

FEATURES IN PATIENT AFFECTED WITH HEMIFACIAL MICROSOMIA AND CLASSIFICATION METHODS

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Abstract

Hemifacial Microsomia (HFM) is one of the most common congenital conditions treated in craniofacial centers worldwide. This condition is variably associated with anomalies of the jaws, ears, facial soft tissue, orbits, and facial nerve function and can be associated with extracranial anomalies. Diagnosis, treatment, and outcome assessment in HFM is challenging due to the wide phenotypic spectrum observed in this condition. Surgical treatment requires a coordinated team approach involving multiple specialties, which can include plastic surgery, craniofacial surgery, orthognathic surgery, and microsurgery.

Introduction

Hemifacial Microsomia (HFM) is a facial deformity second for frequency only to cleft palate with a variable incidence of 1: 4000 to 1: 5600 born alive, characterized by an asymmetry due to the hypodevelopment of the skeletal, nerve, muscular and vascular structures that derive embryologically from the 1st and 2nd brachial arches. Although bilateral hypoplasia has been noticed in 5 to 30% of the cases, when it occurs, it is generally asymmetric. Many studies have revealed a unilateral male predominance of the right hand side, while others have found equivalent left and right cases in both genders.Â Â

Review

Clinically, HFM presents an extremely variable phenotype. The main clinical evidence is facial asymmetry, due to hemimandibular hypodevelopment, which may present different stages of severity and be associated to hypoplasia of the rest of the facial skeleton (upper jaw, zygomatic and temporal bones), of the medium and external ear, of soft facial tissues and cranial nerves. Although these anatomical districts may present different levels of Hypoplasia, the face has typical clinical features. For long, the mandibule

has been considered the cornerstone of HFM, and has always been variably involved. Mandibular Hypoplasia can go from slight flattening of the condylar head to total agenesis of the condyle, of the upward ramus and glenoid fossa. The variable level of Hypoplasia in these structures can determine an altered relation between the articular heads at a temporo-mandibular articulation level, that vary from slight condyle malpositioning in the glenoid cavity, due to an abnormal cranial base articulation, to total relation absence in case of severe Condyle Hypoplasia. The mandibular body may be reduced in all its dimensions, with frequent increase of the gonial angle. In cases where Hypoplasia regards the zygomatic area, a variable single lateral Hypoplasia can be found in the orbital-zygomatic area, which can determine orbital dystopia. Upper jaw Hypoplasia combined with mandibular deficiency frequently determines dental malocclusions (class II occlusal relationships on the affected side and deep bite), and in proportion to the severeness of maxillo-mandibular deficiency, it is responsible for an upper occlusal plane on the affected side, which clinically determines the front shifting of the occlusal plane. The secondary involvement of skeletal structures not deriving from, but strictly linked to the first and second brachial arches, is inevitably due to their position. Therefore, it may regard the squamous and tympanic parts of the temporal bone, the styloid and mastoid processes and the pterygoid process of the sphenoid. The other clinical feature of HFM phenotype is soft tissue deficiency, associated with skeletal Hypoplasia. Cutaneous and subcutaneous connective tissue and neuromuscular deficiency are evident in the external ear and eye areas, and in the temporal, malar and masseteric areas of the face. Periocular anomalies can vary from slight inferior dislocation of the lateral part and/or of the palpebral rim to Microphthalmia or Anophthalmia.Â Iris or lashless eyelid colobomas can be present. Lack of cutaneous and subcutaneous mass contributes to the characteristic emptying of the temporal area and malar flattening, more visible if seen from an under chin perspective. These features may be highlighted by Hypoplasia of chewing muscles such as masseter, external temporalis pterygoid and internal pterygoid. The function of chewing muscles on

the affected side may also be compromised. The damaged lateral pterygoid on the affected side, in combination with a severe ipsilateral maxillo-mandibular Hypoplasia, worsens the dental occlusion and it contributes to the deviation of the chin from the affected side. Macrostomia or Cleft may be present in the oral commissure and Hypoplasia of the parotid gland. Neuromuscular Hypoplasia may be present in muscles controlling facial expressions. Facial paralyses are observed in 22% to 45% of patients and are caused by a great variety of muscular and neural anomalies. Considering the common embryonal origin of parts of the external and medium ear and those of the mandibula, it is not surprising that, auricular and preauricular malformations are not only fundamental, but necessary characteristics of this syndrome. When auricular malformations, such as Microtia and preauricular, skin or sinus malformations, are present as isolated evidence, they may represent the minor form of HFM. If case of isolated Dysplasia or simply one element of the whole phenotype, the ear malformations noticed in HFM, vary like the ones present in the other characteristics of the syndrome. External ear Hypoplasia varies from minor damage of the auricular architecture to complete external auricular Agenesis, with an Atresia of the ear canal. In severe cases, the only observable evidence of external ear development is the presence of an auricular residue, which is totally absent in the most severe cases. Variable Hypoplasia of the medium ear is also a common characteristic. External and internal ear Dysplasia may cause conductive hearing loss in 75% of the patients. In literature, a wide range of extra-cranial anomalies has been reported, among which skeletal, cardiovascular, renal, gastrointestinal and lung abnormalities. Increased proof of extra-craniofacial anomalies seems to be connected to the severeness of the facial malformation. Due to the variety of anomalies described and the numerous terms used to define the same clinical condition, it is easy to understand the nosological problems encountered in the attempt to set rigid diagnostic criteria, and the importance of a correct diagnostic and classification chart of the various pathological frames, in order to decide on the proper therapy. In the past, various clinical classification methods examined only one or two features of the syndrome. In 1960, Prunzansky et al. accurately described skeletal anomalies, classifying HFM mandibular anomalies into three levels, according to the increasing Hypoplasia of both the mandibular ramus and condyle, but it was incomplete in the description of other HFM phenotype features. The SAT (skeletal, auricle, soft tissues) clinical classification method used by David et al.

