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Hemifacial Microsomia – An Intermediate Form in Oculo-Auriculo-Vertebral Disorder Spectrum

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Abstract

Oculo-auriculo-vertebral spectrum (OAVS) refers to three rare disorders that many clinicians believe to be intimately related to one another and which represent the range of severity of the same disorder. These disorders are apparent at birth (congenital). As the name suggests, they involve malformations of the eyes, ears and spine. Oculo-auriculo-vertebral disorder (OAVD) represents the mildest form of the disorder, while Goldenhar syndrome presents frequently as the most severe form. Hemifacial microsomia appears to be an intermediate form. The disorder is characterized by a wide spectrum of symptoms and physical features that may vary greatly in range and severity from case to case. However, such abnormalities tend to involve the cheekbones, jaw, mouth, ears, eyes, and/or bones of the spinal column (vertebrae). Although, in most cases (about 60%), such malformations affect one side of the body (unilateral), approximately 10 to 33 percent of affected individuals have such malformations on both sides of the body (bilateral), with one side typically more affected than the other (asymmetry). In the majority of such cases, the right side is more severely affected than the left. In most cases OAVS appears to occur randomly, with no apparent cause (sporadic). However, in some cases, family histories suggest autosomal dominant or recessive inheritance. In addition, some researchers suggest that the disorder may be caused by the interaction of many genes, possibly in combination with environmental factors (multifactorial inheritance).

Our case is part of this spectrum having facial asymmetry (microsomia) with vertebral anomalies.

Keywords: Oculo-auriculo-vertebral spectrum (OAVS); Goldenhar syndrome; Hemifacial microsomia; Preauricular Tags; Tessier facial cleft; Trachiomalacia; Hypospadias with chordee.

Introduction

Hemifacial microsomia is a congenital disorder that affects the development of the lower half of the face, most commonly the ears, the mouth and the mandible. It can occur on one side of the face or both. If severe it can lead to difficulties in breathing, obstructing the trachea and requiring a tracheotomy. It is the second most common facial birth defect after clefts, with an incidence in the range of 1 in 3500 to 4500.[1] Hemifacial microsomia shares many similarities with Treacher Collins syndrome.

The condition is also known by various other names:

- Lateral facial dysplasia
- First and second branchial arch syndrome
- Oral-mandibular-auricular syndrome
- Otomandibular dysostosis
- Craniofacial microsomia

The condition develops during the fetal stage of pregnancy at approximately 4 weeks of gestation, when some form of vascular problem leads to clotting and a poor supply of blood to the face. This can be caused by a

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Fig 1: Preauricular Tags - 3 Pre Auricular Tags, Pre Auricular Pits on Left Side with Microtia and 1 Pre Auricular Tag on Right Side of the Face

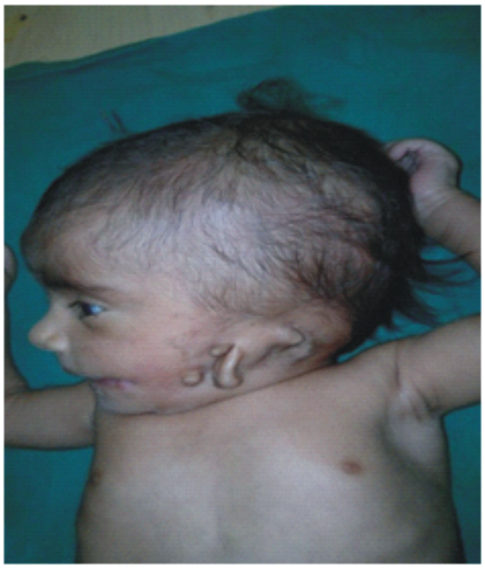
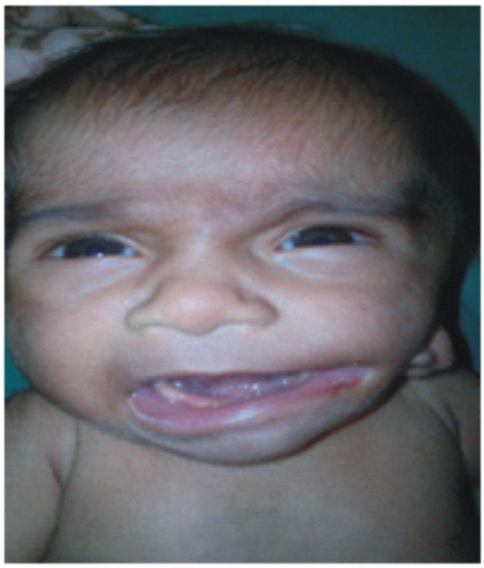


