Supplementary information

Table S1. A list of references from which 184 published families were selected for penetrance analysis.

Table S2. Detailed information of 184 published families. For each family, we documented the related reference, the *PRRT2* variant, the pathogenicity classification of variants, the family history, the ethnic origin, the total number of variant carriers and affected ones, the total number of obligate carriers and affected ones.

Table S3. Detailed information of 353 obligate carriers in 184 published families. For each obligate carrier, we recorded the related reference and family ID, the *PRRT2* variant, the gender, the phenotype, and the origin of the variant (parental transmission), if available.

Table S4. Detailed information of 100 variant carriers identified in our cohort. For each variant carrier, we recorded the family history, the *PRRT2* variant, the gender, the phenotype, the parental transmission and whether an obligate carrier or not.