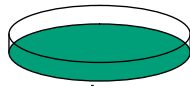
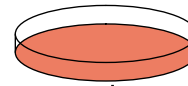


Het1A-  
Parental



Het1A-  
STE-8M



DNA extraction, library preparation and sequencing

Input  
format

FASTQ

FASTQ

BAM/SAM

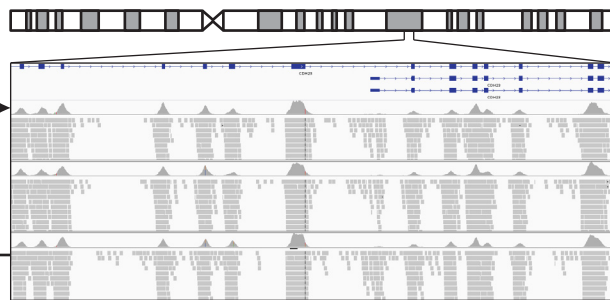
Output  
format

FASTQ

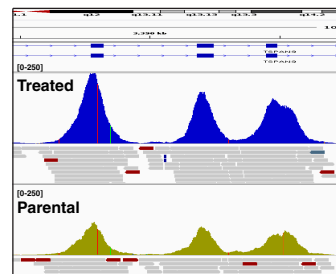
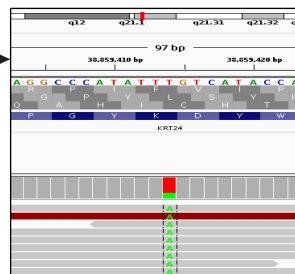
BAM/SAM

VCF

Raw reads acquired from Illumina HiSeq 2500  
Quality assessment of raw reads  
using FastQC



Alignment of reads against human reference  
genome hg19 using BWA-MEM and preprocess  
BAM file with GATK package to improve  
alignment



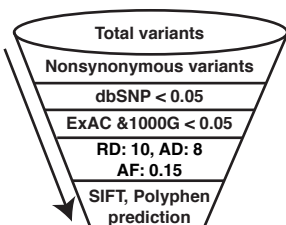
Single nucleotide variants identification using Strelka  
and copy number alteration using OncoCNV

Annotation of variants using Varimat and OncoMD;  
Filter high confidence variants;  
data visualization and literature survey



Copy number alterations  
identified with pvalue < 0.000001  
Amplification >= 3  
Deletion <= 0.5

CNA filtration criteria



SNV filtration criteria