Fig 3: Micrognathia with Cleft between Angle of Mouth and Microtic Ear (Tessier Facial Cleft)



physical trauma, though there is some evidence of it being hereditary.[2] This restricts the developmental ability of that area of the face. Currently there are no definitive reasons for the development of the condition.

The clinical presentation of hemifacial microsomia (HFM) can be quite variable. In some children, just the ear is affected (microtia or a small, underdeveloped ear), and in other children, multiple parts of the face and body can be affected. While there is no grading scale that is in use universally, the OMENS scale (standing for Orbital, Mandible, Ear, Nerves and Soft tissue) was developed to help describe the heterogeneous phenotype that makes up this malformation. Children with HFM are at higher risk for hearing loss, and should have their hearing tested. A proportion of children with HFM will have extra-cranial anomalies, including

Fig 2: Facial Asymmetry with Left Ear Lying Below Level of Right

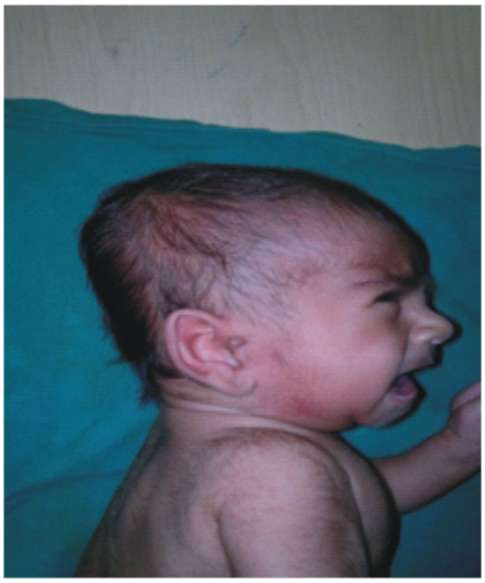


Fig 4: Tracheomalacia

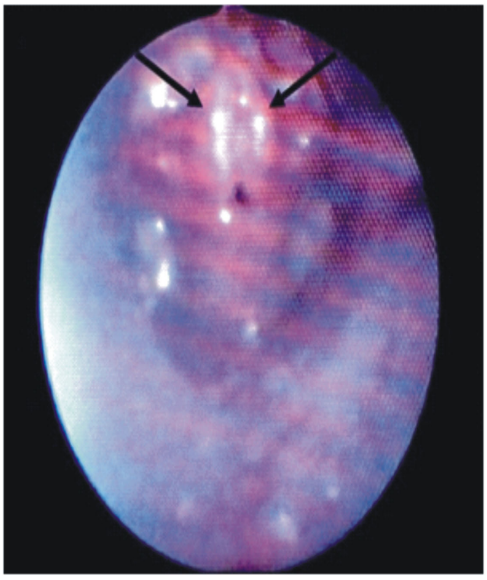
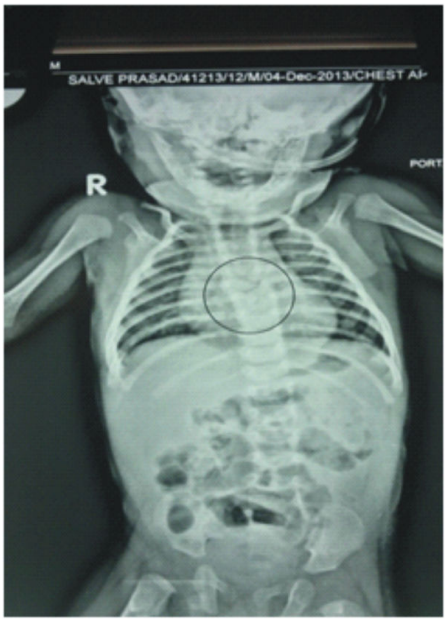


Fig 5: Vertebral Scoliosis



abnormalities of the kidneys and cervical spine, which are important to detect early in childhood. Cognition and development is usually typical in children with HFM.

