

Supplementary methods

VEP annotation:

GRCh37 26 October 2018

BCBIO software versions:

bamtools,2.4.0
bcbio-nextgen,1.0.9a0
bcbio-variation,0.2.6
bcftools,1.6
bedtools,2.27.1
biobambam,2.0.87
bioconductor-bubbletree,2.8.0
bowtie2,2.2.8
break-point-inspector,1.5
bwa,0.7.17
chanjo,
cnvkit,0.9.3
cufflinks,2.2.1
cutadapt,1.16
fastqc,0.11.7
featurecounts,1.4.4
freebayes,1.1.0.46
gatk4,4.0.2.1
gemini,0.20.1
grabix,0.1.8
hisat2,2.1.0
htseq,0.9.1
lumpy-sv,0.2.13
manta,1.3.2
metasv,0.4.0
mirdeep2,2.0.0.7
mutect,1.1.5
novoalign,3.07.00
novosort,V3.00.02
oncofuse,1.1.1
phylowgs,20150714
picard,2.17.11
platypus-variant,0.8.1.1
preseq,2.0.2
qualimap,2.2.2a
rna-star,
rtg-tools,3.8.4
sailfish,0.10.1
salmon,0.9.1
sambamba,0.6.6
samblaster,0.1.24
samtools,1.7
scalpel,0.5.3
seqbuster,3.1

snpeff,4.3i
vardict,2017.11.23
vardict-java,1.5.1
variant-effect-predictor,
varscan,2.4.3
vcflib,1.0.0_rc1
vt,2015.11.10
wham,1.7.0.311

BCBIO data versions:

genome,resource,version
hg19,seq,broad-20120813
hg19,twobit,broad-20120813
hg19,GA4GH_problem_regions,20160916
hg19,capture_regions,20161202
hg19,MIG,20150730
hg19,prioritize,20160215
hg19,dbsnp,150-20170710
hg19,hapmap,3.3
hg19,1000g_omni_snps,2.5
hg19,ACMG56_genes,20160629
hg19,1000g_snps,2.8
hg19,mills_indels,2.8
hg19,clinvar,20170905
hg19,cosmic,68-20180114
hg19,ancestral,20141010
hg19,qsiganture,20140703
hg19,genesplicer,2004.04.03
hg19,effects_transcript,2017-03-16
hg19,vcfanno,20171008
hg19,viral,2017.02.04
hg19,transcripts,2014-07-17
hg19,RADAR,v2-20180202
hg19,srnaseq,20180122
hg19,giab-NA12878,v3_3_2
hg19,platinum-genome-NA12878,v8_0_1
hg19,giab-NA24385,v3_3_2-sv_v0.5.0
hg19,giab-NA24631,v3_3_2