**S1 Table.** Sample metrics for exome sequencing of a trio from the family (Fig. 1).

|  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| Pedigree No. | Disease Status (sex) | Total Mapped Reads  (% total reads) | % Mapped Reads in Exome | Mean Mapped Read Depth for Exome (X) | % Exome ≥10X Coverage | Total Exome SNPs | Non-synonymous SNPs | Novel SNPs | Unexpected Gaps |
| I:1 | Affected (M) | 44,856,239 (99.01) | 89.24 | 88.8 | 88.09 | 46,889 | 9,350 | 3,048 | none |
| II:4 | Affected (M) | 50,632,041 (99.41) | 86.02 | 100.27 | 92.41 | 48,139 | 9,374 | 2,783 | none |
| II:9 | Unaffected (F) | 45,123,376 (98.97) | 86.48 | 89.36 | 91.87 | 48,270 | 9,465 | 3,036 | chrY: 141 exons |