Supplementary Table S2. Filtering steps in the whole exome sequencing on patient 2

	Variant number
Total variant (exon +/- 30 bps)	5712
Homozygous	2150
In-house =< 5/575	111
ExAC =< 0.005	60
ESP6500 = < 0.005	59
HGVD =< 0.005	59
Remove synonymous	44
Know disease gene match to patient's phenotype	1 (*DSE*)

Footnote: The common SNP (MAF > 1%) has been already removed in our pipeline.