SnpEff: Variant analysis

Contents

Summary

Genome	hg19
Date	2022-06-20 23:48
SnpEff version	`SnpEff 5.1d (build 2022-04-19 15:49), by Pablo Cingolani`
Command line arguments	`SnpEff hg19 Variants/SLGFSK.vcf`
Warnings	3,204
Errors	0
Number of lines (input file)	19,928
Number of variants (before filter)	19,985
Number of not variants (i.e. reference equals alternative)	0
Number of variants processed (i.e. after filter and non-variants)	19,985
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	71,599
Genome total length	3,137,161,265
Genome effective length	395,962,365
Variant rate	1 variant every 19,812 bases

Variants rate details

Chromosome	Length	Variants	Variants rate
5	180,915,260	5,930	30,508
12	133,851,895	7,134	18,762
17	81,195,210	6,921	11,731
Total	**395,962,365**	**19,985**	**19,812**

Number variants by type

Type	Total
SNP	17,952
MNP	0
INS	972
DEL	1,061
MIXED	0
INV	0
DUP	0
BND	0
INTERVAL	0
Total	**19,985**

Number of effects by impact

Type (alphabetical order)	Count	Percent
HIGH	153	0.214%
LOW	5,506	7.69%
MODERATE	3,497	4.884%
MODIFIER	62,443	87.212%

Number of effects by functional class

Type (alphabetical order)	Count	Percent
MISSENSE	3,445	44.229%
NONSENSE	48	0.616%
SILENT	4,296	55.155%

Missense / Silent ratio: 0.8019

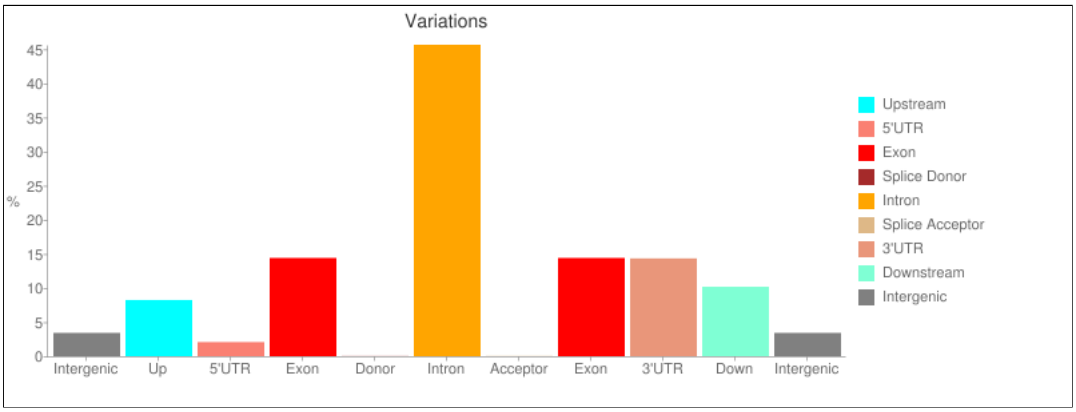
Number of effects by type and region

Type

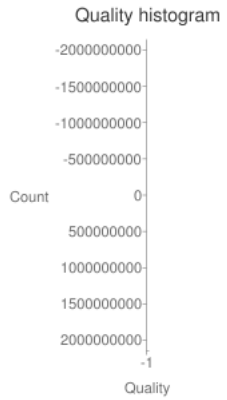
Type (alphabetical order)	Count	Percent
3_prime_UTR_variant	10,271	14.093%
5_prime_UTR_premature_start_codon_gain_variant	178	0.244%
5_prime_UTR_variant	1,309	1.796%
conservative_inframe_deletion	7	0.01%
conservative_inframe_insertion	9	0.012%
disruptive_inframe_deletion	29	0.04%
disruptive_inframe_insertion	15	0.021%
downstream_gene_variant	7,293	10.007%
frameshift_variant	56	0.077%
intergenic_region	2,453	3.366%
intron_variant	33,729	46.28%
missense_variant	3,437	4.716%
non_coding_transcript_exon_variant	2,542	3.488%
splice_acceptor_variant	24	0.033%
splice_donor_variant	21	0.029%
splice_region_variant	1,247	1.711%
start_lost	8	0.011%
stop_gained	48	0.066%
stop_lost	4	0.005%
stop_retained_variant	8	0.011%
synonymous_variant	4,289	5.885%
upstream_gene_variant	5,903	8.1%

