Genomics Report for ukE3E2DF

1 Summary

Thank you for donating your genomic information to the Personal Genomes Project - United Kingdom. We have compiled this genome report from the data and we hope you will find it useful.

This is the genome report for participant ukE3E2DF . It was produced using collaborative research tools, including SNPedia and GetEvidence. This summary shows an overview of all the variants identified in your donation.

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.

There are several different types of genomic variants. The most common are single nucleotide variants (SNV) that correspond to the change of a single nucleotide in the DNA. 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. The types of variants being looked at in this report depend on the type of data donated to PGP-UK. Some sections of the report or variant types might be omitted if the donated data is not whole genome sequencing data.

Variants can be found throughout the genome. "Overlapped genes" refers to variants that were found in a region of the genome containing a gene. "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. The diagram in Figure 1 is a simplification of the usual gene structure.

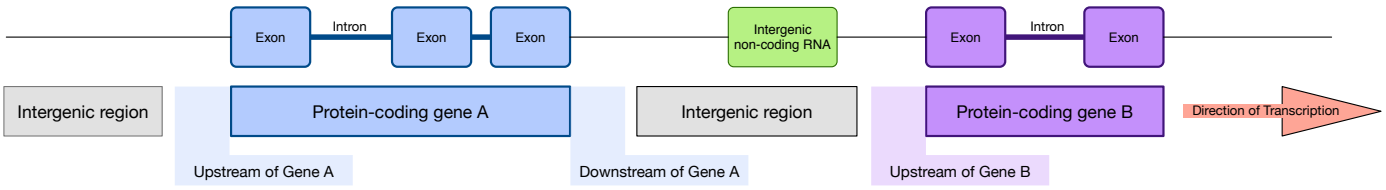

Figure 1: Diagram of gene structure indicating locations of potential variants

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.

