Differential Diagnosis and Treatment Approaches of Hemifacial Microsomia: A Review

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Abstract

Hemifacial microsomia (HFM) is the second most common congenital anomaly of the head and neck region after cleft lip and palate (1-3). Since this condition has a wide range of phenotypic expression, different terminologies have been used for HFM in the literatures (4). The most obvious clinical finding in HFM is deficiency of hard and soft tissue on one side of the face (1-4), although bilateral involvement is seen in 10 to 15% of cases (2). Jaws, temporal, malar, orbital bones, masticatory muscles, ears (ranging from anotia to a mildly dysmorphic ear), nervous system, eyes (blepharoptosis, palpebral shortening, anophthalmia, microphthalmia, epibulbar dermoids, colobomas of the upper lid) and soft tissue are affected anatomic region (1, 2). Extent of facial asymmetry depends on the amount of ears abnormality and skeletal support. Facial asymmetry and chin deviation to affected side are obvious. The asymmetry may not be apparent in the infancy or childhood period but can be expressed by the age and developmental growth, usually becoming apparent by age four (5). There are controversies about the progressive nature of deformity in HFM (6).

For infants, with isolated microtia, complete craniofacial examination is recommended because isolated microtia can be an early clinical marker for HFM (7).

Incidence of HFM is approximately 1 in 3500 to 6000 live births (there are variations in different references) (1, 3-8). The male-to-female ratio and also right-side versus left-side involvement ratio is said to be 3:2 (3). Although some references have stated that these statistics may not be accurate (2).

The exact etiology of HFM has not yet been determined, but an old theory states damage to the stapedial artery can cause hematoma formation in the first and second branchial arches, which results in abnormal formation of the mandible (2, 8). Recently, laboratory studies suggest that the specific factor, responsible for the clinical presentation of HFM is early loss of first branchial arch neural crest cells (9-12). Environmental causes for HFM have been proved (including thalidomide, primidione, and retinoic acid administered during the organogenesis (2, 11, 13, 14) and multiple maternities (15) and recently some documents has confirmed the possibility of its genetic inheritance too (16-19).

The purpose of this paper is to explain the differential diagnosis of HFM and the possible treatment approaches for different grades of this anomaly.

Keywords: Asymmetry, Differential Diagnosis, Hemifacial Microsomia, Treatment

1. Introduction

Hemifacial microsomia (HFM) is the most common form of isolated facial asymmetry (1) and the second most common congenital anomaly of the head and neck region after cleft lip and palate (1-3). Since this condition has a wide range of phenotypic expression, different terminologies have been used for HFM in the literatures (4). The most obvious clinical finding in HFM is deficiency in the amount of hard and soft tissue on one side of the face (1-4), although bilateral involvement is seen in 10 to 15% of cases (2). Jaws, temporal, malar, orbital bones, masticatory muscles, ears (ranging from anotia to a mildly dysmorphic ear), nervous system, eyes (blepharoptosis, palpebral shortening, anophthalmia, microphthalmia, epibulbar dermoids, colobomas of the upper lid) and soft tissue are affected anatomic region (1, 2). Extent of facial asymmetry depends on the amount of ears abnormality and skeletal support. Facial asymmetry and chin deviation to affected side are obvious. The asymmetry may not be apparent in the infancy or childhood period but can be expressed by the age and developmental growth, usually becoming apparent by age four (5). There are controversies about the progressive nature of deformity in HFM (6).

For infants, with isolated microtia, complete craniofacial examination is recommended because isolated microtia can be an early clinical marker for HFM (7).
2. Grading of HFM

Different grading systems have been described for HFM. We preferred to use a simple one which describes HFM in three grades of severity. Of course grade two, includes two groups of mild and severe.

Grade I: glenoid fossa, condyle, ramus and soft tissues are normal but small on the affected side.

Mild grade II (in some literatures is described as type IIA): The ramus, glenoid fossa, condyle and soft tissue are present but more hypoplastic and malformed.

