**S2 Table. Exome sequencing results**

| Sample | Raw sequences (Gb)[a] | Mapped reads (M)[b] | Mean coverage[c] | % of bases covered at least 20X | Variants | SNPs | Indels | Ti/Tv[d] |
|---|---|---|---|---|---|---|---|---|
| Patient N° 1 | 3.6 | 23.4 | 56.8 | 77.4 | 47 955 | 45 182 | 3 247 | 2.57 |
| Patient N° 2 | 4.4 | 28.1 | 70.7 | 82.1 | 47 739 | 44 276 | 3 839 | 2.60 |
| Patient N° 3 | 5.3 | 34.9 | 82.4 | 86.0 | 50 655 | 47 902 | 3 079 | 2.54 |
| Patient N° 4 | 6.9 | 45.7 | 108.0 | 92.9 | 51 628 | 48 879 | 3 155 | 2.50 |
| Patient N° 5 | 6.9 | 46.2 | 106.5 | 91.1 | 50 848 | 48 104 | 3 091 | 2.49 |
| Patient N° 6 | 6.0 | 39.2 | 94.7 | 90.2 | 50 642 | 48 046 | 2 966 | 2.52 |
| Patient N° 7 | 9.2 | 53.9 | 148.4 | 94.4 | 51 553 | 48 760 | 3 166 | 2.55 |

[a] total bases sequenced;

[b] number of sequencing reads aligning to the reference genome;

[c] mean depth of coverage across the target region;

[d] transitions over Transvertions ratio.