

**Table 1** Exome sequencing filtering strategy

<b>Criteria for filtering</b>	<b>No. of variants</b>
Total No. of variants	78,801
Number of rare variants (MAF $\leq$ 0.02)	11,700
Exonic and splicing variants	3,397
Exclusion of synonymous variants	2,562
Variants following segregation pattern of autosomal recessive/X-linked inheritance	211
Exclusion of variant present in-house WES data	32
Phenotypic correlation	1 in <i>MED12</i>

Abbreviations: MAF, minor allele frequency; *MED12*, mediator complex subunit 12; WES, whole exome sequencing.