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The clinical phenotype and heterogeneity of homozygous mutation in PTRH2 gene

PTRH2 is an evolutionarily highly conserved mitochondrial protein that belongs to a family of peptidyl-tRNA hydrolases. Recently, patients from two consanguineous families with mutations in the PTRH2 gene were reported. Global developmental delay associated with microcephaly, growth retardation, progressive ataxia, distal muscle weakness with ankle contractures, demyelinating sensorimotor neuropathy, and sensorineural hearing loss were present in all patients, while facial dysmorphism with widely spaced eyes, exotropia, thin upper lip, proximally placed thumbs, and deformities of the fingers and toes were present in some individuals. Here, we report a new family with three siblings affected by sensorineural hearing loss and peripheral neuropathy. Autozygosity mapping followed by exome sequencing identified a previously reported homozygous missense mutation in PTRH2 (c.254A>C; p. (Gln85Pro)). Sanger sequencing confirmed that the variant segregated with the phenotype. In contrast to the previously reported patient, the affected siblings had normal intelligence, milder microcephaly, delayed puberty, myopia, and moderate insensitivity to pain. Our findings expand the clinical phenotype and further demonstrate the clinical heterogeneity related to PTRH2 variants.

Recent Publications

1. Doe J, Kaindl AM, Jijiwa M, de la Vega M, Hu H, Griffiths G et al (2017) PTRH2 gene mutation causes progressive congenital skeletal muscle pathology. Hum Mol Genet 26(8):1458-1464.
2. Hu H, Matter ML, Issa-Jahns L, Jijiwa M, Kraemer N, Kaindl AM et al (2016) Mutations in PTRH2 cause novel infantile-onset multisystem disease with intellectual disability, microcephaly, progressive ataxia, and muscle weakness. Ann Clin Transl Neurol 1(12):1024-35.
3. Picker-Minh S, Mignot C, Doummar D, Hashem M, Faqeih E, Josset P, Dubern B, Alkuraya FS, Kraemer N and Kaindl AM (2016) Phenotype variability of infantile-onset multisystem neurologic, endocrine and pancreatic disease (IMNEPD). Orphanet J Rare Dis. 11(1):52.
4. Alazami AM, Patel N, Shamseldin HE, Anazi S, Al-Dosari MS, Alkuraya FS et al (2015) Accelerating novel candidate gene discovery in neurogenetic disorders via whole-exome sequencing of prescreened multiplex consanguineous families. Cell Rep 10(2):148-161.
5. Ishii T, Funakoshi M and Kobayashi H (2006) Yeast PTRH2 is a UBL domain-binding protein that participates in the ubiquitin-proteasome pathway. EMBO J 25(23):5492-5503.

Biography

Muhammad Mahajnah is an Associate Professor of Pediatrics and Pediatric Neurology at the Technion Faculty of Medicine, Israel. He completed his Medical degree in 1992 at Bruce and Ruth Rappaport Faculty of Medicine, Technion, Israel and his PhD degree in 1998. He was trained in Pediatrics at Carmel Medical Center and completed fellowship in Pediatric Neurology at Schneider Children Medical Center, Tel Aviv. He has worked with children neurological disorder for about 20 years and has special interest in neurodevelopmental disorders and neuro genetic disorders and devotes his time to both clinical work and research.

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