Supplementary Table 1. Sequencing quality statistics of NGS on the targeted region

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| --- | --- | --- | --- | --- |
| Sample | Number of targeted genes | Coverage of target region (%) | Average sequencing depth on target（X）  | Fraction of target covered with at least 20x (%)  |
| Proband | 22,000 | 99.59 | 107.76 | 96.54 |
| Father | 22,000 | 99.91 | 118.55 | 97.67 |
| Mother | 22,000 | 99.62 | 106.11 | 97.67 |