Supplementary Table 1. Clinical and genetic information of the patients without PPRT2 mutation.

|  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| Family | Patient | Inheritance | Gender | Age(y) | PKD(y) | IC(m) | Duration of attacks(s) | Frequency | Phenotype and laterality |
| 13 | I:1 | AD | M | 39 | 12 | None | <5 | 1-3 attacks/d | Bilateral dystonic movements |
|  | II:1p | AD | F | 12 | 10 | None | 5-10 | 2-5 attacks/d | Bilateral dystonic movements |
| 14 | I:2 | AD | F | 41 | 14 | None | 15-20 | 5-10 attacks/d | Bilateral dystonic movements |
|  | II:1p | AD | M | 18 | 15 | None | 5-10 | 5-10 attacks/d | Right-sided dystonic movements |
| 15 | I:1 | AD | M | 45 | 11 | None | <5 | 2-5 attacks/d | Bilateral dystonic movements |
|  | II:1p | AD | M | 16 | 8 | None | <5 | 1-2 attacks/d | Left-sided dystonic movements |
| 16 | I:1 | AD | F | 41 | 8 | 6 | 10-15 | 5-10 attacks/d | Bilateral dystonic movements |
|  | II:1p | AD | F | 20 | 15 | None | <5 | 2-5 attacks/d | Left-sided dystonic movements |
| 17 | I:2 | AD | F | 53 | 13 | None | <5 | 5-10 attacks/d | Left-sided dystonic movements |
|  | II:1 | AD | F | 32 | 12 | None | <5 | 2-5 attacks/d | Left-sided dystonic movements |
|  | II:3 | AD | M | 29 | 13 | None | <5 | 5-10 attacks/d | Bilateral dystonic movements |
|  | III:1p | AD | M | 10 | 9 | None | <5 | 5-10 attacks/d | Bilateral dystonic movements |
| 18 | Case 44 | Sporadic | M | 20 | 18 | None | <5 | 20-30 attacks/d | Left-sided dystonic movements |
| 19 | Case 45 | Sporadic | M | 12 | 11 | None | <5 | 5-10 attacks/d | Right-sided dystonic movements |
| 20 | Case 46 | Sporadic | M | 12 | 12 | None | 5-10 | 5-10 attacks/d | Right-sided dystonic movements |
| 21 | Case 47 | Sporadic | M | 16 | 13 | None | 5-10 | 2-5 attacks/d | Right-sided tonic movements |
| 22 | Case 48 | Sporadic | M | 21 | 21 | None | <5 | 10-20 attacks/d | Left-sided dystonic movements |
| 23 | Case 49 | Sporadic | M | 12 | 12 | None | 5-10 | 1-2 attacks/d | Bilateral dystonic movements |
| 24 | Case 50 | Sporadic | M | 17 | 14 | None | <5 | 2-5 attacks/d | Bilateral dystonic movements |
| 25 | Case 51 | Sporadic | M | 23 | 13 | None | <5 | 2-5 attacks/d | Right-sided dystonic movements |
| 26 | Case 52 | Sporadic | M | 20 | 14 | None | <5 | 1-2 attacks/d | Bilateral dystonic movements |
| 27 | Case 53 | Sporadic | F | 22 | 15 | None | 5-10 | 5-10 attacks/d | Bilateral dystonic movements |
| 28 | Case 54 | Sporadic | M | 26 | 14 | None | <5 | 2-5 attacks/w | Bilateral dystonic movements |
| 29 | Case 55 | Sporadic | M | 20 | 12 | None | 5-10 | 2-5 attacks/d | Right-sided dystonic movements |
| 30 | Case 56 | Sporadic | M | 18 | 18 | None | 5-10 | 5-10 attacks/d | Left-sided dystonic movements |
| 31 | Case 57 | Sporadic | M | 15 | 10 | None | 5-10 | 2-5 attacks/d | Left-sided tonic movements |
| 32 | Case 58 | Sporadic | M | 15 | 14 | None | <5 | 5-10 attacks/d | Right-sided dystonic movements |
| 33 | Case 59 | Sporadic | M | 14 | 13 | None | 5-10 | 1-2 attacks/d | Right-sided dystonic movements |
| 34 | Case 60 | Sporadic | F | 18 | 11 | None | <5 | 1-2 attacks/d | Right-sided dystonic movements |
| 35 | Case 61 | Sporadic | M | 15 | 12 | None | <5 | 2-5 attacks/d | Bilateral dystonic movements |
| 36 | Case 62 | Sporadic | F | 32 | 12 | None | 5-10 | 10-15 attacks/d | Left-sided dystonic movements |
| 37 | Case 63 | Sporadic | F | 16 | 9 | None | <5 | 1-2 attacks/d | Bilateral dystonic movements |
| 38 | Case 64 | Sporadic | F | 15 | 10 | None | 5-10 | 2-5 attacks/d | Bilateral dystonic movements |
| 39 | Case 65 | Sporadic | F | 17 | 14 | None | <5 | 5-10 attacks/d | Left-sided dystonic movements |
| 40 | Case 66 | Sporadic | M | 23 | 20 | None | 5-10 | 1-2 attacks/d | Bilateral tonic movements |
| 41 | Case 67 | Sporadic | F | 14 | 11 | None | <5 | 2-5 attacks/d | Bilateral dystonic movements |
| 42 | Case 68 | Sporadic | M | 18 | 14 | None | <5 | 5-10 attacks/d | Bilateral dystonic movements |
| 43 | Case 69 | Sporadic | M | 11 | 10 | None | <5 | 2-5 attacks/d | Bilateral dystonic movements |
| 44 | Case 70 | Sporadic | M | 24 | 8 | None | 20-30 | 10-20 attacks/d | Left-sided dystonic movements |
| 45 | Case 71 | Sporadic | M | 17 | 10 | None | <5 | 1-2 attacks/d | Bilateral dystonic movements |
| 46 | Case 72 | Sporadic | M | 28 | 13 | None | 10-20 | 2-10 attacks/d | Bilateral dystonic movements |
| 47 | Case 73 | Sporadic | M | 12 | 11 | None | 5-10 | 2-5 attacks/w | Bilateral dystonic movements |
| 48 | Case 74 | Sporadic | M | 15 | 10 | None | 20-30 | 10-20 attacks/d | Bilateral dystonic movements |
| 49 | Case 75 | Sporadic | M | 16 | 10 | 10 | <5 | 5-10 attacks/d | Left-sided dystonic movements |
| 50 | Case 76 | Sporadic | M | 14 | 8 | None | 20-30 | 5-10 attacks/d | Bilateral dystonic movements |
| 51 | Case 77 | Sporadic | M | 31 | 15 | None | <5 | 2-5 attacks/d | Bilateral dystonic movements |

AD, autosomal dominant; p, proband.