

Exercise – 1 : Task 1 Solution

BGvar

Small Group Assignment
Genetics Student Accounts

About BGvar

BGvar is a comprehensive compilation of human blood group alleles belonging to International Society of Blood Transfusion (ISBT) approved human blood group systems. The resource integrates relevant data from a range of public sources, encompassing a total of 44 human group systems (updated till October, 2022), 349 antigens, 1873 approved alleles. BGvar also enlists about 1509 blood group alleles predicted from 1000 Genomes Project and compiled from literature reports. The compiled variants are systematically annotated for their HGVS nomenclature, Global Allele Frequencies and corresponding clinical significance collated from literature evidences.

ISBT approved Alleles

Predicted and curated alleles

p.Thr193Met

Example Search :

Gene Name

ABOBCAM

Allele Name

RAPH*01.02ABO*A2.06

Nucleotide Change

c.243T>Ac.505G>A

Protein Change

p.Asn81Lysp.Ala169Thr

dbSNP ID

rs1008708453rs28399653

Blood groups list

ABOABCC1AUGUSTINECD59CHIDO/RODGERSCOLTONCROMERCTL2DIEGODOMBRUCKDUFFYEMMERFORSGATA1GERBICH

GILGLOBHIINJMHJUNIORKANNOKELLKIDDKLFIKNOPSKXLANGEREISLEWISLUTHERANLWMAMMNSOK

P1PKPELRAPHRRHAGSCIANNASIDVELXGYT

Gene Name : KEL

Gene Id : ENSG00000197993

Protein Id : P23276

Variant details

Gene Alias :

CD238, ECE3

ISBT Version :

v7.0 30-NOV-2021

Variant phenotype :

KEL:1,-2 or K+k-

Phenotype category :

NA

Phenotype Description :

NA

Variant Type :

Single Nucleotide Variation

Reference Allele name :

KEL*02

Allele name :

KEL*01.01

Reference allele :

G

Alternate allele :

A

Nucleotide_Change :

c.578C>T

Protein Change :

p.Thr193Met

HGVS nomenclature :

| Nucleotide | Transcript | Protein |
|-----------------------------|----------------------|-------------------------|
| NC_000007.13:g.142655008G>A | NA | NP_000411.1:p.Thr193Met |
| NC_000007.14:g.142957921G>A | NM_000420.2:c.578C>T | |

External Variation ID :

NA

| | |
|--------------------|------|
| Gene name | KEL |
| Blood group system | KELL |
| Blood phenotype | K+k- |
| Chromosome | 7 |


| | |
|------------------|-----------|
| Genomic position | 142655008 |
| Variant type | SNV |
| Variation ID | NA |

Exercise – 2 : Task 2 Solution

Variant Effect Predictor

New job

Species:

 Homo_sapiens X

Assembly: GRCh38.p14
[Change species](#)
If you are looking for VEP for Human GRCh37, please go to [GRCh37 website](#).

Name for this job (optional):

Input data:

Either paste data:

rs2071699
rs5030865
rs5030862
rs206069
rs206079
rs144848
rs1057128

Run instant VEP for current line >

Examples: [Ensembl default](#), [VCF](#), [Variant identifiers](#), [HGVS notations](#), [SPDI](#)

Or upload file:

Choose file No file chosen

Or provide file URL:

