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MICPCH syndrome – a rare cause of NPMD

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Abstract

MICPCH syndrome (mental retardation and microcephaly with pontine and cerebellar hypoplasia) is a very rare (according to Orphanet, prevalence is under one in a million cases) X-linked dominant disease produced by heterozygous pathogenic mutations in CASK (Xp11.4), a gene involved in brain development. The main features of this syndrome are microcephaly with pontocerebellar hypoplasia, severe global developmental delay, limb spasticity, dystonic movements, stereotypic behaviours, seizures, various ocular manifestations, sensorineural deafness, as well as non-specific facial dysmorphism. Up to the present time, more than 50 females and a very small number of males with this syndrome have been reported. We aim to review the main features of this syndrome and to present the case of a three year old girl with normal family history admitted to our hospital for medical recovery who presented a MICPCH phenotype; genetic testing revealed a never before reported intronic variant in CASK gene. Given the overlap between the patient's phenotype and the CASK gene mutations phenotype, we believe this variant might be pathogenic.

Keywords: *MICPCH, neuropsychomotor developmental delay, CASK gene*