



DNA isoaltion

WGS/WES

Sequence variants

Structural variants

Custom filtering

Gene list S2A
Protein coding/splice site
Synonymous if splicing effect
< 0.1% globally
Allele fraction > 30%
ClinVar not benign/likely benign

Genmod/Scout

Gene list S2B+S2C
Protein coding/splice site
< 3 occurences locally and < 0.1% globally
ClinVar not benign/likely benign

Individual cancer syndrome gene list

Gene lists S3
Protein coding and non-coding
< 0.1% globally
ClinVar not benign/likely benign

Scout

Gene lists S2C
Protein modifying
< 5 occurances locally
Not likely artifacts