What is Hemifacial Microsomia?

By Pravin K. Patel, MD and Bruce S. Bauer, MD
Children’s Memorial Hospital, Chicago, IL
773-880-4094

Early in the child’s embryonic development the structures destined to become the various parts of the face may not develop normally. The left and right side of the face may not grow equally or the entire lower jaw may not keep pace with the rest of the developing face. Not uncommonly these deformities of the jaw are also associated with the underdevelopment of the ear and other soft tissues, the overlying facial muscles and skin of the face.

The term Hemifacial or Craniofacial Microsomia is used to describe the condition when one side of the child’s face is smaller and malformed and the term microtia (micro means small and otia means ear) is used. The term Goldenhar Syndrome may be used to describe this group of deformities when the eye (epibulbar dermoids) and spine (hemivertebra) are also affected. There is a broad spectrum to this condition and there are many names associated with it: Tessier Number 7 cleft, oral-mandibular – auricular syndrome, first and second branchial arch syndrome, lateral facial dysplasia, and others. Although it is described as involving only one side (unilateral condition) of the face, it may also involve both sides (bilateral) of the face to different extent resulting in an asymmetrical bilateral hemifacial microsomia.

How common is it and How or Why does it occur?

At most craniofacial centers, the second most common deformity seen, after children with cleft lip and palatal deformities, is hemifacial Microsomia. It is said to occur in approximately 1 in 3500 to 1 in 5000 births. For families with one child affected with hemifacial microsomia, the likelihood of a second child with the same condition is less than 1%, and the risk is 3% for an adult with hemifacial microsomia to pass this on to his children. There are some variants of hemifacial microsomia where the likelihood of passing this to their children is significantly greater and a discussion with a geneticist is valuable.

A genetic cause has not yet been identified and it is believed that most cases occur because of an event in the developing embryo that disrupts the normal development of the ear and jaw. In the first six weeks of embryonic life, the ear forms from the coalescence of six small bumps, and closely associated with the formation of the ear is the development of the lower jaw. Because of this close relationship, ear deformities are frequently associated with deformities of the lower jaw, the mandible. While how it may occur can be understood, exactly why it occurs remains unknown today.

What are the anatomic and functional deformities?

Each of the structures is involved to varying degrees of severity. In some children only the ear deformity is evident while in others the ear is normal but the jaw is affected. In most severe cases, all the soft tissue and bony structures are hypoplastic.

The Soft Tissue Deformity

The extent of soft tissue involvement may vary from the barely perceptible to severe deficiency. The structures affected include the skin, the muscles and nerve of facial expression, the salivary gland and the ear.

It is the appearance of an abnormal external ear that is the most noticeable feature. Frequently it is no more than a ‘lump’ on the side of the face and bears little resemblance to the structure we recognize as an ear. Along with the external ear deformity, the ear canal and the internal structures of the ear that allow for hearing may also be affected. While children with unilateral involvement have problems locating the direction from which sound
comes, most children do not need hearing aides as long as the unaffected ear has normal hearing. In some children
the various branches of the facial nerve and the associated muscles which allow for facial expression may
be affected. Various degrees of facial paralysis are seen. The muscles that work the lower jaw and allow
mastication are also affected and occasionally the muscles of the soft palate and tongue on the same side as well.
The thickness of the skin and underlying tissue is deficient of hypoplastic. In some children there is a lateral cleft
of the lip extending from the corner of the mouth toward the ear resulting in macrostomia, an enlarged opening of
the mouth.

The Bony Deformity

Even though the upper facial bones (maxilla, zygoma, orbit and temporal bones) may be involved, it is the lower
jaw, the mandible that is believed to be the “keystone” to this deformity. With continued asymmetrical growth
of the mandible, the facial deformity worsens. The amount of deficiency (hypoplasia) of the mandible varies
from being minimally involved to where there is complete absence of the vertical portion (ramus) and the
various bony structures (zygomatic arch and condyle) of the temporomandibular joint. The chin is deviated to
the involved side and becomes more noticeable with opening and closing of the mouth. This hypoplasia of the
mandible affects the normal downward growth of the upper jaw, the maxilla, and this results in a cant to the
teeth. In addition, the cheek bone (zygoma) may be deficient and the orbit may be malpositioned.

How is it treated?

These varied deformities require treatment in order to improve a child’s ability to breathe, eat, speak, and hear.
Because of the degree and the complexity of various structures involved, a coordinated approach is needed to
reconstruct anatomically and functionally the individual elements if the face. Caring for children with
hemifacial microsomia requires a multidisciplinary approach. This means a close co-operation of a number of
pediatric specialists – plastic surgeons, otolaryngologists, dentists, orthodontists, audiologists, speech and
language pathologists, geneticist. Because of the great variability of presentation, treatment will necessarily
vary and must be individualized. Nevertheless, the goals remain the same: to restore the normal shape and
contour of the face by both correcting the bony deformity and the external ear and the soft tissue deficiency and
the external ear and to restore the normal function of hearing, speech and bite to the extent possible.

