

SnpEff: Variant analysis

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Summary

|                                      |   |
|--------------------------------------|---|
| Genome                               | reference_db  |
| Date                                 | 2025-12-06 23:21  |
| SnpEff version                       | SnpEff 5.2b (build 2023-11-28 15:56), by Pablo Cingolani                                      |
| Command line arguments               | SnpEff -o results/variant/vcf/SnpEff.html reference_db results/variants/filtered_variants.vcf |
| Warnings                             | 0   |
| Errors                               | 0   |
| Number of lines (input file)         | 13  |
| Number of variants (before filter)   | 14  |
| Number of non-variants               | 0   |
| (i.e. reference equals alternative)  |   |
| Number of variants processed         | 14  |
| (i.e. after filter and non-variants) |   |
| Number of known variants             | 0 (0%)  |
| (i.e. non-empty ID)                  |   |
| Number of multi-allelic VCF entries  | 1   |
| (i.e. more than two alleles)         |   |
| Number of annotations                | 15  |
| Genome total length                  | 13,455  |
| Genome effective length              | 13,455  |
| Variant rate                         | 1 variant every 961 bases   |

Variants rate details

| Chromosome  | Length | Variants | Variant rate |
|-------------|--------|----------|--------------|
| hg_007457.2 | 13,455 | 14       | 961          |
| Total       | 13,455 | 14       |              |

Number variants by type

| Type     | Total |
|----------|-------|
| SNP      | 14    |
| MNP      | 0     |
| INS      | 0     |
| DEL      | 0     |
| MIXED    | 0     |
| INV      | 0     |
| DUP      | 0     |
| END      | 0     |
| INTERVAL | 0     |
| Total    | 14    |

Number of effects by impact

| Type (alphabetical order) | Count | Percent |
|---------------------------|-------|---------|
| MODERATE                  | 1     | 0.25%   |
| MODIFIER                  | 13    | 67.50%  |

Number of effects by functional class

| Type (alphabetical order) | Count | Percent |
|---------------------------|-------|---------|
| MISSENSE                  | 1     | 100%    |

Missense / Silent ratio: 0

Number of annotations and region counts

| Annotation                |       |         | Region                    |       |         |
|---------------------------|-------|---------|---------------------------|-------|---------|
| Type (alphabetical order) | Count | Percent | Type (alphabetical order) | Count | Percent |
| downstream_gene_variant   | 1     | 0.25%   | DOWNSTREAM                | 1     | 0.25%   |
| intergenic_region         | 1     | 0.25%   | EXON                      | 1     | 0.25%   |
| intragenic_variant        | 1     | 0.25%   | INTERGENIC                | 1     | 0.25%   |
| intron_variant            | 1     | 0.25%   | INTRON                    | 1     | 0.25%   |
| missense_variant          | 1     | 0.25%   | TRANSCRIPT                | 1     | 0.25%   |
| upstream_gene_variant     | 1     | 0.25%   | UPSTREAM                  | 1     | 0.25%   |

Quality:

|                    |   |
|--------------------|---|
| Min                | 32  |
| Max                | 295                                       |
| Mean               | 95.615                                    |
| Median             | 75  |
| Standard deviation | 67.778                                    |
| Values             | 32,41,45,54,61,70,102,109,115,121,122,295 |
| Count              | 1,1,1,1,1,1,1,1,1,1,1                     |

Insertions and deletions length:

|                    |     |
|--------------------|-----|
| Min                | 1   |
| Max                | 1   |
| Mean               | 1   |
| Median             | 0.5 |
| Standard deviation | 0   |
| Values             | 1   |
| Count              | 1   |

Base changes (SNPs)

|   |   |   |   |   |
|---|---|---|---|---|
|   | A | C | G | T |
| A | 0 | 0 | 0 | 0 |
| C | 0 | 0 | 0 | 1 |
| G | 0 | 0 | 0 | 0 |
| T | 0 | 1 | 0 | 0 |

TbTv (transitions / transversions)

Note: Only SNPs are used for this statistic.  
Note: This TbTv ratio is a 'raw' ratio (ratio of observed events).

|               |      |
|---------------|------|
| Transitions   | 12   |
| Transversions | 2    |
| TbTv ratio    | 3.25 |

All variants:

Sample , sample1, Total  
Transitions : 12, 12  
Transversions : 2, 2  
TbTv : 3.250, 3.250

Only known variants (i.e. the ones having a non-empty ID field):  
No results available (empty input?)

Allele frequency

|                    |        |
|--------------------|--------|
| Min                | 50     |
| Max                | 100    |
| Mean               | 73.077 |
| Median             | 50     |
| Standard deviation | 25.944 |
| Values             | 50,100 |
| Count              | 7,6    |

Allele Count

|                    |       |
|--------------------|-------|
| Min                | 1     |
| Max                | 2     |
| Mean               | 1.462 |
| Median             | 1     |
| Standard deviation | 0.519 |
| Values             | 1,2   |
| Count              | 7,6   |

Hom/Het per sample

sample\_names , sample1  
Reference , 0  
Het , 1  
Hom , 6  
Missing , 0

Codon changes

How to read this table:  
- Rows are reference codons and columns are changed codons. E.g. Row 'AAA' column 'TAA' indicates how many 'AAA' codons have been replaced by 'TAA' codons.  
- Red background colors indicate that more changes happened (heat-map).  
- Diagonals are indicated using grey background color  
- WARNING: This table may include different translation codon tables (e.g. mammalian DNA and mitochondrial DNA).

|     |     |     |
|-----|-----|-----|
|     | ACC | GCC |
| ACC |     |     |
| GCC | 1   |     |

Amino acid changes

How to read this table:  
- Rows are reference amino acids and columns are changed amino acids. E.g. Row 'A' column 'E' indicates how many 'A' amino acids have been replaced by 'E' amino acids.  
- Red background colors indicate that more changes happened (heat-map).  
- Diagonals are indicated using grey background color  
- WARNING: This table may include different translation codon tables (e.g. mammalian DNA and mitochondrial DNA).

|   |   |   |
|---|---|---|
|   | A | T |
| A |   |   |
| T |   |   |

Variants by chromosome

Details by gene

[Here](#) you can find a sub-segregated table.