Hemifacial Microsomia : A Case Report

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Abstract

Hemifacial microsomia is a congenital malformation in which there is a deficiency in the amount of hard and soft tissue on one side of the face. It is primarily a syndrome of the first branchial arch, involving underdevelopment of the temporomandibular joint, mandibular ramus, masticatory muscles and the ear. The affected ear may have an external soft-tissue malformation in addition to being lower set than on the contra lateral side. Hearing loss may result from underdevelopment of the osseous components of the auditory system and a diminished or absent external auditory meatus. Occasionally, second branchial arch defects involving the facial nerve and facial muscles coexist with Hemifacial microsomia. Radiographic examination in case of Hemifacial microsomia is of limited value because of superimposition of normal and abnormal bony structures. The skeletal and soft-tissue findings of a patient with Hemifacial microsomia are presented here to improve our knowledge and diagnostic skill of this uncommon entity.

Keywords: Hemifacial microsomia, Hypoplasia, External auditory meatus, Facial nerve.

Introduction

Hemifacial microsomia is a developmental syndrome in which one side of the face is underdeveloped, usually including the bony jaws and overlying soft tissues; usually (but not always) the ear is involved (microtia).Hemifacial microsomia is the second most common developmental craniofacial anomaly after cleft lip and palate and affects one of every 5600 live births.[1-2] More than 15 terms, including Goldenhar's syndrome and oculoauriculovertebral dysplasia, have been applied to this disease, with each term representing the perspectives of different specialists. Diagnostic imaging is important in the presurgical evaluation of patients with this anomaly; however, the broad spectrum of abnormalities encountered in patients with hemifacial microsomia can be confusing.[2]

Hemifacial microsomia was first described by German physician Carl Ferdinand Von Arlt in 1881. Gorlin et al. used the term Hemifacial microsomia 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.[3] Some studies have shown that the mesiodistal dimensions of the mandibular second primary molar and the mandibular permanent first molar teeth on the affected side in hemifacial microsomia were significantly smaller compared with those of normal teeth. Furthermore, in the maxillary and mandibular first permanent molars and the maxillary and mandibular first and second primary molars, the teeth in the apparently "normal" side of hemifacial microsomia were also significantly reduced in the mesiodistal dimensions. Comparison of overall dimensions revealed that all primary and permanent molars in hemifacial microsomia were significantly smaller in the mesiodistal dimensions compared with control teeth.[4] A general gradient effect was observed, with the most posterior tooth in each arch being the most severely affected and no effect being seen in the canines and the incisors. These findings suggest that the dental lamina in hemifacial microsomia is affected, and support the hypothesis that its pathogenesis involves an abnormality of the neural crest.

Case Report

A 7-year-old male patient reported to our department with the complaint of pain in lower right second deciduous molar. Pain in the tooth was dull and intermittent in nature and more severe at night. On Extraoral examination face of the patient was found to be bilaterally asymmetrical and lower jaw deviation was towards the right side (Fig.1). Right ear was deformed with complete absence of external auditory meatus. (Fig.2). Orthopantomogram revealed slight under development of the right side ramus of mandible with normal appearing condyle (Fig.3). On palpation the parotid gland seemed hypoplastic on right side and fullness of cheek was also absent. No abnormality was seen in eyes, nose, lips, neck moments and skin. Left side of the face was apparently normal. (Fig.4) Intra oral examination showed carious mandibular right deciduous second molar. (Fig. 5) Dental treatment was completed at the Department of Paediatric 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 term Hemifacial microsomia was first used by Gorlin to refer to patients with unilateral microtia, macrostomia, and failure of formation of the mandibular ramus and condyle.[1,5] The incidence of Hemifacial microsomia is about 1 in 5,600 live births[6]. Males appear to be more frequently affected than females (3:2) and the right side is affected more often than the left side.[7] It is usually unilateral (70%) and always asymmetrical if it exhibits bilaterally.[3] While the exact etiology of Hemifacial microsomia 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 Hemifacial microsomia. It is the second most common craniofacial malformation after cleft lip and palate. In the past, Hemifacial microsomia has been the perview of various medical specialists, each preoccupied with 1 or 2 anatomic areas and each with a particular technical expertise.

Reconstructive surgeons have struggled with the external ear anomalies, microphthalmia, soft tissue and muscle defects.

Otolaryngologists have been concerned with hearing disorders, middle ear anomalies and airway obstruction.

Oral surgeons and orthodontists have focused on occlusal and jaw abnormalities. These specialists, all concerned with Hemifacial microsomia, are beginning to work together in craniofacial teams. Prosthodontists also become members of craniofacial teams to offer expertise in prosthetic reconstruction of the external ear using craniofacial implants as a support or as a retention system for prosthetic rehabilitation.