The key to reconstructing the facial skeletal deformity is correcting the cant of the mandibular asymmetry.
While in some cases orthodontic devices can sometimes be used to stimulate mandibular growth, more often
when the deformity is significant surgical procedures are needed. This means surgically repositioning the lower
jaw to correct the cant and bring the chin to the center to a more symmetrical position to match the other side.
On the side with the bony deficiency, the mandible can be lengthened by using a rib or by using a newer
technique called distraction osteogenesis that gradually lengthens the bone by using a special device. Distraction
osteogenesis may provide a means of treating many of these deformities both at an earlier age and with the
potential for better long-term results than conventional treatment of using a rib graft typically done in early
childhood. When the mandible is lowered, this creates what is called an “open bite”, since the upper jaw
remains in its uncorrected canted position, the teeth of the upper jaw do not meet the teeth of the lower jaw on
the affected side. The upper jaw is then gradually brought down to meet the lower jaw. Even with correction of
the occlusal cant in childhood, both upper and lower jaw may need to be simultaneously repositioned in
adolescence at a time when facial growth is completed. This requires cutting the bones of the upper and lower jaw,
realigning the various elements of the facial bones and fixing them into their position with plates and screws.

Reconstructing the ear is a surgical challenge. The ear is built from the child’s own rib cartilage and requires a
number of stages, frequently three to four operations. Reconstruction of the ear typically begins between ages 5
and 6 when most of the growth if the normal ear on the other side is nearly complete and the child’s chest wall
cartilage is large enough to sculpt the ear framework. The size and position of the normal ear is used as a template
for the new ear.
The ear framework is placed in either a skin pocket buried under a thin layer of vascularized tissue from the scalp and then covered with a skin graft. The lobule remnant frequently present is repositioned more symmetrically and the tragus is constructed from the concha or the bowl of the other ear. Each of these may be either a separate surgery or integrated with other surgical procedures. Both the ear reconstruction and jaw reconstruction can frequently be integrated in childhood.

Even after re-alignment of the facial skeleton there is not infrequently a soft tissue deficit. This requires transplanting a soft tissue from another part of the body to restore the symmetry in volume. Frequently the skin from the upper back (scapular region) is used. In order for this tissue to live, the small vessels measuring between 1 to 2 mm in size must be connected to the vessels in the face. This is done using a microscope and is called microvascular surgery.
A child born with craniofacial / hemifacial microsomia, the left side of his face is smaller than the right. The left ear failed to develop, and the eye and jaw are canted. Reconstruction involves restoring the symmetry to the facial skeleton and soft tissue volume, along with building an ear. The CT scan illustrates the skeletal asymmetry. The left zygomatic arch and mandible are underdeveloped. The left upper jaw (maxilla) and orbit are affected as well.

Reconstructing the ear involves several operations beginning typically between the ages of five and six. The child’s chest wall cartilage is carved to give the overall form of the ear. Further surgery involves, elevating the ear from the side of the face and adding other finer elements to recreate the complicated structure we recognize as the external ear.
FIGURE 3 A, B, C [Clinical Photographs]
Before and after in a child with microtia without the jaw asymmetry.

FIGURE 4 A, B [Clinical Photographs]
Before and after in a child with left hemifacial microsomia. In children with hemifacial microsomia in addition to microtia, the ear reconstruction is more complex and symmetry difficult to obtain because of the underlying bony deficiency.

FIGURE 5 A, B, C, D, E [Clinical Photographs and Line Drawing]
This child with Goldenhar Syndrome, a variant of hemifacial microsomia. Her eyes and cervical spine are involved. In older children orthognathic surgery is used to correct the facial skeletal asymmetry with various osteotomies of the jaws. The bones are repositioned, and when additional bone is needed, rib is used. Reconstruction must often wait until adolescence when facial growth is completed. [Line drawings are from Ian Munro, Operative Techniques in Plastic and Reconstructive Surgery, 1994.]
Distraction osteogenesis is used to lengthen the lower jaw similar to the way in which orthopedic surgeons correct a child’s legs of different length by slowly ‘stretching’ the bone that is there. This new technique in some children replaces the need for bone grafting and allows skeletal correction at an earlier age in a child.

This child was born with asymmetric hemifacial microsomia. Because of severe mandibular deficiency, she had difficulty breathing and needed a tracheostomy.
FIGURE 8 A, B  [Clinical Photographs]

Lengthening the lower jaw using distraction osteogenesis allowed her tracheostomy tube to be removed and helped to correct the skeletal asymmetry. She then underwent staged ear reconstruction.

FIGURE 9 A, B, C  [Line Drawing and Clinical Photographs]

Using microvascular surgery, the skin and underlying tissues from her back was transferred to correct the soft tissue volume deficiency.
Surgical Reconstruction

1. Macrostomia (cleft lip) repair 2-4 months of age
2. Ear lobe repositioning (auricular dystopia) 2-4 months of age
3. Mandibular distraction 2-8 years of age
4. Ear reconstruction, staged 5-8 years of age
   1. Costal cartilage ear framework graft with temporoparietal fascial flap if needed 3 to 4 operations to reconstruct an ear
   2. Helical rim elevation, skin grafting
   3. Tragal reconstruction and additional detailing
5. Orthognathic surgery 4-18 years of age
   Midface osteotomy and asymmetric repositioning
   Mandibular ramal osteotomy and repositioning
   Anterior mandibular osteotomy and repositioning
6. Microvascular soft tissue augmentation 8 to 18 years of age

[Additional procedures may include middle ear reconstruction in appropriate situations.]

Staging, timing and specific surgical procedure will depend on the anatomic deformity, functional severity and psychosocial factors.