

SnpEff: Variant analysis

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Summary

|  |  |
|--|--|
| Genome   | vitis  |
| Date   | 2021-06-06 00:28   |
| SnpEff version   | SnpEff 5.0e (build 2021-03-09 06:01), by Pablo Cingolani |
| Command line arguments   | SnpEff vitis SNPs_matrixeqtls.recode.vcf                 |
| Warnings   | 12,719   |
| Errors   | 0  |
| Number of lines (input file)   | 7,842  |
| Number of variants (before filter)                                   | 7,931  |
| Number of not variants<br>(i.e. reference equals alternative)        | 0  |
| Number of variants processed<br>(i.e. after filter and non-variants) | 7,931  |
| Number of known variants<br>(i.e. non-empty ID)                      | 0 ( 0% )   |
| Number of multi-allelic VCF entries<br>(i.e. more than two alleles)  | 89   |
| Number of effects  | 20,219   |
| Genome total length  | 513,581,148  |
| Genome effective length  | 426,176,009  |
| Variant rate   | 1 variant every 53,735 bases                             |

Variants rate details

| Chromosome  | Length      | Variants | Variants rate |
|-------------|-------------|----------|---------------|
| NC_012007.3 | 23,037,639  | 143      | 161,102       |
| NC_012008.3 | 18,779,844  | 92       | 204,128       |
| NC_012009.3 | 19,341,862  | 361      | 53,578        |
| NC_012010.3 | 23,867,706  | 475      | 50,247        |
| NC_012011.3 | 25,021,643  | 572      | 43,744        |
| NC_012012.3 | 21,508,407  | 291      | 73,912        |
| NC_012013.3 | 21,026,613  | 300      | 70,088        |
| NC_012014.3 | 22,385,789  | 402      | 55,686        |
| NC_012015.3 | 23,006,712  | 433      | 53,133        |
| NC_012016.3 | 18,140,952  | 282      | 64,329        |
| NC_012017.3 | 19,818,926  | 626      | 31,659        |
| NC_012018.3 | 22,702,307  | 676      | 33,583        |
| NC_012019.3 | 24,396,255  | 376      | 64,883        |
| NC_012020.3 | 30,274,277  | 352      | 86,006        |
| NC_012021.3 | 20,304,914  | 538      | 37,741        |
| NC_012022.3 | 22,053,297  | 443      | 49,781        |
| NC_012023.3 | 17,126,926  | 311      | 55,070        |
| NC_012024.3 | 29,360,087  | 684      | 42,924        |
| NC_012025.3 | 24,021,853  | 574      | 41,849        |
| Total       | 426,176,009 | 7,931    | 53,735        |

Number variants by type

| Type     | Total |
|----------|-------|
| SNP      | 7,431 |
| MNP      | 0     |
| INS      | 253   |
| DEL      | 247   |
| MIXED    | 0     |
| INV      | 0     |
| DUP      | 0     |
| BND      | 0     |
| INTERVAL | 0     |
| Total    | 7,931 |

Number of effects by impact

| Type (alphabetical order) | Count  | Percent |
|---------------------------|--------|---------|
| HIGH                      | 181    | 0.895%  |
| LOW                       | 506    | 2.503%  |
| MODERATE                  | 875    | 4.328%  |
| MODIFIER                  | 18,657 | 92.275% |

Number of effects by functional class

| Type (alphabetical order) | Count | Percent |
|---------------------------|-------|---------|
| MISSENSE                  | 902   | 65.936% |
| NONSENSE                  | 72    | 5.263%  |
| SILENT                    | 394   | 28.801% |

