Table e-1: Summary of previously reported pathogenic variants in XPNPEP3 and their associated phenotypes. *Reduced activity in muscle and fibroblasts. **Reduced activity in fibroblasts. aThis patient has pancreatic cysts whereas his sibling b has hepatopathy. Like our patient, patients A1 and A3 also have gout.

Key: ET: essential tremor; Complex I: CI; Complex IV: CIV; No: N; Yes: Y; Not reported: NR; Ragged red fibers: RRF; Seizures: SZ.

References
