

Supplementary Table 1. Proband's whole-exome sequencing in detail

Exome Capture Statistics	Proband
Raw reads (bp)	229789072
Clean reads (bp)	229158346
GC (%)	52.09
Initial bases on target (bp)	60456963
Total effective reads (bp)	201325227
Total effective bases (Mb)	19884.90
Effective sequences on target (Mb)	14486.50
Capture specificity (%)	72.85
Mapping rate on genome (%)	99.91
Duplicate rate on genome (%)	12.16
Mismatch rate in target region (%)	0.42
Average sequence depth on target (X)	239.62
Fraction of target region covered $\geq 1X$ (%)	99.71
Fraction of target region covered $\geq 4X$ (%)	99.66
Gender	Female