Congenital facial nerve aplasia: MR depiction of a rare anomaly

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Abstract

We report two infants presenting with unilateral congenital facial palsy since birth. Magnetic resonance imaging (MRI) in both the cases revealed complete unilateral aplasia of facial nerve. To our knowledge, this is the first reported MR depiction of nonsyndromic isolated facial nerve aplasia. Imaging features and the pertinent anatomy is discussed along with a brief review of literature.

Key words: Agenesis; congenital facial nerve palsy; facial nerve aplasia; magnetic resonance imaging

Introduction

Congenital facial nerve palsy is clinically defined as unilateral or bilateral paralysis of facial nerve in neonatal period. Etiology includes intrapartum injury or various developmental disorders. Facial nerve aplasia is an extremely rare anomaly that usually occurs in association with Moebius syndrome. Even rarer is the occurrence of isolated aplasia of facial nerve, with only few cases reported in the literature.[1] Although aplasia or hypoplasia of the facial nerve is rare, their recognition is clinically important for its prognostic and medicolegal implications. The advent of high resolution three-dimensional constructive interference steady state (3D CISS) magnetic resonance (MR) sequence enables us to achieve detailed and precise evaluation of cranial nerves because they provide excellent contrast resolution between cerebrospinal fluid (CSF) and nerves owing to their ability to generate a strong signal in tissues having a high T2/T1 ratio such as CSF.

Case Reports

Case 1

A 6-month-old boy, born at full term by normal vaginal delivery, presented to us with facial asymmetry since birth. Magnetic resonance imaging (MRI) in both the cases revealed complete unilateral aplasia of facial nerve. To our knowledge, this is the first reported MR depiction of nonsyndromic isolated facial nerve aplasia. Imaging features and the pertinent anatomy is discussed along with a brief review of literature.

**Access this article online**

Quick Response Code:

Website: www.ijri.org

DOI: 10.4103/0971-3026.195791

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and diffusion weighted imaging. In addition, 3D-CISS was also obtained to evaluate facial nerve and other cranial nerves. Brain stem, bilateral cerebral, and cerebellar parenchyma revealed no significant abnormality on conventional sequences. However, 3D CISS images showed absence of right-sided facial nerves throughout its course with presence of only vestibulocochlear nerve in the cisternal portion of internal auditory canal [Figure 2]. Facial nerve was normally seen on the contralateral side. Rest of the cranial nerves were normal. Parotid glands were normally seen. On the basis of the clinical and MRI picture, a diagnosis of congenital facial nerve agenesis was made and explained to the parents. Interestingly, mild but obvious improvement in the facial functions was observed on 3-months follow-up in the form of decreased left-sided deviation of the angle of mouth. The degree of eye closure, however, did not improve.

**Case 2**

A 2-month-old boy presented with facial asymmetry and inability to close the left eye while sleeping. There was no history of birth trauma or birth asphyxia. Child cried normally after birth. On examination, child was unable to close the left eye and showed deviation of the angle of mouth toward the right side. Absent left nasolabial fissure was seen [Figure 3A]. Rest of the neurological and systemic examinations were normal. A diagnosis of congenital facial nerve palsy was made and MRI was performed to look for the cause using routine institutional protocol with the addition of 3D CISS sequences to evaluate cranial nerves. Conventional MR sequences of the brain showed no significant abnormality. 3D-CISS sequences revealed only one cranial nerve, consistent with vestibulocochlear nerve, in left the cerebellopontine angle entering the left internal acoustic meatus [Figure 3B-D]. Left facial nerve was absent. On the right side, vestibulocochlear and facial nerves were normally visualized. Nerves in bilateral internal auditory canal could not be delineated clearly due to the small size of the canal. On 2-months follow-up, the patient did not show clinical improvement.

**Discussion**

Congenital facial nerve palsy is a rare condition with estimated incidence of approximately 0.8–2.1 per 1000 live births.\(^2,3\) Various causes of congenital facial palsy have been broadly classified in two categories—traumatic and developmental. Facial nerve can get injured because of its relatively superficial extracranial course. These causes include intrapartum trauma either due to forceps during assisted delivery or compression of the nerve against maternal bony protuberance during birth,\(^4\) or abnormal intrauterine posture leading to compression of the nerve between jaw and shoulder.\(^5\) Developmental causes include isolated cases of aplasia/hypoplasia of facial nerve or their nuclei or various syndromic associations such as Poland syndrome (congenital facial palsy with absent pectoralis major muscle), Goldenhaar’s syndrome (unilateral facial hypoplasia), Moebius syndrome (variable degree of agenesis/hypoplasia of the 6th and 7th cranial nerves/nuclei), and cardiofacial syndrome (weakness of the facial muscles).\(^6\)