Severe grade II (in some literatures is described as type IIB): The ramus, glenoid fossa, condyle and soft tissue are severely hypoplastic and malformed, the TMJ has an anterior, inferior, medial location.

Grade III: there is no glenoid fossa, the condyle and the ascending ramus are absent, either completely or partly, soft tissues are deficient too. So mandible is almost floating (11, 20, 21) (Figure 1).

3. Differential Diagnosis of HFM

Patient with HFM must be distinguished from those with Treacher Collins syndrome, traumatic postnatal deformity, Goldenhar syndrome, hemi mandibular elongation, Parry-Romberg syndrome, juvenile rheumatoid arthritis, Nager acrofacial dysostosis syndrome, post axial acrofacial dysostosis, muscle dysfunction, branchio-oto-renal syndrome (BOR) and Maxillofacial dysostosis.

Treacher Collins syndrome (mandibulofacial dysostosis) shows a well-defined pattern of inheritance and unlike HFM, it is always bilateral. Zygomatic area and lateral orbital rim are deficient and the medial lower eyelashes are absent (2, 11, 22). The other distinguishing features are deficient or absent condyles and muscles, short ramus and retrognatia (11) (Figure 2).

The most common reason of asymmetric mandibular deficiency is an old condylar fracture (23). Trauma and infection within the temporomandibular joint could result in ankylosis of condyle to temporomandibular bone (24). Asymmetry due to trauma and mild HFM may be misdiagnosed. If a short ramus and a deficient condyle are present but ears and soft tissues are normal, an old fracture is suspected, in these cases the deformity is not present at birth and there is a history of trauma (11, 22) (Figure 3).

There is no obvious guideline to diagnose Goldenhar’s syndrome. However, patient with this syndrome have ear malformations (including microtia and accessory tragi), HFM, vertebral anomalies and epibulbar dermoids and/or eyelid colobomas (5). Between these features, multiple accessory tragi in a preauricular/mandibular distribution is an important clinical marker (25). Facial asymmetry is reported in 65 - 75% of cases (26) (Figure 4).

Figure 1. The Phenotypic Spectrum of Hemifacial Microsomia is Displayed in These Three Patients.

A. Grade I of HFM, deficiency of hard and soft tissue on left side of the face; B. Grade II of HFM, obvious chin deviation to right, severe hypoplasia of right condyle and soft tissues, ear malformation; C. Grade III of HFM, absence of left condyle and ramus, severe soft tissue deficiency, bilateral ear malformation.
**Figure 2.** Treacher Collin Syndrome

[Image: Reprinted from Proffit et al. (23).]

**Figure 3.** Condylar Fracture

[Image: Reprinted from Proffit et al. (23).]

**Figure 4.** Goldenhar Syndrome

[Image: Reprinted from Proffit et al. (23).]
In hemi mandibular elongation, chin and mandibular midline deviates to unaffected side. The form and size of condyle is normal, the neck can be either slender or normal but there is a long ramus (28, 29). This abnormality usually manifest itself after growth spurt but it can happen before and during growth spurt (11), unlike HFM that is present from the birth (Figure 5).

Juvenile rheumatoid arthritis can cause asymmetric deficiency in children. The TMJ often is affected in the multi-articular form of this disease. Asymmetry develops in two thirds to three fourths of the children because the disease progression on one side is faster than the other (11). In these patients, there is a limitation of mouth opening. The symptoms are mainly pain and morning stiffness of TMJ but do not exist in all patients (30) (Figure 6).

Progressive hemifacial atrophy (Parry-Romberg syndrome) is a slow progressing facial atrophy of subcutaneous fat and the associated skin, cartilage, and bone which is often on one side of the face. This disorder is self-limited, starts with an active progressive phase (2 to 10 years) then the atrophic process ceases and subsequent stability happens. Unilateral nature of this abnormality and subsequent facial asymmetry makes it one of the differential diagnosis of HFM. The acquired characteristics of progressive hemifacial atrophy distinguish it from congenital HFM (31). Some patients diagnosed with Parry-Romberg syndrome had atrophy or neural involvement of the arm, chest, and back (5) (Figure 7).