| | A | B | C | D | E | F | G | H | I | J | K | L |
|----|------------|-------------|--------|-------------|----------|---------|------|---------|---------|---------|------|----------|
| 1 | #Upload | Location | Allele | Consequ | IMPACT | SYMBOL | Gene | Feature | Feature | BIOTYPE | EXON | INTRON |
| 8 | rs2298719 | 18:45736484 | G | missense_va | MODERATE | SLC14A1 | | | | | | 07-Nov - |
| 9 | rs2298719 | 18:45736484 | G | missense_va | MODERATE | SLC14A1 | | | | | | 07-Nov - |
| 10 | rs2298719 | 18:45736484 | G | missense_va | MODERATE | SLC14A1 | | | | | | 05-Sep - |
| 11 | rs2298719 | 18:45736484 | G | missense_va | MODERATE | SLC14A1 | | | | | | 05-Sep - |
| 12 | rs2298719 | 18:45736484 | G | missense_va | MODERATE | SLC14A1 | | | | | | 04-Aug - |
| 13 | rs2298719 | 18:45736484 | G | missense_va | MODERATE | SLC14A1 | | | | | | 06-Oct - |
| 14 | rs2298719 | 18:45736484 | G | missense_va | MODERATE | SLC14A1 | | | | | | 07-Nov - |
| 15 | rs2298719 | 18:45736484 | G | missense_va | MODERATE | SLC14A1 | | | | | | 03-Jul - |
| 16 | rs2298719 | 18:45736484 | G | missense_va | MODERATE | SLC14A1 | | | | | | 07-Nov - |
| 17 | rs2298719 | 18:45736484 | G | missense_va | MODERATE | SLC14A1 | | | | | | 06-Oct - |
| 18 | rs2298719 | 18:45736484 | G | missense_va | MODERATE | SLC14A1 | | | | | | 07-Oct - |
| 19 | rs2298719 | 18:45736484 | G | missense_va | MODERATE | SLC14A1 | | | | | | 08-Dec - |
| 20 | rs2298719 | 18:45736484 | G | missense_va | MODERATE | SLC14A1 | | | | | | 07-Oct - |
| 22 | rs2071699 | 19:48751247 | A | missense_va | MODERATE | FUT1 | | | | | | 04-Apr - |
| 23 | rs2071699 | 19:48751247 | T | missense_va | MODERATE | FUT1 | | | | | | 04-Apr - |
| 26 | rs2071699 | 19:48751247 | A | missense_va | MODERATE | FUT1 | | | | | | 05-May - |
| 27 | rs2071699 | 19:48751247 | T | missense_va | MODERATE | FUT1 | | | | | | 05-May - |
| 28 | rs2071699 | 19:48751247 | A | missense_va | MODERATE | FUT1 | | | | | | 02-Feb - |
| 29 | rs2071699 | 19:48751247 | T | missense_va | MODERATE | FUT1 | | | | | | 02-Feb - |
| 47 | rs7683365 | 4:143999443 | A | missense_va | MODERATE | GYPB | | | | | | 04-Jun - |
| 48 | rs7683365 | 4:143999443 | C | missense_va | MODERATE | GYPB | | | | | | 04-Jun - |
| 49 | rs7683365 | 4:143999443 | T | missense_va | MODERATE | GYPB | | | | | | 04-Jun - |
| 50 | rs7683365 | 4:143999443 | A | missense_va | MODERATE | GYPB | | | | | | 03-May - |
| 51 | rs7683365 | 4:143999443 | C | missense_va | MODERATE | GYPB | | | | | | 03-May - |
| 52 | rs7683365 | 4:143999443 | T | missense_va | MODERATE | GYPB | | | | | | 03-May - |
| 53 | rs7683365 | 4:143999443 | A | missense_va | MODERATE | GYPB | | | | | | 03-May - |
| 54 | rs7683365 | 4:143999443 | C | missense_va | MODERATE | GYPB | | | | | | 03-May - |
| 55 | rs7683365 | 4:143999443 | T | missense_va | MODERATE | GYPB | | | | | | 03-May - |
| 56 | rs7683365 | 4:143999443 | A | missense_va | MODERATE | GYPB | | | | | | 04-Jun - |
| 57 | rs7683365 | 4:143999443 | C | missense_va | MODERATE | GYPB | | | | | | 04-Jun - |
| 58 | rs7683365 | 4:143999443 | T | missense_va | MODERATE | GYPB | | | | | | 04-Jun - |
| 59 | rs16879498 | 6:49612534 | - | missense_va | MODERATE | RHAG | | | | | | 06-Oct - |
| 60 | rs16879498 | 6:49612534 | - | downstream | MODIFIER | RHAG | | | | | | - |

Sort

Ascending Descending

By colour: None

Filter

By colour: None

Choose One

Search

☐ CYP2D6

☐ FGF21

☒ FUT1

☒ GYPB

☐ IZUMO1

☐ KCNQ1

☐ LOC105372093

☐ NDUFA6-DT

☒ RHAG

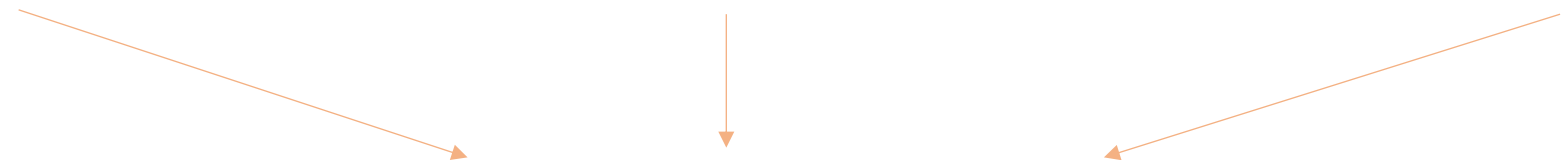
☒ SLC14A1

Clear Filter

Solution – rs2298719, rs2071699, rs7683365 and rs16879498

Exercise – 3 : Task 2 Solution

| Variant | Zygosity | Associated Phenotype |
|-------------------|-------------------------|----------------------|
| Chr17:42328621G>A | Homozygous (1/1) | Di(a+) |



Interpretation Two copies of alternate allele (**Presence of only alternate antigen**)

Proposed genotype **DI*01/DI*01**

 Alternate allele name

Proposed phenotype **Di(a+b-)**

Exercise – 4 : Task 4 Solution

| | | |
|----------------------|---------------------------------------|---------------------------------|
| Variant | Chr7:30951658C>T | Chr12:14993439T>C |
| Zygosity | Heterozygous (0/1) | Homozygous (1/1) |
| Associated Phenotype | <i>Co(b+)</i> | <i>Do(b+)</i> |
| Interpretation | One reference + One alternate antigen | Two copies of alternate antigen |
| Proposed genotype | CO*01/CO*02 | DO*02/DO*02 |
| Proposed phenotype | Co(a+b+) | Do(a-b+) |