Project 5 H+, or how to build a perfect human.

The work was performed by Maria Uzun and Alisa Fedorenko.

Human genome analyzed in this study is encrypted for privacy and security purposes.

1. File conversion

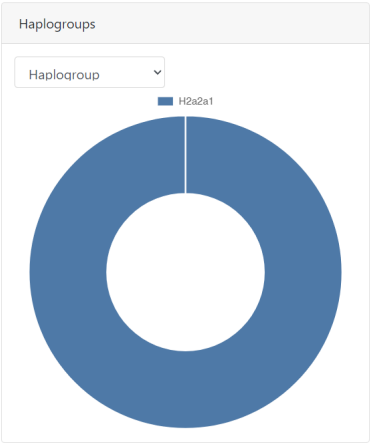
The file conversion has been already done, so we skip this step.

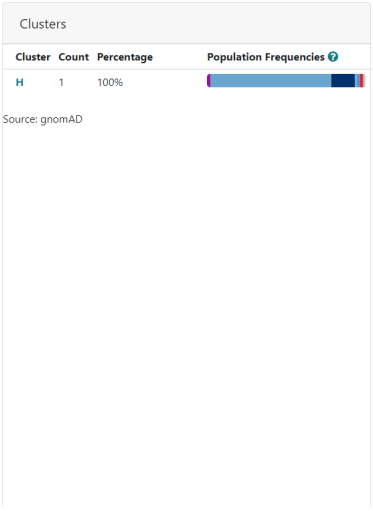
2. Origins, haplogroups

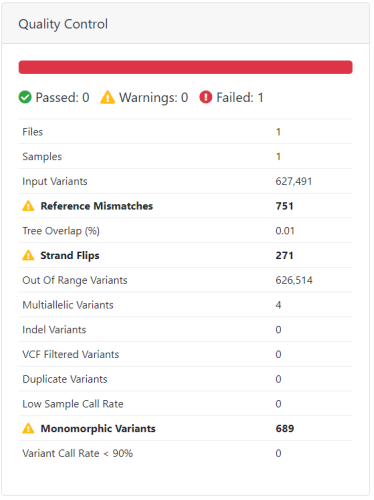
Haplogrep 3 - Haplogrep 3 3.2.1 (i-med.ac.at) used vcf file to detect haplogroups.

The found haplogroup is H2a2a1.

“H2, the subhaplogroup of haplogroup H, originated in Central Asia and Eastern Europe between 10,300 and 13,300 years ago, then diverged into haplogroup H2a between 8,900 and 12,2000 years ago, and then into haplogroup H2a2 between 7,300 and 11,000 years ago. H2a2a1, a subgroup of H2a2, is thought to be 5,000-9,100 years old (Behar, Harmant, et al., 2012; Behar, van Oven, et al., 2012)”







3. Annotation - sex and eye colour, health features

The genome contains X-chromosome, and do not contains Y, thus we are working with female human genome.

For annotation of SNPs associated with phenotypic traits (eye, hair, skin color) we used IGV genome browser. For finding SNPs associated with significant health features the Variant Effect Predictor was used. The following results are presented in the Table 1 below.

