;G) | 1.2x higher risk for migraines | | | |
| 1.2 | rs2814707 | (A;A) | >1.2x increased risk for ALS | | Link | |
| 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 | rs498872 | (C;T) | 1.2x higher risk for glioma development | | Link | |
| 1.2 | rs6010620 | (A;G) | 1.2x higher risk for glioma development: 1.17x ... | | 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 | |
| 1.17 | rs3802842 | (A;C) | 1.17x increased risk of colorectal cancer | | Link | |
| 1.15 | rs748404 | (C;T) | Very slightly increased risk (1.15) for lung ca... | | Link | |
| 1.1 | rs11110912 | (C;C) | 1.3x high blood pressure risk | | | |
| 1.1 | rs13387042 | (A;G) | 1.12x increased risk for breast cancer | | Link | |
| 1.1 | rs34516635 | (G;G) | Less longevity for Ashkenazi Jewish women. | Link | | Link |
| 1.1 | rs688034 | (C;T) | 1.1x risk higher risk for coronary artery disea... | | Link | |
| 1.1 | rs7412 | (C;C) | More likely to gain weight if taking olanzapine... | Link | Link | Link |
| 1.1 | rs889312 | (A;C) | Very slightly higher risk for breast cancer | | Link | |
| 1.1 | rs997669 | (G;G) | Very slightly increased (1.18x) increased breas... | | | |
| 1.05 | rs2291834 | (C;T) | Very slightly higher risk for myocardial infarc... | | | |
| 1 | rs10504861 | (G;G) | Major allele: normal risk of migraine | | | |
| 1 | rs1143674 | (A;A) | 1.3x increased autism risk | Link | | |
| 1 | rs2228000 | (T;T) | Statistically significant: but slight: increase... | Link | Link | Link |
| 1 | rs2273697 | (A;G) | Adverse reaction more likely to carbamazepine i... | Link | Link | Link |
| 1 | rs2546890 | (A;A) | Higher risk of multiple sclerosis | | | |
| 1 | rs6932590 | (T;T) | 1.1x increased risk for schizophrenia | | Link | |
| 0 | rs1061646 | (C;C) | 1.16x increased risk for breast cancer | 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 | rs4293393 | (T;T) | 1.25x Increased Risk of CKD for T allele in ... | | | |
| 0 | rs6314 | (C;C) | Higher risk for RA | Link | Link | |
| 0 | rs6684865 | (A;A) | 1.5x risk of rheumatoid arthritis | | | |
| 0 | rs7787082 | (G;G) | 7x less likely to respond to certain antidepres... | | Link | |
| 0 | rs855791 | (T;T) | 0.2 g/dL lower hemoglobin on average | Link | Link | Link |

### 3.3 Genosets (Multi-variant Phenotypes)

| Magnitude | Identifier | Summary |
|---|---|---|
| 3.2 | gs238 | Red hair |
| 3 | gs241 | Lighter green: brown or hazel eye color |
| 2.5 | gs102 | ALS risk |
| 2.5 | gs155 | CYP3A5 non-expressor |
| 2.5 | gs281 | Part of the 88% of the population claimed not t... |
| 2.4 | gs297 | Lower heart attack risk than average |
| 2 | gs101 | Probably able to digest milk |
| 2 | gs103 | Restless legs syndrome risk |
| 2 | gs154 | NAT2 Slow metabolizer |
| 2 | gs179 | CYP2D6*41 |
| 2 | gs246 | APOE3/APOE3 |
| 1.5 | gs185 | The beta blocker metoprolol is effective with 1... |
| 1.5 | gs186 | HLA-B*5801 heterozygosity is possible: unfortun... |
| 1.5 | gs247 | Parkinson's Disease Risk |
| 1 | gs163 | CYP2D6*2A |
| 0 | gs158 | CYP1A2 normal metabolizer |

# 4 Raw Data

The raw data used to create this report has been assigned the identifier ERS1176599 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/ERS1176599

# 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.