

68257 detected variants from proband WES data



Not present in Father



DP \geq 20 & QUAL \geq 100 *



Zygoty : Heterozygous



Pathogenicity in any :
PolyPhen2 prediction, HumDiv, HumVar : probably damaging, possibly damaging,
MutationTaster prediction : disease causing automatic, disease causing,
MutationAssessor prediction : High, Medium,
PROVEAN prediction : damaging



Coding effect : all except synonymous



Population Frequency as rare :
ESP6500 < 1%
ExAC < 1%
1000G Phase 1 < 1%
1000G Phase 3 < 1%



125 selected variants for manual curation