snp	chromosome	position	variant	feature	Suggested changes

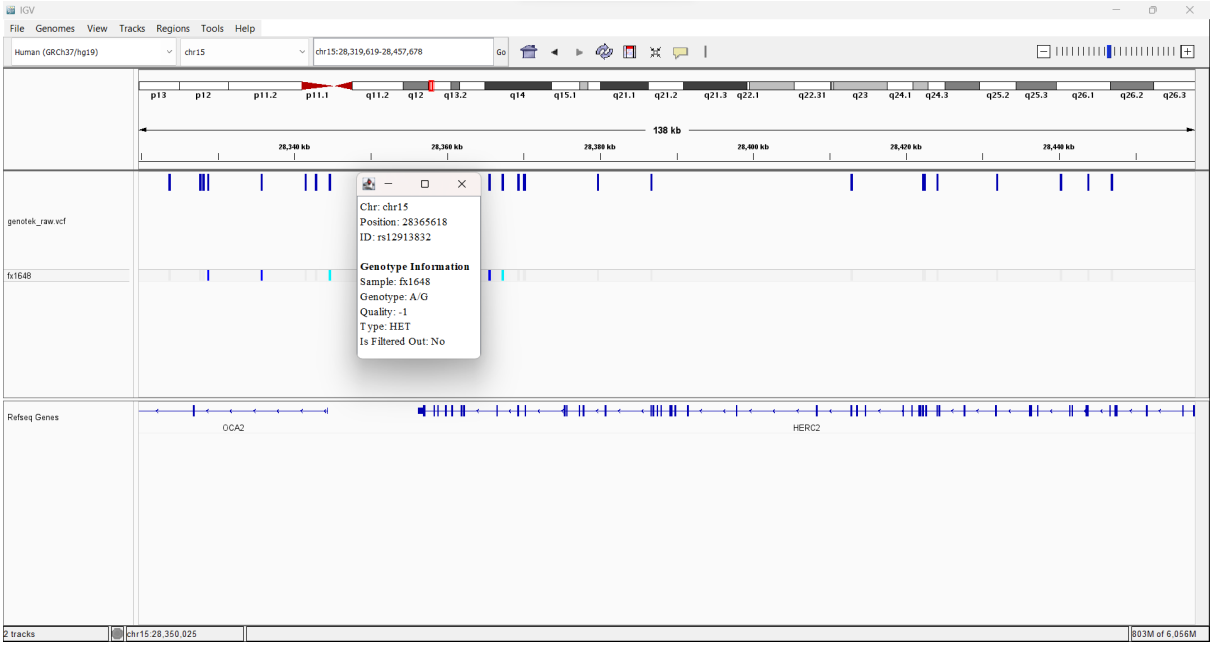
eye color					
rs12913832	15	28365618	A/G	not blue	-
rs12203592	6	396321	C/T	Primarily in Europeans; likely presence of freckles, brown hair and high sensitivity of skin to sun exposure.	-
rs16891982	5	33951588	G/G	Generally European; Light skin; Possibly an increased risk of melanoma	-
rs12896399	14	92773663	G/G	Brown eyes	-
risks of diseases					
rs6548238	2	634905	C/C	Risk of obesity	C/T
rs887829	2	234668570	T/T	HYPERBILIRUBINEMIA I; HYPERBILIRUBINEMIA, ARIAS TYPE; Gilbert Disease;	T/C
rs7385804	7	100235970	C/A	decreased iron status	C/C
rs1801133	1	11856378	G/A	decreased efficiency in processing folic acid	G/G
rs7850258	9	97786731	G/G	Slightly higher odds of developing primary hypothyroidism.	A/A

Supplementary information

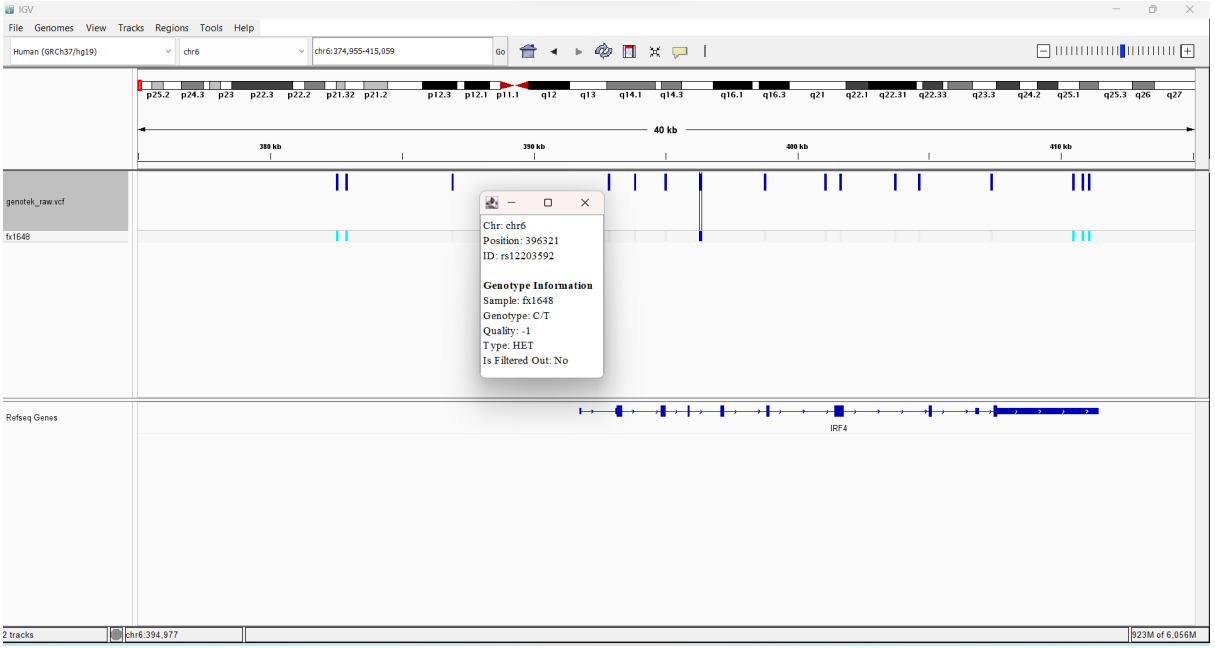
1. The determination polymorphisms by applying IGV.

