

Supplementary table 2 Summary of whole exome sequencing data from two patients with the SYCP2L variant.

Sample ID	Family P0001	Family P0005
	IV-2	IV-2
Raw bases (Mb)	19890.63	19270.79
Clean bases (Mb)	19878.65	18218.58
Clean data rate (%)	99.94	94.54
Clean read1 Q20 (%)	97.98	98.52
Clean read2 Q20 (%)	95.91	96.46
Clean read1 Q30 (%)	91.33	95.80
Clean read2 Q30 (%)	87.08	91.40
Bases in target region (bp)	60456963	60456963
Total effective bases (Mb)	11546.40	84346114
Effective sequences on target (Mb)	7934.22	8058.02
Capture specificity (%)	68.72	64.49
Mapping rate on genome (%)	99.86	99.42
Average sequencing depth on target	131.24	133.29
Fraction of target region covered $\geq 4X$ (%)	99.60	99.58
Fraction of target region covered $\geq 10X$ (%)	99.26	99.24
Fraction of target region covered $\geq 20X$ (%)	97.94	97.15
GC (%)	51.53	53.95