



### Hemifacial Microsomia: A Case Report and Overview

#### Hemifasyal Mikrozozi: Olgu Sunumu ve Genel Bakış

Medhini Madi<sup>1</sup>, Shishir Ram Shetty<sup>1</sup>, Subhas G Babu<sup>1</sup>, Sonika Achalli<sup>1</sup>

<sup>1</sup>NITTE University, A.B. Shetty Memorial Institute of Dental Sciences. Department of Oral Medicine and Radiology. Mangalore. İNDIA.

*Cukurova Medical Journal 2014;39(3):625-635.*

#### ABSTRACT

Hemifacial Microsomia is a developmental craniofacial anomaly typically displaying reduced growth and development of half of the face as a result of abnormal development of first and second branchial arches. The patients with Hemifacial Microsomia usually exhibit unilateral involvement of the face but occasionally might involve both the sides. Cases which show vertebral anomalies and epibulbar dermoids have been considered to form a separate category within this condition. The condition is now known to be extremely complex and heterogenous. Here we present a case of Hemifacial Microsomia with its characteristic clinical and radiographic features that will help us in diagnosing and differentiating this rare entity from other closely related syndromes.

**Key Words:** Anomaly, developmental, hemifacial microsomia, branchial arches.

#### ÖZET

Hemifasyal mikrozozi tipik olarak birinci ve ikinci brankial arkların anormal gelişimi sonucu yüzün yarısında eksik büyüme ve gelişme gösteren gelişimsel kraniofasial bir anomalidir. Hemifasyal mikrozozi genellikle hastalarda yüzün tek bir tarafında ortaya çıkmaktayken nadiren her iki tarafıda içerdiği vakalarda mevcuttur. Vertebral anomali ve epibulbar dermoid gösteren vakalar ise bu durumlar içerisinde farklı bir kategoride değerlendirilir. Bu yapı oldukça kompleks ve heterojenite gösteren bir durum olarak bilinir. Burada; bilinen yakın ilişkili sendromlardan farklı olarak, bu nadir durumunun ayırt edilmesi ve teşhisi için bize yardımcı olacak karakteristik klinik ve radyografik özellikli Hemifasyal mikrozoimli bir vaka sunulmaktadır.

**Anahtar Kelimeler:** Anomali, gelişimsel, Hemifasyal mikrozozi, brankial arklar

#### INTRODUCTION

In the 1960's, Hemifacial Microsomia was defined as a condition affecting primarily aural, oral, and mandibular development. The disorder varied from mild to severe, and involvement was limited to one side in many cases, but bilateral involvement was also known to occur, with more severe expression on one side. Goldenhar syndrome was considered a variant of this complex, characterised additionally by vertebral

anomalies and epibulbar dermoids<sup>1</sup>. The condition is now known to be extremely complex and heterogenous. Thus, the term oculo-auriculo-vertebral spectrum is employed<sup>2</sup>.

The many terms used for this complex indicate the wide spectrum of anomalies described and emphasized by various authors. The complex has been known as Hemifacial Microsomia, Oculo-auriculo-vertebral dysplasia, Goldenhar syndrome, Goldenhar-Gorlin syndrome, first arch syndrome,

first and second branchial arch syndrome, lateral facial dysplasia, unilateral craniofacial microsomia, otomandibular dysostosis, unilateral mandibulofacial dysostosis, unilateral intrauterine facial necrosis, auriculo-branchiogenic dysplasia, and facio-auriculo-vertebral malformation complex<sup>3</sup>.

Hemifacial Microsomia is the second most common developmental craniofacial anomaly after cleft lip and palate and affects one of every 5600 live births<sup>4,5</sup>. Hemifacial Microsomia results from the abnormal development of the first and second branchial arches and the first branchial membrane. These arches are the mounds of tissue that contribute to the development of facial structures (cheek bones, upper and lower jaws and ear). Neural crest cells migrate to the developing arches and are responsible for the correct formation of these structures. Damage to, or disruption of, these cells result in the facial abnormalities of hemifacial microsomia and related syndromes<sup>6</sup>.

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<sup>7</sup>. Here we present a case of Hemifacial Microsomia with its characteristic clinical and radiographic features that will help us in diagnosing and differentiating this rare entity from other closely related syndromes.

