Hemifacial Microsomia Associated with Facial Palsy and Oculo-Auricular Malformation: A Rare Case Report

Abstract

Hemifacial microsomia (HFM) is the most frequently encountered form of isolated facial asymmetry. It is a congenital facial deformity involving the structures of the first and second pharyngeal arches: maxilla, mandible, external and middle ear, facial and trigeminal nerves, muscles of mastication, and overlying soft tissue. After cleft lip and palate, HFM is the most common craniofacial malformation. We report a rare case of HFM in association with facial palsy, oculoauricular malformation. The case had variable presentations ranging from the facial asymmetry and ear deformity to the most severe and unusual form with facial nerve paralysis. The article provides a discussion on the etiology, diagnosis and treatment of HFM.

Keywords: Facial palsy, Goldenhar syndrome, Hemifacial microsomia

INTRODUCTION

Hemifacial microsomia (HFM) is a highly heterogeneous condition with an estimated rate of 1 in 5,600-20,000 births,1 perhaps making it the most significant asymmetric craniofacial disorder. Gorlin used the term HFM to describe a spectrum of abnormalities such as macrostomia, unilateral microtia, and underdeveloped mandibular ramus, and condyle. These patients have differences in the size and shape of facial structures between the right and left sides of the face (facial asymmetry). Patients with HFM show different clinical manifestations thus identified by various nomenclature such as Goldenhar syndrome, oculo-auriculo-vertebral spectrum, facio-auriculo-vertebral dysplasia, and first and second branchial arch syndrome. Wide spectrum of anomalies associated with HFM involves one or both ears, eyes, facial nerve, mandibular nerve, soft-tissue and vertebral abnormalities. Mild-to-severe ear findings may involve an underdeveloped or absent external ear (microtia, or anotia), growths of skin (skin tags) in front of the ear (preauricular tags), or closed or absent ear canal; these abnormalities may lead to hearing loss. Sensorineural hearing loss (SNHL) is uncommon in HFM but can be devastating at a time. A rare case of HFM is presented in this article with its characteristics clinical and radiological features that will add a new dimension to the literature.

CASE REPORT

A 21-year-old male patient reported to Department of Oral Medicine and Radiology complaining of facial asymmetry since birth. No significant positive family history was present. Extra-oral examination revealed underdevelopment of the left side of face involving malformation of ipsilateral eye and ear (Figure 1). Ear examination showed cup shape pinna, low-lying deformity of the pinna also profound SNHL of the left side (Figure 2). The patient was unable to draw back the left corner of his mouth on smiling (Figure 3) and was not able to wrinkle his forehead on the same side (Figure 4). All the features were suggestive of facial nerve paralysis of the left side. Eye examination revealed that the patient was
also unable to close his left eye completely (lagophthalmos) leading to exposure keratitis, iris coloboma of left eye, and when he attempted to close his left eye, the eyeball rotated upwards, demonstrating Bell’s sign.

On intraoral examination unilateral cross bite of right side anterior and posterior, rotated right side incisors (Figure 5) and gingival hyperplasia on the palatal aspect of maxillary posterior region of the left side was noted (Figure 6).
Orthopantomogram revealed a hypoplastic mandible and absence of condylar head and neck, reduced ramus height and width (Figure 7). Lateral cephalometric radiograph depicted increased angle of mandibular ramus & hypoplastic ramus on left side (Figure 8). A frontal skull radiograph (posterior-anterior view) showed the degree of osseous asymmetry of the face (Figure 9). Transpharyngeal view of temperomandibular joint showed absence of condylar head and neck with reduced ramus width on left side (Figure 10). Cone beam computed tomography depicted Facial asymmetry, hypoplastic maxilla and mandible on left side (Figure 11). Cytogenetic investigation revealed normal male karyotype 46 and XY without chromosomal aberration.

DISCUSSION

Craniofacial microsomia was first described by German physician Carl Ferdinand Von Arlt in 1881. The facial characteristics in HFM typically include underdevelopment of one side of the upper or lower jaw (maxillary or mandibular hypoplasia), which can cause dental problems and difficulties with feeding and speech. In severe cases of mandibular hypoplasia, breathing may also be affected. HFM is the most common congenital facial anomaly, secondary to facial clefts, predominantly seen in males and commonly affects the right side of the face. Goldenhar syndrome (oculo-auriculo-vertebral dysplasia) was described as a variant with vertebral anomalies, epibulbar dermoids, and other anomalies that may also occur in association.

