Hemifacial Microsomia

This pamphlet was written to provide information and guidance for parents of a child with **Hemifacial Microsomia (HFM)**. It includes a description of a HFM, its possible causes (what is known), and treatment. A glossary of medical terms (in bold type) is appended. This pamphlet is intended as an aid, not as a substitute, for discussion between parents and medical specialists. Parents are encouraged to ask all their questions and to be certain they receive understandable answers from members of the craniofacial team.

Introduction

HFM is the most familiar of several terms for a well-recognized congenital facial difference involving both bone and soft tissue. In some medical textbooks it is referred to as either oculoauriculo-vertebral syndrome, craniofacial microsomia, of first and second branchial arch syndrome: it is sometimes called by an old term, "Goldenhar syndrome." HFM is the second most frequent craniofacial abnormality; the most common is cleft lip/palate. A child with HFM typically has underdevelopment of the lower jaw (**mandible**). There can also be abnormal formation of the eyes, ears, **cranial nerves**, and other facial structures. Some children have abnormalities in other parts of the body.

Clinical Features

The hallmark of HFM is asymmetry, i.e., the involved side of the face is smaller than the normal side. The most common feature is a small lower jaw (mandible) on one side. As a result, the chin is rotated to the involved side and the plane of the teeth tilts up on this short side. The ear can also be affected. Often there is an extra tag of skin in front of the ear. If the external ear is small, this is called **microtia.** The soft tissues, fat and muscles of the face, can be underdeveloped so that the affected cheek is smaller. Some children have a **dermoid cyst** of the eyebrow, nose, scalp, or around the eye. The soft palate can pull to the normal side of the face when the child speaks; this is due to underdevelopment of the palatal muscles. Rarely, however, is speech affected in HFM. The eye on the affected side can be small, (**microphthalmia**) and/or downwardly displaced (**orbital dystopia**). The forehead can be flat on the involved side. About 20 – 45% of children with HFM have a weak 7th cranial nerve causing diminished facial movement. The degree of nerve weakness correlates with the involvement of the ear and soft tissues. Weakness of the 12th cranial nerve to the tongue is sometimes seen; absent sensation of the eye (5th cranial nerve) is very rare. **Macrostomia,** a cleft in the skin and muscles at the corner of the mouth, occurs in 30 – 40% of children with HFM. Some children with HFM have cleft lip/palate (about 15 – 18%). About 15 – 20% of children have HFM on both sides of the face; this is called bilateral or craniofacial microsomia. One side is usually more affected than the other so that the face is asymmetric.

It is important for parents to understand that HFM is not a progressive condition. The craniofacial differences that are present at birth do not become worse with time. We classify the involvement of the eye, mandible, ear, nerves, and soft tissues using the OMENS system. Each letter of the acronym indicates one of the five major presentations of HFM:

O = orbital asymmetry
M = mandibualr underdevelopment
E = ear deformity
N = nerve involvement
S = soft tissue deficiency

In addition to craniofacial anomalies, HFM can be associated with abnormalities outside the head and neck area. This is called the "expanded HFM spectrum" or "OMENS Plus". The most common extra craniofacial abnormalities involve the skeletal system (40%): usually a fusion of the bones in the cervical spine (neck), abnormally shaped ribs, and/or scoliosis (curvature of the spine). Cardiac malformations occur in about 25 – 40% of children with HFM. Minor central nervous system problems are found in about 15% of the children. Other rare malformations with HFM (10% or less) involve the kidneys, genito-urinary system and/or lungs.

Etiology

Some textbooks state that there is a genetic or familial basis for HFM; however, there is very little evidence supporting this belief. Our craniofacial team has seen only one family with several members with HFM. Most studies suggest that HFM is a spontaneous occurrence and not caused by genetic mutation. It is likely that HFM is due to an environmental factor; there are two theories. One is a vascular cause. There is evidence in animal models that a drug can cause bleeding and bruising on the side of the fact in the developing embryo. The haphazard nature of the expanding **hematoma** could account for the variable extent and severity of the facial deformities. Another theory states that an environmental factor, possibly a drug, causes disruption of migration and distribution of cells that are critical for development of the facial bones and soft tissues.

