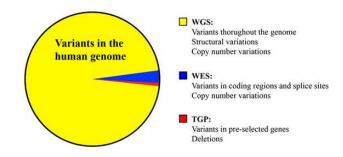
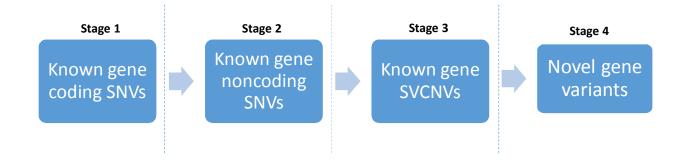
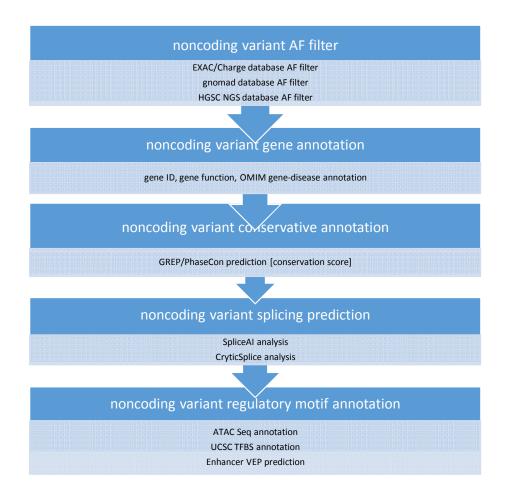
WGS analytic pipeline

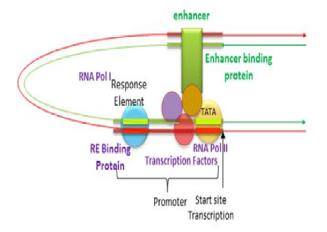
- TGP/WES pipeline limitation:
- 1. structural variants;
- 2. copy number change variants;
- 3. non-coding variants;
- 4. non-coding regulatory elements;
- 5. non-annotated genes.





noncoding variant analysis





SVCNV variant analysis

SVCNV variant calling

CNVnator [calling copy number change by read depth]

delly / lumpy / manta [calling DEL/DUP/INS/INV by junction pair-end reads]

MELT [calling transposon by MEI Consensus sequence]

SVCNV variant QC filter

svtyper QC summary [score based on variant read number, junction read count, read map quality, confidence interval of read ends , copy number estimate, etc]

Svtyper score filter [hard-cutoff>100]

CNV depth distribution filter [q0 <= 0.5]

SVCNV variant merge

DEL/DUP [merge by partial overlapping of either start/end position]

INV [merge by overlapping of both start/end position]

INS [merge by the distance of insert position]

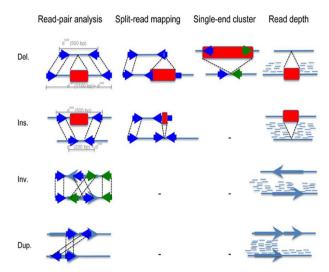
TRS [merge by the distance of both transposition junction end position]

SVCNV variant annotation

gene ID, gene function, OMIM gene-disease annotation, affected exon numbers, regulatory element annotation

SVCNV variant AF filter

DGV database filter
HGSC SV WGS database AF and filter



Novel gene variant analysis