Region

Type (alphabetical order)	Count	Percent
DOWNSTREAM	7,293	10.186%
EXON	10,316	14.408%
INTERGENIC	2,453	3.426%
INTRON	32,696	45.665%
SPLICE_SITE_ACCEPTOR	24	0.034%
SPLICE_SITE_DONOR	17	0.024%
SPLICE_SITE_REGION	1,139	1.591%
UPSTREAM	5,903	8.245%
UTR_3_PRIME	10,271	14.345%
UTR_5_PRIME	1,487	2.077%

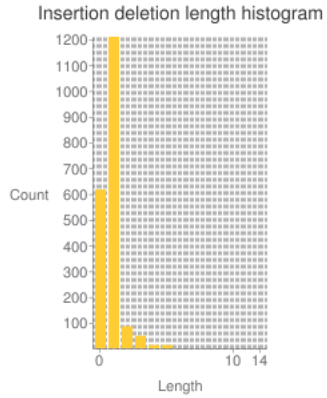


Quality:



Insertions and deletions length:

Min	0
Max	14
Mean	0.95
Median	1
Standard deviation	1.197
Values	0,1,2,3,4,5,6,7,8,9,10,11,14
Count	618,1210,86,58,17,19,5,4,6,2,2,5,1



Base changes (SNPs)

	A	C	G	T
A	0	630	3,028	398
C	711	0	896	3,235
G	3,402	876	0	708
T	427	3,021	620	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	35,021
Transversions	14,448
Ts/Tv ratio	2.4239

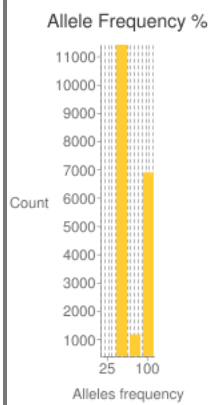
All variants:

```
Sample ,NORMAL,TUMOR,2:NORMAL,2:TUMOR,Total
Transitions ,17269,17752,0,0,35021
Transversions ,7116,7332,0,0,14448
Ts/Tv ,2.427,2.421,NaN,NaN,2.424
```

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

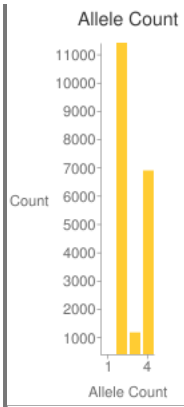
```
No results available (empty input?)
```

Allele frequency



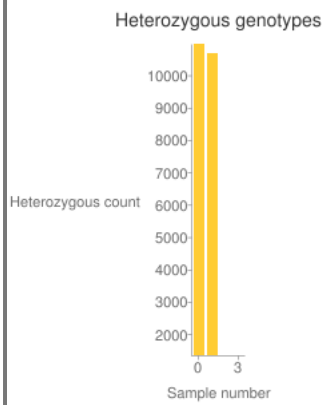
Min	25
Max	100
Mean	68.357
Median	50
Standard deviation	24.057
Values	25,50,75,100
Count	391,11404,1242,6891

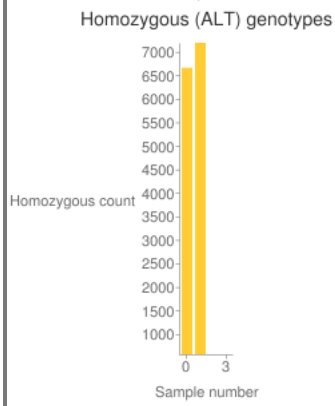
Allele Count

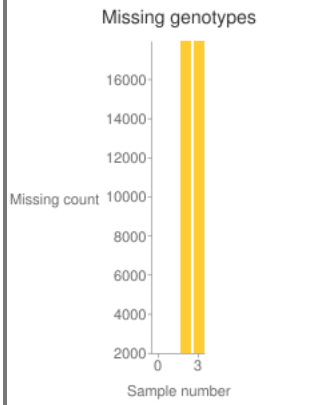


Min	1
Max	4
Mean	2.734
Median	2
Standard deviation	0.962
Values	1,2,3,4
Count	391,11404,1242,6891

