Case Report

Magnetic resonance imaging of congenital unilateral hypoplasia of the face

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Abstract

Facial asymmetry may originate from abnormalities of facial musculature, facial innervation, Lateral facial dysplasias or otomandibular dysostosis. We describe Magnetic Resonance Imaging findings of congenital unilateral hypoplasia in a teenager. **Case Report:** A 15 Year old female referred from plastic surgery department for assessment of congenital unilateral asymmetry of face. Data included relevant history and physical examination and the basic diagnostic work up. The child presented with hypoplastic appearance of facial features on the left side compared to the right side. Asymmetry was more pronounced when she smiled and cried. On examination, the face appeared asymmetric at rest. The left hemiface appeared smaller and showed diffuse deficiency of soft tissue compared to the right side. However the overlying skin appeared normal. No neurological deficit was found in nervous system examination. Magnetic resonance imaging findings revealed diffuse reduction of the subcutaneous fat and muscle tissue on the left side. The left orbital, nasal, maxillary, mandibular bones showed hypoplasia. Muscle bulk was reduced on the left side with smaller submandibular and parotid salivary glands. **Conclusions:** Congenital unilateral hypoplasia of face is a rare anomaly that causes asymmetric of face. Pediatricians and otolaryngologists need to be cognizant of cardiac, head and neck, and central nervous system anomalies associated with congenital unilateral hypoplasia of face. The objective of the treatment is to restore facial symmetry and proper function. Teamwork is essential with a board of members including plastic surgeon, otolaryngologist, orthodontist and prosthodontist.

Keywords: Hemifacial hypoplasia, microsomia of face, unilateral hypoplasia of face

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INTRODUCTION

Facial asymmetry may occur due to many causes, unilateral hypoplasia being a rare cause. We describe the magnetic resonance (MR) imaging findings in a young girl with unilateral hypoplasia of the face. She had hypoplasia of the soft tissues as well as the bone on the affected side. No previous documentation on MR imaging findings on congenital unilateral hypoplasia of face was found in our literature search.

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CASE REPORT

A 15-year-old female child born to nonconsanguineous parents with uneventful birth history was referred for MR examination from the Department of Plastic Surgery. She presented with complaints of less growth of facial features on the left side as compared to the right since birth. On examination, the left hemiface appeared smaller and showed diffuse deficiency of the soft tissues [Figure 1a] as compared to the right side. However, the overlying

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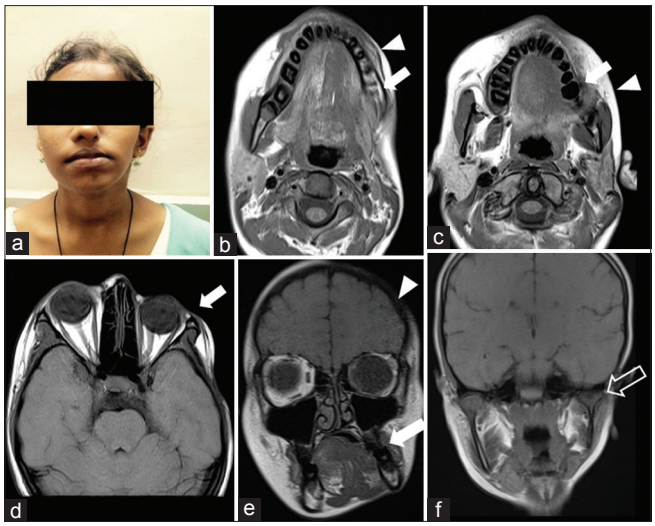

Figure 1: (a) Clinical photograph showing deficiency of the soft tissues in the left side of face. (b-e) Axial, (f) coronal T1-weighted images showing hypoplasia of the maxilla, mandible (arrow) with associated hypoplasia of the muscle, and subcutaneous fat (arrowhead). There is also irregularity of the articular surfaces of the left temporomandibular joint (open arrow)

skin appeared to be normal with no skin discoloration. No neurological deficit was found in her nervous system examination. Her systemic examination was unremarkable.

