1. Alignment stats

Seq ID	Sample Name	Number of reads	Reads mapped	Percent reads aligned	Reads duplicated	Percent duplicate reads	Percent reads on target	Percent reads off target	Avg. coverage over the entire kit	Avg. coverage of the targeted regions
D11	1N	80041732	79881534	99.7998569	13596982	16.98736604	91.7655	8.2345	115.495093	116.661764
D11	1T	161788416	161504566	99.82455481	32252802	19.93517385	91.4695	8.5305	227.013754	228.926347
D12	2N	78452928	78330167	99.84352273	8610697	10.97562222	92.8889	7.1111	121.716394	122.706875
D12	2T	149233692	148997293	99.8415914	30031523	20.12382231	90.699	9.301	206.754309	208.493539
D13	3N	78955518	78769717	99.76467636	7781711	9.855816537	92.7647	7.2353	123.248583	124.269927
D13	3T	156277388	155962325	99.79839502	39625697	25.3560016	87.9608	12.0392	189.981679	191.598477

2. Variant VCF file

#CHROM	POS	ID	REF	ALT	QUAL	FILTER	INFO	FORMAT	Germline	Tumor
1	809319	.	C	T	.	PASS	ANN=T|u	GT:AD:BQ	0:34,1:.:34	0/1:27,6:30:33:0.182:2
1	1407616	.	C	G	8.5	PASS	AB=0.2307	GT:AD:AO	0/0:18,0:0	0/1:20,6:6:26:8:45,0,494:173:633:20
1	2585315	.	A	G	14.54	PASS	AB=0.2727	GT:AD:AO	0/0:12,0:0	0/1:8,3:3:11:15:51,0,175:90:233:8
1	2586384	.	A	C	15.02	PASS	AB=0.2;AB	GT:AD:AO	0/0:28,0:0	0/1:16,4:4:20:15:50,0,287:124:457:16
1	2586415	.	C	A	8.68	PASS	AB=0.2;AB	GT:AD:AO	0/0:23,0:0	0/1:16,4:4:20:9:47,0,289:120:480:16
1	6763809	.	G	A	100.63	PASS	AB=0.6;AB	GT:AD:AO	0/0:15,0:0	0/1:4,6:6:10:75:136,0,77:181:118:4
1	7890024	rs2016629	T	G	161	PASS	ANN=G|m	GT:AD:AD	0/0:54,1:0	0/1:144,55:0:0.2083:11,44:2,2:264:0.2727:52.8:4.2:1.31:12.8:1:1:28:23,121:0.53017:6.857:55
1	7890026	rs1999473	A	G	174	PASS	ANN=G|m	GT:AD:AD	0/0:52,1:0	0/1:138,64:0:0.2397:13,51:2,2:267:0.3261:53.8:3.8:1.34:12.8:1:1:29:22,116:0.43306:15:64
1	7890053	.	G	A	1178.47	PASS	AC=1;AF=(	GT:AD:AO	0|0:327,0:	0|1:303,117:117:420:99:1554,0,6345:3274:8713:303
1	7890055	.	T	A	1178.47	PASS	AC=1;AF=(	GT:AD:AO	0|0:327,0:	0|1:303,117:117:420:99:1554,0,6345:3274:8713:303
1	7890064	.	A	G	410.5	PASS	AB=0.192(	GT:AD:AO	0/0:383,0:	0/1:365,87:87:453:99:677,0,7985:2389:10664:365
1	7890069	.	C	T	54.42	PASS	AB=0.1516	GT:AD:AO	0/0:395,1:	0/1:386,69:69:455:54:283,0,7999:1929:10646:386
1	8385485	.	GA	G	55.08	PASS	AC=1;AF=(	GT:AD:AO	0|0:58,0:0	0|1:121,22:22:149:55:108,0,2616:597:3384:121
1	10719645	.	A	G	344.53	PASS	AB=0.6785	GT:AD:AO	0/0:25,1:1	0/1:9,19:19:28:99:431,0,137:569:243:9
1	12907385	.	G	A	71.78	PASS	AB=0.2137	GT:AD:AO	0/0:132,9:	0/1:114,31:31:145:72:164,0,2672:929:3453:114
1	16890570	.	G	C	679.48	PASS	AB=0.2263	GT:AD:AO	0/0:152,7:	0/1:188,55:55:243:99:729,0,4477:1672:5902:188
1	16913094	.	G	A	8.43	PASS	AB=0.1714	GT:AD:AO	0/0:23,0:0	0/1:29,6:6:35:8:43,0,600:178:804:29
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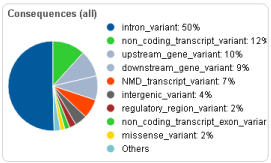
3. VEP annotation

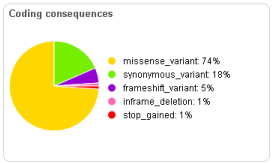
Variant Effect Predictor results

Job details

Summary statistics

Category	Count
Variants processed	667
Variants filtered out	0
Novel / existing variants	384 (57.6) / 283 (42.4)
Overlapped genes	580
Overlapped transcripts	2476
Overlapped regulatory features	77





Results preview

Navigation — Page: 1 of 134 | Show: 1 5 10 50 All variants

Filters — Uploaded variant is defined Add

Download — All: VCF VEP TXT; BioMart: Variants Genes

Show/hide columns

Show All entries — Filter

Uploaded variant	Location	Allele	Consequence	Impact	Symbol	Gene	Feature type	Feature	Biotype	Exon	Intron	HGVSc	HGVSp	cDNA position	CDS position	Protein position	Amino acids
rs1534952	1:1650628-1650628	A	intron_variant	MODIFIER	CDK11A	ENSG00000008128	Transcript	ENST00000356200	protein_coding	-	3/18	-	-	-	-	-	-
rs1534952	1:1650628-1650628	A	intron_variant, non_coding_transcript_variant	MODIFIER	CDK11A	ENSG00000008128	Transcript	ENST00000356937	retained_intron	-	2/16	-	-	-	-	-	-
rs1534952	1:1650628-1650628	A	intron_variant	MODIFIER	CDK11A	ENSG00000008128	Transcript	ENST00000357760	protein_coding	-	4/19	-	-	-	-	-	-
rs1534952	1:1650628-1650628	A	intron_variant	MODIFIER	CDK11A	ENSG00000008128	Transcript	ENST00000358779	protein_coding	-	4/19	-	-	-	-	-	-
rs1534952	1:1650628-1650628	A	intron_variant	MODIFIER	CDK11A	ENSG00000008128	Transcript	ENST00000378633	protein_coding	-	4/19	-	-	-	-	-	-
rs1534952	1:1650628-1650628	A	intron_variant	MODIFIER	CDK11A	ENSG00000008128	Transcript	ENST00000378635	protein_coding	-	4/10	-	-	-	-	-	-
rs1534952	1:1650628-1650628	A	intron_variant	MODIFIER	CDK11A	ENSG00000008128	Transcript	ENST00000378638	protein_coding	-	4/19	-	-	-	-	-	-
rs1534952	1:1650628-1650628	A	downstream_gene_variant	MODIFIER	CDK11A	ENSG00000008128	Transcript	ENST00000401096	protein_coding	-	-	-	-	-	-	-	-