Joubert Syndrome: A Rare Cause of Hypotonia and Developmental Delay in Infancy and Childhood

Pediatricians not uncommonly encounter children with hypotonia and developmental delay. Joubert syndrome is a rare autosomal recessive condition that is associated with hypotonia, developmental delay, abnormal eye movement, breathing problems and cerebellar dysfunction. Joubert syndrome was first described by French neurologist Marie Joubert in 1969.[1] Joubert syndrome is an autosomal recessive disorder with significant genetic heterogeneity that is associated with cerebellar vermis hypoplasia and the ponto-mesencephalic molar tooth sign, deepening of the interpenduncular fossa with thick and straight superior cerebellar penduncles. It is associated with hypotonia, ataxia, and characteristic breathing abnormalities including episodic apnea and hyperpnea, global developmental delay, nystagmus, strabismus and oculomoter apraxia.[2]

A two years old male child was brought to us for evaluation of development delay and abnormal eye movements. He was unable to stand or walk without support. He was the first child, born at term, to nonconsanguineous parents by cesearian section in view of meconium stained liquor,. His birth weight was 3.5 kgs and was admitted to neonate intensive care unit (NICU) for about 10 days in veiw of respiratory distress. There was no history of seizures and feeding or swallowing difficulty. He had gross developmental delay in all the fields. He was not able to walk and even stand, but he was able to hold his head and roll over and his developmental motor age was approximately 6 months. He was able to speak monosyllables only.

On examination, he looked alert and awake. His head circumference was within normal limits. His vitals were normal. His ocular examination was normal except bilateral horizontal nystagmus. There were no neurocutaneous markers. Neurological examination revealed hypotonia and sluggish deep tendon reflexes. Respiratory and cardiac examinations were normal. Echocardiography and ultrasound abdomen were normal. CT scan brain was done which showed enlarged fourth ventricle and elongated cerebellar penduncles with vermian hypoplasia, i.e. “Molar tooth sign”. Based on these clinical features and neuroimaging a diagnosis of Joubert's syndrome was made.

Joubert's syndrome is an autosomal recessive disorder characterized by hypotonia, poor body balance, and developmental delay, respiratory abnormalities characterized by tachypnea followed by apnea.[3] This syndrome was first described by Joubert and colleagues as a familial agenesis of the cerebellar vermis. The Diagnostic criteria of Joubert's syndrome have not been established, the clinical features frequently mentioned as essential for the diagnosis of classic Joubert's syndrome comprise - 1) Hypotonia in infancy 2) Developmental delay/ Mental retardation 3) One or both of the following (not absolutely required, but helpful for diagnosis) A) Irregular breathing pattern in infancy (Intermittent tachypnea and/or apnea) B) Abnormal eye movement.[4]

Neuroimaging of head in the axial plane demonstrate the “Motor tooth sign”, deep posterior interpeduncullar fossa, thick and elongated superior cerebellar penduncles and hypoplasia or aplasia of vermis. Amin et al and Singh et al also reported similar finding that is developmetal delay, hypotonia, and nystagmus.[5,6] Therefore, in any child presenting with hypotonia, developmental dealy and characteristic neuroimaging findings, diagnosis of Joubert syndrome should be suspected.

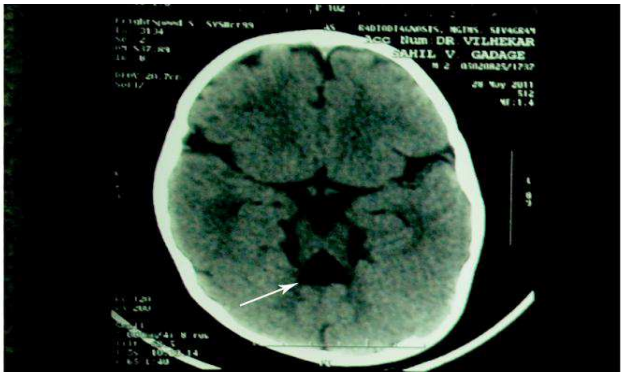

Figure 1: CTscan- Enlarged fourth ventricle and enlongated cerebrellar penduncles with vermian hypoplasia –Molar Tooth Sign.

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