ISSN 0972-3757
International Journal of
HUMAN GENETICS

PRINT: ISSN 0972-3757 ONLINE: 2456-6360
Int J Hum Genet, 13(4): 195-199 (2013)
DOI: 10.31901/24566330.2013/13.04.04

Early Diagnosis of the Chromosomal Deletion 5q14.2-q21.3 in a Preterm Newborn: Case Report

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KEYWORDS 5q Deletion Syndrome. Deletion 5q14.2-q21.3. Epilepsy. Cerebellar Hypoplasia. Psychomotor Retardation. Horseshoe Kidney. Ventricular Septal Defect. Prematurity. *MEF2C*. Newborn

ABSTRACT The researchers report a case of a premature baby diagnosed early in the postnatal period with an exceptionally long chromosomal deletion 5q14.2-q21.3 associated with dysmorphic features, epilepsy, cerebellar hypoplasia, psychomotor retardation, ventricular septal defect and a horseshoe kidney. According to clinical signs a standard cytogenetic examination on peripheral blood was performed and revealed an interstitial deletion of chromosome 5. A single nucleotide polymorphism array (SNP) analysis subsequently confirmed the 22.3 Mb long deletion of the long arm of chromosome 5. Within the deleted region 59 RefSeq genes were identified. Thirteen of these were expressed in specialized brain regions during fetal or adult stages of brain development. Among these the *MEF2C* gene seems to be the most limiting in relation to the normal development of brain and muscle. Only a few cases with this deletion have been reported in the literature to date. All cases manifested with severe mental retardation. Epilepsy and dysmorphic features in a newborn should lead to further genetic examination. New molecular genetic techniques such as the SNP array provide a quick and accurate diagnostic tool.