Phenotypic traits:

rs12913832 - A/G

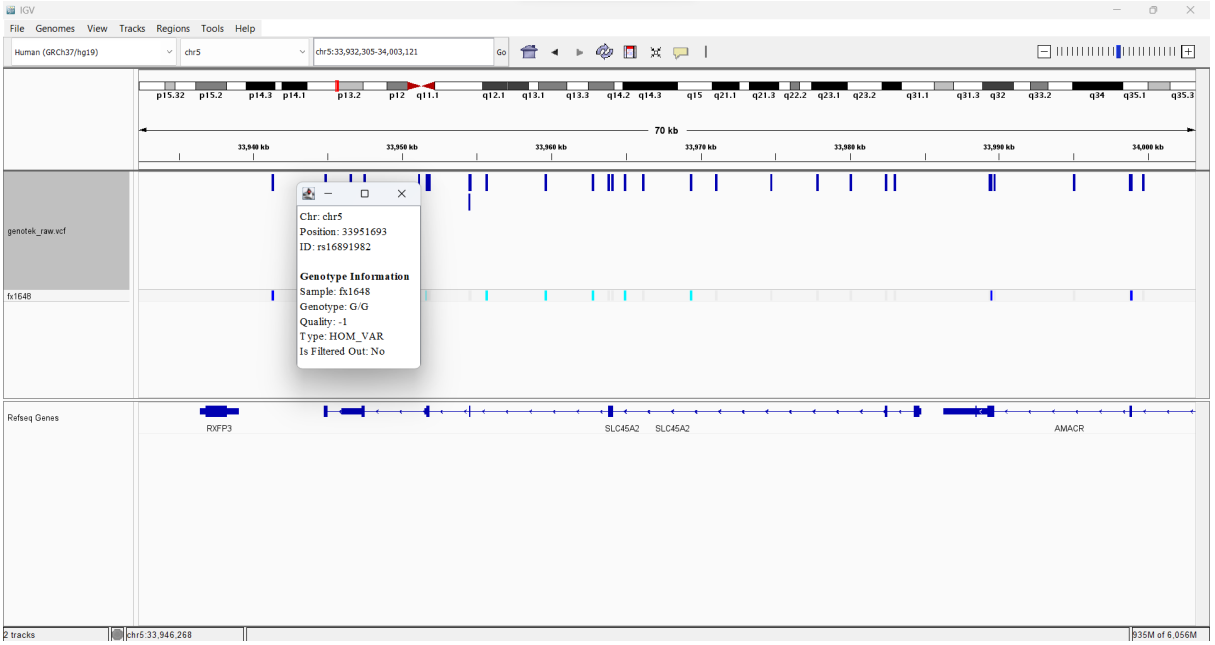


rs12203592 - C/T



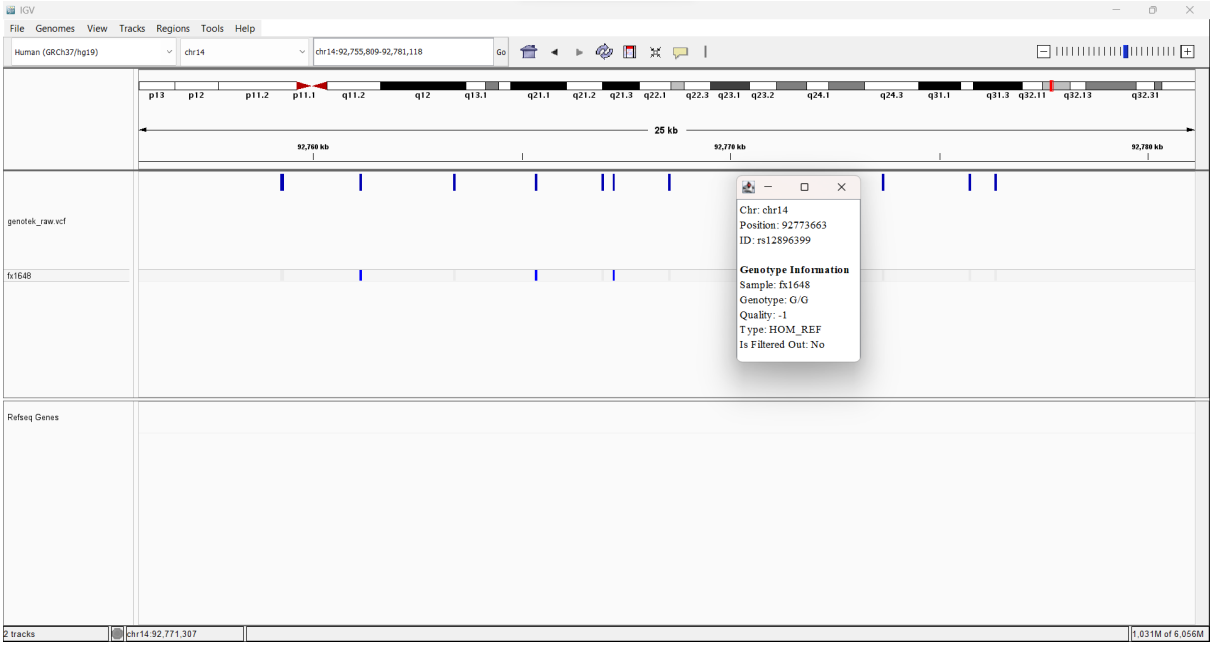
Primarily in Europeans; likely presence of freckles, brown hair and high sensitivity of skin to sun exposure.

