S2 Table. Summary of whole exome sequencing (WES) metrics in the three affected individuals

	Individual III-7	**III-8**	**IV-2**
Total number of reads	136,048,746	127,298,302	114,750,902
Non-duplicated reads	90,255,052	97,195,484	85,612,771
Reads aligned to target	82,045,320 (90,9%)	83,570,035 (86%)	74,665,623 (87,2%)
Mean target coverage (%)	88.12	79.88	71.04
Total number of variants	22,318	22,566	22,531