

Hemifacial microsomia: a clinicoradiological report of three cases

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Abstract: Hemifacial microsomia is a congenital malformation in which there is deficiency in the amount of hard and soft tissues on one side of the face. It is primarily a syndrome of first and second branchial arches involving underdevelopment of the temporomandibular joint, mandibular ramus, masticatory muscles, ears and occasionally defects in facial nerve and muscles. Here, we report three cases of hemifacial microsomia diagnosed based on clinical and radiographic findings. All three cases had variable presentations ranging from the mildest form that included facial asymmetry and ear deformity to the most severe and unusual form with facial nerve paralysis and spine deformity. (J Oral Sci 52, 319-324, 2010)

Keywords: hemifacial microsomia; facial nerve paralysis; spine deformity.

Introduction

Hemifacial microsomia (HFM) is the second most common congenital facial anomaly after cleft lip/palate, with a reported incidence of about 1 in 5,600 live births (1). HFM was first described by German physician Carl Ferdinand Von Arlt in 1881. Gorlin et al. used the term HFM to describe patients with unilateral microtia, macrostomia and malformation of mandibular ramus and

condyle, whereas Goldenhar syndrome was described as a variant, with vertebral anomalies and epibulbar dermoids. The name, craniofacial microsomia, was proposed by Converse et al. when cranial deformities were included. Other synonyms include first arch syndrome, first and second branchial arch syndrome, otomandibular dysostosis, oculo-auriculovertebral dysplasia and lateral facial dysplasia (2). Three case reports of hemifacial microsomia with variable and uncommon features are discussed here.

Case Reports

Case 1

A 6-year-old female patient presented to our department complaining of facial asymmetry since birth. Extraoral examination revealed underdevelopment of the left side of the face. The corner of the mouth on the left side was posteriorly placed when compared to the other side, leading to macrostomia (Fig. 1). The ear on the same side was deformed with the presence of a preauricular skin tag along the line that joined the corner of the mouth and tragus (Fig. 1). Lipoepidermoids were also present in the contralateral eye (Fig. 2). Intraoral examination was not significant except for deviation of the tongue to the opposite side on protrusion, indicating hypoglossal nerve paralysis (Fig. 3). Orthopantomogram revealed a hypoplastic mandible and shortened ramus height with complete absence of the condyle on the left side (Fig. 4). After the dental treatment, the patient was referred to the Departments of Oral Surgery and Plastic Surgery for the management of macrostomia, preauricular skin tags and hypoplastic mandible.

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Case 2

A 5-year-old male patient reported to the outpatient clinic complaining of toothache. Extraoral examination revealed underdevelopment of the right side of the face with low set pinna and preauricular skin tags (Fig. 5). A preauricular skin tag was also present on the left side demonstrating bilateral involvement (Fig. 6). Intraoral examination revealed presence of anterior and left posterior crossbites (Fig. 7) and inflammatory enlargement of tonsils. The orthopantomogram showed an underdeveloped right side mandible with short ramus and complete absence of condyle (Fig. 8). Pulpectomies and root canal treatments were performed for the pulpally involved carious teeth at the Department of Pediatric Dentistry. The patient was then referred to the Department of Orthodontics for correction of the anterior and posterior crossbites.



Fig. 1 The corner of the mouth on the left side is placed posterior compared to the normal side with presence of preauricular skin tags.



Fig. 2 Lipoepidermoid below the pupil in the right eye (arrow).

Case 3

A 7-year-old male patient reported to the out patient clinic with the complaint of toothache. Extraoral examination revealed a deformed right ear with complete absence of external auditory meatus. A sinus opening was also present in the postauricular region (Figs. 9 and 10). The patient was unable to draw back the right corner of his mouth on smiling and was not able to wrinkle his forehead on the same side. The patient was also unable to close his right eye completely and when he attempted to close his right eye, the eyeball rotated upwards, demonstrating Bell's sign (Fig. 11). All the features were suggestive of facial nerve paralysis of the right side. Orthopantomogram revealed missing mandibular central incisors and slight underdevelopment of the right side ramus of mandible with normal appearing condyle (Fig. 12). Computed tomography also revealed the same with partial stenosis of right side ear canal and polypoidal soft tissue mass on the contralateral



Fig. 3 Deviation of the tongue to the right side on protrusion, demonstrating hypoglossal nerve paralysis.



Fig. 4 Orthopantomogram depicts shortened ramus height with complete absence of condylar head on the left side (arrow).



Fig. 5 Low set pinna and preauricular skin tags (arrow) present on the right side along the line joining tragus and corner of mouth.



Fig. 7 Presence of anterior and left posterior crossbite.



Fig. 6 Preauricular skin tag just anterior to tragus of the ear on the left side.