Nager acrofacial dysostosis (NAD) is a disorder of facial, limb, and skeletal morphogenesis which is inherited. Limb anomalies are the main sign of NAD (32, 33) radial ray abnormalities are seen in most of patients with Nager syndrome (Figure 8).

The faces of patients with post axial acrofacial dysostosis look like those with Treacher Collins syndrome but the
The difference is the existence of post axial limb deficiencies, absence or incomplete development of the fifth digital rays in the limbs and accessory nipples which are found in most of the patients (5, 34) (Figure 9).

Figure 8. Patients with Nager Syndrome

A, Nager acrofacial dysostosis syndrome; B, missing phalanges and syndactyly; C, absence of thumbs. Reprinted from Gorlin et al. (5).

Figure 9. A, Post Axial Acrofacial Dysostosis; Band C, post Axial Agenesis of Digits. Reprinted from Gorlin et al. (5).
Muscle dysfunction can cause facial asymmetry. Examples are muscle atrophy and contraction. Damage to the motor nerve of facial musculature causes muscle atrophy, as a result, deficiency of soft tissue and underlying skeleton happens. (Figure 10) Also, excessive muscle contraction can limit the growth. Torticollis is a clear example, in which excessive contraction of the neck muscle on one side, results in restriction of growth on that side and facial asymmetry (23), (Figure 11).

Branchio-oto-renal syndrome (BOR) is an autosomal dominant genetic disorder. The features are hearing loss, preauricular pits, branchial fistulae, pinnae deformities, malformed middle or inner ear, lacrimal duct external stenosis/aplasia, and renal anomalies (35) (Figure 12).

Maxillofacial dysostosis is a rare autosomal dominant disorder. The features are delayed onset of speech, and poor development of language skills without associated hearing loss. Down slanting of the palpebral fissures and minor auricle abnormalities are seen in some of patients. In this disorder, unlike HFM the main area of involvement is the maxilla, and ear anomalies are minimal (5) (Figure 13).
4. Treatment Approaches for Different Grades of HFM

Treatment plan of patients with grade one and mild grade two: Growth modification can help these patients (11, 36, 37). If the patient has potential growth, with the simulation provided by a hybrid functional appliance and stretching the soft tissue, growth is possible on the affected side.

The point that should be considered before beginning the treatment is that the child must be able to open his mouth about 20 mm to ensure that the translation of condyles is possible because only in this way mandible growth occurs.

An asymmetric functional appliance (hybrid) is used to guide the growth of dentoalveolar area on the affected side. This appliance directs the developmental growth of mandible in three planes of space. It guides mandible to a new postural position, controls the eruption of teeth and inhibits the soft tissue pressure (11).

Role of the orthodontist: An orthodontist should monitor the eruption of primary and permanent teeth. To improve facial asymmetry, mandibular deficiency and maxillary canting, an asymmetric functional appliance should be used. At the age of 12 to 15 years, after the adolescent growth spurt, a desirable occlusion can be achieved by means of full fix appliances. In a few cases, orthognathic surgery, rhinoplasty, soft tissue augmentation, might be needed to correct the remaining facial esthetic problems at older ages (11, 38).

5. Treatment Plan of Patients With Severe Grade Two

These patients used to be candidates for early surgery either to lengthen the affected ramus or to construct a condyle/ramus unit (11). Construction of a condyle and ramus should be reserved for children in whom the proximal portion of the mandible is completely missed. As long as there is a functional condyle, regardless of TMJ region morphology, we keep it (11, 39).

Two approaches are available to achieve the goal lengthening the ramus height:
1. surgical techniques with bone grafting; 2. distraction osteogenesis (DO).

