SNV Germline Analysis Pipeline

ANNOTATIONS NCBI / RefSeq Gene Ontology GeneRIF KEGG Reactome Interactions OMIM ProteinAtlas (expr. Normal / expr. cancer / subcellular location) Gene Families Pathways pLI (from ExAC)

MANUAL FILTERS

- AD column avoid false positives Indels (DP and sum of AD values should be similar)
- Strand bias IGV
- Repetitive regions IGV
- Function / Domain annotations

