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CASE REPORT

HEMIFACIAL MICROSOMIA - A CASE REPORT WITH REVIEW OF LITERATURE

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ABSTRACT

Hemifacial microsomia (HFM) is a congenital deformity characterised by unilateral deficiency in the volume of hard and soft tissue of face. It is a syndrome predominantly affecting first branchial arch featuring poorly developed temporomandibular joint, mandibular ramus, muscles of mastication and ear. People with HFM may present with an underdeveloped ear (microtia) or absent ear (anotia) on the affected side of the face. The incidence of this disorder is 1:3000-26,000, which is usually seen at birth. Here we present a case of hemifacial microsomia in a 12-year-old girl with hearing difficulties on the affected side with review of literature.

Key words; Hemifacial, microtia, syndrome

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Introduction

The occurrence of hemifacial microsomia has been found to be about 1 in every 5600 live births and thus one of the most significant craniofacial disorders with accompanying facial asymmetry[1]. Hemifacial microsomia is the second most common defect of craniofacial structures after cleft lip and palate. The term hemifacial microsomia was coined by a German physician Carl Ferdinand von Arlt when he came across a case with an asymmetrical face, eye, and ear in 1881. It was first reported as a condition in the 1960s with aural, oral and mandibular developmental defects[2]. It arises due to developmental defects in first and second branchial arches with unilateral underdeveloped bones of the jaws and associated soft tissues[3]. The facial deformities accompanying hemifacial microsomia result due to destruction or disturbance of the neural crest cells[4]. According to Converse et al if cranial defects are associated with hemifacial microsomia the condition should be called as craniofacial microsomia.

A variant of hemifacial microsomia having additional features of vertebral anomalies and epibulbar dermoids in addition to unilateral microtia, microsomia, mandibular condyle and ramus malformation was described as Goldenhar syndrome by Gorlin et al[2,4].

A careful assessment of wide spectrum of abnormalities, etiology, and features associated with hemifacial microsomia is needed to manage the functional incapacities and to improve aesthetics of patients with this disorder. We present a case of hemifacial microsomia with its characteristic clinical and radiographic findings with an effort towards improving the knowledge and an attempt to differentiate it from other similar syndromes.

Case Report

A 12-year-old female patient reported to the Department of Oral Medicine and Radiology with a chief complaint of defect of the right side of the face since birth. The patient was born full term to non-consanguineous parents. The patient had no significant family and antenatal history. The patient complained of hearing deficit on the right side. There was no family history of the similar condition in other family members. There was no history of prior intervention. The patient had consulted a dentist for a toothache a few months back and was advised analgesics for the same. On general examination, the patient was found to be well nourished with moderate built and normal mental status. On extra oral examination, there was facial asymmetry detected on the right side of the face due to the defect since birth (Figure 1a). There was an absence of prominence over the right ramus and mandibular angle area due to the hypoplastic right mandibular body. The mandible was retrognathic and deviated to right side upon opening (Figure 1b). The patient had a normal mouth opening with no clicking noticed in temporomandibular joints. The examination of muscles of mastication revealed no significant findings. The right side of face showed a primitive ear, microtia (Figure 1c) and there was a small preauricular tag on left side of the face (Figure 1d). The right side of the patient exhibited hearing deficit. Intraorally a V-shaped palate with high arch was appreciated. There was crowding of teeth in the mandibular arch. The gingiva was soft and oedematous with the maxillary right first permanent molar, and mandibular first permanent molars were grossly decayed. The patient exhibited Angle's Class I malocclusion bilaterally. Based on the chief complaint and the clinical features a provisional diagnosis of hemifacial microsomia of right side was made. Hypoplasia of the ramus,

condyle, coronoid process and mandibular body on the right side was evident on panoramic radiograph along with prominent antegonial notch. The absence of third molar tooth buds in first, third and fourth quadrants could also be appreciated on panoramic radiograph (Figure 2a).

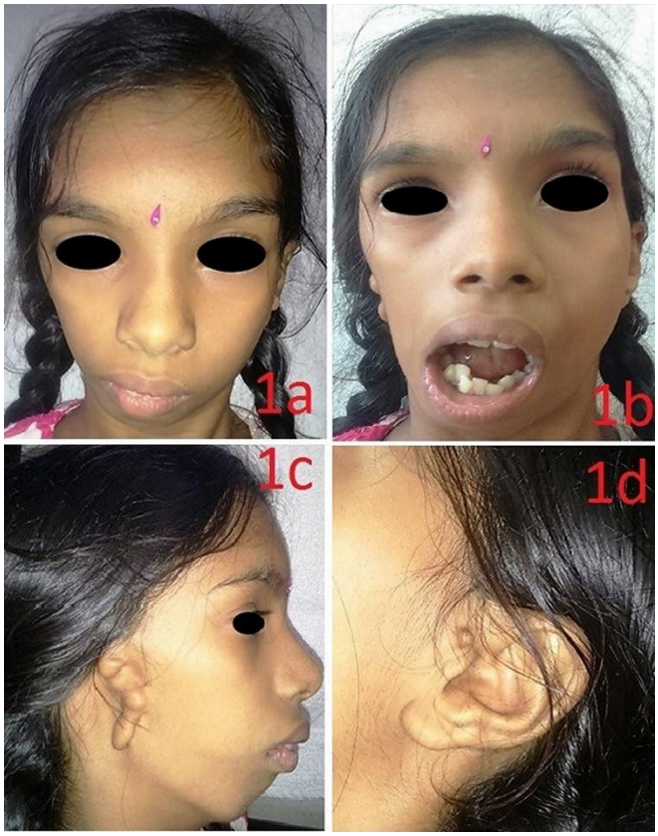

Figure 1. Clinical pictures:
Figure 1a. Clinical photograph of the patient showing defect on the right side of face.
Figure 1b. Clinical photograph of the patient showing deviation of mandible on the right side on opening.
Figure 1c. Clinical photograph of the patient showing microtia of the right ear.
Figure 1d. Clinical photograph of the patient showing ear tag on the left side.

