MitoSeek Parameters

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| Parameter | Description |
| - i [bam] | Input bam file |
| - t [input type] | Type of the bam files, the possible choices are 1=exome, 2=whole genome, 3= RNAseq, 4 = mitochondria only |
| - b | Save mitochondria only bam files, default =on |
| - a | Produce allele count file for input bam(s), default =on |
| - ch | Produce circus plot input files and circus plot figure for heteroplasmic mutation, default = off |
| - hp [int] | Heteroplasmy threshold using [int] percent alternative allele observed, default = 5 |
| - ha [int] | Heteroplasmy threshold using [int] allele observed, default = 0 |
| - A | If - A is used, the total read count is the total allele count of all allele observed. Otherwise, the total read count is the sum of major and minor allele counts. Default = off |
| - mmq [int] | Minimum map quality, default =20 |
| - mbq [int] | Minimum base quality, default =20 |
| - sb [int] | Remove all sites with strand bias score in the top [int] %, default = 10 |
| - cn | Estimate relative copy number of input bam(s), does not work with mitochondria targeted sequencing bam files. |
| - s [bam1] [bam2] | Compute somatic mutation between bam1 and bam2 |
| - sp [int1][int2] | Somatic mutation detection threshold, int1 = percent of alternative allele observed in normal, int2 = percent of alternative allele observed in tumor, default int1=0, int2=5 |
| - sa [int1][int2] | Somatic mutation detection threshold, int1 = number of alternative allele observed in normal, int2 = number of alternative allele observed in tumor, default int1=0, int2=3 |
| - cs | Produce circus plot input files and circus plot figure for somatic mutation, default = off |
| - L [bed] | A bed file that contains the regions MitoSeek will perform analysis on |
| - r [ref] | The reference used in the bam file, the possible choices are HG19 and rCRS, default=HG19 |
| - R [ref] | The reference used in the output files, the possible choices are HG19 and rCRS, default=HG19 |
| - QC | Produce QC result |

Output

- a will produce allele count file with allele count on both forward and reverse strands and strand bias scores for each location

- c will produce circus plot input file and circus plot figure

- cn will produce summary information on estimated copy number of input bam(s)

A heteroplasmy summary file will also be generated