Joubert Syndrome Mimicking Hypotonic Cerebral Palsy

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To the Editor: Joubert syndrome (JS) is a rare autosomal recessive disorder frequently presented as infantile hypotonia, mental retardation, abnormal breathing patterns and abnormal eye movements [1]. Hypotonic cerebral palsy is a rare type of cerebral palsy (CP), mainly due to a non-progressive insult to the developing brain, presents in children with reduced tone and delayed motor milestones [2].

An 8-mo-old girl was referred as a case of hypotonic CP for further management. She was born to a non-consanguineous parents after a full-term pregnancy with no significant antenatal/neonatal history. She had hypotonia, squint, nystagmus and abnormal rotatory eye movements. Her developmental age was equivalent to 4 mo. The family history was negative for similar illness. Her Ultrasound abdomen, complete blood count, renal function and liver function tests were normal. The brain MRI showed *molar tooth appearance* with deep interpenducular fossa and thickened superior cerebellar peduncle surrounding on oblong shaped fourth ventricle and small dysmorphic clefted vermis. All findings were consistent with Joubert syndrome.

Joubert syndrome is a heterogenous condition with long-term developmental and neurological consequences. Retinal dysplasia, coloboma, multicystic dysplastic kidney, hepatic fibrosis and polydactyly are other systemic associations in Joubert syndrome [3]. The presence of the molar tooth sign

and vermian hypoplasia is a hallmark for the diagnosis of Joubert syndrome [3].

Since there are overlapping clinical presentations in Joubert syndrome and hypotonic CP, Joubert syndrome should be considered as differential diagnosis of hypotonic CP despite its low incidence. The brain MRI is important to make a diagnosis of JS. The prompt and early diagnosis of JS will help in identification of other associated abnormalities and management.

Compliance with Ethical Standards

Conflict of Interest None.

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