## CASE REPORT

A 14 year old male patient reported to the Department of Oral Medicine and Radiology with the chief complaint of decay in the lower left and right back teeth since 3 months. He was accompanied by his parents. The patient noticed the decay a year back. At the time he noticed it

first he had h/o food lodgement in that region. Three months back the patient developed pain in the mandibular right and left back teeth. Pain was intermittent in nature and dull aching type. Since 1 month the patient has no pain but has food lodgement. It was patients first visit to the dentist. Medical history revealed that the patient had absence of right ear and hearing loss on one side and asymmetry of the face since childhood for which he had consulted a physician. No treatment was instituted. The patient was born to a non consanguineously married couple at term. Family history revealed that the child was the only child of the parents and none of the family members were affected by this disease. Patient was conscious, co-operative and well oriented to time, place and person. On extraoral examination, the patients face showed marked facial asymmetry. There was deviation of the lower jaw towards the right side. The midsagittal plane of the patients face was curved towards the right side (Fig 1). Facial anteroposterior and vertical dimensions were reduced on the affected side, especially in the lower face towards the otocephalic centre. The maxillary, temporal and malar bones on the right side are reduced in size and flattened. There was also hypoplasia of the mandibular ramus and the condyle on the right side (Fig 1,2 and 3). The right ear was malformed with absence of external auditory meatus. There was a rudimentary right pinna and preauricular skin tag (Fig 4). The left ear was normal. (Fig 5). There was no epibulbar dermoids and coloboma. There was mild strabismus. (Fig 1). There was no abnormality seen in eyes, nose, lips, ribs, neck movements and skin. The patient had hearing loss on the right side. No mental retardation was found. Intraoral examination revealed high palatal vault, crowding w.r.t the maxillary anterior due to which the lateral incisors are lingually tipped and canines are labially placed, chronic pulpitis w.r.t 16, 17,11 and dentinal caries wrt 22 and 26 (Fig 6). Mandibular arch also showed crowding in the anterior region due to which the mandibular left first premolar was

lingually placed out of the arch and canine on the left side was mildly rotated and labially tipped. There was chronic pulpitis w.r.t 36,45, 46 and 47 (Fig 7). The gingival was soft and edematous. The patient was subjected to Extra oral radiographs. Panoramic radiograph revealed hypoplastic ramus, condyle and coronoid process on the right side with a prominent antegonial notch. Panoramic radiograph also reveals radiolucency involving the enamel dentin and approximating the pulp w.r.t 16,17,11,45,46,47 and 36 and hazy radiolucency at the apex of 36 suggestive of chronic periapical abscess. Radiolucency involving the enamel and

the dentin w.r.t 22 and 26 suggestive of dentinal caries. Erupting teeth buds of 18,28,38, and 48 are also visible (Fig 8). Posteroanterior skull view revealed deviated nasal septum, deviated maxilla and mandible towards the right side. It also shows the lack of development of ramus, coronoid process and the condyle on the right side (Fig 9). The Lateral Cephalogram showed underdevelopment of the mandible. The height of the ramus and the mandibular length was markedly short and the mandibular plane was steep (Fig 10).



**Figure 1.** Clinical photograph of the patients face showing marked facial asymmetry, deviation of the lower jaw towards the right side and the midsagittal plane of the patients face curved towards the right side



**Figure 2.** Clinical photograph showing the lateral view on the right side of the patient revealing the maxillary, temporal and malar bones on the right side which are reduced in size and flattening and hypoplasia of the mandibular ramus and the condyle on the right side



**Figure 3.** Clinical photograph of the lateral view on the left side



**Figure 4.** Clinical photograph of the right ear showing malformed ear with absence of external auditory meatus, with a rudimentary right pinna and preauricular skin tag.



**Figure 5.** Clinical photograph revealing normal left ear



**Figure 6.** Intraoral photograph of the maxillary arch showing high palatal vault, crowding w.r.t the maxillary anterior due to which the lateral incisors are lingually tipped and canines are labially placed, chronic pulpitis w.r.t 16, 17,11 and dental caries wrt 22 and 26.



**Figure 7.** Intraoral photograph of the mandibular arch showing crowding in the anterior region due to which the mandibular left first premolar was lingually placed out of the arch and canine on the left side was mildly rotated and labially tipped and chronic pulpitis w.r.t 36,45, 46 and 47.



**Figure 8.** Panoramic radiograph revealing hypoplastic ramus, condyle and coronoid process on the right side with a prominent antegonial notch, radiolucency involving the enamel dentin and approximating the pulp w.r.t 16,17,11,45,46,47 and 36 and hazy radiolucency at the apex of 36 suggestive of chronic periapical abscess, radiolucency involving the enamel and the dentin w.r.t 22 and 26 suggestive of dental caries and erupting teeth buds of 18,28,38, and 48 are also visible.



