

Hemifacial Microsomia with bilateral ear involvement

Ananya Madiyal, Vidya Ajila, Shruthi Hegde, Subhas Babu, Priyadharshini Alva

Department of Oral Medicine and Radiology A. B. Shetty Memorial Institute of Dental Sciences Nitte University, Karnataka, India

Address for correspondence:
Ananya Madiyal, Department of Oral Medicine and Radiology A. B. Shetty Memorial Institute of Dental Sciences Nitte University, Karnataka, India
ananyamadiyal@gmail.com

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ABSTRACT

Hemifacial microsomia is a rare form of craniofacial anomaly displaying abnormalities of the derivatives of the first and second branchial arches. A diagnosis of hemifacial microsomia is made if the patient presents with microtia, hypoplastic mandible, maxilla or the zygomatic arch on one side of the face. Bilateral involvement is rare. Here we present a rare form of the syndrome which presented with bilateral ear involvement and hearing deficit. Radiographs helped in diagnosing the case as hemifacial microsomia. The condition is extremely complex and heterogenous. Accurate diagnostic workup is necessary to differentiate this condition from other conditions of the first and second arch derivatives. We describe here the characteristic clinical and radiographic features which help to improve the knowledge on the disorder.

KEY WORDS: Hemifacial microsomia, hypoplasia, microtia

INTRODUCTION

Hemifacial microsomia (HFM) is the second most common craniofacial developmental defect after cleft lip and palate. It affects about one in every 5600 live births [1]. It was first described as a condition affecting the aural, oral and mandibular development in the 1960s [2]. It is a branchial arch developmental deformity syndrome in which one side of the face is underdeveloped including bony jaws and the overlying soft tissues [3]. It results due to abnormal development of the first and second branchial arches and the first branchial membrane which contribute to the development of facial structures. Damage to or disruption of the neural crest cells causes the facial abnormalities associated with hemifacial microsomia [2].

The German physician Carl Ferdinand von Arlt in 1881 first described a case of characteristic asymmetrical malformations of the face, eye and ear and later gave the term hemifacial microsomia. Converse et al stated that the term craniofacial microsomia should be used when cranial deformities are included. Gorlin et al described patients with unilateral microtia, macrostomia and mandibular condyle and ramus malformation as hemifacial microsomia and the variant with vertebral anomalies and epibulbar dermoids as Goldenhar syndrome [2,4].

Broad spectrum of abnormalities associated with HFM is misleading and results in delayed treatment initiation.

Better understanding of the etogenesis and features are essential in recognising and managing the disorder in order to improve the esthetic and functional deficit associated with this condition. We describe here a case of hemifacial microsomia with its characteristic clinical and radiographic features in an attempt to improve the knowledge and help in differentiating it from other similar syndromes.

CASE REPORT

A 15 year old male patient was reported with a chief complaint of defect in the jaws since birth. The patient was the older of 2 siblings and was born to non-consanguineous parents at full term. Antenatal history and family history was non-contributory. Medical history revealed that the patient had hearing deficit on one side and the patient had visited a physician for the facial asymmetry but no treatment was instituted for the same.

General examination revealed that the patient was well nourished with normal mental status. There was facial asymmetry of the right side (figure 1). Right side of the mandible was found to be hypoplastic with absence of the prominence over the right angle of the jaw. Facial antero-posterior and vertical dimensions were found to be reduced on the affected side. On mouth opening, there was deviation of the mandible towards the right side. Microtia was seen on the right and left side with rudimentary pinna

(figure 2). Hearing deficit was elicited in the right ear. Slight drooping of the lateral canthus of the right eye was seen. Mouth opening was limited. Intra-oral examination revealed high arched V-shaped palate with anterior open bite (figure 3) and crowding due to which there was palatal eruption of 23 with soft and edematous gingiva. Right maxillary lateral incisor was found to be missing along with retained deciduous root stump in the region of 53.



Figure 1. Extra-oral photograph showing facial asymmetry on the right side.

