

Table 3 Number of effects by type and region

Variation type	Count	Percent	Variation region	Count	Percent
3' UTR variant	7 906	1.09 %	Downstream	174 403	24.22 %
5' UTR premature start codon gain	485	0.07 %	Exon	25 443	3.53 %
5' UTR variant	2 773	0.38 %	Intergenic	276 549	38.40%
Downstream gene variant	174 403	24.13 %	Intron	59 074	8.20%
Initiator codon variant	2	0 %	Splice site acceptor	97	0.01%
Intergenic region	276 549	38.27 %	Splice site donor	60	0.01%
Intron variant	61 125	8.46 %	Splice site region	2 273	0.32%
Missense variant	13 422	1.86 %	Transcript	356	0.05%
Non coding transcript exon variant	289	0.04 %	Upstream region of gene (within 5k)	170 706	23.71%
Non coding transcript variant	356	0.05 %	Utr_3_prime	7 906	1.10%
Splice acceptor variant	97	0.01 %	Utr_5_prime	3 258	0.45%
Splice donor variant	60	0.01 %	-	-	-
Splice region variant	2 541	0.35 %	-	-	-
Start lost	24	0.00%	-	-	-
Stop gained	292	0.04 %	-	-	-
Stop lost	20	0.00%	-	-	-
Stop retained variant	19	0.00 %	-	-	-
Synonymous variant	11 625	1.61 %	-	-	-
Upstream gene variant	170 706	23.62 %	-	-	-