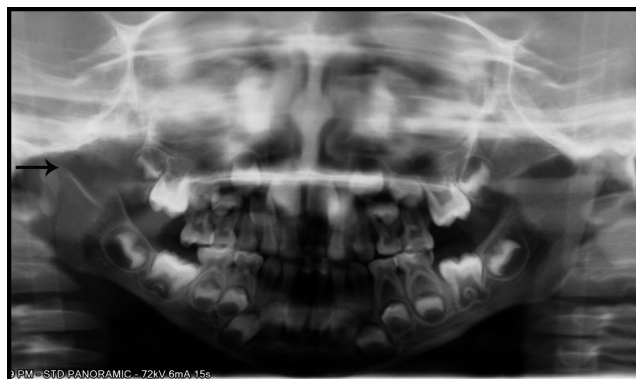


Fig. 8 Orthopantomogram depicts reduced ramus height and absence of condylar head on the right side (arrow).

Table 1 The different clinical features demonstrated in the three cases suggesting that HFM has diverse and variable presentations, which range from the mildest form with facial asymmetry and ear deformity to the most severe and unusual form with cranial nerve paralysis and spine deformities

Features	Case 1	Case 2	Case 3
Underdevelopment of facial structures	+	+	+
Small or malformed ears	+	+	+
Preauricular skin tags	+	+	-
Macrostomia	+	+	-
Intraoral problems	+	+	+
Cranial nerve involvement	+	-	+
Eye defects	+	-	-
Vertebral defects	-	-	+
Hearing loss	-	-	+
Missing teeth	-	-	+
Bilateral involvement	-	+	-
OMENS classification	O ₀ M ₁ E ₁ N ¹² S ₂	O ₀ M ₁ E ₁ N ₀ S ₂	O ₀ M ₁ E ₃ N ⁷ ₃ S ₁



Fig. 9 Frontal profile of the patient depicting the underdeveloped right side of face.



Fig. 12 Orthopantomogram depicts slight flattening of the right angle of mandible (arrow) and missing 31 and 41.

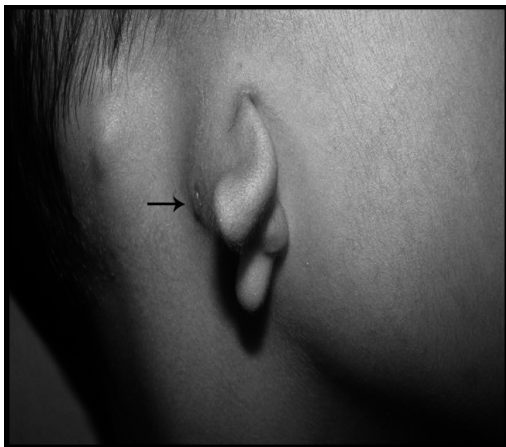


Fig. 10 Malformed right ear with absence of external auditory meatus and post-auricular sinus (arrow).

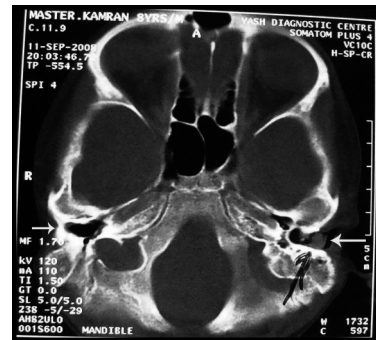


Fig. 13 Axial computed tomography depicts partial stenosis of the right auditory meatus (small arrow) and presence of polypoidal soft tissue mass in the left auditory meatus (large arrow).

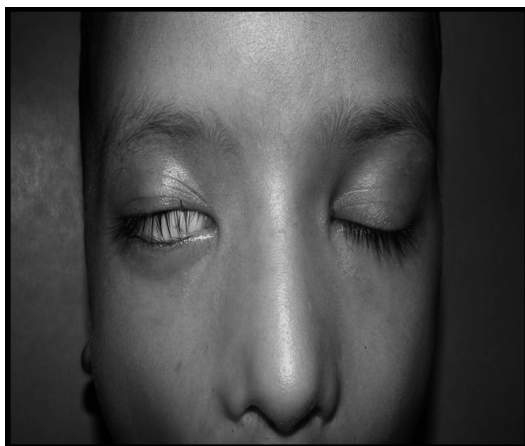


Fig. 11 Bell's sign, demonstrating right facial nerve paralysis.

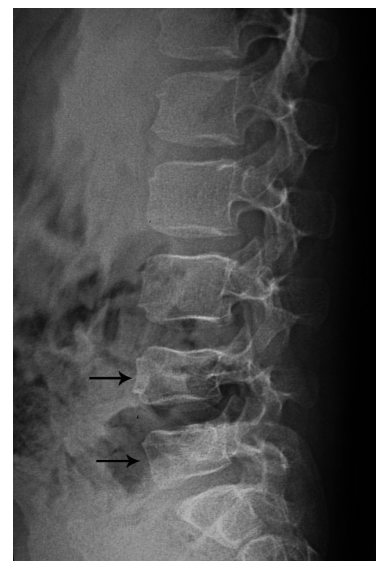


Fig. 14 Cropped lateral spine radiograph depicting spina bifida of L5 and reduced height of L4 and L5 vertebrae (arrows).

side (Fig. 13). Spine radiograph demonstrated spina bifida of lumbar 5 and reduced height of lumbar 4 and 5 vertebrae (Fig. 14). Dental treatment was completed at the Department of Pediatric Dentistry. The patient was then referred to the Departments of Oral Surgery and Prosthetics for surgical correction and prosthetic rehabilitation of the malformed right ear.

