Genomics Report for uk6D0CFA

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 uk6D0CFA . 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.

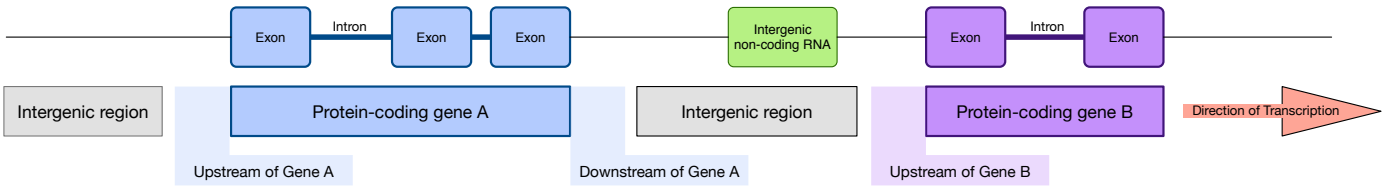

Figure 1: Diagram of gene structure indicating locations of potential variants

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.

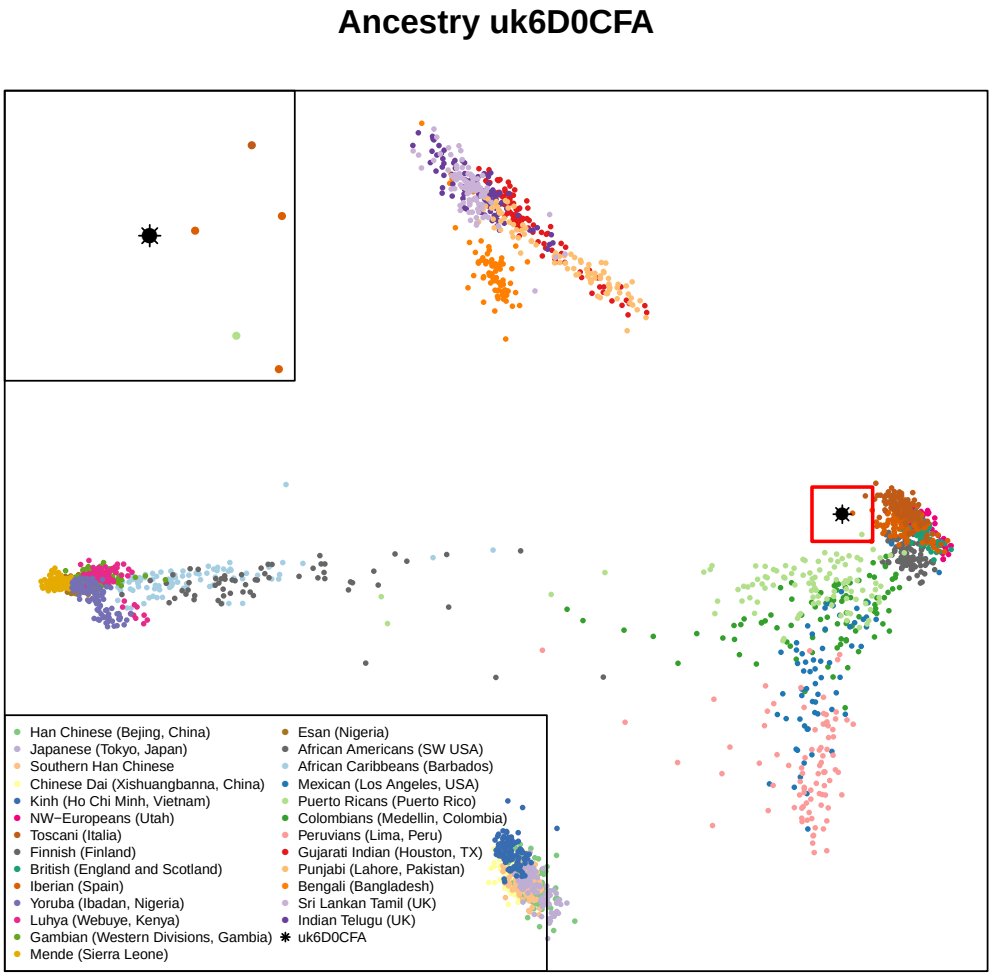

Figure 2: Ancestry Principal Component Analysis

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
2.1	rs3775291	(A;G)	0.71x decreased risk for dry age related macula...	Link	Link	Link
2	rs1136410	(C;T)	0.80x reduced risk for glioblastoma	Link	Link	
2	rs1864163	(G;G)	Associated with higher HDL cholesterol		Link	
2	rs2235015	(G;T)	Somewhat more likely to respond to certain anti...	Link	Link	
2	rs261332	(A;A)	Associated with higher HDL cholesterol			
2	rs2707466	(A;A)	Stronger bones	Link	Link	
2	rs2908004	(T;T)	Stronger bones	Link	Link	
2	rs3819331	(T;T)	Lower risk of autism	Link		Link
1.8	rs1128535	(A;G)	0.77x risk for Crohn's disease			
1.8	rs1800588	(C;T)	Higher HDL-C levels	Link	Link	
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	rs2278206	(C;C)	Possibly less susceptible to asthma	Link	Link	
1.5	rs2007153	(A;A)	Decreased risk of schizophrenia in limited stud...			
1.5	rs5888	(C;C)	Higher HDL cholesterol but lower risk for age-r...	Link		
1.5	rs610932	(A;A)	A allele associated with reduced risk of Alzhei...			
1.5	rs6427528	(A;A)	For rheumatoid arthritis patients: better respo...			
1.4	rs2294008	(C;C)	Lower risk of gastric and bladder cancer	Link	Link	
1	rs1800547	(G;G)	MAPT H2/H2 diplotype	Link		Link
1	rs182549	(C;T)	Can digest milk.			Link
1	rs2235067	(A;A)	7x more likely to respond to certain antidepres...			
1	rs800292	(C;T)	1% decreased risk of macular degeneration	Link	Link	Link
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	
0.1	rs891512	(G;G)	Lower blood pressure than those with an A allel...	Link		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

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
0	rs16990018	(A;A)	PrP Codon 171 Asn - Non-pathogenic variant	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	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	

