CASE REPORT

Hemifacial microsomia
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Abstract

Hemifacial microsomia (HFM) is a condition in which the lower half of one side of the face is underdeveloped and does not grow in a normal pattern. After clefts, this is the second most common facial birth defect. Etiology of HFM is unknown, but prenatal exposures of some drugs and genetic abnormalities may be associated with the condition. Diagnosis and treatment of HFM is challenging due to a wide spectrum of deformities. Here, we present a case of a 10-year-old boy, who complained of facial asymmetry on left side of the face and deformity of the left ear since birth.

Keywords
Craniofacial microsomia, hemifacial microsomia, left facial asymmetry

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Introduction

Hemifacial microsomia (HFM) was first described in 1881 by a German physician named Carl Ferdinand Von Arlt. Later, Gorlin et al. used this term to describe the patients with unilateral microtia, macrostomia and malformation of mandibular ramus and condyle. Its synonyms are first arch syndrome, first and second branchial arch syndrome, otomandibular dysostosis, oculo-auriculo-vertebral dysplasia, and lateral facial dysplasia. Converse et al. proposed the term “craniofacial microsomia” which is used when cranial deformities are also present. Hence, hereby presenting a case of hemifacial microsomia (HFM) in a 10-year-old male patient with its characteristic clinical and radiological features which will help us to understand and diagnose this rare entity in a better way.

Case Report

A 10-year-old male patient has reported to the department with the chief complaint of facial asymmetry on left side of the face and malformed left ear since birth. There was no history of pain and clicking sound while opening and closing the mouth, no history of trauma, and no history of hearing loss. The patient was born to non-consanguineous parents with normal delivery. There was no significant family and antenatal history. Last surgical history revealed that patient has completed one session out of three sessions of the left ear reconstruction surgery owing to malformed left ear. General physical examination revealed patient to be moderately built and nourished and well oriented to time, place and person. Extra-oral examination revealed facial asymmetry with flattening of the face on the left side [Figure 1a], convex facial profile with retrognathic mandible which on opening the mouth deviated to the left side. Ear tag was evident inferiorly, and partially reconstructed ear was evident superiorly on the left ear region. Eyes were apparently normal [Figure 1b]. On temporomandibular joint (TMJ) examination right lateral movements were not evident. Left TMJ showed asymmetrical movements as compared to the right side. No preauricular tenderness or clicking sound was evident. Muscles of mastication were nontender and mouth opening was about 28 mm [Figure 2]. Intra-oral examination revealed gingiva to be soft, edematous and inflamed. Mesio-occlusal caries was evident w.r.t 84. Anterior deep bite and mandibular anterior crowding with Angle’s Class I molar relation on the right and left side was evident. Based on the history and clinical features a provisional diagnosis of HFM on the left side was considered. Orthopantomogram (OPG), PA skull and lateral cephalogram were taken. OPG showed hypoplasia of the mandibular body, ramus, coronoid process, and condyle on the left side with prominent antegonial notch [Figure 3a]. PA skull showed asymmetry and deviation of the mandible toward the left side [Figure 3b]. Lateral cephalogram showed reduced width of
Discussion

The incidence of HFM is reported between 1:5,000 and 1:5,600 live births with predilection for males than females (3:2). It is usually unilateral (70%), and the right side is affected more commonly than the left side. In this case, left side of the face was affected. Its exact etiology is still unknown, but studies suggest that an early loss of neural crest cells may be the specific factor responsible for its clinical presentation. Zielinski and colleagues investigated the largest CFM pedigree to date and found that a duplication in chromosome 14q22.3 (coding for OTX2) was present in all affected individuals.

Most commonly used classifications for HFM are the skeletal-auricular-soft tissue and the orbital asymmetry-mandibular hypoplasia-ear malformation-nerve dysfunction-soft tissue deficiency classification. In this case, orbit and nerve involvement were not present. Its clinical presentation varies from slight asymmetry of the face to severe underdevelopment of one half of the face with orbital implications, a partially formed ear or even its total absence. The chin and midline of the face are off-centered and deviated to the affected side. Other symptoms include unilateral hypoplastic maxillary and temporal bones, a unilateral shorter zygomatic arch and malformed external and internal parts of the ear. Patients may have minimal to complete aplasia of mandibular condyle and/or ramus of the mandible. Introral deformity includes hyperplastic or aplastic teeth and enamel with delayed and missing dentition on the affected side. The following minimum diagnostic criteria for HFM as proposed by Cousley in 1993 are:

1. Ipsilateral mandibular and ear defects (external/middle)
2. Asymmetrical mandibular or ear defects (external/middle) along with:
   a. Two or more indirectly associated anomalies, or
   b. A positive history of HFM in the family.

Its differential diagnosis includes Pierre Robin syndrome, Moebius syndrome, and Treacher Collins syndrome. An OPG provides an excellent view of the osseous structures of the complete maxillofacial complex. A lateral cephalogram helps to establish the relationship of the mandible and maxilla to the cranial base. A frontal skull radiograph (posterior-anterior view) shows the degree of osseous asymmetry of the face. Computed tomography provides both a 3-D view of the soft tissue of the face and an image of the underlying bone. Further, hearing evaluation, phonics tests, laryngoscopic inspection and vocalization analysis can help to establish each patient’s anatomical, neurological and functional status.

Treatment options include a combined surgical-orthodontic approach. In the last, osteotomies followed by acute orthopedic movement and osseous fixation were used, but the results are compromised by the risk of relapse caused by the inability of muscles to be acutely stretched. An alternative procedure called distraction osteogenesis is now widely accepted in which new bone is formed between the surfaces of bone segments that are gradually separated by incremental traction. This is a gradual method of creating bone after a surgical corticotomy sectioning of the mandibular body on the left side. The patient was taken for oral prophylaxis, restoration w.r.t 84, corrective surgery, orthodontic treatment, and kept under follow-up.
cortical plates. Nevertheless, prosthetic ear reconstruction can also be done in cases of partially formed or deformed ears.

**Conclusion**

HFM is a craniofacial malformation which can affect both soft tissue and hard tissue components of the maxillofacial region unilaterally. Hence, a proper diagnosis should be made based on history, clinical and radiological features. Its treatment involves multidisciplinary team to start with oral medicine and radiologist followed by pedodontist, orthodontist, pediatric surgeons, and plastic surgeon to maintain the normal functioning in the patient. Further best care can be provided by feeding therapy, speech therapy, psychological support, ophthalmologic assessment, and audiological evaluation.

**References**