Variant Summary Table Key

Variant Report Tab

Name	Description
Sample_ID	User defined library ID, derived from FASTQ file nameUser defined library ID, derived from FASTQ file name
Chromosome	Chromosome number in human genome where variant occurs
Position	Base position on given chromosome where variant occurs (hg19)
REF	Sequence found in human reference genome (hg19)
ALT	Variant sequence found in sample
Location	Genomic coordinates of variant (hg19)
Variant_Type	SNV=single nucleotide variant; Deletion=one or more base deletion; Insertion=one or more base insertion; Delins="deletion-insertion", variant characterized by both a deletion of one or more bases AND an insertion of one of more bases on the same allele. i.e. AGACTA ---> ATTCTA resulting from deletion of GA and insertion of TT or AGACTA ---> ATCTA resulting from deletion of GA and insertion of T
Variant_Length	Number of affected bases
Amplicon_ID	Amplicon in assay in which variant occurs
Variant_Read_Frequency_(%)	Occurrence of variant in sequencing data, as a percentage of total sequenced reads in the given segment. (Variant_Coverage / Total_Coverage)*100
Variant_Coverage	Number of reads that contain variant in segment

Total_Coverage	Total number of reads in segment
Variant_Quality	Measure of variant call quality. Score is from 0 (low quality) to 40 (highest quality)
Variant_Read_Direction_Ratio	Measure of strand bias in variant calling. Value of 0 indicates variant was found on both the positive strand and negative strand at equal rates (ideal). Values greater than zero or less than zero indicate percentage of bias towards the negative strand (negative values) or positive strand (positive values)
Zygosity	Germline mutation panels only. HETEROZYGOUS=one out of two alleles contain variant; HOMOZYGOUS=both alleles contain variant
Consequence	Type of mutation. Examples: frameshift_variant=variant produces an mRNA transcript that is out of the normal reading frame. synonymous_variant=variant does not affect amino acid sequence of final protein due to redundancy of codons. missense_variant=variant leads to change in amino acid sequence. inframe_deletion=deletions of 3, or multiples of 3, which remove whole codon sequences. intron_variant=variant occurs in intron. splice_region_variant=variant in splice site, may lead to splicing error in final mRNA transcript. stop_gained=variant results in premature stop codon in mRNA transcript. 3_prime_UTR_variant=variant occurs in 3" untranslated region (UTR).
Impact	Impact of variant on normal gene function
Gene_Symbol	Gene name
Gene_ID	NCBI gene ID number
Feature	NCBI transcript identifier
HGVSC	HGVSC variant name. Describes position and variant change in gene (nucleic acid) sequence in the Feature
HGVSP	HGVSP variant name. Describes position and variant change in protein (amino acid) sequence in the Feature

Exon	Affected exon number in Feature out of total number of exons. Applies to variants within exon regions only. (affected exon # / total # of exons)
Intron	Affected intron number in Feature out of total number of introns. Applies to variants within intron regions only. (affected intron # / total # of introns)
CDS_Position	cDNA sequence position in Feature
Protein_Position	Affected amino acid number in Feature
Amino_Acids	Amino acid resulting from variant (reference amino acid / variant amino acid)
Codons	Position in codon where variant occurs and resulting codon with variant
Co-located_Known_Variation	Variants that occur at the same position in publicly available databases
Strand	Transcribed strand. -1=negative strand, 1=postive strand
Repeat	Number of repeat bases, including repeat sequence (if applicable). i.e. 14G indicates a repeat of 14 Gs.
HGVS_Offset	Correction factor to synchronize genomic base position for indels in repeat regions in negative strand genes. Read alignment uses left-align paradigm (left most base assumed to be indel; first genomic position) while HGVS uses right-align paradigm (right most base assumed to be indel; first cDNA position).
SIFT	Output from SIFT (v5.2.2). PREDICTION(SIFT score). Predicts impact of variant on protein function. Prediction possibilities: tolerated, deleterious. SIFT score is a range from 0 - 1. Scores > 0.05 are tolerated, scores of <0.05 are deleterious.
POLYPHEN	Output from PolyPhen (v2.2.2). PREDICTION(PolyPhen score). Predicts impact of variant on protein function. Prediction

	possibilities: benign, possibly damaging, probably damaging. PolyPhen score is a range from 0 (benign) - 1 (probably damaging) and is the probability of variant leading to a damaging mutation. Benign=variant unlikely to cause damage to protein function. possibly damaging=variant likely to cause damage to protein function but prediction is with low confidence. probably damaging=variant likely to cause damage to protein function and prediction is with high confidence.
AF	Allele frequency of variant found in global population (1000 Genomes Phase 3, dbSNP v147)
AFR_AF	Allele frequency of variant found in African populations (dbSNP v147)
AMR_AF	Allele frequency of variant found in American populations (dbSNP v147)
EAS_AF	Allele frequency of variant found in East Asian populations (dbSNP v147)
EUR_AF	Allele frequency of variant found in European populations (dbSNP v147)
SAS_AF	Allele frequency of variant found in South Asian populations (dbSNP v147)
CLIN_SIG	Clinical significance (ClinVar v201610)

Overall Stats Tab

Name	Description
Statistic	Description
Total Reads	Total number of sequencing reads for the library
Overall:Q=30	Percentage of bases with Q score greater than or equal to 30

Overall:Q=20	Percentage of bases with Q score greater than or equal to 20
Properly Paired Read	Number of read mates properly paired, from paired end sequencing
Properly Paired Read (%)	Percentage of total reads properly paired
Mapped Reads	Percentage of total reads that map to human genome (hg19)
Mapping Rate (%)	Percentage of total reads that map to human genome (hg19)
On Target Reads	Reads that map to target amplicon regions of interest (ROIs)
On Target Rate (%)	Percentage of mapped reads that map to target amplicon regions of interest (ROIs)
Insert Size Mean	Mean size of the library insert (varies based on panel)
Insert Size Median	Median size of the library insert
Insert Size Std Dev	Standard deviation of library insert size
Coverage_Mean	Mean base coverage of all bases within the defined ROI; with paired end sequencing, merged paired-end reads (forward and reverse) create a coverage of 1x
STDEV	Standard deviation of mean base coverage
Coverage_Median	Median base coverage of all bases within the defined ROI; with paired end sequencing, merged paired-end reads (forward and reverse) create a coverage of 1x
Coverage_Max	Maximum base coverage of all bases within the defined ROI
Coverage_Min	Minimum base coverage of all bases within the defined ROI
Base_Coverage_Depth_>_(Nx)	Percent of bases that have a minimum base coverage greater than or equal to Nx (absolute coverage)
Base_Coverage_Depth_>_(Nx)_Relative_to_Mean_Coverage	Percent of bases that obtain at least Nx*mean base coverage, usually described as percent of mean base coverage. Used to determine uniformity of base coverage across ROIs in panel. Value

	of 100 indicates 100% of bases in all ROI are above the given Nx relative to the Coverage_Mean.