

## HEMIFACIAL MICROSOMIA

### Description

After clefts, hemifacial microsomia is the second most common craniofacial condition. Categorized under this term are: Goldenhar Syndrome (see additional information), Oculo-Auriculo-Vertebral Dysplasia and Facio-Auriculo-Vertebral Dysplasia. Each of these conditions involves underdevelopment of tissues on one side of the face along with eye, ear and vertebral anomalies.

The variability in deformity in hemifacial microsomia (also known as “first and second branchial arch syndrome” and “lateral facial dysplasia” is very wide. However, in most instances, there is mal-development of the ear and mandible.

In the most severe cases of hemifacial microsomia there is underdevelopment of: the external and middle ear, the side of the skull, thickness of cheek tissue, the upper and lower jaws, teeth and some facial nerves.

In moderate cases only some of the structures are affected and to a lesser degree and in some patients, the facial asymmetry is so mild that it can hardly be seen.

Hemifacial microsomia is a spectrum of conditions whose abnormalities are most often unilateral to the right side, but can include both sides.

Most commonly seen are facial asymmetry and external ear abnormalities ranging from underdevelopment (microtia) to complete absence of the ear (anotia).

Underdevelopment of the upper and lower jaw on the affected side is marked in hemifacial microsomia. The portion of the mandible (lower jaw) that rises toward the ear may be short or absent. There is a tendency of the chin to deviate toward the affected side. Teeth on the involved side are higher than on the opposite side of the face giving the mouth an appearance of slanting upward toward the involved side.

Facial and skull bone deformities may include: a flattened forehead or cheekbone and the eye socket (orbital) may be smaller or displaced downward.

The absence of certain cheek muscles or nerves, which supply those muscles, can result in an asymmetrical smile. Cheek tissue (fat and muscle) is often underdeveloped which makes one side of the face fuller than the other.

Ear deformities range from mild to almost a complete absence of the external ear (see additional information on *Microtia*). The ear canal is often absent or blind-ended (aural atresia).

### Prevalence/Causes

Hemifacial microsomia occurs in approximately 1:5,000-6,000 live births. There is no known definitive cause for this condition. Research is leading in the direction of a disruption in the blood supply to the first and second branchial arches in the first 6-8 weeks of pregnancy. These arches are mounds of tissue that contribute to the development of facial structures (cheek bones, upper and lower jaws and ear). Neural crest cells migrate to the developing arches and are responsible for the correct formation of these structures. Damage to, or disruption of, these cells result in the facial abnormalities of hemifacial microsomia and related syndromes. There are no known genetic or external causes yet identified.

### Treatment

The goal of surgeries is to improve facial symmetry and functioning. Due to growth and development issues, treatment can be broken down into age segments:

- *Newborn*: Vital functions – breathing, eating and sleeping must be assessed immediately. Once the airway is secure (in severe cases this requires a temporary tracheostomy), attention is paid to feeding to ensure that the infant can thrive (a temporary feeding tube may be required).
- *2-4 years*: Mildly affected children require no treatment during this time. In those cases where the lower jaw (mandible) is severely

underdeveloped, it is reconstructed using a rib bone graft or lengthened with a bone distraction device. Bone distraction requires only a short operation to insert the device with parents turning a screw at home to gradually lengthen the mandible. This is a painless process avoiding painful bone grafts and scarring.

Following lengthening of the mandible, the gap between the upper and lower teeth on that side requires an orthodontist to insert a retainer to allow the upper teeth to grow into the gap and contact the lower teeth. In this way, the mouth and teeth are leveled.

- *6-8 Years:* Reconstruction of the external ear may take place in patients who are not severely affected. Severe facial asymmetry requires reconstruction of the jaw and cheekbone contour, delaying ear reconstruction.
- *8-10 Years:* In terms of appearance, this is crucial time in the treatment program. Soft tissue and its blood supply from another part of the body (i.e. back or abdomen) are transferred to the cheek to create fullness.
- *Teenage Years:* Jaw surgery may be performed on those patients whose mild condition did not require it in early childhood. The lower jaw growth that occurs in adolescence may require those severely affected to undergo further surgery. There is both pre- and post surgical orthodontic treatment as a result of surgical movement of the jaws.

#### Associated Conditions

- Skin tags are often present and may extend from the ear to the corner of the mouth. Eyes may be uneven and lids notched.
- Cleft lip or palate is seen as well as underdevelopment/paralysis of tongue muscles and salivary glands.
- Vertebrae (backbones) display a range of abnormalities from bone underdevelopment to bone fusions. The cervical spine (neck area) can be affected.
- Partial weakness in movement on affected side of the face.

