

Hemifacial Microsomia – A Case Report and Review of Literature

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ABSTRACT

Hemifacial microsomia is a congenital deformity in which there is a deficient amount of skeletal and soft tissues on one side of the face. It is a syndrome affecting mainly first and second branchial arches, which include underdeveloped temporomandibular joint, mandibular ramus, masticatory muscles, ears and defects in facial nerve and muscles. The soft tissue malformation is present in the external ear, and the affected ear is placed lower as compared to the contralateral side. In addition to this, underdevelopment of osseous components leads to hearing loss with diminished external auditory meatus. The incidence of this disorder is 1:3000-26,000, which is usually seen at birth. Here, we present a case of 20-year-old girl, who complained of facial asymmetry on the left side of the face and deformity of left ear since birth with loss of hearing.

Keywords: Hemifacial microsomia, Hypoplasia, Microphthalmia, Microtia

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INTRODUCTION

Hemifacial microsomia (HFM) was first described by German physician Carl Ferdinand Von Arlt in 1881. HFM comprises of unilateral microtia, malformation of the mandibular ramus, condyle with macrostomia.¹ Goldenhar syndrome is a variant of HFM, which includes vertebral anomalies and epibulbar dermoids. This is known as craniofacial microsomia when there is involvement of cranial deformities (Converse *et al.*). The second most common congenital facial anomaly is HFM after cleft lip/palate. The reported incidence cases of HFM are about 1 in 5600 live births.^{2,4} It manifests as a highly variable phenotype and the structures which are derived from the first and second pharyngeal arches are most commonly affected. HFM commonly affects unilaterally as compared to bilateral anomalies.⁵ During the development of the jaw, the neural crest cells migrate to the first pharyngeal arch from the posterior mesencephalic fold and from the first rhombomeres, which gives rise to the skeletal maxillo-mandibular component.⁶ Damage or disruption of these neural crest cells result in HFM and some related syndromes.⁷ The HFM is characterized by asymmetrical defects of first pharyngeal arch derivatives. The most commonly affected structures include, ascending ramus of the

mandible, temporo-mandibular joint, zygomatic arch and external and middle ear, which includes the incus, the malleus, and the tympanic bone. There are chances of deviation of the mandible accompanied by malocclusion and hearing defect.^{4,8}

CASE REPORT

A 20-year-old female patient, reported to the Department of Oral Medicine and Radiology, Bharati Vidyapeeth Dental College, Pune with a chief complaint of malaligned teeth in upper and lower front and back teeth region of the jaw since 10 years. Patient also complains of pain in upper front teeth region since 2 years. Patient experiences difficulty in chewing food due to malaligned teeth. Pain was sudden on onset, mild, dull aching type and intermittent in nature. Patient's mother observed the asymmetry of patient's face, and there was difficulty in hearing and perceiving and malformation of left ear since birth. The medical, dental, family, and habit histories were non-contributory. No other family members had similar findings.

On general examination, the patient was moderately built, well nourished, cooperative and well oriented to time and place. The gait and posture were normal. Extraoral

examination revealed, on inspection, facial asymmetry on the left side of the face that appeared short as compared to the right side and flattened with hollowing of cheek. The zygomatic bone appeared prominent on the left side and short. The angle of mandible was prominent, and the body and ramus of mandible are short on the left side as compared to the right side of the face. The chin and midline were deviated to the left side. The ala of the nose and corner of the mouth were placed higher on the left side. The corner of the mouth was deviated to the left side (Figure 1). The pinna and lobules of left ear were deformed, the lobule was bifurcated with loss of hearing (Figure 2). The lips were competent, and the temporomandibular joint revealed no abnormality except for the deviation on opening to the left side following normal mouth opening (40 mm) (Figure 3). On palpation, the left masseter and temporalis muscle was deficient. Intraoral examination revealed a high arched palate and mixed dentition with retained 65, root stump of 54 and missing 25, 36, 42, 46. The teeth were of normal size

and shape, and there was Angle's Class III occlusion on both sides with edge to edge anterior bite. There was no abnormality detected on examining the tongue and the maxillary and mandibular ridge. The patient was diagnosed with HFM or facial hemiatrophy on the left side of the face. The differential diagnosis included were Pierre Robin syndrome, Treacher Collin Syndrome, Parry-Romberg syndrome, Down's syndrome, hypoplasia of condyle on the left side and congenital unilateral ankylosis on the left side. The patient was subjected for investigations, panoramic radiograph (Figure 4) and posteroanterior cephalometric radiograph (Figure 5) were carried out. The radiographs revealed retained 65, root stump of 54, mesioangular impacted 25 and missing 36, 46. There was deficiency on left side compared to right side such as short ramal height and width, small condylar head and reduced depth of sigmoid notch, reduced height of body of mandible, absence of antegonial notch, high level of external oblique ridge, superiorly placed inferior alveolar canal. The proposed treatment for the patient was distraction osteogenesis to bring about the growth of mandible to the desired size, ear abnormalities to be corrected by maxillofacial prosthetics and correction of hearing by hearing aids and alignment of teeth with extraction of retained deciduous teeth.