**Figure 9.** Posteroanterior skull view revealing deviated nasal septum, deviated maxilla and mandible towards the right side, the lack of development of ramus, coronoid process and the condyle on the right side.



**Figure 10.** The Lateral Cephelogram showing underdevelopment of the mandible. The height of the ramus and the mandibular length was markedly short and the mandibular plane was steep.

## DISCUSSION

Hemifacial Microsomia ( HFM) was first described by German physician Carl Ferdinand Von Arlt in 1881<sup>7</sup>. The term Hemifacial Microsomia was first used to refer to patients with unilateral microtia, macrostomia, and failure of formation of the mandibular ramus and condyle<sup>4,5,6</sup>. The incidence of Hemifacial Microsomia is between 1:5000 and 1:5600 live births<sup>2</sup>. Males appear to be more frequently affected than females (3:2) and the right side is affected more often than the left side<sup>8</sup>. While there are no agreed upon minimal diagnostic criteria, the facial phenotype is characteristic when enough manifestations are present. In some instances, isolated microtia or auricular or preauricular abnormality may represent the mildest manifestation<sup>2</sup>. Unilateral microtia or ear abnormality, including preauricular tags, has been suggested as a mandatory feature by some authors<sup>9</sup>.

While the exact etiology of HFM has not yet been determined, there are many theories based on embryologic, clinical and laboratory studies. The pathogenesis HFM is incompletely understood<sup>10</sup>. A widely accepted theory for the pathogenesis of HFM is that haemorrhage associated with the formation of the stapedia arterial system during embryogenesis disrupts normal development of the first, and also second, arch derivatives<sup>11</sup>. Laboratory studies suggest that an early loss of neural crest cells may be the specific factor responsible for the clinical presentation of HFM. Additional problems associated with HFM, such as cleft palate (seen in as many as 10 percent of the cases) and cardiac anomalies (seen in as many as 50 percent of the cases) also have been associated with an early loss of neural crest cells. The extent of the neural crest cell loss is reflected in the degree of severity



of the facial deficiency and, therefore, is thought to dictate the severity of the clinical presentation<sup>12,13</sup>.

The clinical features of HFM include flattening of the face on the affected side due to hypoplastic maxillary and malar bones and aplasia of the mandibular ramus and condyle. The eye may thus appear to be on the lower level than that of the affected side. Malformation of the external ear may vary from complete aplasia to a crumpled, distorted pinna. 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. There can be hypoplasia of muscles such as the masseter, temporalis, pterygoids and muscles of facial expression on the involved side<sup>14</sup>. Sensorineural hearing loss and facial nerve dysfunction are common in HFM. Auditory problems are present in 30-50% of the patients<sup>15</sup>. In oral manifestations, there is macrostomia, aplasia of the mandibular ramus and condyle. Agenesis of the third molar and second premolar may be present on the affected side as well as there can be the presence of supernumerary teeth, enamel malformations, delay in tooth development and hypoplastic teeth<sup>14</sup>.

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 deficiency (OMENS) classifications<sup>16</sup>. The OMENS classification is the most comprehensive one and, therefore, it is one of the most commonly used systems<sup>17</sup>.

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<sup>18</sup>.

The management of HFM necessitates a multidisciplinary approach. The surgery may be done during growth phase or after the growth phase is over. Excisions of the preauricular skin tags and cartilage remnants can remove certain amount of social stigma which is associated with this condition. Treatment options may include, limited autogenous bone grafting of deficient portions of the craniofacial skeleton, a bilateral mandibular advancement in patients with mild to moderate mandibular micrognathia, a combined Le Fort I osteotomy, a bilateral mandibular osteotomy, genioplasty, microvascular free flaps for augmenting the soft tissue of the face on the affected side and costo-chondral grafts which can be used to provide a new growth centre for treating this anomaly<sup>16,19</sup>.

The purpose of this article was to stress upon the clinical and the radiographic features of Hemifacial microsomia and how it is different from the other very similar syndromes. This article would be incomplete without mentioning about the Goldenhar syndrome and how that is different from Hemifacial microsomia although both are considered under Oculo-auriculo vertebral spectrum. Goldenhar syndrome. This is a variant of hemifacial microsomia which in addition to asymmetrical bone and tissue development of the face and microtia (missing or abnormally developed ear), features vertebral skeletal anomalies such as a form of scoliosis or defects of the cervical spine (neck). Children with Goldenhar syndrome display ear tags, benign tumors at the rim of the cornea that leads to astigmatism or lazy eyes and colobomas or missing eyelids. Malformations of the spinal column that set this condition apart from other craniofacial syndromes

include: open spine (spina bifida), fusion of the top of the spine to the lower edge of the skull, incomplete asymmetric spinal column development and more than the normal number of vertebrae<sup>20</sup>.

