

Supplementary information, Table S2

Table S2 Genetic variants identified through whole-exome sequencing

Sample	Total number of variants	Number of variants in target regions	Number of non-synonymous variants
III:6	25561	15561	6755
II:5	23407	14254	6235
III:4	22470	13673	5939
IV:1	22529	13667	5886
IV:2	20754	12780	5455
Case-specific variants	771	484	203
Case-specific novel variants*	3	3	2

* novel variants were defined as mutations that were not detected in the SNP data sets from dbSNP 132 and the 1000 Genome project.