included the data, regarding both ear and soft tissues, assessing them according to the severity level of each of the three districts. To today, the method offering the most accurate clinical classification frame is, of course, the OMENS system, developed by Vento et al, thanks to a 1991 study on 154 patients affected with HFM. It focuses on each of the five Hemifacial Microsomia anatomic cases, according to Dysformia severity on a scale from 0 to 3. Each case corresponds to a letter of the acronym: O - Orbital Asymmetry, M - Mandibular Hypoplasia, E - Ear Malformation, N - Nerve Dysfunction and S- Soft Tissue Deficiency. The assessment is determined on the basis of conventional x-rays, among which postero-anterior, submittal and lateral tele-radiography and orthopantomography on both clinical examinations and diagnostic imaging. The orbital case reflects both dimension and position. When the latter is abnormal, there is an arrow marking superior or inferior dislocation. The mandibular case assessment is based on x-rays using the Pruzansky and Murray et al system. Mandibular anomalies are divided into three levels (I-III) according to the increasing Hypoplasia degree regarding both mandibular ramus and condyle. A normal, unaltered contralateral hemymandibula is considered means of comparison for all types of Microsomia. Mandibular type I is defined by normal morphological features of the ramus and condyle, but inferior in size. Mandibular type II shows significative architectural and dimensional distortions of the ramus, the condyle and semilunar incisure. Lastly, mandibular type III is characterized by severe distortions of the ramus or its agenesis. Subsequently, Kaban and colleagues divided type II into two separate categories reflecting the architecture and the temporomandibular articulation function; type II-A presents an acceptable anatomy and position of the glenoid fossa compared to the normal side. Classification regarding external ear anomalies partly uses the Marx and Meurman system. External ear malformations are divided into three levels of growing severity, which go from slight destruction of the auricular architecture to nearly total auricular aplasia. The system adds level zero which reflects the absence of any observable malformation. The category, regarding the nervous facial rami, includes the zygomatic and temporal rami in one group, and the vestibular, marginal mandibular and cervical rami in another, therefore dividing the face into upper and lower halves. Level zero refers to a non-nerve involvement, while level three indicates a panemifacial paralysis regarding all rami. Rating lack of soft tissues uses a modified version of the Murray and colleagues system; muscular /subcutaneous deficiency is defined as absent, slight, moderate or

severe. Classification for each side of the face is determined separately in cases of bifacial Microsomia. The Omens system represents a means to assess anomalies that form the very accessible, flexible, global and mainly objective Hemyfacial Microsomia spectrum. The classification system within each category comprises the entire range of Displasia severeness, and it defines each anatomic malformation in a very simple and reproducible way. The use of number rating is necessary to give objectivity, within certain limits, to the naturally subjective features of this disorder; in doing so, it helps the analysis of this population within the institutions. Using Coehn's words: the OMENS assessment used for Hemyfacial Microsomia is a pleasant addition to literature. At the same time, Coehn states that the OMENS system disconsidered extra-craniofacial anomalies. Therefore, in 1995, Horhan et al. modified the system, allowing the optional addition of an asterisk or a plus sign [OMENS (+)] to indicate the presence of associated extra-craniofacial anomalies. A supplementary critique is focused on the stated presence of orbital dystopia; indeed, Cousley and Calvert suggested this definition be bettered so as to determine the quantity of diagnostic imaging, necessary to classify the orbit, abnormal in both size and position (denomination 03). The original study performed by Vento and colleagues on 154 patients and on 65 patients by Poon et al. are, as we know of, the only studies that so far classify Hemyfacial Microsomia patients following the OMENS chart. In addition, the original study by Vento and colleagues is the only one that links the mandibular Hypoplasia level to the other four fundamental features of the classification system (orbital, auricular, nerve and soft tissue morphology). Their research found a positive connection between mandibular Hypoplasia and all other anatomic features of the acronym; this statement underlines the importance of mandibular anomalies in the syndrome. It would be convenient to classify the numerous cases of Hemyfacial Microsomia, according to the OMENS chart. The above mentioned studies, as Vento and colleagues suggest, would allow the independent analysis of the different Hemyfacial Microsomia anatomic features, many of which would reveal possible relations among the various cranio-facial and extra-craniofacial characteristics of this complex and variable syndrome.

Conclusions

Considering the complex nature of this condition,

patients affected with HMF are treated by a multidisciplinary team able to provide coordinated and specialized treatment; besides orthodontic treatment and orthognathic surgery, it includes nutritional therapy, speech therapy, plastic surgery, psychological therapy and an audiological and ophthalmological evaluation. Team approach helps to develop a realistic treatment plan, which allows to achieve the objectives set, improving patients' quality of life and expectations. We understand that the OMENS assessment system is probably, so far, the most complete, allowing us to have a global picture of pathology severeness, but it doesn't include a category defining the range regarding malar/third medium skeletal Hypoplasia. The nerve category doesn't indicate the presence of a single ramus paresis. We hope that, until a more inclusive system is developed, the variables included in the modified OMENS system (*), will allow to assess the incidence of these specifics.

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