The observations of anatomic variables of HFM are supported by the pathogenic theory of axial mesodermal dysplasia. Findings from a recent ongoing case-control study are consistent with the vascular disruption hypothesis. However, links between HFM risk and other pathogenetic processes such as oxygen free radical generation, maternal diabetes or exposure to teratogens and finally, assisted reproductive technologies have been postulated by many authors. Teratogenic and genetic components have been examined by many investigators. Reports indicate that several teratogenic agents, such as retinoic acid, primidone, and thalidomide, have produced HFM in infants born to pregnant women exposed to those agents. The most useful in clinical practice are the Pruzansky’s and Kaban’s classifications. In Kaban’s...
modification of Pruzansky’s grading system, in a Kaban Type IIA, the temporomandibular joint, ramus and glenoid fossa are hypoplastic, malformed and malpositioned, but the deformed joint is adequately positioned for symmetric opening of the mandible. Reported case can be classified into this category based on these features.

The clinical picture of HFM varies from a little asymmetry in the face, to severe under-development of one facial half, with orbital implications, a partially formed ear or even a total absence of the ear. This extreme variability of expression for HFM is especially recognized by facial asymmetry. This is due in part to the absence, hypoplasia, and/or displacement of the pinna, but the degree of involvement is markedly variable. Malformation of the external ear may vary from a complete aplasia to a crumpled, distorted pinna that is displaced anteriorly and inferiorly.1 Maxillary, temporal, and malar bones on the involved side are somewhat reduced in size and flattened.

Strömland et al. 200711 performed prospective multidisciplinary study on Swedish patients with the oculo-auriculo-vertebral spectrum. Most frequent systemic malformations included, ear abnormalities (100%), ocular malformations (72%) was noted. In his study, only 28% patients found to be associated with both visual and hearing impairment which were present in this patient. Consistent findings like vertebral deformities (67%), cerebral anomalies (50%), and rare features such as congenital heart defects (33%), mental retardation (39%), and severe autistic symptoms (11%) were not reported in the present case.

The degree of underdevelopment of the bone is directly related to the hypoplasia of the muscle to which they are attached.12 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 side13 in present patient hypoplasia of malar bone, maxilla and ramus on the left side were present.

Terzis and Anesti14 in a review on developmental facial paralysis reported that facial palsy was present in 22-50% of patients with HFM. Dysmorphogenesis of the temporal bone and its effects on the facial nerve are most likely implicated in the cause of facial weakness. Congenital malformations associated facial palsy has a poor prognosis for recovery. Facial palsy affects the eye, causing lagophthalmos, ectropion, decreased tear production and corneal damage. In this case, lagophthalmos resulted in exposure keratosis of left eye. In these cases, great attention should be paid to prevent corneal drying, especially at night. Tarsorrhaphy is suggested for severe forms of lagophthalmos. Unilateral colobomas of the superior lid is a common finding. Coloboma of the iris of left eye was noted in the present case.

Mild-to-severe ear findings have been reported, including flattened helical rim off pinna, preauricular skin tags, microtia, external auditory canal atresia, ossicular malformations, and anotia.8,10-12 Conduction deafness due to middle ear abnormalities and/or absence or deficiency of the external auditory meatus has been noted. No such ear tags were found in this patient, but profound SNHL was noted on audiometric examination.

The incidence of delayed tooth development with HFM is proportional to the extent of mandibular deformity15 intraoral deformities include delay of tooth development on the affected side hyperplastic or aplastic teeth and enamel. Chalky opacifications of enamel are occasionally found on the maxillary central and lateral incisors of the underdeveloped side as a marker of development for HFM. No such intraoral findings were seen in the present case.

Skeletal alterations are other common anomalies of HFM; HFM patients exhibits occipitalization of the atlas, cuneiform vertebra, cervical complete or partial synostosis of 2 or more vertebrae, supernumerary vertebrae, spinal bifida, and anomalous ribs.16 No such findings were noted in the present case.
Treatment

It must be recognized that there is no prescribed treatment program for the child with craniofacial microsomia. The factors such as growth and development and prior therapy, must be considered before recommending an individualized treatment program. Surgical correction of the unilateral deformity is challenging. In patients older than 15 years of age, surgery is often indicated in the period of skeletal maturity. Consequently, all treatment plans must be customized according to the needs and age of the individual patient. When craniofacial growth and development are almost complete, limited autogenous bone grafting of deficient portions of the craniofacial skeleton could be considered.

CONCLUSION

The purpose of this article is to add one new interesting case with typical clinical and radiological features of HFM and to point out that whenever such case is encountered, the dental radiologist should critically evaluate the temporal bone as inner ear anomalies though rare, can occur in association with HFM.

REFERENCES