

**Table S2.** Detail information about NGS data of proband IV8.

<b>Detail information about NGS data</b>	<b>Value or ratio</b>
Raw data bases(Mb)	5144.76
Clean data bases(Mb)	5066.03
Aligned bases (Mb)	5057.47
Aligned	99.83%
Initial bases on target	2680649
Base covered on target	2678606
Coverage of target region	99.92%
Effective bases on target	1491661469
Fraction of effective bases on target	29.49%
Average sequencing depth on target	556.46
Fraction of target covered with at least 4X	99.88%
Fraction of target covered with at least 10X	99.80%
Fraction of target covered with at least 20X	99.63%
Duplication rate	22.94%