The posterior anterior view of the skull showed a reduced height of ramus on the right side along with a shift of midline towards the right side. The nasal septum and maxilla were also seen to deviate towards the right side (Figure 2b). Lateral cephalogram showed an underdeveloped mandible along with a steep mandibular plane with a marked reduction in the height of ramus (Figure 2c). The patient was referred to an otolaryngologist for the hearing deficit. The patient was advised extraction of grossly decayed teeth and oral prophylaxis. The patient was also advised corrective surgery and correction of malocclusion, ear prosthesis for ear abnormalities and use of hearing aids to facilitate hearing.

Figure 2. Radiographic images:
Figure 2a. Panoramic radiograph showing hypoplasia of the ramus, condyle, coronoid process and mandibular body on the right side.
Figure 2b. Posterior-anterior view of skull showing reduced height of ramus on the right side along with a shift of midline towards the right side.
Figure 2c. Lateral cephalogram showing an underdeveloped mandible along with a steep mandibular plane.

Discussion

Hemifacial microsomia (HFM) is a congenital disorder of rare entity with wide phenotypic diversity. HFM has been seen to affect males more than females usually in the ratio of 3:2 and right side of the face is more affected than left side[5]. The case presented here is of a female patient with right side involvement. Early loss of neural crest has been reported to result in certain factors leading to the clinical presentation of hemifacial microsomia[4]. Certain teratogens, gene defects and anomalies of the vascular system, acting individually or collectively cause derangement of usual normal development and in turn result in features seen in these patients. Poswillo postulated that maternal intake of 10mg/kg Thalidomide causes rupture of the stapedial artery and results in its total or incomplete development. Derivatives of first and second branchial arches are also seen to suffer localized necrosis. According to researchers, this could lead to compromised blood supply to first and second branchial arches during the first 6-8 weeks of pregnancy[4,6]. The characteristic diagnostic features of hemifacial microsomia include asymmetrical hypoplasia of facial structures like hypoplasia of mandible, microtia, and presence of preauricular tags with or without periauricular skin tags[7]. The shifting of the chin and facial midline towards the affected side results in an oblique lip line with the corner of the mouth higher on one side than the other[8]. According to some authors, it is mandatory to have unilateral microtia or any kind of ear abnormality such as preauricular tags in such cases to be diagnosed as hemifacial microsomia[4]. Our case presented with the features of unilateral mandibular hypoplasia because of reduction in height of mandibular ramus, a shift in the midline resulting in oblique lip line with one corner of the mouth placed higher than the other and unilateral microtia which helped us to arrive at the diagnosis of hemifacial microsomia. The present case also had a preauricular tag on the normal side of the face. Treacher-Collins syndrome a different syndrome shows bilateral ear abnormalities in addition to micrognathia[9]. This could have led to a diagnostic confusion

in our case but as there was neither a sign of hypoplasia of zygomatic arches nor any downward slanting of palpebral fissures, therefore we diagnosed our case as hemifacial microsomia. Such distinction helps to supplement any efforts and improves knowledge about the wide spectrum of features associated with hemifacial microsomia. The ear canal in HFM patients is either absent or its end is blinded on the affected side[10]. The middle ear abnormalities result in conduction defects in 30 to 50% of HFM cases [3,4]. This feature coincided with our case. Various features on intra-oral exam seen in HFM cases include missing teeth on affected side (mandibular second premolar), absent mandibular third molars, delay in tooth development and susceptibility for aplastic and hypoplastic teeth[10,11]. The present case showed the absence of third molar tooth buds in first, third and fourth quadrants. The various advanced imaging tools used for diagnostic and treatment purposes in HFM include Cone Beam Computed Tomography (CBCT), Multi- Slice Computed Tomography (MSCT), Magnetic Resonance Imaging (MRI) and Three Dimensional (3D) Surgical stimulation models[8]. The aim of surgery is to correct facial asymmetry and restore function[12]. There is a need for collaborative efforts between different medical specialities for the wide spectrum of disabilities involving the various structures of the craniofacial area and occasionally other regions of the body such as associated defects in kidneys or heart in the management of HFM[13]. The management should begin early in life and should involve a team of specialists which include geneticists, audiologists, speech pathologists, physicians, plastic surgeons, maxillofacial surgeons, pedodontists, prosthodontists and orthodontists[5]. The management of HFM involves synergistic approach from specialists of different medical disciplines. Pre-and post-growth phase surgeries, use of grafts for deficit regions of the craniofacial framework, osteotomy for protraction of deficient mandible in mild cases of micrognathia of the mandible are among the treatment options in the management of HFM. Costochondral grafts can be given to provide new growth sites and soft tissue augmentation of the involved side can be done using the procedures of genioplasty and microvascular free flaps[14].

Conclusion

Hemifacial microsomia is a developmental abnormality in which there is a deficiency in both soft and skeletal tissues of the maxillofacial region on one side of the face. Early diagnosis and treatment should be planned with a team of experts which include dental radiologists, orthodontists, pedodontists, speech therapist, surgeons and paediatric surgeons for efficient clinical care to restore normal functions in the patients affected with hemifacial microsomia.

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