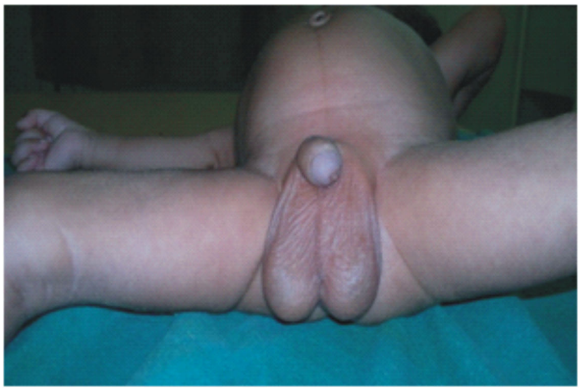
Some have hypothesized that the severity of hemifacial microsomia depends on the extent of the physical injury (the area with a short supply of blood) and the gestational age of the fetus – the earlier the injury, the greater the chance of wide-scale problems. This has not yet been proven in an animal or human model.

Case Summary

A 9 month old male baby admitted in our PICU for "Dettol poisoning" due to accidental ingestion of dettol (5 ml) at home when his examination revealed following findings on examination

- Preauricular Tags - 3 pre auricular tags, pre auricular pits on left side with microtia and 1 pre auricular tag on right side of the face.
- Facial asymmetry with left ear lying below level of right
- Micrognathia with cleft between angle of mouth and microtic ear (Tessier facial cleft).

Fig 6: Hypospadias with Chordee



- Tracheomalacia
- Vertebral scoliosis.
- Genital examination revealed hypospadias with chordee

A Baby is 1st product of 2nd degree consanguineous marriage with mother having previous 1 spontaneous abortion delivered by full term vaginal delivery.

Discussion

Craniofacial microsomia, a variable hypoplasia of the skeleton as well as of the overlying soft tissue, is the second most common congenital syndrome of the head and neck region, with an incidence as high as 1 in 3,500 live births.

The deformity has been known by a variety of terms. In continental Europe, the term dysostosis otomandibularis has been used. Gorlin and Pindborg preferred hemifacial microsomia, but this term implies that the syndrome is unilateral and that the deformity is confined to the face. I prefer the term unilateral craniofacial microsomia or, when there is bilateral involvement, bilateral craniofacial microsomia.[3]

Craniofacial microsomia can be confused with Treacher Collins syndrome, but the latter shows a well-defined pattern of inheritance and, unlike bilateral craniofacial microsomia, the pathology is symmetrical. Treacher Collins syndrome has other distinguishing features

(absence of the medial lower eyelashes and antegonial notching of the mandible), findings that are absent in craniofacial microsomia. Likewise, craniofacial microsomia should be distinguished from micrognathia of the developmental or posttraumatic type. In the latter, the underdevelopment is restricted to the mandible and there is no evidence of facial paralysis, ear anomalies, or soft- tissue hypoplasia of the cheeks.[4]

Etiology

The hypoplasia can be variably manifest in any of the structures derived from the first and second brachial arches (Table 26.1), accounting for the wide spectrum of deformity observed in this syndrome.

There is no evidence of genetic transmission of the syndrome. In a series of 102 affected patients, only four had a sibling or parent with evidence of craniofacial microsomia; in- deed, only a few pedigrees of the syndrome have ever been reported. Despite the possibility of an occasional autosomal dominant transmission, only a 2% to 3% recurrence rate was found in a study of first-degree relatives.

The most commonly accepted is a teratogen theory of a vas- cular insult, with hemorrhage and hematoma formation in the developing first and second branchial arches and subsequent maldevelopment of the latter. The stapedial artery is a temporary embryonic collateral of the hyoid artery, which forms connections with the pharyngeal artery, only to be replaced by the external carotid artery. Defects of this temporary vessel may result in hemorrhage, accounting for injury to the developing first and second branchial arches.