Missense / Silent ratio: 2.2893

Number of effects by type and region

| Type   |       |         | Region                    |       |         |
|--|-------|---------|---------------------------|-------|---------|
| Type (alphabetical order)                      | Count | Percent | Type (alphabetical order) | Count | Percent |
| 3_prime_UTR_variant                            | 209   | 1.028%  | DOWNSTREAM                | 5,508 | 27.242% |
| 5_prime_UTR_premature_start_codon_gain_variant | 27    | 0.133%  | EXON                      | 1,557 | 7.701%  |
| 5_prime_UTR_variant                            | 157   | 0.772%  | INTERGENIC                | 2,596 | 12.839% |
| conservative_inframe_deletion                  | 9     | 0.044%  | INTRON                    | 4,772 | 23.602% |
| conservative_inframe_insertion                 | 3     | 0.015%  | SPLICE_SITE_ACCEPTOR      | 8     | 0.04%   |
| disruptive_inframe_deletion                    | 5     | 0.025%  | SPLICE_SITE_DONOR         | 5     | 0.025%  |
| disruptive_inframe_insertion                   | 3     | 0.015%  | SPLICE_SITE_REGION        | 94    | 0.465%  |
| downstream_gene_variant                        | 5,508 | 27.08%  | UPSTREAM                  | 5,286 | 26.144% |
| frameshift_variant                             | 49    | 0.241%  | UTR_3_PRIME               | 209   | 1.034%  |
| intergenic_region                              | 2,596 | 12.763% | UTR_5_PRIME               | 184   | 0.91%   |
| intron_variant                                 | 4,853 | 23.859% |                           |       |         |
| missense_variant                               | 857   | 4.213%  |                           |       |         |
| non_coding_transcript_exon_variant             | 131   | 0.644%  |                           |       |         |
| splice_acceptor_variant                        | 8     | 0.039%  |                           |       |         |
| splice_donor_variant                           | 5     | 0.025%  |                           |       |         |
| splice_region_variant                          | 115   | 0.565%  |                           |       |         |
| start_lost                                     | 4     | 0.02%   |                           |       |         |
| stop_gained                                    | 77    | 0.379%  |                           |       |         |
| stop_lost                                      | 44    | 0.216%  |                           |       |         |
| stop_retained_variant                          | 19    | 0.093%  |                           |       |         |
| synonymous_variant                             | 375   | 1.844%  |                           |       |         |
| upstream_gene_variant                          | 5,286 | 25.988% |                           |       |         |



Quality:

|                    |   |
|--------------------|---|
| Min                | 19  |
| Max                | 999   |
| Mean               | 547.31  |
| Median             | 473   |
| Standard deviation | 350.582   |
| Values             | 19,20,21,22,23,24,25,26,27,28,29,30,31,32,33,34,35,36,37,38,39,40,41,42,43,44,45,46,47,48,49,50,51,52,53,54,55,56,57,58,59,60,61,62,63,64,65,66       |
| Count              | 13,9,13,12,10,12,19,10,10,10,11,8,6,8,15,9,11,13,8,12,11,10,5,10,14,7,11,12,12,7,15,5,10,12,9,6,10,5,17,12,10,9,8,8,11,4,7,10,5,11,9,9,14,10,9,10,9,8 |



Insertions and deletions length:

|                    |  |
|--------------------|--|
| Min                | 0  |
| Max                | 24   |
| Mean               | 2.09   |
| Median             | 1  |
| Standard deviation | 3.113  |
| Values             | 0,1,2,3,4,5,6,7,8,9,10,11,13,14,16,17,20,22,24 |
| Count              | 80,285,36,21,12,13,11,12,5,7,4,1,3,5,1,1,1,1,1 |



Base changes (SNPs)

|   | A   | C   | G     | T     |
|---|-----|-----|-------|-------|
| A | 0   | 296 | 1,043 | 343   |
| C | 304 | 0   | 213   | 1,462 |

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snpEff\_summary.html

|   |       |       |     |     |
|---|-------|-------|-----|-----|
| G | 1,513 | 198   | 0   | 360 |
| T | 371   | 1,043 | 285 | 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   | 34,206 |
| Transversions | 15,848 |
| Ts/Tv ratio   | 2.1584 |

All variants:

Sample , ../align\_stepwise/Barberanera\_mkdup.bam, ../align\_stepwise/CabernetSauvignon\_mkdup.bam, ../align\_stepwise/Garganega\_mkdup.bam, .  
Transitions ,2808,3401,3059,3223,3293,3030,2907,3173,3148,3204,2960,34206  
Transversions ,1340,1596,1375,1454,1533,1394,1335,1501,1494,1454,1372,15848  
Ts/Tv ,2,096,2,131,2,225,2,217,2,148,2,174,2,178,2,114,2,107,2,204,2,157,2,158

Only known variants (i.e. the ones having a non-empty ID field):

No results available (empty input?)

