

Sequencing & Alignment



High-Quality Filtering Criteria

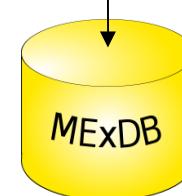
- Depth ≥ 15
- QUAL ≥ 150
- AF $\geq 20\%$
- Coding Novel SNV
- Clinical rs (dbSNP132)
- Filtered out HLA genes
- Filtered out homologous genes
- Internal exome DB filtering

53120 novel SNVs

10188 novel coding SNVs

3325 novel coding HQ-SNVs
(1153 syn + 2172 non-syn)

SIFT prediction tool



gBrowse

QUAL= SNV Call PHRED quality;
AF= Variation Allele Frequency

1770 genes with at least
1 non-syn HQ-SNV

ATTCACCACGGATGTAACTACCTTACTTAGAGTAGCAGCA
[Genomic sequence with green and red tracks]