Development of hydrocephalus in a patient with Joubert syndrome

Sir, Joubert syndrome is a rare autosomal-recessive condition characterized by dysgenesis of the cerebellar vermis, hypotonia, ataxia, abnormal eye movements, mental retardation, episodic hyperpnoea and apnoea.1

We report a patient diagnosed as Joubert syndrome during the neonatal period who developed cystic dilation of the posterior fossa communicating with the fourth ventricle with hydrocephalus at one year of age. He was evaluated as Joubert-plus anomaly and this patient is reported to emphasize the rare development of hydrocephalus during follow-up.

A two-day-old neonate was admitted for evaluation of abnormal breathing started at two hours after his birth. He was born with caesarean section from a 21-year-old primiparous mother and had a birth weight of 3190 g. His parents were first-degree relatives, his two aunts were mentally retarded and his uncle had lack of vision.

On physical examination the weight was 3170 g (25-50 percentile), length was 49 cm (25 percentile) and head circumference was 35 cm (25-50 percentile), heart rate was 142 per minute and blood pressure was 70/40 mmHg. He had an abnormal breathing pattern with apnoea periods lasting more than 20 seconds alternating with periods of hyperpnoea increasing up to 100 per minute. During sleep, this respiratory abnormality was absent. Neurological evaluation revealed irregular, jerky eye movements, nystagmus and muscular hypotonia. Fundus was normal.

The laboratory investigations including haemogram, urinalysis, blood sugar, electrolytes, liver and renal function tests, and acid-base values were normal. Chest X-ray, electroencephalogram, electrocardiography, echocardiography and abdominal ultrasonography were normal. In his cranial computerised tomography scanning, the fourth ventricle was in normal localization but showed an abnormal triangular shape. He had deeper than normal posterior interpeduncular fossa and cerebellar vermis was not visualized.

In the differential diagnosis of this patient with an abnormal breathing pattern, infections, cardiovascular and respiratory system problems, thermoregulation and electrolyte disturbances were investigated. The patient was diagnosed as Joubert syndrome including clinical features of hypotonia, abnormal breathing pattern, nystagmus and vermic agenesis. Having first-degree relativity between the parents and two mentally retarded aunts supported the diagnosis. The patient could not be followed up until one year of age. His physical examination at one year revealed macrocephaly, severe motor and mental retardation, hypotonia, vertical and horizontal nystagmus. Cranial magnetic resonance imaging showed dilation of the posterior fossa communicating with the fourth ventricle, ventricular dilation and agenesis of the cerebellar vermis. Paediatric neurosurgery consultation was obtained and ventriculoperitoneal shunt was planned for hydrocephalus.

Hypotonia, ataxia and developmental delay with cognitive impairment are three key clinical features of the Joubert syndrome. Ocular and oculomotor abnormalities are common. In the neonatal period most children have episodic apnoea alternating with tachypnoea which is helpful in making a diagnosis.1 Neuroimaging of the head in the axial plane demonstrates the molar tooth sign with deep posterior interpeduncular fossa, thick and elongated superior cerebellar peduncles, and hypoplastic or aplastic superior cerebellar vermis.2

Other central nervous system abnormalities, including a hypoplastic brainstem, cortical atrophy, corpus callosum agenesis, delayed myelination and enlarged fourth ventricle have been described in the Joubert syndrome.3,4 The Joubert-plus anomaly has been defined as the Joubert malformation plus additional anomalies of either the mesencephalon or the caudal fourth ventricle.2 Anderson et al reported a patient with clinical and radiographic features consistent with Joubert syndrome who presented with congenital hydrocephalus.3 Maria BL et al have stressed that 1 in 10 patients with Joubert syndrome has abnormal cerebrospinal fluid collections misdiagnosed as Dandy-Walker variants. In these patients genetic heterogeneity or epigenetic factors have accounted for abnormal cerebrospinal fluid collections.5 In our patient who was diagnosed as Joubert syndrome during the neonatal period, cystic dilation of the posterior fossa communicating with the fourth ventricle developed at one year of age and this observation was further evaluated as Joubert-plus anomaly. This patient has been reported to emphasize the rare development of hydrocephalus that could be confused with Dandy-Walker variants in patients with Joubert syndrome.

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