## **Variant Effect Predictor results**

## Job details

Job summary VEP analysis of group5 ngs zain chr15 in Homo sapiens

Assembly GRCh38

Options summary 1000 Genomes continental allele frequencies: Enabled

1000 Genomes global minor allele frequency: Enabled

Ancestral allele<sup>(p)</sup>:

APPRIS: Enabled

BLOSUM62<sup>(p)</sup>: Enabled

Buffer size: 5000

**DisGeNET**<sup>(p)</sup>: Enabled

Filter by frequency: Disabled

Find co-located known variants: Enabled

Gene Ontology<sup>(p)</sup>:

Gene symbol: Enabled

gnomAD (exomes) allele frequencies: Enabled

MANE: Enabled

Phenotypes<sup>(p)</sup>: Enabled

PolyPhen: Prediction and score

Protein: Enabled

Protein matches: Enabled

PubMed IDs for citations of co-located variants: Enabled

Get regulatory region consequences: Yes

Restrict results: Disabled

Right align variants prior to consequence calculation: Disabled

SIFT: Prediction and score

Transcript biotype: Enabled

Transcript database to use: Ensembl transcripts

Transcript support level: Enabled

Transcript version: Enabled

UniProt: Enabled

Upstream/Downstream distance (bp): 5000

(p) = functionality from <u>VEP plugin</u>

VEP and data version

**1000genomes** phase3

Assembly GRCh38.p14

Cache 111\_GRCh38

**ClinVar** 202306

COSMIC 98

Database homo\_sapiens\_core\_111\_

**dbSNP** 156

**GENCODE** GENCODE 45

Genebuild 2014-07

gnomADe r2.1.1

gnomADg v3.1.2

HGMD-PUBLIC 20204

Polyphen 2.2.3

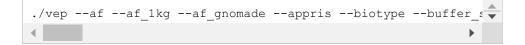
Regbuild 1.0

**SIFT** 6.2.1

Time 2024-03-13 15:54:17

**VEP** v111

Command line equivalent



Loading component

## Loading component

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