Because of an extraordinarily wide range of phenotypic expression, various nomenclature are applied to Hemifacial microsomia such as Goldenhar-Gorlin syndrome, first arch syndrome, lateral facial dysplasia, unilateral craniofacial micro-somia, otomandibular dysostosis, oculoauri-culovertebral dysplasia, auriculo-branchiogenic dysplasia, and oculoauri-culovertebral spectrum. Though there is extreme variability of expression for Hemifacial microsomia, it is especially recognized by facial asymmetry. This is due in part to absence, hypoplasia, and/ or displacement of the pinna, but the degree of involvement is markedly variable. Maxillary, temporal, and malar bones on the involved side are somewhat reduced in size and flattened. Malformation of the external ear may vary from a complete aplasia to a crumpled, distorted pinna that is displaced anteriorly and inferiorly. Occasionally, bilateral anomalous pinnas are noted.

Approximately 40% of patients with microtia have varying degrees of the

syndrome. Conduction deafness due to middle ear abnormalities and/or absence or deficiency of the external auditory meatus has been noted in 30% to 50% of cases. Supernumerary ear tags may occur anywhere from the tragus to the angle of the mouth. Patients may have minimal under-development of the condyle to unilateral aplasia of the mandibular ramus and/ or condyle with absence of the glenoid fossa; 50% to 70% of Hemifacial microsomia patients have agenesis of the ramus on the affected side.

The maxilla is narrowed on the involved side with decreased palatal width. Associated cleft lip and/ or palate is found in 7% of Hemifacial microsomia patients. Hypoplasia of facial muscles, such as the masseter, temporalis, pterygoids, and those of facial expression on the involved side has also been observed. Narrowing of the palpebral fissure occurs on the affected side in about 10% of patients. Clinical microphthalmia or anophthalmia has been reported and the ipsilateral eye may be at a lower level than that on the opposite side. Unilateral colobomas of the superior lid is a common finding.

The differential diagnosis of this condition includes Pierre Robin syndrome, Moebius syndrome and Treacher Collins syndrome. Unlike Hemifacial microsomia, 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. Hemifacial microsomia usually does not lead to ocular muscle paralysis and nerve involvement occurs unilaterally.

Most of the features of Treacher Collins syndrome mimic Hemifacial microsomia; however, the latter occurs unilaterally and it is sporadic in a vast majority of cases. In designing the course of treatment, the dental occlusion must be considered in conjunction with the underlying skeletal condition. Typically, a combined surgicalorthodontic approach is taken.

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.[8]

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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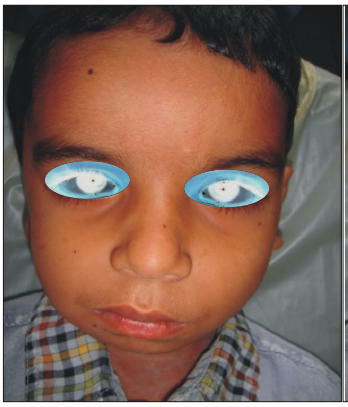

Fig. 1

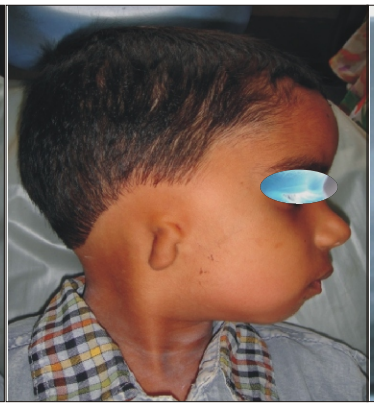

Fig. 2

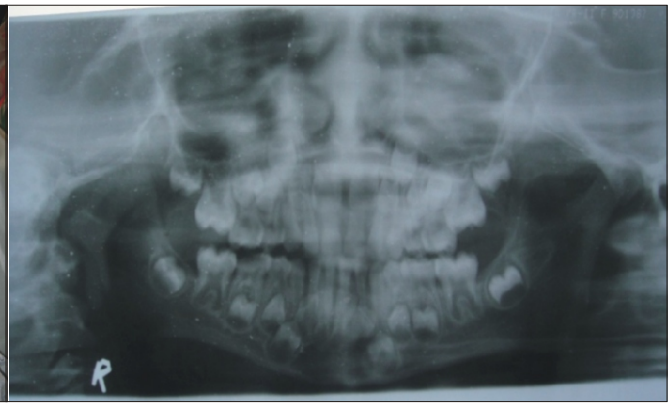

Fig. 3

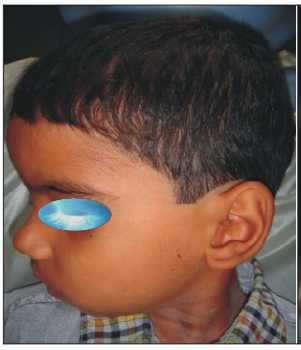

Fig. 4

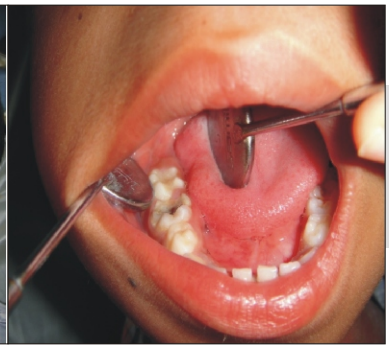

Fig. 5

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