- Possibly Harmful Traits

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
3	rs1801282	(C;G)	Unconfirmed higher risk of cardiovascular disea...	Link	Link	Link
3	rs258322	(T;T)	2x increased risk of Melanoma		Link	
3	rs3738579	(T;T)	1.5x - 2x increased risk for cervical cancer: H...			
3	rs3803662	(T;T)	1.6x increased risk for breast cancer		Link	
2.5	rs12536657	(A;A)	Hypermetropia risk - longsightedness	Link		
2.5	rs13266634	(C;T)	Increased risk for type-2 diabetes	Link	Link	Link
2.5	rs1799971	(A;G)	Stronger cravings for alcohol. if alcoholic: na...	Link	Link	Link
2.5	rs2241880	(C;C)	2x-3x increased risk for Crohn's disease in Cau...	Link	Link	Link
2.5	rs2254958	(C;C)	1.61x increased risk for Alzheimer's			
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	rs2494732	(C;C)	Greater odds of cannabis-associated psychosis	Link	Link	
2.1	rs5186	(A;C)	~1.4x increased risk of hypertension	Link	Link	Link
2.1	rs5751876	(T;T)	Significantly higher anxiety levels after moder...	Link		
2	rs10096097	(G;G)	Increased Anorexia Nervosa risk			
2	rs1042838	(G;T)	1.28x risk for endometrial ovarian cancer; over...	Link	Link	
2	rs1045642	(C;T)	Slower metaboliser for some drugs	Link	Link	Link
2	rs1169300	(A;A)	~2x increased lung cancer risk			
2	rs11983225	(T;T)	7x less likely to respond to certain antidepres...		Link	
2	rs144848	(G;G)	Very slightly increased breast cancer risk	Link	Link	Link
2	rs16942	(A;G)	Very slightly increased breast cancer risk	Link	Link	Link
2	rs17576	(A;G)	Higher risk for MI and lung cancer: and COPD in...	Link	Link	Link
2	rs1994090	(G;G)	Increased risk of developing Parkinson's Diseas...		Link	
2	rs2070676	(G;G)	CYP2E1*1B homozygote			
2	rs2230201	(G;G)	>1.4x risk of lupus	Link		Link
2	rs2235040	(G;G)	7x less likely to respond to certain antidepres...	Link	Link	
2	rs2274223	(A;G)	1.5x increased risk for stomach and esophageal ...	Link	Link	Link
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	rs326	(A;A)	Lower HDL cholesterol		Link	Link
2	rs3746444	(C;T)	~1.2x increased risk for cancer	Link		
2	rs4633	(T;T)	Higher risk for endometrial cancer	Link	Link	Link
2	rs4792311	(A;G)	Increased risk of prostate cancer	Link	Link	Link
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	rs965513	(A;A)	3.1x increased thyroid cancer risk		Link	
2.0	rs1434536	(A;A)	1.94x increased breast cancer risk			Link
1.8	rs1136287	(C;T)	1.5x increased risk of wet ARMD in a Taiwanese ...	Link	Link	Link
1.6	rs11523871	(A;C)	1.6x increased breast cancer risk for women ove...	Link	Link	
1.6	rs1800450	(A;A)	Mannose binding deficiency but of low clinical ...	Link	Link	Link