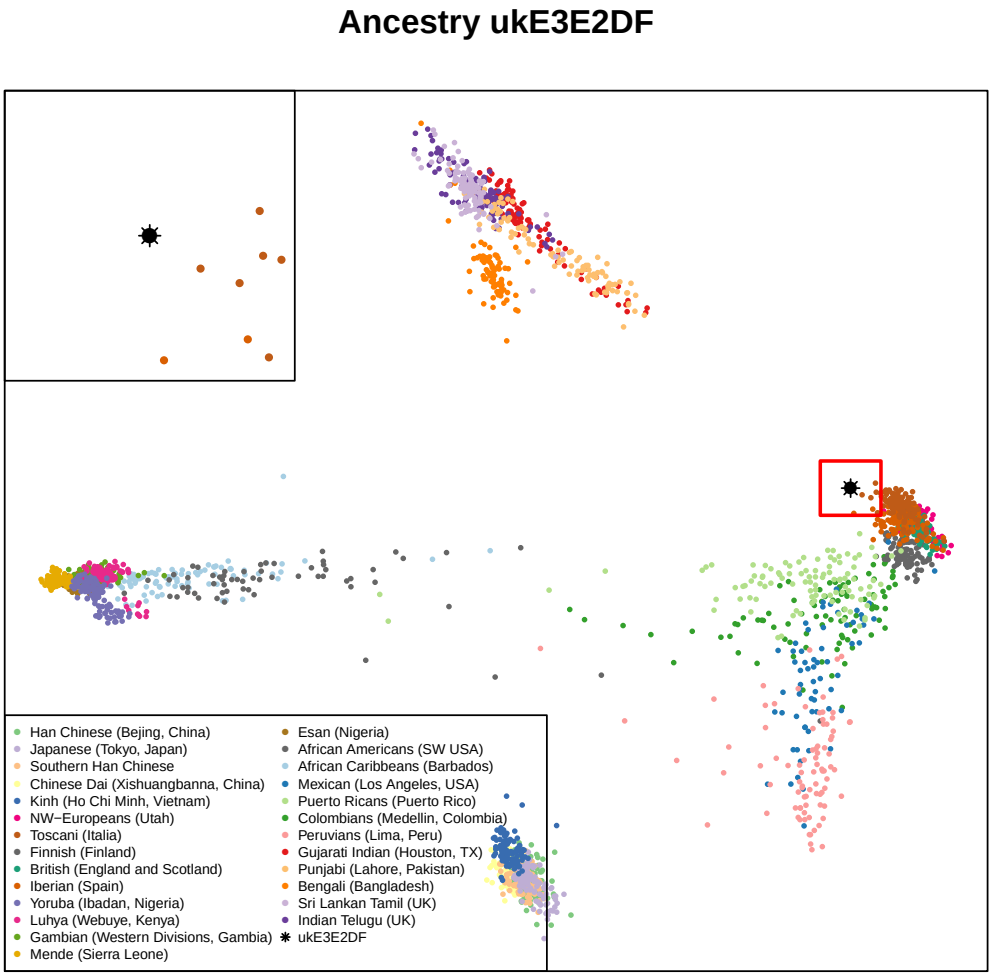

Figure 2: Ancestry Principal Component Analysis

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.

- Possibly Beneficial Traits

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
2.4	rs2802288	(A;A)	Longer lifespan			
2.1	rs547154	(A;C)	0.47x decreased risk for AMD			Link
2.1	rs6505162	(A;A)	0.43x decreased risk for esophageal cancer	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	(G;G)	Associated with higher HDL cholesterol		Link	
2	rs2060793	(A;A)	Lower serum levels of vitamin D			
2	rs261332	(A;A)	Associated with higher HDL cholesterol			
2	rs3819331	(T;T)	Lower risk of autism	Link		Link
2	rs4149268	(G;G)	Associated with higher HDL cholesterol		Link	
2	rs505922	(T;T)	Blood type O		Link	
2	rs7216389	(C;C)	0.69x lower risk of Childhood Asthma.		Link	
2	rs9642880	(G;G)	Slightly lower risk of Bladder Cancer.		Link	
1.6	rs10801935	(C;C)	0.3x decreased risk of breast cancer			
1.6	rs3775948	(C;C)	Slightly lower risk for gout			
1.5	rs3851179	(A;A)	0.85x decreased risk for Alzheimer's disease		Link	
1.5	rs4149274	(C;C)	Associated with higher HDL (good) cholesterol.			
1.5	rs610932	(A;A)	A allele associated with reduced risk of Alzhei...			
1.5	rs6427528	(A;A)	For rheumatoid arthritis patients: better respo...			
1.5	rs9939609	(T;T)	Lower risk of obesity and Type-2 diabetes		Link	
1.2	rs4686484	(G;G)	Slightly decreased risk for celiac disease			
1.2	rs6048	(G;G)	Slightly lower risk (10-20%) of deep vein throm...	Link	Link	Link
1	rs182549	(C;T)	Can digest milk.			Link
1	rs7850258	(A;G)	Typical odds of developing primary hypothyroidi...			
1	rs8179183	(G;G)	Less likely to gain weight if taking risperidon...	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

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
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	Link
0	rs1799945	(C;C)	Not a H63D hemochromatosis carrier.	Link	Link	Link
0	rs1800562	(G;G)	Not a C282Y hemochromatosis carrier.	Link	Link	Link
0	rs28933385	(G;G)	Prion protein Codon 200 (E) - Non pathogenic va...			Link
0	rs5065	(A;A)	1.12x risk on diuretic; if hypertensive: better...	Link	Link	Link
0	rs6259	(G;G)	Best inverse correlation between tea-drinking: ...	Link	Link	
0	rs74315403	(G;G)	PrP codon 178 (D) - non pathogenic variant			Link
0	rs7495174	(A;A)	Blue/gray eyes more likely		Link	
0	rs9394492	(C;C)	<0.76x risk for restless legs			