Laboratory phenocopies of craniofacial microsomia have been created following the administration of triazine to the developing mouse and thalidomide to the monkey. Histo- logic studies demonstrated hematoma formation with varying amounts of hemorrhage before formation of the stapedial artery. The spectrum of the pathology varied depending on the volume of hemorrhage, ranging from involvement of only the external ear and auditory ossicles to a larger defect involving the zygomatic complex and the entire mandible on the affected side of the mouse model. Moreover, the laboratory finding was supported by the clinical documentation in Germany of approximately 1,000 severe cases and an additional 2,000 less severe cases of craniofacial microsomia following the widespread use of thalidomide as a tranquilizer in pregnant women.[3,4]

Tessier, in a classification system of orbitofacial clefts, in voked a clefting mechanism as he described three types of clefts involving the orbitozygomatic complex in patients with craniofacial microsomia.

Clinical Findings

There is a wide variety of pathologic expression of craniofacial microsomia in the following anatomic regions: jaws, other skeletal components, muscles of mastication, ears, nervous system, and soft tissue BLE 26.1

Jaws

The most obvious deformity is the mandible, especially the ascending ramus, which can be absent or reduced in the vertical dimension. The size of the condyle usually reflects the

Figure 1: Patient with Left-Sided Craniofacial Microsomia Demon - Strating the Characteristic Occlusal Cant Upward on the Affected Side with Associated Cheek Hypoplasia and Ear Anomaly

Table 1

Structures Derived from the First and Second Branchial Arches and the Otic Capsule

First branchial arch	
Maxillary process	**Maxilla Palatine bone Zygoma**
Mandibular process	**Trigeminal nerve** **Anterior part of auricle Mandible** **Head of malleus Body of incus Tympanic bone** **Sphenomandibular ligament**
First branchial groove	**External auditory meatus** **Tympanic membrane**
First pharyngeal pouch	**Eustachian tube** **Middle car cavity**
Second branchial arch	**Facial nerve** **Posterior part of auricle Manubrium of malleus, long process of incus, stapedial superstructure, tympanic surface** **Stapedial artery, styloid process, stylohyoid ligament** **Lesser cornu of hyoid**
Otic capsule	**Vestibular surface of stapes, internal acoustic meatus** **Inner ear**

Modified from Pearson, A. A., and Jacobson, A. D. The development of the ear. In: Manual of the Am. Academy of Ophthmology & Otolaryngology, Portland: University of Oregon Printing Dept., 1967 and from Converse, J. M. *Reconstructive Plastic Surgery*. Philadelphia: Saunders, 1977.

degree of hypoplasia of the ramus. Involvement of the temporomandibular joint (TMJ) can range from mild hypoplasia to only a pseudoarticulation at the cranial base. In addition to being short, the ramus is usually displaced toward the midline. Because of the hypoplastic ramus, the mandibular plane angle is increased and the body of the affected mandible can show an increased horizontal dimension.[5]

The chin is deviated toward the affected side and there is a corresponding cant of the mandibular occlusal plane, which is paralleled in the corresponding planes of the floors of the maxillary sinuses and the pyriform apertures. Similarly, the maxillary and mandibular dentoalveolar complexes are also reduced in the vertical dimension on the affected side. In addition to crowding, there is often delayed eruption of the deciduous and permanent teeth; the molars can also be absent.

Pruzansky proposed a classification of the mandibular deficiency, which was later modified by Mulliken and Kaban (Fig 1):

Type I: Mild hypoplasia of the ramus, and the body of the mandible is minimally or slightly affected.

Type II: The condyle and ramus are small; the head of the condyle is flattened; the glenoid fossa is absent; the condyle is hinged on a flat, often convex, infratemporal surface; the coronoid may be absent.

Type III: The ramus is reduced to a thin lamina of bone or is completely absent. There is no evidence of a temporomandibular joint.