rs16891982 - G/G



Generally European; Light skin; Possibly an increased risk of melanoma

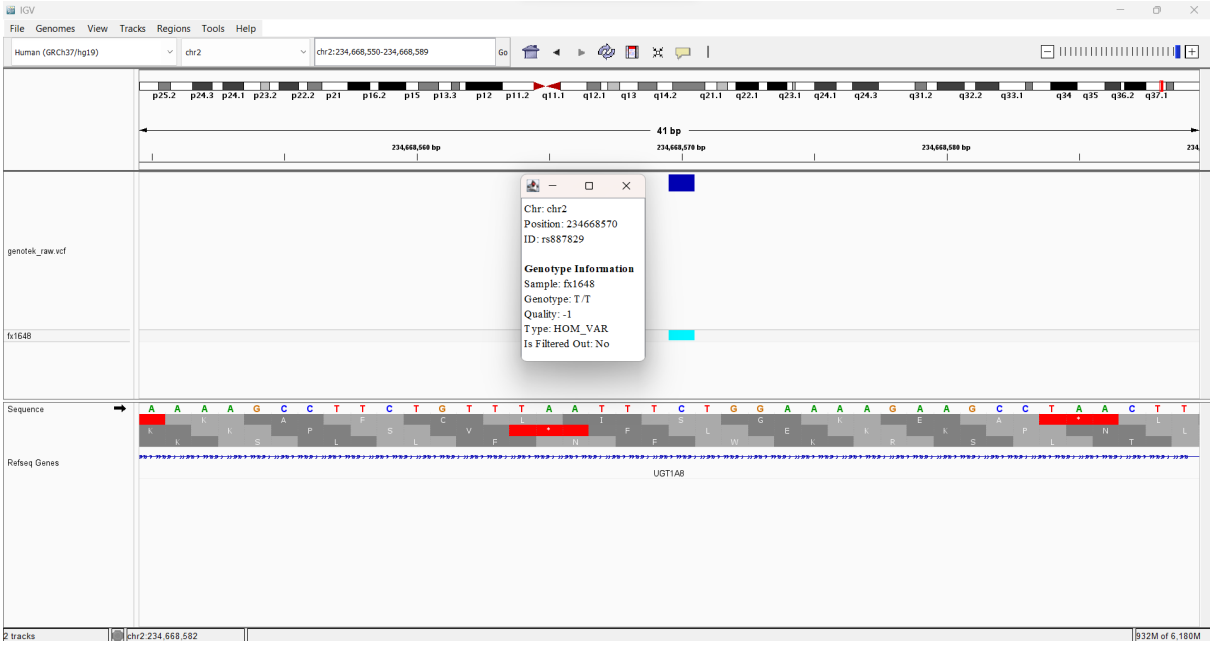
rs12896399 G/G



Diseases/problems:

HYPERBILIRUBINEMIA I; HYPERBILIRUBINEMIA, ARIAS TYPE; Gilbert Disease;

rs887829 position 234,668,570 - patient variant C→T **NM_000463.3(UGT1A1):c.1091C>T (p.Pro364Leu)]**



2. Risks associated with SNPs and identified by VEP tool.

Uploaded variant	Location	Allele	Consequence	Feature type	Feature	Biotype	Existing variant	REF ALLELE	UPLOADED ALLELE	Feature strand	AF	Clinical significance	Motif name
rs6548238	2:634905-634905	C	regulatory_region_variant	RegulatoryFeature	ENSR00001222132	TF_binding_site	rs6548238	T	T/C	-	0.8770	risk_factor	-
rs6548238	2:634905-634905	C	TF_binding_site_variant	MotifFeature	ENSM00321844616	-	rs6548238	T	T/C	1	0.8770	risk_factor	ENSPFM0238
rs6548238	2:634905-634905	C	TF_binding_site_variant	MotifFeature	ENSM00332358802	-	rs6548238	T	T/C	1	0.8770	risk_factor	ENSPFM0260
rs6548238	2:634905-634905	C	intergenic_variant	-	-	-	rs6548238	T	T/C	-	0.8770	risk_factor	-