Lengthening the ramus height through usual orthognathic surgery is so hard (11). Since, DO was introduced, gained great popularity in the treatment of craniofacial deformities. Because in this technique, the need for surgical intervention is minimal and not only can lengthen the affected ramus, but also augments the associated soft tissue and muscles of mastication (40). More recent studies have shown facial symmetry gained by DO is not stable in the long-term, especially of the affected ramus height (6, 41, 42), and no improvement have been seen in the soft-tissue (6, 41). However, DO treatment improves facial proportions and aesthetic for short time and has psychological advantages (42, 43). Because of the questionable stability of early DO treatment, it has been suggested to postpone any surgical intervention to the permanent dentition. If a large mandibular advancement is needed and there is not enough bone volume, a unidirectional corpus distraction, 1 year before the facial surgery, or an interpositional bone grafting can help getting bone volume needed (41). The common surgical treatment is mandibular osteotomy for lengthening ramus and corpus on the affected side and maxillary osteotomy for canting and midline correction (11, 39).

Role of the orthodontist: It seems functional appliances can improve minor skeletal and soft tissue asymmetries in grade I and mild grade II patients but there are controversies about the efficacy of functional appliances in severe grade of HFM (44, 45). So, an orthodontist should evaluate, whether it is worthwhile to subject patients with severe grade II or grade III of HFM to functional treatment (44). These patients usually, get orthognathic surgery, at the age 12 to 15 (11, 38). An orthodontist must prepare the dentition in a form that the surgeon can place skeletal segments in predetermined position. Therefore presurgical orthodontics includes removal of dental compensations, tooth leveling and aligning, and arch coordination (46).
6. Treatment Plan of Patients With Grade Three

In patients with grade III, glenoid fossa, condyle and ramus do not exist, therefore, the first treatment option for these patients, is reconstructive surgery of these issues with costochondral grafting (11, 41, 47). Reconstruction of TMJ is advised to be performed at very young age (47, 48). In addition to its psychosocial advantages, since there is potential growth, the growth of the affected side will be similar to the normal side, and facial asymmetry correction, will be stable (47). However, in some cases, sufficient growth of graft, may not happen or even, overgrowth may occur (47, 48). If the zygomatic arch is missing or severely deficient, it also can be reconstructed in this stage. These patients often need a second phase of surgery at the age 12 to 15 (11, 41). This stage of surgery is the same as the previous group, mandibular advancement is often necessary, and usually vertical elongation of the affected ramus should be done and also an asymmetric inferior border osteotomy is done to correct chin deviation, which improves both lip function and esthetics. Whatever the first stage of surgery is more successful, the need for maxillary canting correction will be less in second stage.

A third surgery, may be needed to improve bone and soft tissue contours, in the late teens (11). Free flap and fat graft are common tissue augmentation techniques, which are used to offset the remaining defects (49).

Depending on the severity of ear malformation, ear reconstruction may be undertaken at the time of any of the surgical stages (11).

Role of the Orthodontist: these patients are undergoing surgery at an early age, the presence of an orthodontist for monitoring is essential. After costochondral graft construction of the condyle, a posterior open bite is created on the affected side, an immediate functional hybrid appliance is advised to guide the dentoalveolar eruption on the affected side (50). Like severe grade II patients, these patients also need presurgical and postsurgical orthodontic treatments.

7. Conclusions

The first step to treating patients with HFM, is an accurate diagnosis. Many of the craniofacial anomalies can be misdiagnosed, although the treatment of some of these anomalies is like HFM. Decision making for treatment planning of patients with HFM, is highly dependent on the severity of the deformity and patients age. In mild grades of the anomaly, functional therapy can improve facial and occlusal symmetry in young ones, but in more severe grades, imposition of orthopedic treatment, may be undesirable and waste of time. Early surgical interventions to encourage the growth in the affected condyle may be helpful in severe cases, however, Consultation with the surgeon is advised to determine the patients who need early surgery. Distraction osteogenesis is a controversial treatment modality, which still needs more long-term studies.

References