The above classification was subsequently modified by sub- dividing the type II mandible according to the pathology of the temporomandibular joint region. In type IIA, although the ra- mus and condyle are abnormal in size and shape, the glenoid fossa–condyle relationship is maintained because the glenoid fossa has a position in the temporal bone similar to that of the contralateral side. Temporomandibular joint function is almost normal. In contrast, in Type IIB, the condyle is hypoplastic and malformed and displaced toward the midline relative to the contralateral side. Patients open with restricted hinge like functioning of the mandible on the ipsilateral side.[14]

Other Skeletal Components

The maxilla is reduced in the vertical dimension and, depending on the degree of hypoplasia of the mandible, there is a corresponding cant of the occlusal surface of the maxillary dentition. The maxillary molars are consequently slow to erupt.

The zygomatic complex can be reduced in all of its dimensions; the zygomatic arch can be decreased in length or even absent. These findings, combined with soft-tissue deficiency of the cheek, result in a reduction or shortening in the distance between the oral commissure and tragus (often rudimentary) on the affected side.

The temporal bone can also be involved, although the petrous portion is usually spared. The mastoid process can be hypoplastic and there can be partial or complete lack of pneumatization of the mastoid air cells. The styloid process can be shortened or absent. The orbit is often reduced in all dimensions, and occasional patients have microphthalmos. The frontal bone can be flattened, giving the illusion of a plagiocephaly without radiographic evidence of synostosis of the ipsilateral coronal structure.

Malformations of the cervical vertebrae are not uncommon and include the presence of hemivertabrae, fused vertebrae, and even a basilar impression syndrome. Goldenhar described a variant of craniofacial microsomia characterized by epibulbar *dermoids/ lipodermoids, associated vertebral (usually cervical)*, and occasional rib anomalies.

Muscles of Mastication

The syndrome is not restricted to the skeleton; the associated muscles of mastication are hypoplastic. The deficiency is not always proportional to the skeletal deficiency. A three dimensional computed tomography (CT) scan study compared the volume of the mandibular deformity to that of the adjacent muscles of mastication and noted that there was not always a 1:1 relationship in the degree of pathologic involvement.[9]

Muscle function is impaired, as is especially evident with lateral pterygoid muscle function on the affected side. The lateral pterygoid muscle is responsible for movement of the mandible and chin point to the contralateral side. Consequently, in patients with unilateral craniofacial microsomia who attempt a protrusive chin movement, the chin deviates to the affected side during opening and during forceful protrusion. The hypoplastic lateral pterygoid muscle on the affected side is overpowered by its unaffected counterpart. Moreover, mouth opening is also adversely affected by the hypoplastic ramus and malpositioned temporomandibular joint

Ears

Involvement of the auricle occurs in most patients. Meur- mann proposed a classification of the external ear deformities: Grade I, distinctly smaller malformed auricle but all components are present; Grade II, only a vertical remnant of cartilage and skin with complete atresia of the external auditory canal; Grade III, almost complete absence of the auricle except for a small remnant, usually a soft tissue lobule. There is not a direct correlation between the auricular deformity, as classified using Meurmann's proposal, and the hearing function as measured by audiometry and temporal bone tomography. [9]

Nervous System

Cerebral abnormalities, although rare, can occur and include hypoplasia of the cerebrum and corpus callosum, as well as hydrocephalus of the communicating and obstructive types. The brainstem can be involved secondarily because of anomalies of the cervical vertebrae, resulting in disturbances such as impression of the brainstem.

The most common cranial anomaly is a facial palsy of varying degrees, attributed to the following (alone or in combination): absence of the intracranial portion of the facial nerve and nucleus in the brainstem, aberrant pathway of the nerve in the temporal bone, or angenesis of the facial muscles. Absence of

facial nerve function in the distribution of the marginal mandibular branch is seen in approximately 25% of patients, with weakness of other components, such as the buccal and zygomatic branches, occurring in a smaller percentage.

Soft Tissue

On the affected side, preauricular skin tags are common, and the skin and subcutaneous tissue of the cheek region show varying degrees of hypoplasia. The muscles of mastication are also involved, and hypoplasia or aplasia of the parotid gland has been documented. The soft-tissue deficiency is multidimensional and may result in a marked reduction in the distance between the oral commissure and the rudimentary ear on the affected side.