Allele frequency

Min

Max

Mean

Median

Standard deviation

Values

Count

5

95

30.486

18

27.778

5,6,8,9,10,11,12,13,15,16,18,20,22,25,27,30,31,33,35,36,40,45,50,54,55,59,60,61,63,65,66,68,70,72,75,77,80,81,83,85,86,90,91,92,94,95

9,1,1,2056,81,14,1,1435,90,9,804,25,444,33,306,18,231,2,9,138,206,216,106,161,2,53,2,3,72,3,3,150,7,60,3,53,11,208,1,12,119,254,1,1,1,427

Allele Count

Min

Max

Mean

Median

Standard deviation

Values

Count

1

21

6.754

4

6.105

1,2,3,4,5,6,7,8,9,10,11,12,13,14,15,16,17,18,19,20,21

11,2152,1540,824,478,324,240,144,205,216,107,167,59,83,153,70,62,219,131,242,415

Hom/Het per sample

Sample\_names , ../align\_stepwise/Barberanera\_mkdup.bam, ../align\_stepwise/CabernetSauvignon\_mkdup.bam, ../align\_stepwise/Garganega\_mkdup.bam, .  
Reference , 4546, 3689, 4427, 4105, 3828, 4320, 4540, 3962, 4135, 4258, 4086  
Het , 2133, 2713, 2129, 2541, 2941, 2197, 2125, 2749, 2337, 2242, 2702  
Hom , 1134, 1292, 1283, 1195, 1073, 1235, 1175, 1096, 1297, 1341, 958  
Missing , 29, 148, 3, 1, 0, 90, 2, 35, 73, 1, 96

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).

|     |   |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     |
|-----|---|-----|-----|-----|-----|-----|-----|-----|-----|-----|-----|-----|-----|-----|-----|-----|-----|-----|-----|-----|-----|-----|-----|
|     | - | AAA | AAC | AAG | AAT | ACA | ACC | ACG | ACT | AGA | AGC | AGG | AGT | ATA | ATC | ATG | ATT | CAA | CAC | CAG | CAT | CCA | CCC |
| -   |   | 4   | 2   |     |     |     |     |     |     |     |     |     | 2   | 2   |     |     | 1   |     |     |     |     |     |     |
| AAA | 3 | 1   | 1   | 7   | 1   | 2   |     |     |     | 8   |     |     |     | 3   |     |     |     | 1   |     |     |     |     |     |
| AAC | 2 | 1   |     | 1   | 7   |     | 1   |     |     | 1   | 4   |     |     |     | 1   |     |     |     | 1   |     |     |     |     |
| AAG | 3 | 11  | 4   |     | 4   |     |     |     |     |     |     | 2   |     |     |     | 1   |     |     |     | 1   |     |     |     |
| AAT | 4 | 2   | 3   | 1   |     |     |     |     | 2   |     |     |     | 7   |     |     |     | 4   |     |     |     | 1   |     |     |
| ACA | 1 | 2   |     |     |     | 1   |     | 2   | 4   | 1   |     |     |     | 4   |     |     |     |     |     |     |     | 2   |     |
| ACC |   |     |     |     |     | 1   |     | 1   | 1   |     | 1   |     |     |     | 8   |     |     |     |     |     |     |     |     |
| ACG |   |     |     |     |     | 5   |     |     | 3   |     |     | 1   |     |     |     | 6   |     |     |     |     |     |     |     |
| ACT |   |     |     |     |     |     | 3   | 1   |     |     |     |     | 1   |     |     |     | 6   |     |     |     |     |     |     |
| AGA | 2 | 7   |     |     |     | 3   |     |     |     | 3   | 3   | 4   | 2   | 3   |     |     |     |     |     |     |     |     |     |
| AGC |   |     | 4   |     |     |     |     |     |     | 1   |     |     | 6   |     | 2   |     |     |     |     |     |     |     |     |
| AGG | 1 |     |     | 5   |     |     |     |     |     | 8   | 1   |     | 2   |     |     | 4   |     |     |     |     |     |     |     |
| AGT | 2 |     |     |     | 3   |     |     |     | 1   | 4   |     | 2   | 1   |     |     |     | 1   |     |     |     |     |     |     |
| ATA | 1 | 3   |     |     |     | 4   |     |     |     | 4   |     |     |     | 1   | 2   | 6   |     |     |     |     |     |     |     |
| ATC |   |     |     |     |     |     | 4   |     |     |     |     |     |     |     | 4   |     | 5   |     |     |     |     |     |     |
| ATG |   |     |     | 1   |     |     |     | 6   |     |     |     | 4   |     | 5   | 1   |     | 2   |     |     |     |     |     |     |
| ATT | 3 |     |     |     | 2   |     |     |     | 7   |     |     |     | 3   | 1   | 1   | 1   |     |     |     |     |     |     |     |