Discussion

The incidence of HFM is between 1:5,000 and 1:5,600 live births (3). Males appear to be more frequently affected than females (3:2) and the right side is affected more often than the left side (4). It is usually unilateral (70%) and always asymmetrical if it exhibits bilaterally (2). While the exact etiology of HFM has not yet been determined, there are many theories based on embryologic, clinical and laboratory studies. Laboratory studies suggest that an early loss of neural crest cells may be the specific factor responsible for the clinical presentation of HFM (5). Although 'hemifacial' refers to one half of the face, the condition is bilateral in 31% of cases, with one side being more affected than the other. In 48% of cases, the condition is part of a larger syndrome such as Goldenhar syndrome (6). The clinical picture of HFM varies from slight asymmetry in the face to severe underdevelopment of one facial half with orbital implications, a partially formed ear or even total absence of the ear. The chin and the facial midline are off-centered, and deviated to the affected side. Often, one corner of the mouth is situated higher than the other, giving rise to an oblique lip line. Other asymmetric symptoms are the unilateral hypoplastic maxillary and temporal bones, a unilateral shorter zygomatic arch and malformation of the external and internal parts of the ear (7). Sensorineural hearing loss and facial nerve dysfunction are common in HFM. Auditory problems are present in 30-50% of patients (8). Intra-oral structures can also be affected in this condition: agenesis of the third molar and second premolar may be present on the affected side, as well as supernumerary teeth, enamel malformations, delay in tooth development and hypoplastic teeth. The masseter, temporal 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 (7). In most cases, there is an underdeveloped condyle, but aplasia of the mandibular ramus and/or condyle with the absence of one glenoid fossa may also occur. In these cases, the maxilla is hypoplastic on the affected side (8). The two most frequently used classifications are the skeletal-auricular-soft tissue (SAT) and the orbital asymmetry-mandibular hypoplasia-ear malformation-

nerve dysfunction-soft tissue (OMENS) deficiency classification (9,10). The OMENS system, a newer and revised HFM classification system, assesses five major dysmorphic manifestations and allows each to be graded separately, unlike the SAT system. The orbit is assessed independently from the mandible, and nerve involvement has been added to the system (4).

A panoramic radiograph provides an excellent overview of the osseous structures of the mandible and maxillofacial complex. Since a cleft palate is often associated with HFM, an occlusal radiograph is needed. The relationship of the mandible and maxilla to the cranial base can be established initially with a lateral cephalometric radiograph. A frontal skull radiograph (posterior-anterior view) can be used to depict the degree of osseous asymmetry of the face. Computed tomography (CT) can provide both a three-dimensional rendition of the soft tissue of the face and an image of the underlying bone. Information on comparative muscle development can be assessed through CT or magnetic resonance imaging on a case-by-case basis. Hearing evaluation, phonics tests, laryngoscopic inspection and vocalization analysis help establish each patient's anatomical, neurological and functional status (5).

The differential diagnosis of this condition includes Pierre Robin syndrome, Moebius syndrome and Treacher Collins syndrome. Unlike HFM, Pierre Robin syndrome always consists of cleft palate, micrognathia and glossoptosis. Moebius syndrome is a nonfamilial deficient development of cranial muscles consisting of facial diplegia with bilateral paralysis of the ocular muscles, particularly those supplied by abducens. HFM usually does not lead to ocular muscle paralysis and nerve involvement occurs unilaterally. Most of the features of Treacher Collins syndrome mimic HFM; however, the latter occurs unilaterally and it is sporadic in a vast majority of cases (11).

In designing the course of treatment, the dental occlusion must be considered in conjunction with the underlying skeletal condition. Typically, a combined surgical-orthodontic approach is taken. In the past, growth-directing devices such as functional appliances were used to encourage growth and to minimize the extent of orthognathic surgery needed once the child has finished growing. Osteotomies followed by acute orthopedic movement and osseous fixation were used in the past. Unfortunately, the inherent risk of relapse caused by the inability of muscles to be acutely stretched often compromised the results. When autogenous costochondral grafting was used in more severe deformities, infection, pain and donor site morbidity posed important postoperative concerns (7).

Use of an alternative procedure called distraction

osteogenesis is now widely accepted. It is a process 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 cortical plates. Prosthetic ear reconstruction can also be done for deformed ears (7).

Interestingly, the three cases reported here showed variation from common features such as macrostomia, underdeveloped mandible and deformed ears to uncommon and unusual features like nerve paralysis, lipodermoids and vertebral defects. Dental surgeons should be aware of variable presentations of this syndrome which help to distinguish it from other syndromes so that proper treatment can be planned.

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