Panoramic radiograph showed hypoplasia of the ramus, condyle and coronoid process on the right side with prominent antegonial notch (figure 4). Developing third molar tooth buds were seen in all quadrants and 12 was found to be missing. Posterior-anterior cephalogram showed decreased ramus height of the right side with shift in midline towards the right. Right orbit also appeared to be hypoplastic. Deviated nasal septum and deviation of the maxilla towards the right side were also seen (figure 5). Lateral cephalogram shows underdevelopment of the mandible. Mandibular plane was found to be steep with marked reduction in the ramus height and mandibular length (figure 6). Based on the clinical and radiographic features provisional diagnosis was given as hemifacial microsomia involving the right side.



Figure 2. Extra-oral photograph showing deformed pinna.



Figure 3. Intra-oral photograph showing anterior open bite.



Figure 4. Panoramic radiograph showing hypoplastic condyle, coronoid, ramus and mandibular body on right side.

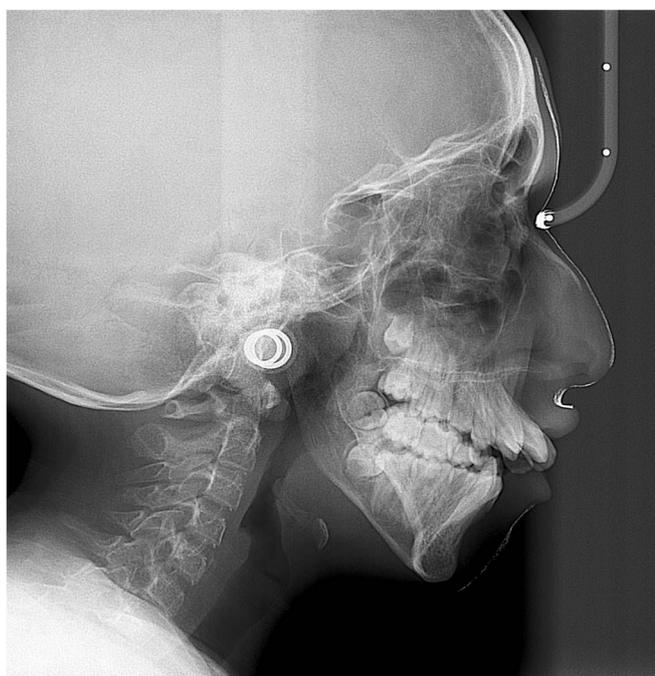


Figure 5. Postero-anterior cephalogram showing deviation of midline towards the right side with decreased ramus height.

DISCUSSION

Hemifacial microsomia is a rare congenital craniofacial abnormality where the phenotype is diverse. Males appear to be more commonly affected than females (3:2) and the right side is more often involved than the left [5]. The exact etiology of HFM is unknown but is clearly heterogeneous. Early loss of neural crest cells that form the facial structures is responsible for the characteristic features of this syndrome [3]. Other features seen with the syndrome like cleft palate (10% of the cases) and heart anomalies (50% of the cases) are consistent with early loss of neural cells [2]. Though familial inheritance is seen in some cases, several reports suggest

that HFM is caused due to focal hematoma formation and necrosis by anastomosis of the stapedia artery at the first and second branchial arches or teratogen activity like retinoic acid, thalidomide and primidone in the first 6-8 weeks of pregnancy [6].

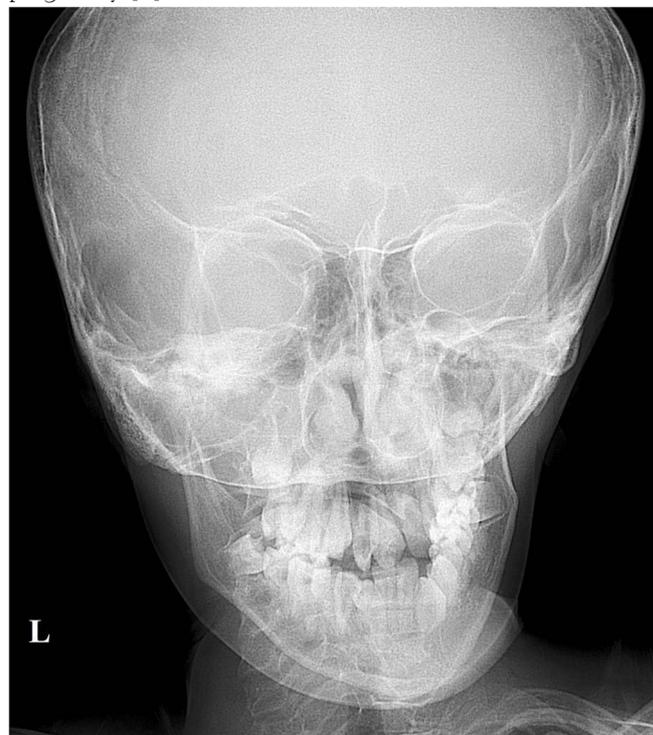


Figure 6. Lateral cephalogram showing steep mandibular plane.

