

Patient	Performed Test	Result
Case 1	WES	Homozygous
Case 2	Targeted Mutation	Homozygous
Father	Targeted Mutation	Heterozygous
Mother	Targeted Mutation	Heterozygous
Unaffected Brother	Targeted Mutation	Heterozygous

**Supplementary 1.** Genetic testing results identifying a likely pathogenic variant in the *PTRH2* gene (NM\_001015509.2: c.114dup, p. Gly39Trpfs\*16) according to American College of Medical Genetics classification.