## REFERENCES

- Gorlin RJ, Jue KL, Jacobsen U, et al. Oculoauriculovertebral dysplasia. *J Pediatr.* 1963;63:991-9.
- Gorlin RJ, Cohen MM, Levin LS. Syndromes of the head and neck. 3<sup>rd</sup> edition, McGrawHill.Inc., Oxford university Press.,1990;641-55.
- Jongbloet PH. Goldenhar syndrome and overlapping dysplasias. *J Med Genet.* 1987;24:616-20.
- Munjaj D. Hemifacial microsomia: A case report and review of literature. *J Indian Assoc Oral Maxillofac Radiol.* 2006;18:64-9.
- Monahan R, Seder K, Patel P, Alder M, Grud S, O'Gara M. Hemifacial microsomia: Etiology, diagnosis and treatment. *Am Dent Assoc.* 2001;132:1402-8.
- Raymond WS, Angelisa MP, Samson L, Michael LC. Hemifacial microsmia in pediatric patients: Asymmetric abnormal development of the first and second branchial arche. *AJR.* 2002;178:1523-30.
- Kalsotra P, Chowdhary A, Bhagat DR, Parihar SS, Prabhakar R. Craniofacial microsomia. *J K Sci.* 2006;8:168-70.
- Wang RR, Andres CJ. Hemifacial microsomia and treatment options for auricular replacement: A review of literature. *J Prosthet Dent.* 1999;82:197-204
- Rollinck BR, Kaye CI. Hemifacial microsomia and variants:Pedigree data. *Am J Med Genet.* 1983;15:233-53.
- Poswillo DE. Etiology and pathogenesis of first and second branchial arch defects: the contribution of animal studies. In: Symposium on Diagnosis and Treatment of Craniofacial Anomalies. Eds.: McCarthy JG, Wood-Smith D. New York University. 1976;20.
- Poswillo D. The pathogenesis of the first and second branchial arch syndrome. *Oral Surg. Oral Med. Oral Pathol.* 1973;35:302-28.
- Johnston M.C, Bronsky P.T. Animal models for human craniofacial malformations. *J Craniofac Genet Dev Biol.* 1991;11:277-91.
- Thomas J.T, Frias J.L. The heart in selected congenital malformations: a lesson in pathogenetic relationships. *Ann Clin Lab Sci.* 1987;17:207-10.
- Chaudhari SY. Craniofacial microsomia: A rare case report. *J Oral Maxillofac Radiol.* 2013;1:70-4.
- Carvalho GJ, Song CS, Vargervik K, Lalwani AK. Auditory and facial nerve dysfunction in patients with hemifacial microsomia. *Arch Otolaryngol Head Neck Surg.* 1999;125:209-12.
- Romsee MC, Verdonck A, Schoenaers J, Carels C. Treatment of hemifacial microsomia in a growing child: the importance of co-operation between the orthodontist and the maxillofacial surgeon. *J. Orthod.* 2004;31:190-200.
- Vento AR, LaBrie RA, Mulliken JB. The O.M.E.N.S. classification of hemifacial microsomia. *Cleft Palate Craniofac J.* 1991;28:68-77.
- Rajendran R, Sivapathasundaram B. Shafer's textbook of oral pathology. 5th ed, Elsevier, New Delhi. 2006;986:1189-90.
- McCarthy JG. Craniofacial microsomia. Joseph Grabb and smith's plastic surgery sixth edition by Charles H. Thorne. Chapter 2007;26:248-55.
- Kapur RA, Kapur RB, Sheikh SC, Jindal SD, Kulkarni SE. Hemifacial microsomia: A case report. *J Indian Soc Pedod Prevent Dent.* 2008;34-40.

**Yazışma Adresi / Address for Correspondence:**

Dr. Medhini Madi  
Postgraduate Student  
Department of Oral Medicine and Radiology  
A.B. Shetty Memorial Institute of Dental Sciences  
NITTE University  
Mangalore  
Email: medhkat@gmail.com.

Geliş tarihi/Received on:18.11.2013

Kabul tarihi/Accepted on:25.12.2013