Figure 1: Frontal view of patient showing reduced growth of the mandible and deviation of the corner of the mouth to the left side



Figure 2: The pinna and lobules of the left ear are deformed, the lobule is bifurcated

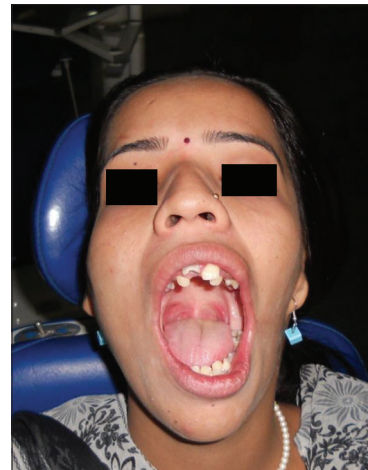


Figure 3: Deviation of the mandible to the left side on opening the mouth



Figure 4: Panoramic radiograph showing short ramal height and small condylar head on left side

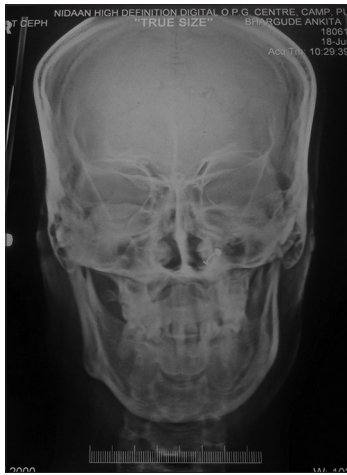


Figure 5: Posteroanterior cephalometric radiograph showing short ramal height, small condylar head, decreased width of ramus absence of antegonial notch on left side

DISCUSSION

There are many theories given for the HFM based on embryologic, clinical and laboratory studies, but the exact etiology is yet not known, but pathogenically it is heterogeneous. The laboratory studies suggest that there are certain factors that are responsible for the clinical presentation of HFM, which can be due to early loss of neural crest cells.⁴ The defective genes, teratogens and vascular anomalies are singly or collectively responsible for the disruption of normal development, which leads to anomalies seen in these patients. HFM affects the infants, which are born to exposed pregnant women by these teratogens. Poswillo did the laboratory investigations by maternal intake of 10 mg/kg thalidomide, and he believed that either there is total or incomplete development of the stapodial artery that is caused by rupture. A localized necrosis in the derivatives of first and second branchial arches is also seen. The researchers suggest that there is possible etiological factor, which could cause disruption in the blood supply to the first and second branchial arches in the first 6-8 weeks of pregnancy.^{4,9}

The various studies have reported that HFM commonly affects males than the females and common on the right side of the face as compared to the left side (3:2).¹⁰ However in the present case, HFM was seen in a female patient, and the left side of the face was affected, which is inconsistent with the literature.

The literature reveals that HFM varies clinically from slight asymmetry to severe underdevelopment of one side of the face with involvement of orbital, internal and external ear malformation with or without total absence of the ear. Auditory problems such as hearing loss and facial nerve dysfunction are commonly present in 50% of patients. The most common symptoms are the

unilateral hypoplastic maxillary and temporal bones with or without unilateral shorter zygomatic arch. The chin and the facial midline are always deviated to the affected side with one corner of the mouth situated higher than the other, giving rise to an oblique lip line. In the present case, there was gross facial asymmetry, the left side of the face was underdeveloped with prominent and short zygomatic arch. The left ear was deformed with loss of hearing. The chin and midline were deviated to the left side with the corner of the mouth situated slightly higher on the left side compared with the right side, which was consistent with the literature given. In intra-oral examination, the most commonly affected structures are third molar and second molars, in which agenesis is seen with the presence of supernumerary teeth. There is a presence of enamel malformations, anterior teeth are inclined towards affected side, delay in tooth development and hypoplastic teeth¹¹ but in present case there were no supernumerary teeth, teeth malformations but there was agenesis of 18, 38, 48 and missing 36, 46, delayed eruption of 25, there was inclination of 12, 15, 22, 32 lingually and 11, 13, 23 labially, which is inconsistent with literature. In HFM, there is a deficiency of masseter, temporalis and pterygoid muscles, and the muscles of facial expression are hypoplastic on the affected side. The degree of underdevelopment of the bone is directly related to the hypoplasia of the muscle to which they are attached. But in the present case, the masseter and temporalis muscle were hypoplastic on palpation, which is consistent with given literature. In most cases, there is an underdeveloped condyle with the absence of one glenoid fossa and aplasia of the mandibular ramus. But in the present case additional features like reduced depth of the sigmoid notch, absence of antegonial notch, high level of oblique ridge and superiorly placed alveolar canal, which is inconsistent with given literature. The severity of the clinical presentation only depends on the extent of loss of neural crest cell which in turn reflects in the degree of severity.^{11,12}

The differential diagnosis of HFM includes Pierre Robin syndrome and Treacher Collins syndrome. Unlike HFM, Pierre Robin syndrome always consists of cleft palate, micrognathia and glossoptosis, which is not seen in the present case. Most of the features of Treacher Collins syndrome include hypoplasia of facial bones especially malar and mandibular bones, malformation of external, middle and internal ear, macrostomia, high palatal arch which mimic HFM, however, the other features like antimongoloid palpebral fissures, deficiency of eyelashes, blind fistula between angles of the ear and mouth with facial cleft and skeletal deformities with characteristic appearance of face is "birdlike or fishlike"¹³ which were not seen in present case.

The advanced imaging modalities used in diagnosis and treatment planning of craniofacial abnormalities include multislice computed tomography, cone-beam computed tomography, magnetic resonance imaging and three dimensional surgical stimulation models. The management of HFM is a multidisciplinary approach, which include surgery done during growth phase or after the growth phase is over, limited autogenous bone grafting of deficient portions of the craniofacial skeleton, bilateral mandibular jaw advancement in patients with mild to moderate mandibular micrognathia with a combined Le Fort I osteotomy. The genioplasty and microvascular free flaps can be used for augmenting the soft tissue of the affected side of face and costo-chondral grafts which can be used to provide a new growth center for treating this anomaly.¹³

CONCLUSION

HFM is a developmental malformation in which there is a deficiency in both skeletal and soft tissues of the maxillofacial region on one side of the face. Hence, early diagnosis and treatment should be done for proper functioning and esthetics of the orofacial structures, which will lead to a better prognosis.

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