

SnEff: Variant analysis

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Summary

Genome	hg38
Date	2025-01-16 14:48
SnEff version	SnEff 4.3t (build 2017-11-24 10:18), by Pablo Cingolani
Command line arguments	SnEff -i vcf -o vcf -stats /data/jwd05e/main/077/887/77887124/outputs/dataset_d68aa172-dd4d-4d5f-9bc1-8c0cfaea8bd1 hg38 /data/dnb10/galaxy_db/files/4/2/d/dataset_42d298d7-f61a-4da3-9447-e677e7bc4cde.dat
Warnings	912
Errors	0
Number of lines (input file)	16,988
Number of variants (before filter)	16,991
Number of not variants (i.e. reference equals alternative)	0
Number of variants processed (i.e. after filter and non-variants)	16,991
Number of known variants (i.e. non-empty ID)	0 (0%)
Number of multi-allelic VCF entries (i.e. more than two alleles)	3
Number of effects	41,944
Genome total length	3,209,286,106
Genome effective length	3,033,239,033
Variant rate	1 variant every 178,520 bases

Variants rate details

Chromosome	Length	Variants	Variants rate
1	248,956,422	1,860	133,847
2	242,193,529	1,471	164,645
3	198,295,559	1,059	187,247
4	190,214,555	837	227,257
5	181,538,259	743	244,331
6	170,805,979	754	226,533
7	159,345,973	719	221,621
8	145,138,636	565	256,882
9	138,394,717	730	189,581
10	133,797,422	769	173,988
11	135,086,622	1,044	129,393
12	133,275,309	861	154,791
13	114,364,328	405	282,381
14	107,043,718	564	189,793
15	101,991,189	515	198,041
16	90,338,345	607	148,827
17	83,257,441	769	108,267
18	80,373,285	331	242,819
19	58,617,616	852	68,800
20	64,444,167	424	151,990
21	46,709,983	234	199,615
22	50,818,468	321	158,312
14_GL000225v1_random	211,173	1	211,173
17_GL000205v2_random	185,591	2	92,795
1_KI270766v1_alt	256,271	2	128,135
21_KI270874v1_alt	166,743	1	166,743
3_KI270779v1_alt	205,312	1	205,312
3_KI270935v1_alt	197,351	2	98,675
4_GL000008v2_random	209,709	3	69,903
9_KI270719v1_random	176,845	1	176,845
M	16,569	9	1,841
Un_KI270438v1	112,505	3	37,501
Un_KI270442v1	392,061	3	130,687
Un_KI270746v1	66,486	2	33,243
X	156,040,895	527	296,092
Total	3,033,239,033	16,991	178,520

Number variants by type

Type	Total
SNP	16,186
MNP	0
INS	400
DEL	404
MIXED	1
INV	0
DUP	0
BND	0
Total	16,991

Type	Total
INTERVAL	0
Total	16,991

Number of effects by impact

Type (alphabetical order)	Count	Percent
HIGH	1,213	2.892%
LOW	8,501	20.267%
MODERATE	5,860	13.971%
MODIFIER	26,370	62.87%

Number of effects by functional class

Type (alphabetical order)	Count	Percent
MISSENSE	5,694	45.288%
NONSENSE	62	0.493%
SILENT	6,817	54.219%

Missense / Silent ratio: 0.8353

Number of effects by type and region

Type			Region		
Type (alphabetical order)	Count	Percent			
3_prime_UTR_variant	1,373	3.164%			
5_prime_UTR_premature_start_codon_gain_variant	124	0.286%			
5_prime_UTR_variant	645	1.486%			
conservative_inframe_deletion	14	0.032%			
conservative_inframe_insertion	21	0.048%			
disruptive_inframe_deletion	19	0.044%			
disruptive_inframe_insertion	13	0.03%			
frameshift_variant	48	0.111%			
initiator_codon_variant	2	0.005%			
intergenic_region	1,823	4.201%			
intron_variant	22,500	51.854%			
missense_variant	5,682	13.095%			
non_coding_transcript_exon_variant	1,236	2.849%			
protein_protein_contact	63	0.145%			
sequence_feature	498	1.148%			
splice_acceptor_variant	26	0.06%			
splice_donor_variant	37	0.085%			
splice_region_variant	1,409	3.247%			
start_lost	8	0.018%			
stop_gained	62	0.143%			
stop_lost	3	0.007%			
stop_retained_variant	1	0.002%			
structural_interaction_variant	968	2.231%			
synonymous_variant	6,816	15.708%			

Type (alphabetical order)	Count	Percent
EXON	14,804	35.295%
INTERGENIC	1,823	4.346%
INTRON	21,319	50.827%
SPLICE_SITE_ACCEPTOR	26	0.062%
SPLICE_SITE_DONOR	36	0.086%
SPLICE_SITE_REGION	1,296	3.09%
TRANSCRIPT	498	1.187%
UTR_3_PRIME	1,373	3.273%
UTR_5_PRIME	769	1.833%



Quality:

Min	30
Max	998
Mean	77.223
Median	58
Standard deviation	48.853
Values	30,31,32,33,34,35,36,37,38,39,40,41,42,43,44,45,46,47,48,49,50,51,52,53,54,55,56,57,58,59,60,61,62,63,64,65,66,67,68,69,70,71,72,73,74,75,76,77,78,79,80,81,82,83
Count	161,230,179,203,132,143,99,105,122,108,114,125,132,153,149,191,226,235,246,311,382,435,756,614,590,875,816,526,459,607,317,395,56,47,40,51,42,58,63,61,47,6



Insertions and deletions length:

Min	0
Max	11
Mean	0.869
Median	1
Standard deviation	0.914
Values	0,1,2,3,4,5,6,8,11
Count	252,471,47,19,8,4,1,1,1



Base changes (SNPs)

	A	C	G	T
A	0	522	2,934	415
C	554	0	724	2,787
G	2,829	777	0	608
T	432	3,050	554	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	21,392
Transversions	8,515
Ts/Tv ratio	2.5123

```
Sample ,unknown,Total
Transitions ,21392,21392
Transversions ,8515,8515
Ts/Tv ,2.512,2.512
```

No results available (empty input?)

Min	0
Max	100
Mean	92.542
Median	100
Standard deviation	17.92
Values	0,50,100
Count	13.2508,14467

Min	0
Max	2
Mean	1.851
Median	2
Standard deviation	0.358
Values	0,1,2
Count	13,2508,14467


```
Sample_names , unknown
Reference , 13
Het , 2508
Hom , 14467
Missing , 0
```

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	CCG	CCT	CGA
-				6	1			3				4		4	2			3		3						
AAA	1		5	77	14	7				46				2				11								
AAC		10		3	169		10				37				7				1							
AAG	7	82	13	6	12			6				43				5				5						
AAT	4	15	125	8					7				50				4				18					
ACA		6					18	161	10	7			3	41								9				
ACC			8			18		20	108		11				27								14			
ACG				9		181	13		12			6				70										
ACT	9				3	22	121	15	2				25				31							6		
AGA		24				7					4	36	8	1											9	
AGC			42				12			6		9	110		2											
AGG				21				3		40	4		9			1										
AGT	7				41				14	2	85	4					5									
ATA		3				38				1					20	18	6									
ATC			5				26		2		1			16		2	118									
ATG	3			5				71						33	9		14									
ATT					3				20				4	10	94	4										
CAA	5	5																	4	91	4	7			25	
CAC			9															2		27	109		10			
CAG				12															5		20			19		
CAT					1														81	8						
CCA	1					10												3				38	173	4	1	
CCC	2						11												5			28	10	111		
CCG																				2		162	12	8		
CCT	1								4												7	9	98	6	2	
CGA										8								52				6				
CGC	1																		57			1			4	
CGG												23								109			1		25	
CGT	1												2								50			11	7	
CTA	1													4				3				17			2	
CTC	2														3				2			10				
CTG																27				5			49			
CTT																	2				1			24		
GAA	2	23																7								
GAC	1		26																11							
GAG	1			34																36						
GAT	4				19																10					
GCA						39																9				
GCC	2						54																6			
GCG								13																2		
GCT									44																24	
GGA	1									15															6	
GGC	1										59	1														

-	AAA	AAC	AAG	AAT	ACA	ACC	ACG	ACT	AGA	AGC	AGG	AGT	ATA	ATC	ATG	ATT	CAA	CAC	CAG	CAT	CCA	CCC	CCG	CCT	CGA
GGG											28														
GGT												25													
GTA													55												
GTC														96											
GTG															96				5						
GTT	1															62									
TAA																	1								
TAC			1															10							
TAG																									
TAT				6																15					
TCA					3																15				
TCC	1					5																22			
TCG																							2		
TCT								7																24	
TGA																									
TGC										1															
TGG											3														
TGT	3											5													
TTA	1												5												
TTC	1													1											
TTG															11										
TTT	5															4									

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
*	1																1						2
-			14			1	4				6	6	3		1		6	4	10	3	5		
?																							
A		2		633		19	18		29							41			34	150	124		
C	6	3		111				6	3									59	25			10	22
D	5		22		473	84		42	21						45						23		5
E	7	3		14		99	242		44			57					43				18		
F		6			14			129			5		55						38		5		9
G		2		16	10	38	28		423									62	84		21	1	
H						10			190				4		10	10	37	94					24
I								6			264	3	17	24	8			1	5	86	190		
K		8					98			2	165		5	44		16	89		13				
L	1	4						44		3	14		782	38		100	8	16	32		61		
M		3								56	5	35								71	100		
N		4				61				19	11	36			294				87	17			13
P		4		47						12			85			661	5	42	71	25			
Q	3	5					33			33		17	8			26	193	140					
R	12	2			62				65	107	1	45	23	1		19	161	290	27	10		34	
S	4	8		32	35			9	48		7		53		83	63		26	709	41		1	11
T		9		176							99	15			70	11	29		13	72	701		
V		1		115		19	16	5	18		213		56	96			5				336		
W	2				2				2				6					33					
Y	27				21	9		10		25					7				6				255