Presence of asymmetrical hypoplasia of facial structures such as mandibular hypoplasia, presence of pre-auricular tags and microtia with or without periauricular skin tags are considered to be diagnostic of HFM [7]. Unilateral microtia or ear abnormality including preauricular tags has been suggested as a mandatory feature by some authors [2]. The maxillary, temporal and malar bones on the affected side may be hypoplastic and flattened [3]. The eye may thus seem to be at a lower level on the affected side. The present case also showed unilateral hypoplastic mandible due to reduced ramus height and drooping of the lateral canthus of the affected eye which helped us to diagnose it as HFM. Malformation of the external ear may vary from complete agenesis to a distorted and displaced pinna [3]. Bilateral involvement which is an unusual finding was seen in our case. Another common syndrome with bilateral malformation of the ear and micrognathia is Treacher-Collins syndrome [8]. But the absence of characteristic hypoplastic zygomatic arches and downward slanting palpebral fissures helped to diagnose our case as HFM which improves the body of knowledge regarding the varied features of HFM. There may be hypoplasia of the masticatory muscles and muscles of facial expression giving rise to an oblique lip line [2]. On the affected side, ear canal is often absent or blind ended [9].

Conduction deafness due to middle ear anomalies is seen in 30-50% of the cases and was consistent with our case [2,3].

Intraoral features seen in HFM include delayed development of teeth on the affected side, hypoplastic and aplastic teeth, absence of mandibular third molar, an increased frequency of missing teeth on the affected side; especially the mandibular second premolar [9,10]. The present case showed missing maxillary lateral incisor on the affected side. Chalky opacification of the enamel may be found on the central and lateral incisors on the affected side as a marker of development for HFM [3].

Owing to the varied clinical features of the syndrome, various classification systems are used to describe HFM. The SAT (skeletal-auricular-soft tissue) classification and the OMENS classification (O- orbital distortion, M- mandibular hypoplasia, E- ear anomaly, N- nerve involvement, S- soft tissue deficiency) are the most comprehensive and extensively followed system for the diagnosis of HFM [11,12,13]. OMENS classification system encompasses skeletal and soft tissue anomalies as well as facial nerve and extra-cranial problems. A diagnostic workup should be structured to each patient to determine the extent of structural anomaly and functional impairment. A panoramic radiograph gives an excellent overview of the maxillofacial complex while a frontal skull radiograph can depict the degree of symmetry. Computed tomography can provide both a three-dimensional image of the soft tissue covering the face as well as an image of the underlying bone [14].

MANAGEMENT

Involvement of the ear, eye, oral and craniofacial structures and sometimes other systems of the body like cardiac and renal justifies a multidisciplinary approach. 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 [7].

Excision of the preauricular tags and cartilage remnants can reduce some amount of social stigma associated with this syndrome [2]. Surgery may be done during the growth phase or after the growth phase is over. Recently, distraction osteogenesis has been tried with success for expansion and correction of muscles and ascending rami [7]. For patients with severe deformities, early surgical intervention with costochondral grafting is indicated while after pubertal growth spurt rotational advancement of mandible as well as alloplastic reconstruction of temporomandibular joint structures may be done [8].

CONCLUSION

HFM is a rare craniofacial abnormality which is heterogenous in nature displaying inconsistent phenotypes involving the

structures originating from the first and second branchial arches. Early recognition and prompt treatment by a team of specialists help in reducing the esthetic and functional disability associated with the syndrome. Counselling of families with sporadic occurrence should be done as HFM is found to have a degree of genetic etiology. All aspects of management must be maximised to provide these patients with optimal treatment.

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