- Possibly Harmful Traits

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
3.8	rs5186	(C;C)	7.3x increased risk of hypertension	Link	Link	Link
3.5	rs1800546	(C;G)	Carrier for high risk of of hereditary fructose...	Link	Link	Link
3.5	rs7574865	(T;T)	1.69x risk of rheumatoid arthritis; 2.4x risk o...		Link	Link
3.1	rs10830963	(G;G)	Increased type-2 diabetes risk; higher gestatio...		Link	
3	rs13266634	(C;C)	Increased risk for type-2 diabetes	Link	Link	Link
3	rs891512	(A;A)	Higher blood pressure than G;G	Link		Link
2.6	rs110419	(A;A)	2.6x increased risk for neuroblastoma			
2.5	rs10484554	(T;T)	~4x increased risk for psoriasis		Link	
2.5	rs10490924	(G;T)	2.7x risk for age related macular degeneration	Link	Link	Link
2.5	rs1057910	(A;C)	CYP2C9*3 carrier; average 40% reduction in warf...	Link	Link	Link
2.5	rs16969968	(A;G)	Slightly higher risk for nicotine dependence: l...	Link	Link	Link
2.5	rs339331	(T;T)	Prostate cancer risk			
2.5	rs3738919	(C;C)	1.94x risk of developing rheumatoid arthritis			
2.5	rs5888	(C;T)	3x higher risk for age-related macular degenera...	Link		
2.4	rs1143679	(A;G)	1.78x increased risk for SLE	Link	Link	
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
2.2	rs3129934	(T;T)	3.3x increased risk for multiple sclerosis		Link	
2.1	rs1050152	(T;T)	2.1x increased risk of Crohn's disease	Link	Link	Link
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	rs380390	(C;C)	Increased risk for ARMD		Link	
2.1	rs4444903	(G;G)	3.5x risk of hep-cancer in cirrhosis patients; ...			Link
2.1	rs4693596	(C;C)	2x odds of myopathy if taking statins			
2.1	rs5751876	(T;T)	Significantly higher anxiety levels after moder...	Link		
2	rs10248420	(A;A)	7x less likely to respond to certain antidepres...		Link	
2	rs1045642	(C;T)	Slower metaboliser for some drugs	Link	Link	Link
2	rs1051730	(C;T)	1.3x increased risk of lung cancer	Link	Link	Link
2	rs10984447	(A;A)	>1.17x increased risk for multiple sclerosis		Link	
2	rs11171739	(C;C)	1.75x risk of developing Type-1 diabetes		Link	
2	rs1160312	(A;A)	1.6x increased risk of Male Pattern Baldness.		Link	
2	rs11650354	(T;T)	8x risk for allergic asthma	Link		
2	rs11983225	(T;T)	7x less likely to respond to certain antidepres...		Link	
2	rs12431733	(T;T)	Increased risk of developing Parkinson's Diseas...		Link	
2	rs12469063	(G;G)	Increased risk of developing restless legs synd...			
2	rs1361600	(G;G)	~2x increased risk for adult-onset asthma in Ja...			
2	rs1544410	(A;A)	Increased risk of low bone mineral density diso...		Link	
2	rs16942	(A;G)	Very slightly increased breast cancer risk	Link	Link	Link
2	rs1734791	(A;A)	1.4x increased risk for lupus			
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	Link
2	rs1800896	(A;A)	1.8x increased prostate cancer risk			
2	rs1801160	(A;G)	Possible 5-fluorouracil toxicity	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	rs2305480	(C;T)	3.5x increase in risk of asthma for Han Chinese...	Link	Link	
2	rs25487	(A;G)	2x higher risk for skin cancer; possibly other ...	Link	Link	Link
2	rs2697962	(A;A)	Increased risk of developing Parkinson's Diseas...			
2	rs2736100	(T;T)	Higher risk of Interstitial lung disease: and t...		Link	Link