file:///Users/pedromartinez/Downloads/snpEff/snpEff\_summary.html

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|     | - | AAA | AAC | AAG | AAT | ACA | ACC | ACG | ACT | AGA | AGC | AGG | AGT | ATA | ATC | ATG | ATT | CAA | CAC | CAG | CAT | CCA | CCC |
|-----|---|-----|-----|-----|-----|-----|-----|-----|-----|-----|-----|-----|-----|-----|-----|-----|-----|-----|-----|-----|-----|-----|-----|
| CAA | 1 | 4   |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     | 3   |     | 2   |     |
| CAC | 1 |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     | 8   |     |     |
| CAG | 1 |     |     | 4   |     |     |     |     |     |     |     |     |     |     |     |     |     | 13  | 2   |     |     |     |     |
| CAT | 1 |     |     |     | 1   |     |     |     |     |     |     |     |     |     |     |     |     |     | 4   |     |     |     |     |
| CCA |   |     |     |     |     | 1   |     |     |     |     |     |     |     |     |     |     |     | 2   |     |     |     |     | 1   |
| CCC | 1 |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     | 1   |
| CCG |   |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     | 2   |     | 5   | 1   |
| CCT | 3 |     |     |     |     |     |     |     | 2   |     |     |     |     |     |     |     |     |     |     |     |     | 3   | 3   |
| CGA | 1 |     |     |     |     |     |     |     |     | 1   |     |     |     |     |     |     |     | 7   |     |     |     | 1   |     |
| CGC |   |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     | 1   |     |     |     | 1   |
| CGG | 1 |     |     |     |     |     |     |     |     |     |     | 1   |     |     |     |     |     |     |     | 6   |     |     |     |
| CGT |   |     |     |     |     |     |     |     |     |     |     |     | 1   |     |     |     |     |     |     |     | 6   |     |     |
| CTA | 3 |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     |
| CTC | 2 |     |     |     |     |     |     |     |     |     |     |     |     |     | 1   |     |     |     | 1   |     |     |     | 2   |
| CTG |   |     |     |     |     |     |     |     |     |     |     |     |     |     |     | 5   |     |     |     | 3   |     |     |     |
| CTT | 2 |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     | 5   |     |     |     | 1   |     |     |
| GAA | 4 | 13  |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     | 1   |     |     |     |     |     |
| GAC |   |     | 5   |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     |
| GAG |   |     |     | 10  |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     | 1   |     |     |     |
| GAT | 3 |     |     |     | 9   |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     | 2   |     |     |
| GCA | 1 |     |     |     |     | 10  |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     | 1   |     |
| GCC | 1 |     |     |     |     |     | 1   |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     | 1   |
| GCG |   |     |     |     |     |     |     | 1   |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     |
| GCT |   |     |     |     |     |     |     |     | 5   |     |     |     |     |     |     |     |     |     |     |     |     |     |     |
| GGA | 3 |     |     |     |     |     |     |     |     | 5   |     |     |     |     |     |     |     |     |     |     |     |     |     |
| GGC | 1 |     |     |     |     |     |     |     |     |     | 4   |     |     |     |     |     |     |     |     |     |     |     |     |
| GGG | 1 |     |     |     |     |     |     |     |     |     |     | 6   |     |     |     |     |     |     |     |     |     |     |     |
| GGT | 4 |     |     |     |     |     |     |     |     |     |     |     | 8   |     |     |     |     |     |     |     |     |     |     |
| GTA | 1 |     |     |     |     |     |     |     |     |     |     |     |     | 2   |     |     |     |     |     |     |     |     |     |
| GTC |   |     |     |     |     |     |     |     |     |     |     |     |     |     | 4   |     |     |     |     |     |     |     |     |
| GTG | 2 |     |     |     |     |     |     |     |     |     |     |     |     |     |     | 4   |     |     |     |     |     |     |     |
| GTT | 1 |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     | 11  |     |     |     |     |     |     |
| TAA | 1 | 3   |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     | 6   |     |     |     |     |     |
| TAC | 1 |     | 3   |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     | 2   |     |     |     |     |
| TAG | 2 |     |     | 2   |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     | 2   |     |     |     |
| TAT | 1 |     |     |     | 2   |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     | 3   |     |     |
| TCA |   |     |     |     |     | 3   |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     | 7   |     |
| TCC | 1 |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     | 1   |     | 2   |
| TCG |   |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     |
| TCT | 1 |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     |
| TGA | 2 |     |     |     |     |     |     |     |     | 1   |     |     |     |     |     |     |     |     |     |     |     |     |     |
| TGC |   |     |     |     |     |     |     |     |     |     | 1   |     |     |     |     |     |     |     |     |     |     |     |     |
| TGG | 2 |     |     |     |     |     |     |     |     |     |     | 3   |     |     |     |     |     |     |     |     |     |     |     |
| TGT | 2 |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     |
| TTA | 4 |     |     |     |     |     |     |     |     |     |     |     |     | 1   |     |     |     |     |     |     |     |     |     |
| TTC | 3 |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     |
| TTG | 1 |     |     |     |     |     |     |     |     |     |     |     |     |     |     | 1   |     |     |     |     |     |     |     |
| TTT | 3 |     |     |     |     |     |     |     |     |     |     |     |     |     |     |     | 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. mamalian DNA and mitochondrial DNA).