Treatment

A newborn with HFM usually does not have any special problems. If there is a macrostomia, the baby may need special help with feeding. If both sides of the lower jaw are very small, the infant may have trouble breathing. It is very important that the child's hearing be carefully tested every 6 to 12 months. If your child cannot hear, he/she will not learn to speak.

The following surgical procedures may be indicated during the first year of life: Removal of skin tag(s) in front of the ears, closure of the cleft of the corner of the mouth (macrostomia), and excision of dermoid cyst(s). As the child grows, treatment is focused

of the small side of the mandible. The first radiographic evaluation of the jaws (a x-ray photograph called a "Panorex") is done when the child is 3 to 5 years old. Usually the teeth erupt normally and the child will have proper occlusion. However, failure of the affected side of the jaw to grow at the same rate as the normal side can interfere with the downward growth of the upper jaw (**maxilla).** This asymmetric growth of the upper and lower jaws causes the teeth to tilt up on the involved side. If jaw asymmetry is obvious, the short side of the lower jaw can be lengthened, rotated toward the midline, and brought forward. The surgical technique is called **distraction osteogenesis** or mandibular distraction. This involves making a cut through the bone of the short side of the mandible, attaching an external mechanical device to each side of the cut, and then gradually turning pins so that the space increases between the bones where the cut was made. This space fills in with new bone, much the same way a bony fracture heals, thus lengthening the bone. A bone graft may be necessary if the child's jaw is very small and there is not enough bone for distraction. Bone and cartilage from rib is used because, when placed on the short side of the lower jaw, it will continue to grow and replicate a mandible. These operations to lengthen the mandible are usually done after eruption of the first permanent molars (after age 6).

Correction of the ear deformity (microtia) is postponed until the child is at least six years of age. This is necessary because rib cartilage used to carve and graft a new ear is not big enough until about age 6 years. Also by age 6 years, the ear on the opposite side is fully-grown and it can be used as a model for the new ear. Generally, three operations are required to complete the construction of the new external ear. Sometimes a prosthetic ear, made of silastic, is useful. Some parents and children select this as a temporary option until the child is old enough to have an ear made from the child's rib cartilage. In some children, an operation can be done to open the ear canal to improve hearing.

Asymmetric facial movement (weakness of the 7^{th} cranial nerve) can often be helped by special training after age 6 to 7 years. Sometimes a complex operation is indicated for facial nerve palsy.

As the child grows, "baby fat" is lost and the face usually becomes thinner by the time he/she reaches adolescence. For this reason, we prefer not to consider adding fatty tissue to the small side of the face until the child reaches adolescence. For most children, this operation is unnecessary.

Orthodontic treatment is completed in the teenage years.

If the child was born with severe underdevelopment of the mandible, in the mid to late teen age years, another operation on both the upper and lower jaw may be necessary to bring the plane of the teeth into a more normal relationship. For some teenagers, a bone graft of prosthetic implant is used to correct residual lower facial asymmetry.

GLOSSARY

Cranial nerve

12 nerves that serve different functions in the head and neck region. In HFM, the most commonly involved cranial nerves are:

- Facial (nerve #7) responsible for facial movement
- Hypoglossal (nerve #12) responsible for tongue movement
- Trigeminal (nerve #5) responsible for sensation (feeling) in the face

Derrmoid cyst

A small bean-like structure consisting of skin elements, most commonly located over the eyebrow but sometimes occurring in the nose. A small dermoid cyst of the eye, called a "limbal dermoid", can occur at the rim of the iris (colored part of the eye), and a fatty variant (lipodermoid) is usually hidden by the eyelid.

Distraction osteogenesis

Incision and gradual separation, using a mechanical device, in order to lengthen a bone

Hematoma

Blood in the tissues from a leak in blood vessels

Hemifacial microsomia (HFM)

Congenital facial difference where one half of the face is smaller

Macrostomia

A cleft in the skin and muscles at the corner of the mouth

Mandible

The lower jaw

Maxilla

The upper jaw

Microphthalmia

An abnormally small eye

Microtia

Small, abnormal external ear. The smaller and more deformed the ear, the greater the likelihood that the inner ear is also abnormal and hearing is affected.

Orbital dystopia

One eye is lower than the other is

Syndrome

A group of several abnormalities in different parts of the body, related by the same cause