Hom/Het per sample





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Sample_names , NORMAL, TUMOR, 2:NORMAL, 2:TUMOR
Reference , 271, 54, 63, 14
Het , 10971, 10706, 1346, 1396
Hom , 6708, 7190, 569, 568
Missing , 1978, 1978, 17950, 17950
```

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).

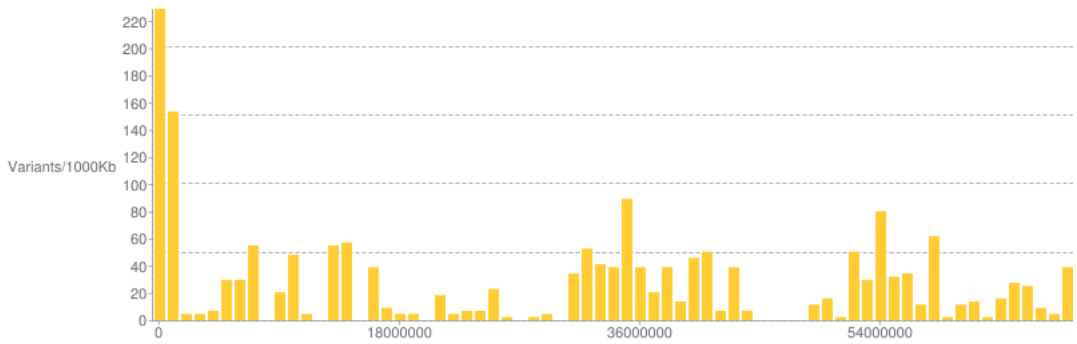
	-	AAA	AAC	AAG	AAT	ACA	ACC	ACG	ACT	AGA	AGC	AGG	AGT	ATA	ATC	ATG	ATT	CAA	CAC	CAG	CAT	CCA	CCC
-			1								2	1	1					2		3	2		4
AAA				22	2	8				28								6					
AAC	4	4	**1**	1	81		5				18				2				6				
AAG	2	56	4		3			3				20				3				5			
AAT	2	2	78						1				47				4				2		
ACA	1						6	92	18	11				11								5	
ACC	1		7			8		24	66		8				5								15
ACG						105	13		4			1				46							
ACT						2	63	9					14				21						
AGA		19				3					3	32											
AGC			25				17			4		9	68		6								
AGG				29		1		3		60	3		4										
AGT					17				5		37	4					2						
ATA		2				17									6	7	3						
ATC	3		2				14				3			10		7	68				4		
ATG	1		2	1	3			33		2		1		10	9		2						
ATT	1				2				14					12	80	2							
CAA	9	7																	1	65	1	17	
CAC	3		7															1		22	114		
CAG	3			2														67	17		1		1
CAT					7													11	47	6			
CCA	1					1												13					7
CCC	4						16												4	3		11	
CCG	5							9												2		75	24
CCT									6												6	8	103
CGA										2								34				1	
CGC																			41				2
CGG												34								79			
CGT																			3		26		
CTA														4				2				10	
CTC	1														2				5				4
CTG																5				6			1
CTT	2																4						
GAA	1	27																7					
GAC	2		18																				
GAG	11			35						1										3			
GAT					10																1		
GCA						30																5	
GCC	4						33																2
GCG								9															
GCT									21														
GGA										9													
GGC	3										17												
GGG												25											
GGT	1												17										
