**Table S3** Family verification results.

|  |  |  |  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
|  | | | | | | | | | | | | | |
| The results of the first-generation sequencing verification are as follows: | | | | | | | | | | | | | |
|  | Gene | Site | Reference gene | II-1  (The index case) | II-2 | II-3 | II-4 | III-1 | III-2 | III-3 | III-4 | III-5 |  |
|  | *AFG3 L2* | c.C316T | CC | CT | CC | CT | CC | CT | CC | CC | CC | CC |
|  | *PPP2 R2B* | rs56261308 2 | GG | GA | GA | GA | GA | GG | GG | GA | GG | GG |
|  | *SLC1 A3* | rs11758869 7 | AA | AG | AA | AG | AA | AA | AA | AA | AA | AA |
|  | *SPG7* | c.C2062T | CC | CT | CC | CT | CT | CC | CC | CC | CC | CC |
|  | *SPG7* | rs12960 | GG | GA | GG | GA | GA | GG | GG | GG | GG | GG |
|  | *SPG7* | c.1150\_1150-1  insCTAC | CC | C/GCT AC | C/GCT AC | C/GC  TAC | C/GCT  AC | C/GC  TAC | C/GC  TAC | CC | C/GC  TAC | C/GC  TAC |