SnpEff: Variant analysis

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Summary

Genome	reference_db
Date	2025-12-06 19:33
SnpEff version	SnpEff 5.1d (build 2022-04-19 15:49), by Pablo Cingolani
Command line arguments	SnpEff -stats KMT2D/results/snpEff/snpEff.html reference_db KMT2D/results/variants/filtered_variants.vcf
Warnings	1
Errors	0
Number of lines (input file)	32
Number of variants (before filter)	32
Number of not variants (i.e. reference equals alternative)	0
Number of variants processed (i.e. after filter and non-variants)	32
Number of known variants (i.e. non-empty ID)	0 (0%)
Number of multi-allelic VCF entries (i.e. more than two alleles)	0
Number of effects	32
Genome total length	43,350
Genome effective length	43,350
Variant rate	1 variant every 1,354 bases

Variants rate details

Chromosome	Length	Variants	Variants rate
NG_027827.1	43,350	32	1,354
Total	**43,350**	**32**	**1,354**

Number variants by type

Type	Total
SNP	29
MNP	0
INS	1
DEL	2
MIXED	0
INV	0
DUP	0
BND	0
INTERVAL	0
Total	**32**

Number of effects by impact

Type (alphabetical order)	Count	Percent
MODIFIER	32	100%

Number of effects by functional class

Type (alphabetical order)	Count	Percent

Missense / Silent ratio: 0

Number of effects by type and region

Type

Type (alphabetical order)	Count	Percent
intron_variant	32	100%

Region

Type (alphabetical order)	Count	Percent
INTRON	32	100%

Quality:

Min	31
Max	922
Mean	156.125
Median	100
Standard deviation	180.549
Values	31,33,35,42,49,55,61,64,80,100,103,130,133,145,154,204,229,233,268,279,283,330,510,922
Count	5,1,3,1,1,1,1,1,1,2,1,1,1,1,1,1,2,1,1,1,1,1,1,1

Insertions and deletions length:

Min	0
Max	1
Mean	0.667
Median	0
Standard deviation	0.577
Values	0,1
Count	1,2

Base changes (SNPs)

	A	C	G	T
A	0	0	8	0
C	0	0	1	6
G	3	1	0	1
T	1	7	1	0

Ts/Tv (transitions / transversions)

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

Transitions	29
Transversions	10
Ts/Tv ratio	2.9

All variants:

```
Sample ,sample1,Total
Transitions ,29,29
Transversions ,10,10
Ts/Tv ,2.900,2.900
```

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	68.75
Median	50
Standard deviation	24.593
Values	50,100
Count	20,12

Allele Count

Min	1
Max	2
Mean	1.375
Median	1
Standard deviation	0.492
Values	1,2
Count	20,12

Hom/Het per sample

```
Sample_names , sample1
Reference , 0
Het , 20
Hom , 12
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. mamalian DNA and mitochondrial DNA).

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. mamalian DNA and mitochondrial DNA).

Variants by chromosome

```
NG_027827.1, Position,0,1000,2000,3000,4000,5000,6000,7000,8000,9000,10000,11000,12000,13000,14000,15000,16000,17000,18000,19000,20000,21000,22000,23000,24000
NG_027827.1,Count,0,0,0,0,0,0,0,0,0,0,0,8,9,4,0,0,0,0,0,0,0,5,5,0,0,0,0,0,0,0,1,0,0,0,0,0,0,0,0,0,0,0
```

Details by gene

Here you can find a tab-separated table.