GTA														10									
GTC															66								
GTG																55							
GTT																	17						
TAA																							
TAC																			15				
TAG																							
TAT	4																				8		
TCA	1																					23	
TCC							18																10
TCG																							
TCT									5														
TGA																							
TGC	2										2												
TGG																							
TGT	1												1										
TTA																							
TTC															1								
TTG	1															4							
TTT																	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
*	**8**												1					1				2	
-			37	5			2		1	2			6		1	5	5	3	3		1		
?																							
A		4		**412**		3	13		30							15			14	93	86		
C		3			**63**			2										54	46				14
D		2		11		**304**	41		7	1					28						2		3
E	1	12		8		30	**142**		19			62					10	1			14		
F					6			**64**			2		45						15		7		9
G		4		35	9	37	13		**316**									38	34		10	2	
H		3				2				**161**			3		14	3	40	48					8
I		4						2		4	**179**	2	19	16	4				3	45	76		
K		2					16					**78**		3	9		11	48		11			
L	1	4						40		5	10		**385**	9		78	8	2	18		41		
M		1									21	1	22		5			3		33	64		
N		6				44				8	6	7			**160**				65	6			
P		10		22						10			67			**427**	18	29	45	32			
Q	25	12					21			20		9	1			29	**132**	73					
R	18				38		1		35	70		48	12			13	113	**290**	10	7		30	
S	1	1		30	26			12	44		8		40		42	53		28	**453**	45		1	6
T		2		97							37			46	7	24		12	37	**410**			
V				69		17	5	10	9		93		40	55							**181**		
W	1				1				2									35	3				
Y	1	4			22	2		3		23									9				**165**

Variants by chromosome



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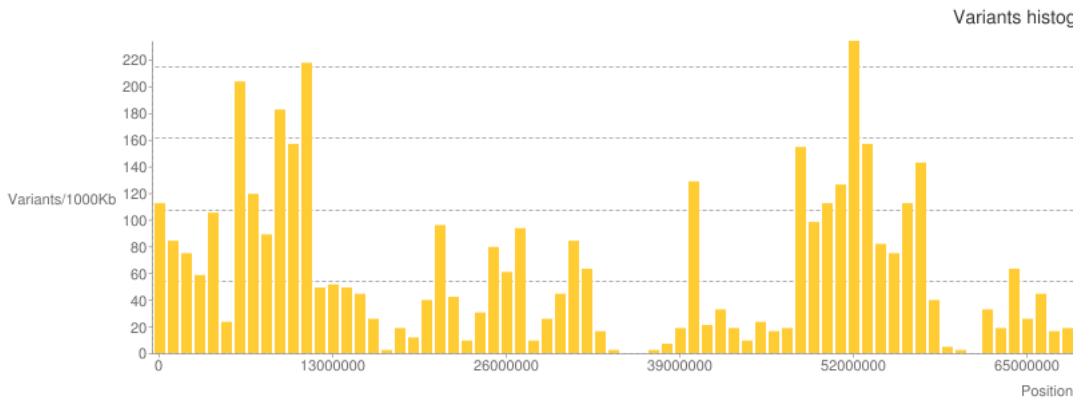

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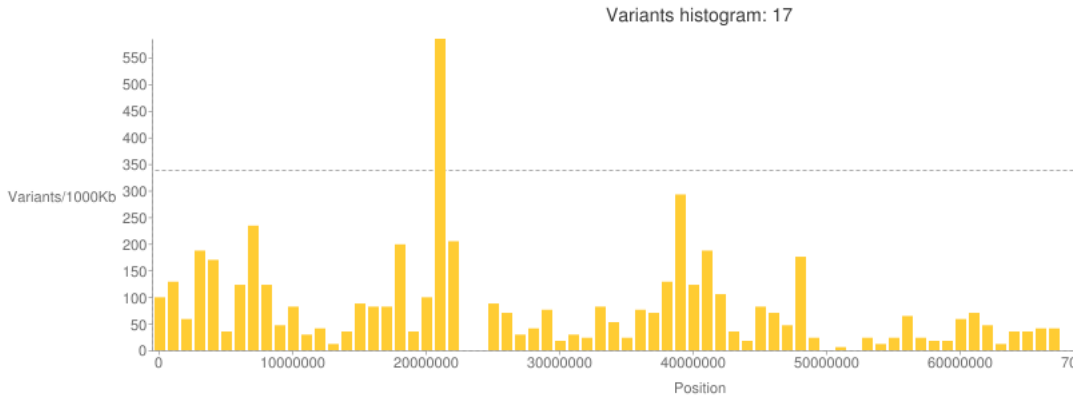


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Details by gene

Here you can find a tab-separated table.