|   | *  | -  | ?  | A  | C  | D | E  | F  | G  | H  | I  | K  | L  | M | N  | P  | Q | R  | S  | T  | V  | W | Y  |
|---|----|----|----|----|----|---|----|----|----|----|----|----|----|---|----|----|---|----|----|----|----|---|----|
| * | 20 | 5  |    |    | 3  |   |    |    | 3  |    |    | 5  | 4  |   |    |    | 8 | 6  | 6  |    |    | 2 | 4  |
| - | 1  |    | 25 | 2  | 1  |   |    | 3  | 1  |    | 3  | 4  | 3  |   | 2  |    |   | 2  | 4  |    | 1  |   | 4  |
| ? |    |    | 1  |    |    |   |    |    |    |    |    |    |    |   |    |    |   |    |    |    |    |   |    |
| A |    | 2  |    | 20 |    |   | 5  |    | 3  |    |    |    |    |   |    | 2  |   |    | 6  | 17 | 24 |   |    |
| C | 3  | 2  |    |    | 14 |   |    | 1  |    |    |    |    |    |   |    |    |   | 7  | 1  |    |    | 2 | 13 |
| D |    | 3  |    | 1  |    | 8 | 5  |    | 8  | 2  |    |    |    |   | 14 |    |   |    |    |    | 5  |   | 3  |
| E | 4  | 4  |    | 2  |    | 3 | 11 |    | 4  |    |    | 23 |    |   |    |    | 2 |    |    |    | 1  |   |    |
| F |    | 6  |    |    |    |   |    | 12 |    |    | 1  |    | 17 |   |    |    |   |    | 13 |    | 3  |   | 3  |
| G | 1  | 9  |    | 5  | 2  | 9 | 6  |    | 26 |    |    |    |    |   |    |    |   | 15 | 12 |    | 7  | 1 |    |
| H |    | 2  |    |    |    | 2 |    |    |    | 12 |    |    | 3  |   | 1  | 1  |   |    |    |    |    |   | 7  |
| I |    | 4  |    |    |    |   |    | 1  |    |    | 14 | 3  | 6  | 7 | 2  |    |   | 4  | 3  | 15 | 17 |   |    |
| K | 3  | 6  |    |    |    |   | 11 |    |    |    | 3  | 19 |    | 1 | 10 |    | 2 | 10 |    | 2  |    |   |    |
| L | 5  | 12 |    | 2  |    |   |    | 16 |    | 2  | 7  |    | 68 | 6 |    | 11 | 3 | 2  | 7  |    | 10 | 1 |    |
| M |    |    |    |    |    |   |    |    |    |    | 8  | 1  | 3  |   |    |    |   | 4  |    | 6  | 11 |   |    |
| N |    | 6  |    |    |    | 6 |    |    |    | 2  | 5  | 5  |    |   | 10 |    |   | 1  | 11 | 3  |    |   | 3  |





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[Here](#) you can find a tab-separated table.