Facial nerve aplasia has been reported as part of the Moebius syndrome, also known as congenital oculofacial paralysis or congenital facial diplegia, characterized by bilateral facial and abducentes nerve palsy.\(^7\) Verzij et al. studied six patients of Mobius syndrome on 3D CISS and magnetization-prepared rapid gradient-echo MR
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Figure 3 (A-D): A 2-month-old boy (A) showing incomplete eye closure, absence of forehead wrinkle, and loss of nasolabial fold on the left side and deviation of angle of mouth toward the right. Axial MRI (B) shows two nerves entering the right IAC (straight arrow) with only one nerve (vestibulocochlear) on the left side (curved arrow). Sagittal image on the right side (C) shows posteriorly placed vestibulocochlear nerve (white arrow) and a small anteriorly placed facial nerve (black arrow). Left-sided sagittal image (D) shows only one nerve (curved arrow), the vestibulocochlear nerve

sequences and confirmed the absence of facial nerve and significant brainstem hypoplasia in all the patients. There are very few cases of isolated facial nerve agenesis/hypoplasia in the literature. Facial nerve hypoplasia has also been reported to be associated with abnormalities of other cranial (especially vestibulocochlear) nerves and ipsilateral parotid gland agenesis. The first case of isolated complete congenital facial nerve agenesis was reported by Jervis et al. where diagnosis was made incidentally during a surgical procedure for an unrelated condition. We extend the scant literature reporting isolated non syndromic facial nerve agenesis, and to the best of our knowledge, provide maiden demonstration of this condition on imaging, with the help of 3D CISS MR sequence.

Facial nerve has a long tortuous course which consists of six segments, namely, cisternal, intracanalicular, labyrinthine, tympanic, mastoid, and extracranial. High resolution computed tomography scan can be used to assess the bony margins and calibre of the internal acoustic canal (IAC). An anteroposterior diameter of less than 2 mm is considered stenotic. MRI is the diagnostic modality of choice to evaluate cisternal and intracanalicular facial nerve segments. It is best evaluated on high-resolution T2-weighted 3D CISS sequences. Within the cerebellopontine angle cistern, facial nerve can be seen anterior to vestibulocochlear nerve extending anteriorly and laterally between the pontomedullary junction and porus acusticus, entering IAC. Evaluation of intracanalicular portion is best done in sagittal oblique images where these nerves can be seen in cross-section, perpendicular to their long axis. The facial nerve lies in the anterior superior aspect of the canal for a length of approximately 7–8 mm. The cochlear nerve travels in the anterior inferior portion of the canal. The superior and inferior vestibular nerves are positioned in their respective portions of the posterior canal [Figure 2]. Absence of or thinning of facial nerve within this canal fetches the diagnosis of aplasia or hypoplasia, respectively. Associated aplasia/hypoplasia of other cranial nerves, especially vestibulocochlear nerve, should also be looked for. It is important to be mindful of the variations in the intracanalicular course of facial nerve which, if overlooked, can ostensibly lead to a diagnosis of aplasia. Notable among these variations are (a) facial nerve ensconced in a completely separate canal parallel to IAC, (b) early exit of the facial nerve form IAC in its middle third and continuation in a separate canal, and (c) the nerve can pass between the temporal bone and the temporal lobe. Occasionally, a fibrous cord can mimic a hypoplastic facial nerve. Clinically, it is important to differentiate between the traumatic and developmental causes of congenital facial palsy because of prognostic and medicolegal implications. Traumatic causes, suggested by other features of birth trauma such as ecchymoses and hemoperitoneum have excellent prognosis where improvement occurs within a few weeks unless the nerve is completely torn. Facial nerve agenesis along with other developmental causes of congenital facial palsy usually ensue poor prognosis. However, in a study of 6 patients of Mobius syndrome, Verzij et al. reported residual function in some facial muscles despite the absence of facial nerves in all of these cases. He attributed this unusual distribution of the facial weakness to aberrant innervations of some of the facial muscles by other cranial nerves such as the trigeminal, hypoglossal, or glossopharyngeal nerves. This phenomenon can explain the clinical improvement observed on follow-up in the first case of the present study.

The present report of two cases shows that MRI plays a crucial role in the diagnosis of developmental etiologies of congenital facial nerve palsy and should be included in the diagnostic algorithm. Knowledge of the MR picture of facial nerve aplasia, complemented with a pedant scrutiny of the MR images can enable the Radiologists and Neurologists to procure this rare diagnosis which helps in clinical management and prognostication.

Declaration of patient consent
The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and
Financial support and sponsorship
Nil.

Conflicts of interest
There are no conflicts of interest.

References