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
2	rs3738579	(C;C)	0.6x decreased risk for cervical cancer: but 1....			
2	rs3746444	(C;T)	~1.2x increased risk for cancer	Link		
2	rs3790565	(C;C)	Increased risk of developing primary biliary ci...			
2	rs3802842	(C;C)	>1.17x increased risk of colorectal cancer		Link	
2	rs4148739	(A;A)	7x less likely to respond to certain antidepres...		Link	
2	rs4242382	(A;A)	1.7x increased risk for prostate cancer		Link	
2	rs4792311	(A;A)	Increased risk of prostate cancer	Link	Link	Link
2	rs486907	(A;A)	2x increased prostate cancer risk	Link	Link	Link
2	rs6601764	(C;C)	1.52x increased risk of developing Crohn's dise...		Link	
2	rs6807362	(C;C)	Increased autism risk	Link	Link	
2	rs6897932	(C;C)	1.08x increased risk for multiple sclerosis	Link	Link	Link
2	rs699	(C;C)	Increased risk of hypertension	Link	Link	Link
2	rs7442295	(A;A)	~4x higher risk for hyperuracemia		Link	Link
2	rs744373	(C;C)	1.17x risk of Alzheimer's			
2	rs7923837	(G;G)	3.2x risk for T2D			
2	rs7961152	(A;A)	1.5x higher risk for hypertension			
2	rs800292	(C;C)	5% higher risk of Age related macular degenerat...	Link	Link	Link
2	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;A)	3.1x increased thyroid cancer risk		Link	
2.0	rs2305795	(A;A)	1.64x higher risk of narcolepsy compared to (G;...			Link
1.8	rs1136287	(C;T)	1.5x increased risk of wet ARMD in a Taiwanese ...	Link	Link	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	
1.6	rs11523871	(A;C)	1.6x increased breast cancer risk for women ove...	Link	Link	
1.6	rs2981745	(C;T)	1.6x increased risk for breast cancer in female...			
1.6	rs3764880	(A;A)	1.2 - 1.8x increased tuberculosis risk	Link	Link	
1.5	rs1169300	(A;G)	~1.5x increased lung cancer risk			
1.5	rs12498742	(A;A)	1.25 increased risk for gout			
1.5	rs13181	(G;T)	1.12x increased risk for cutaneous melanoma	Link	Link	Link
1.5	rs144848	(G;T)	Very slightly increased breast cancer risk	Link	Link	Link
1.5	rs165599	(G;G)	May indicate increased susceptibility to schizo...		Link	
1.5	rs17115100	(G;T)	Slightly increased risk of developing Parkinson...	Link	Link	
1.5	rs1801274	(T;T)	Complex; generally greater risk for cancer prog...	Link	Link	Link
1.5	rs1867277	(A;G)	1.5x increased risk for thyroid cancer			
1.5	rs2007153	(G;G)	Increased risk of schizophrenia in limited stud...			
1.5	rs2076295	(G;G)	Slightly increased risk for pulmonary fibrosis ...			
1.5	rs2240340	(A;A)	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	rs2464196	(C;T)	~1.5x increased lung cancer risk	Link	Link	Link
1.5	rs2881766	(T;T)	Slightly increased risk for pregnancy-induced h...			
1.5	rs393152	(A;A)	Increased risk of both PD and AD	Link	Link	
1.5	rs464049	(T;T)	Increased risk of schizophrenia in limited stud...			
1.5	rs4982731	(C;C)	Possible higher risk of childhood acute lymphob...			
1.5	rs5746059	(A;A)	Slightly higher fat mass			
1.5	rs7341475	(G;G)	1.58x increased schizophrenia risk for women		Link	
1.5	rs872071	(G;G)	~1.5x increased risk for chronic lymphocytic le...		Link	
1.4	rs10865331	(A;A)	1.4x higher risk for ankylosing spondylitis			
1.4	rs1126497	(T;T)	1.4x increased risk for breast cancer	Link	Link	Link
1.4	rs1801157	(A;G)	1.4x higher risk for breast cancer			
1.4	rs2230201	(A;G)	1.4x risk of lupus	Link		Link
1.4	rs3131296	(G;G)	1.4x increased risk for schizophrenia		Link	
1.4	rs3849942	(A;A)	1.4x increased risk for ALS		Link	
1.4	rs4795067	(G;G)	Slight increase in risk for psoriatic arthritis...			