Variants by chromosome



1, Position,0,1000000,2000000,3000000,4000000,5000000,6000000,7000000,8000000,9000000,10000000,11000000,12000000,13000000,14000000,15000000,16000000,17000000,18000000,19000000,20000000,21000000,22000000,23000000,24000000,25000000,26000000,27000000,28000000,29000000,30000000,31000000,32000000,33000000,34000000,35000000,36000000,37000000,38000000,39000000,40000000,41000000,42000000,43000000,44000000,45000000,46000000,47000000,48000000,49000000,50000000,51000000,52000000,53000000,54000000,55000000,56000000,57000000,58000000,59000000,60000000,61000000,62000000,63000000,64000000,65000000,66000000,67000000,68000000,69000000,70000000,71000000,72000000,73000000,74000000,75000000,76000000,77000000,78000000,79000000,80000000,81000000,82000000,83000000,84000000,85000000,86000000,87000000,88000000,89000000,90000000,91000000,92000000,93000000,94000000,95000000,96000000,97000000,98000000,99000000,100000000,101000000,102000000,103000000,104000000,105000000,106000000,107000000,108000000,109000000,110000000,111000000,112000000,113000000,114000000,115000000,116000000,117000000,118000000,119000000,120000000,121000000,122000000,123000000,124000000,125000000,126000000,127000000,128000000,129000000,130000000,131000000,132000000,133000000,134000000,135000000,136000000,137000000,138000000,139000000,140000000,141000000,142000000,143000000,144000000,145000000,146000000,147000000,148000000,149000000,150000000,151000000,152000000,153000000,154000000,155000000,156000000,157000000,158000000,159000000,160000000,161000000,162000000,163000000,164000000,165000000,166000000,167000000,168000000,169000000,170000000,171000000,172000000,173000000,174000000,175000000,176000000,177000000,178000000,179000000,180000000,181000000,182000000,183000000,184000000,185000000,186000000,187000000,188000000,189000000,190000000,191000000,192000000,193000000,194000000,195000000,196000000,197000000,198000000,199000000,200000000,201000000,202000000,203000000,204000000,205000000,206000000,207000000,208000000,209000000,210000000,211000000,212000000,213000000,214000000,215000000,216000000,217000000,218000000,219000000,220000000,221000000,222000000,223000000,224000000,225000000,226000000,227000000,228000000,229000000,230000000,231000000,232000000,233000000,234000000,235000000,236000000,237000000,238000000,239000000,240000000,241000000,242000000,243000000,244000000,245000000,246000000,247000000,248000000,249000000,250000000,251000000,252000000,253000000,254000000,255000000,256000000,257000000,258000000,259000000,260000000,261000000,262000000,263000000,264000000,265000000,266000000,267000000,268000000,269000000,270000000,271000000,272000000,273000000,274000000,275000000,276000000,277000000,278000000,279000000,280000000,281000000,282000000,283000000,284000000,285000000,286000000,287000000,288000000,289000000,290000000,291000000,292000000,293000000,294000000,295000000,296000000,297000000,298000000,299000000,300000000,301000000,302000000,303000000,304000000,305000000,306000000,307000000,308000000,309000000,310000000,311000000,312000000,313000000,314000000,315000000,316000000,317000000,318000000,319000000,320000000,321000000,322000000,323000000,324000000,325000000,326000000,327000000,328000000,329000000,330000000,331000000,332000000,333000000,334000000,335000000,336000000,337000000,338000000,339000000,340000000,341000000,342000000,343000000,344000000,345000000,346000000,347000000,348000000,349000000,350000000,351000000,352000000,353000000,354000000,355000000,356000000,357000000,358000000,359000000,360000000,361000000,362000000,363000000,364000000,365000000,366000000,367000000,368000000,369000000,370000000,371000000,372000000,373000000,374000000,375000000,376000000,377000000,378000000,379000000,380000000,381000000,382000000,383000000,384000000,385000000,386000000,387000000,388000000,389000000,390000000,391000000,392000000,393000000,394000000,395000000,396000000,397000000,398000000,399000000,400000000,401000000,402000000,403000000,404000000,405000000,406000000,407000000,408000000,409000000,410000000,411000000,412000000,413000000,414000000,415000000,416000000,417000000,418000000,419000000,420000000,421000000,422000000,423000000,424000000,425000000,426000000,427000000,428000000,429000000,430000000,431000000,432000000,433000000,434000000,435000000,436000000,437000000,438000000,439000000,440000000,441000000,442000000,443000000,444000000,445000000,446000000,447000000,448000000,449000000,450000000,451000000,452000000,453000000,454000000,455000000,456000000,457000000,458000000,459000000,460000000,461000000,462000000,463000000,464000000,465000000,466000000,467000000,468000000,469000000,470000000,471000000,472000000,473000000,474000000,475000000,476000000,477000000,478000000,479000000,480000000,481000000,482000000,483000000,484000000,485000000,486000000,487000000,488000000,489000000,490000000,491000000,492000000,493000000,494000000,495000000,496000000,497000000,498000000,499000000,500000000,501000000,502000000,503000000,504000000,505000000,506000000,507000000,508000000,509000000,5100000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