

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.

To the best of our knowledge, we report the first report of MR imaging (MRI) depiction of two infants with isolated facial nerve aplasia.

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. Since birth, mother of the child had observed incomplete eye closing on the right side while sleeping. There was no history of birth trauma or facial paralysis in the family. Physical examination showed an obvious facial asymmetry, incomplete eye closing on the right side, deviation of the angle of mouth toward the left side, and right-sided loss of nasolabial furrow [Figure 1]. Examination of the rest of the nervous system did not reveal any other significant abnormality. Systemic examination was normal. Patient was referred to the Radiology department to rule out intracranial etiology. MRI was performed using 1.5 T superconducting system (Magnetom Avanto, Seimens Medical System, Erlangen, Germany). Routine MR sequences were performed as per institutional protocol, which include fast spin echo T1 (axial), T2 (axial, coronal, and sagittal), fluid-attenuated inversion recovery (axial)

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Figure 1: Clinical photograph of the 6-month-old boy with right congenital facial nerve palsy showing facial asymmetry, incomplete eye closing on the right side, and right-sided loss of nasolabial furrow

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.

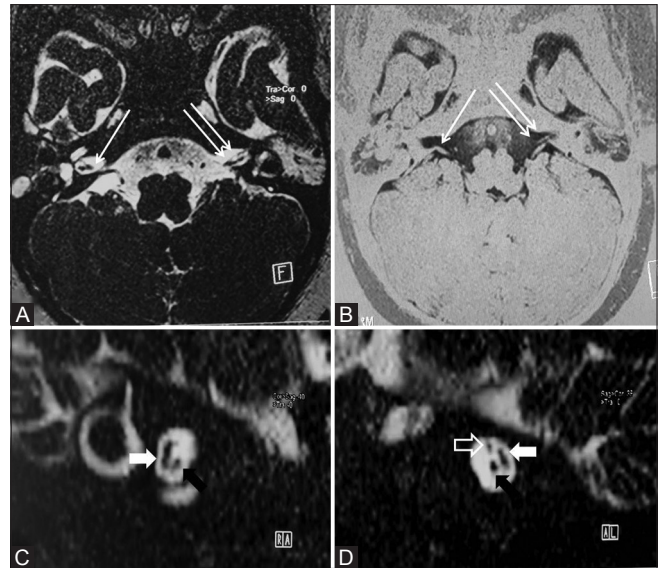


Figure 2 (A-D): Axial MRI (A and B) shows two nerves (facial and vestibulocochlear) entering the left internal acoustic canal (double straight arrows). Only one nerve (vestibulocochlear) is seen on the right side. Oblique sagittal T2-weighted images through the right canal (C) shows the dumbbell shaped with anteroinferior cochlear nerve (solid black arrow). The anterosuperior area of the canal, expected site of facial nerve, is empty. Left side (D) shows all four nerves at expected sites (arrows)

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 abducens nerve palsy.^[7] Verzij *et al.* studied six patients of Moebius syndrome on 3D CISS and magnetization-prepared rapid gradient-echo MR

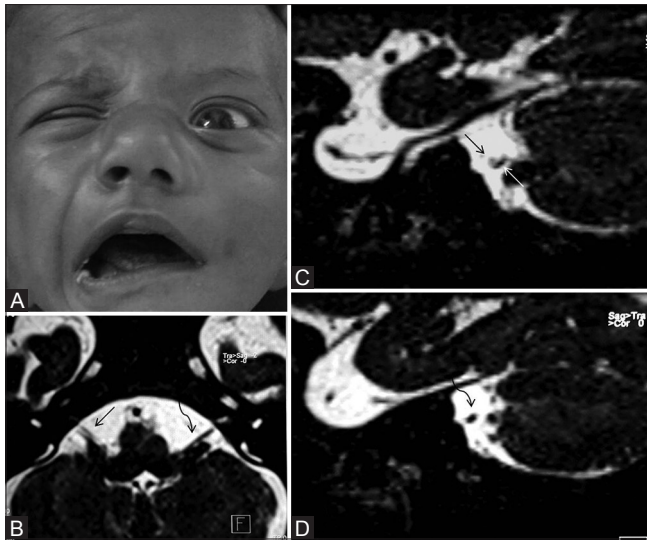


Figure 3 (A-D): A 2-month-old boy (A) showing incomplete eye closure, absence of forehead wrinkle, and loss of nasolabial fold on the left side and deviation of angle of mouth toward the right. Axial MRI (B) shows two nerves entering the right IAC (straight arrow) with only one nerve (vestibulocochlear) 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.^[8] 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^[9] and ipsilateral parotid gland agenesis.^[3] 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.^[1] 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.^[7] 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

acousticus, entering IAC.^[10] 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 from 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.^[10] Occasionally, a fibrous cord can mimic a hypoplastic facial nerve.^[11]

Clinically, it is important to differentiate between the traumatic and developmental causes of congenital facial palsy because of prognostic and medicolegal implications.^[12] 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.^[13] 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.^[8] 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

other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

References

- Jervis PN, Bull PD. Congenital facial nerve agenesis. *J Laryngol Otol* 2001;115:53-4.
- Falcon NA, Ericson E. Facial nerve palsy in the newborn: Incidence and outcome. *Plast Reconstr Surg* 1990;85:1-4.
- Sannagowdara K, Harmelink M, Inglese C, Maheshwari M. Congenital unilateral facial nerve hypoplasia and parotid gland agenesis. *Neurology* 2015;84 (14 Suppl):105.
- Knapp L. Fall von Facialisparese bei einem Neugeborenen nach spontaner Geburt. *Zentralblatt für Gynäkologie* 1896;20:705-8.
- Parmalee AH. Molding due to intra-uterine posture: Facial paralysis probably due to such molding. *Am J Dis Child* 1931;42:1155-9.
- Jemec B, Grobbelaar AO, Harrison DH. The abnormal nucleus as a cause of congenital facial palsy. *Arch Dis Child* 2000;83:256-8.
- Romo LV, Casselman JW, Robson CD. Temporal bone: Congenital anomalies. In: Som PM, Curtin HD, editors. *Head and Neck Imaging*. 4th ed. Vol. 2. St. Louis, Mo: Mosby; 2003. p. 1119-40.
- Verzij HT, Valk J, de Vries R, Padberg GW. Radiologic evidence for absence of the facial nerve in Mobius syndrome. *Neurology* 2005;8:849-55.
- Hamizan AW, Yean KT, Abdullah A. Congenital bilateral facial nerve hypoplasia with sensorineural hearing loss: A case report. *Int J Pediatr Otorhinolaryngol* 2012;76:455-9.
- Curtin HD, Sanelli PC, Som PM. Temporal Bone: Embryology and Anatomy. In: Som PM, Curtin HD, editors. *Head and neck imaging*. 4th ed. St Louis: Mosby Year book; 2003; 1057-1075.
- May M, Schaitkin BM, Shapiro AM. Facial nerve disorders in newborns and children. In: May M, Schaitkin BM, editors. *The Facial Nerve*. New York: Thieme; 2000. p. 358.
- Ozmen OA, Ozgen B, Ozmen S, Sennaroglu L. Congenital facial paralysis: Facial nerve hypoplasia. *Int Adv Otol* 2010;6:282-4.
- Carlo WA, Ambalavanan N. Peripheral nerve injuries. In: Kliegman RM, Stanton BF, St Geme III JW, Schor NF, editors. *Nelson Textbook of Pediatrics*. 20th ed. Philadelphia, PA: Elsevier Inc; 2015. p. 844.