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
1.4	rs498872	(T;T)	1.4x higher risk for glioma development		Link	
1.3	rs10947262	(C;C)	1.3x increased risk for osteoarthritis			
1.3	rs1434536	(A;G)	1.29x increased breast cancer risk			Link
1.2	rs419788	(A;G)	2.0x risk for lupus	Link		
1.2	rs4324715	(C;C)	>1.5x increased testicular cancer risk for men			
1.17	rs17465637	(A;C)	1.17x higher risk for myocardial infarction	Link	Link	
1.15	rs748404	(C;T)	Very slightly increased risk (1.15) for lung ca...		Link	
1.1	rs11037909	(C;T)	1.27x type II diabetes risk	Link		
1.1	rs2653349	(G;G)	2-6x increased risk for cluster headaches	Link	Link	
1.1	rs2828520	(G;G)	1.35x major depressive disorder risk			
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	rs7412	(C;C)	More likely to gain weight if taking olanzapine...	Link	Link	Link
1.05	rs2291834	(C;T)	Very slightly higher risk for myocardial infarc...			
1	rs1143674	(A;G)	1.3x increased autism risk	Link		
1	rs6932590	(T;T)	1.1x increased risk for schizophrenia		Link	
1	rs798766	(T;T)	Increased susceptibility urinary bladder cancer...			
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	rs1042173	(T;T)	Among alcoholics: likely to be heavier drinkers...			Link
0	rs1061646	(C;C)	1.16x increased risk for breast cancer	Link		Link
0	rs10761659	(A;A)	1.5x risk of Crohn's disease		Link	
0	rs1495965	(A;A)	1.2x higher risk for spondylitis			
0	rs4293393	(T;T)	1.25x Increased Risk of CKD for T allele in ...			
0	rs6314	(C;C)	Higher risk for RA	Link	Link	

- Genosets (Multi-variant Phenotypes)

Magnitude	Identifier	Summary
3.1	gs191	Problem metabolizing NSAIDs
2.9	gs192	MTHFR polymorphisms affecting homocysteine
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	gs285	You will lose 2.5x as much weight on a low fat ...
2.4	gs297	Lower heart attack risk than average
2.2	gs280	Light hair color for europeans
2	gs101	Probably able to digest milk
2	gs103	Restless legs syndrome risk
2	gs154	NAT2 Slow metabolizer
2	gs159	CYP1A2 fast metabolizer
2	gs179	CYP2D6*41
2	gs181	CYP2D6*2
2	gs188	One copy of APOE4 is possible: but not certain
1.8	gs1002	Mitochondrial Haplogroup H1
1.5	gs1001	Mitochondrial Haplogroup H
1.5	gs247	Parkinson's Disease Risk
1.2	gs184	Able to taste bitterness.
0.1	gs233	Normal pain sensitivity

4 Report Metadata

Resource	Version	Website
Genome	GRCh37	Link
BWA	0.7.12	Link
SAMtools	1.3	Link
GATK	3.4-46	Link
PLINK	v1.90b3.35	Link
SNPedia	30-Jul-2017	Link
ExAC	v0.3.1	Link
GetEvidence	30-Jul-2017	Link
ClinVar	30-Jul-2017	Link

Table 4: Analysis Pipeline Versions

Report generated on February 22, 2018 (using report generator version 18-053).