1.6	rs33980500	(C;T)	1.6x increase in risk for psoriatic arthritis	Link	Link	Link
1.6	rs3764880	(A;A)	1.2 - 1.8x increased tuberculosis risk	Link	Link	
1.5	rs13181	(G;T)	1.12x increased risk for cutaneous melanoma	Link	Link	Link
1.5	rs1801274	(T;T)	Complex; generally greater risk for cancer prog...	Link	Link	Link
1.5	rs199533	(C;T)	Slightly increased risk of developing Parkinson...	Link		
1.5	rs2240340	(A;A)	Slightly increased (1.5x) risk for RA	Link		
1.5	rs2464196	(C;T)	~1.5x increased lung cancer risk	Link	Link	Link
1.5	rs28694718	(A;G)	2x higher risk for schizophrenia			
1.5	rs401681	(C;C)	~1.2x increased risk for several types of cance...		Link	
1.5	rs5219	(C;T)	1.3x increased risk for type-2 diabetes	Link	Link	Link
1.5	rs619203	(C;G)	Increases susceptibility to Myocardial Infarcti...	Link	Link	
1.4	rs1126497	(C;T)	1.4x increased risk for breast cancer	Link	Link	Link
1.4	rs3184504	(C;T)	Slightly increased risk for celiac disease	Link	Link	

Mag.	Identifier	Genotype	Summary	ExAC	GetEvidence	ClinVar
1.3	rs34330	(C;T)	1.3x higher risk for endometrial cancer (in Chi...			Link
1.25	rs13387042	(A;A)	1.24x increased risk for breast cancer		Link	
1.25	rs748404	(T;T)	Slightly increased risk (1.25) for lung cancer...		Link	
1.2	rs11037909	(T;T)	1.47x type II diabetes risk	Link		
1.2	rs1800693	(A;G)	Slight (1.2x) increase in risk for multiple scl...	Link	Link	Link
1.2	rs3740878	(A;A)	1.46x type II diabetes risk; common	Link		Link
1.2	rs4496877	(T;T)	For type-1 diabetics: 1.6x increased nephropath...			
1.2	rs9858542	(A;G)	1.1x risk Crohn's Disease	Link	Link	
1.1	rs34516635	(G;G)	Less longevity for Ashkenazi Jewish women.	Link		Link
1.1	rs7412	(C;C)	More likely to gain weight if taking olanzapine...	Link	Link	Link
1.07	rs2291834	(C;C)	Very slightly higher risk for myocardial infarc...			
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	rs5326	(A;G)	Possible psychiatric risks			
0.1	rs601338	(G;G)	Susceptible to Norovirus infections	Link	Link	Link
0	rs1061646	(C;C)	1.16x increased risk for breast cancer	Link		Link
0	rs6314	(C;C)	Higher risk for RA	Link	Link	

- Genosets (Multi-variant Phenotypes)

Magnitude	Identifier	Summary
3.1	gs191	Problem metabolizing NSAIDs
3	gs127	Intermediate warfarin metabolizer
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	gs101	Probably able to digest milk
2	gs154	NAT2 Slow metabolizer
2	gs159	CYP1A2 fast metabolizer
2	gs179	CYP2D6*41
2	gs246	APOE3/APOE3
1.5	gs185	The beta blocker metoprolol is effective with 1...
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).