Lateral facial clefts (macrostomia) are common associated findings and also contribute to the overall cheek hypoplasia. Overt clefts of the soft palate are said to occur in 25% of patients, and the soft palate may deviate to the affected side on voluntary function.[9]

Treatment

It must be recognized that there is no prescribed treatment program for the child with craniofacial microsomia. The pathology, as emphasized before, is variable, and other factors, such as growth and development and prior therapy, must be considered before recommending an individualized treatment program. Surgical correction of the unilateral deformity is challenging. Consequently, all treatment plans must be customized according to the needs and age of the individual patient. [11]

Younger than Two Years of Age

Excision of the preauricular skin tags and cartilage remnants is often satisfying to the parents, because it removes some of the stigmata of the syndrome. Likewise, macrostomia can also be corrected by a commissuroplasty on the affected side or on both sides in bilateral craniofacial microsomia. In the occa- sional patient with involvement of the fronto-orbital region, characterized by severe retrusion of the supraorbital bar and frontal bone, a fronto-orbital advancement–cranial vault remodeling can be performed as a combined craniofacial surgical procedure.

Mandibular distraction is indicated in the newborn or infant with sleep apnea (with or without a tracheostomy). It can correct not only the sleep apnea but also the associated alimentary or feeding problems (e.g., swallowing, reflux).[8]

Two to Six Years of Age

In the child with mild deformity, such as Pruzansky type I mandible and a horizontal occlusal plane (Munro and Lauritzen type IA), no surgical treatment is recommended at this age.

In the child with severe reduction in the vertical height of the mandibular ramus (Pruzansky types I and II) and obvious aesthetic deformity, the technique of distraction osteogenesis can be considered after the child has attained at least 2 years of age. Sufficient clinical experience with mandibular distraction has accumulated to demonstrate that this technique not only lengthens the affected ramus, but also augments the associated soft tissue and muscles of mastication. The latter finding and the gradual nature of the distraction process low- ers relapse rates. Studies also demonstrate that the distracted ramus/condyle assumes a more anatomic size, shape, and position.[10]

In the patient with a Pruzansky type III deformity without evidence of a ramus, condyle, and glenoid fossa (or zygomatic arch), a preliminary costochondral rib graft reconstruction should be performed at approximately age 4 years. In this technique, the glenoid fossa, zygomatic arch, and ascending ramus are reconstructed in a singular surgical procedure (Fig 26.6). If there is a persistent mandibular deficiency, distraction, as a secondary procedure, could

be considered

In the child with bilateral craniofacial microsomia (Pruzansky types I and II mandibular deformity) with associated sleep apnea (with or without tracheostomy), bilateral mandibular distraction can be performed after sleep studies have established the diagnosis and the latter has been confirmed by endoscopy. In these children, the treatment can result in removal of the tracheostomy. If there is no evidence of the mandibular rami, bilateral costochondral rib graft reconstruction is the treatment of choice.[12]

Six to Fifteen Years of Age

This is the period of orthodontic treatment, including possible functional appliance therapy to promote eruption and growth of the dentoalveolus on the affected side. Distraction can be considered in the patient with chronic low-grade sleep apnea, and in the patient with severe deformity who has never received treatment. Ear reconstruction is often undertaken during this period. Insertion of a microvascular free flap to augment the facial soft tissue and improve facial contour on the affected side frequently results in considerable aesthetic improvement.

Older than Fifteen Years of Age

Surgery is often indicated in the period of skeletal maturity because of residual deficiency resulting from inadequate growth and development on the affected side, severe malocclusion, or failure of the patient to seek treatment previously.

At this point in time, when craniofacial growth and development are almost complete, the following procedures could be considered:

(a) limited autogenous bone grafting of deficient portions of the craniofacial skeleton;

(b) bilateral mandibular advancement in patients with mild to moderate mandibular micrognathia;

(c) combined Le Fort I osteotomy, bilateral mandibular osteotomy, and genioplasty and

(d) microvascular free flap to augment the soft tissue of the face on the affected side. [13]

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