Pitt-Hopkins Syndrome – A Case Report from Pakistan

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Abstract:

Pitt-Hopkins syndrome (PTHS) is a rare neurodevelopmental disorder caused by loss of function of one allele of the Transcription Factor 4 (*TCF4)* gene. We present the first case report from Pakistan where Chromosomal Microarray (CMA) was utilised for diagnosing PTHS. Haploinsufficiency of *TCF4* gene located at 18q21.2 region was identified by CMA. Haploinsufficiency in the *TCF4* has an important role in the development of the nervous system and the ability of the protein to bind to DNA and initiate neuronal differentiation, contributing to the neurological symptoms seen in PTHS. We present an overview of the clinical presentation and diagnostic workup, ultimately leading to the diagnosis of PTHS.