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A child with delayed milestones and interesting findings on MR

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DESCRIPTION

A 13-month-old male child born of a second degree consanguineous marriage presented with delayed milestones; notably, he was not able to pull to sit or stand and he was swaying from side to side when pulled to sit.

He had bilateral post-axial polydactyly¹ (figure 1). He never experienced any respiratory difficulty and there was no history of apnoeic episodes.

The central nervous system examination was remarkable for generalised hypotonia and gross developmental delay. The child was not able to pull to sit or stand. On attempted sitting he showed truncal ataxia and on approaching an object showed premature grasp and tremulousness. There was no nystagmus. He had started speaking non-specific 'ma' and 'pa'.

Ocular fundi were normal.

Sonography of the abdomen did not show any renal cyst.

MRI of the brain showed features commonly associated with Joubert syndrome and Joubert syndrome related disorders² (figure 2), for example:

dilatation of the fourth ventricle with bat-wing shaped appearance, elongation and stretching of the superior cerebellar peduncles—the molar tooth appearance, absence of the cerebellar vermis.

The distinctive radiological feature of Joubert syndrome in MRI is molar tooth sign, which has been reported in about 85% patients. This results from dysplastic and thick superior cerebellar peduncles, and deep and wide interpeduncular cisterns. The bat-wing appearance emanates from the dilated fourth ventricle and an anteriorly convex floor. The other sign described, is a buttock sign, because the hypoplasia of vermis causes the cerebellar hemisphere to come closer. Hypoplasia of corpus callosum is also described.³

Figure 1 Post-axial polydactyly.

Figure 2 MRI brain showing molar tooth appearance, bat-wing dilatation of the 4th ventricle and absence of cerebellar vermis.

Figure 3 Extra strand of tissue below tongue (oral frenulae).

Moreover, other syndromes with posterior-fossa malformations, including Dekaban-Arima, Senior-Löken and cerebellar vermis hypoplasia/aplasia, oligophrenia, ataxia, coloboma and hepatic fibrosis, frequently lead to diagnostic dilemmas. However, the presence of the same clinical and neuroradiological features is shared by at least eight distinct conditions under the term Joubert syndrome related disorders (JSRD). Presence of ataxia and findings on neuroimaging should lead to suspicion of Joubert syndrome and related disorders, which can be confirmed and typified by genetic testing—this was refused by the family.

The association of primary criteria of Joubert syndrome and polydactyly may be suggestive of JSRD such as oro-facial-digital VI syndrome (Varadi-Papp syndrome). This refers to one type of oro-facial disorder. This child also showed extra strands of tissue below the tongue—the oral frenulae (figure 3). However, this case did not show other orofacial features like cleft palate, or abnormalities of tongue, teeth and other facial dysmorphism.

Oro-facial digital syndrome II (Mohr syndrome), an autosomal recessive disorder, has been described with oral anomalies (Bifid tongue, tongue nodules, enlarged mouth

frenuli, multiple frenuli) and digital anomalies (syndactyly, post axial polydactyly, hallucal abnormalities). Cases of Mohr syndrome with cerebellar atrophy and other cerebellar anomalies are described.⁴ The overlap between Joubert syndrome and oro-facial-digital syndrome has also been described by Egger.⁵

Our case presented with developmental delay, cerebellar signs, MRI having features associated with Joubert syndrome and some oro-facial-digital anomalies. Absence of respiratory symptoms makes this a very interesting case.

Competing interests None.

Patient consent Obtained.

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