Subsequent MR imaging on a 1.5 tesla scanner showed diffuse reduction of subcutaneous fat and muscle tissue on the left side [Figure 1b-f]. The left orbital, nasal, maxillary, and mandibular bones were hypoplastic as compared to the right side [Figure 1b-f]; however, the bones showed normal signal intensities. The left submandibular and parotid glands appeared smaller. There was reduced bulk of the left masseter and pterygoid muscles. Mild malocclusion of teeth was noted on the left side. The articular surfaces of the left temporomandibular joint appeared irregular. The base of tongue, pharynx, valleculae, pyriform fossa, and larynx showed normal configuration. An imaging diagnosis of unilateral hypoplasia of the left half of face was made. She was advised to have cosmetic surgery. The patient promised to come for surgery at a later date

DISCUSSION

Hemifacial hypoplasia was described by Dr. Carl Ferdinand Von Arlt in 1881.[1] Etiology of hemifacial hypoplasia is not known; however, laboratory studies suggest an early loss of neural crest cells resulting in the clinical presentation of hemifacial hypoplasia. Teratogenic and genetic components have also been examined as other contributory factors. Possible pathogenesis could be disruption of the blood supply to the first and second branchial arches in the 6–8 weeks of pregnancy.[2]

Clinical features described in hemifacial hypoplasia are flattening of one side of the face due to underdevelopment of the maxillary, malar bones, mandibular ramus, and condyle. The orbit may be smaller on the affected side. External ear malformations may range from complete aplasia to a distorted pinna. There may be hypoplasia of the muscles of the face, such as masseter, temporalis, pterygoids, and muscles of facial expression. Other ear deformities described are flattened helical rim, absence of the auricle, atresia of the external auditory canal, and abnormal development of the middle ear ossicles. Patients may often present with conductive deafness. Dentation anomalies such as agenesis of the second premolar/third molar tooth and supernumerary teeth may be present.[3]

Many classifications have been proposed for hemifacial hypoplasia.

Pruzansky in 1969 described a classification based on the mandibular deficiency.[4]

1. Grade I - minimal hypoplasia of the mandible
2. Grade II - functioning but deformed temporomandibular joint with anteriorly and medially displaced condyle
3. Grade III - absence of the ramus and glenoid fossa.

Kaban *et al*. in 1988 later modified the classification.[5] They subdivided Type II into A and B: (1) In Type II A, the mandibular ramus, condyle, and TM joint are present but hypoplastic and abnormal in shape.(2) In Type II B, the mandibular ramus is hypoplastic and markedly abnormal in form and location. There is no articulation with the temporal bone.

OMENS classification of craniofacial microsomia was proposed by Vento *et al*. in 1991.[6] Five major areas of involvement in craniofacial microsomia go by the pneumonic: O-orbital, M-mandibular, E-ear, N-facial nerve, and S-soft tissue.

Plain radiographs in unilateral hypoplasia of face may reveal decreased size of the mandibular ramus, condyle, maxillary, and zygomatic bones. Computed tomography (CT) is useful for planning of orthodontic treatment and orthognathic surgery. CT, especially with three-dimensional reconstruction, is also useful in the evaluation of the external auditory canal, middle ear structures and temporomandibular joints. MR imaging (MRI) shows hypoplasia of the facial muscles, subcutaneous fat, and bones and also aids in the study of facial nerve.

Differential diagnosis for hemifacial hypoplasia is Treacher Collins syndrome, and Goldenhar syndrome.

Treacher Collins syndrome[7] is a genetic abnormality due to malformation of the first and second branchial arches bilaterally. It has an autosomal dominant mode of inheritance arising due to mutation of tuberous sclerosis gene on chromosome 5. It is characterized by (a) dental and mandibular deformities such as retrognathia, micrognathia, and macrostomia, (b) malformed or underdeveloped zygomatic arch, (c) otic deformities such as microtia and conductive hearing loss, (d) nasal deformities such as hypoplasia of the alar cartilages and paranasal sinuses, and (e) ocular deformities such as absent eyelids in lower inner third of the eye, notched iris, notched choroid, and colobomas. Goldenhar syndrome[8-9] is a complex congenital anomaly characterized by (a) ear anomalies include preauricular appendages/tags, (b) hemifacial microsomia, (c) ocular anomalies such as unilateral microphthalmia, (d) transverse facial clefts, and (e) asymmetry of the skull and spinal anomalies. Goldenhar syndrome can be associated with abnormal amniotic fluid volumes, congenital genitourinary anomalies, and congenital cardiac anomalies. Our patient had affected features confined to one side of the face and showed Grade IIA unilateral hypoplasia of the face.

The objective of the treatment is to restore facial symmetry and proper function. Teamwork is essential and members of the team should include plastic surgeons, otolaryngologists, orthodontists, and prosthodontist.[10] In conclusion, congenital unilateral hypoplasia of the face is a rare condition, characterized by hypoplasia of the soft tissues as well as the bone on the affected side. Clinical examination and imaging, especially MRI, can establish the diagnosis. Surgery is done for cosmetic reasons.

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.

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