# Hemifacial Microsomia

## A GUIDE FOR FAMILIES

Boston Children's Hospital
Plastic and Oral Surgery

# Table of Contents



# Introduction

### Welcome to the Boston Children's Hospital

This guide is meant to provide you with important information about hemifacial microsomia (also known as Goldenhar Syndrome, craniofacial microsomia, and oculo-auriculo-vertebral syndrome), as well as related issues that children and families often face. It also includes helpful suggestions for locating the many resources available to you and your family.

In addition to providing the highest quality medical and surgical care for our patients, our program is committed to supporting families throughout the treatment process. We are always here to address your questions and concerns. Please do not hesitate to contact us at any time by calling: 617-355-6309

Thank you for entrusting us with your child's care. We hope the information on the following pages reaffirms the reasons you chose Boston Children's.

To schedule an appointment or speak to a member of our team, please call our program coordinator at 617-355-6309.

# Meet Our Team

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# Meet Our Team

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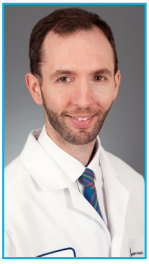

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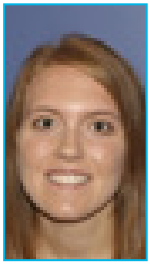


Samantha Hall
Craniofacial Clinic Coordinator
Department of Plastic & Oral Surgery

# Overview of Hemifacial Microsomia

## Overview of the condition

Hemifacial microsomia (HFM), also called craniofacial microsomia or sometimes "Goldenhar syndrome". "Hemifacial" means one side of the face. "Microsomia" means smallness. This a condition in which one side of the face is underdeveloped and does not grow normally.

The deformity in hemifacial microsomia can range from mild to severe, and the areas of the face that are affected vary greatly from child to child. Usually the first sign of the condition is a deformity in the ear causing it to be misshaped, improperly located, or smaller than the other ear. Other more visible signs of the condition are underdeveloped upper and lower jaws on one side of the face. It may appear that your child's mouth slants upward toward the affected side. Often the cheek is flattened on the affected side, and one eye may be smaller than normal. Areas of the face that may be underdeveloped in HFM include:

- Ear
- Eye
- Side of the skull movement
- Cheek tissue
- Upper and lower jaws
- Mouth
- Some of the nerves that allow facial movement

The degree to which areas of the face are affected varies widely, and some areas may not be affected at all.

The cause of hemifacial microsomia is unknown. This condition is believed to occur because of a disruption in a baby's fetal development between the first and sixth week of gestation. It is not known if environmental factors are involved, and genes that may be involved in hemifacial microsomia are not yet known. It is important to know that there is nothing you (or your child's other parent) did — or did not do — that caused this condition to occur.

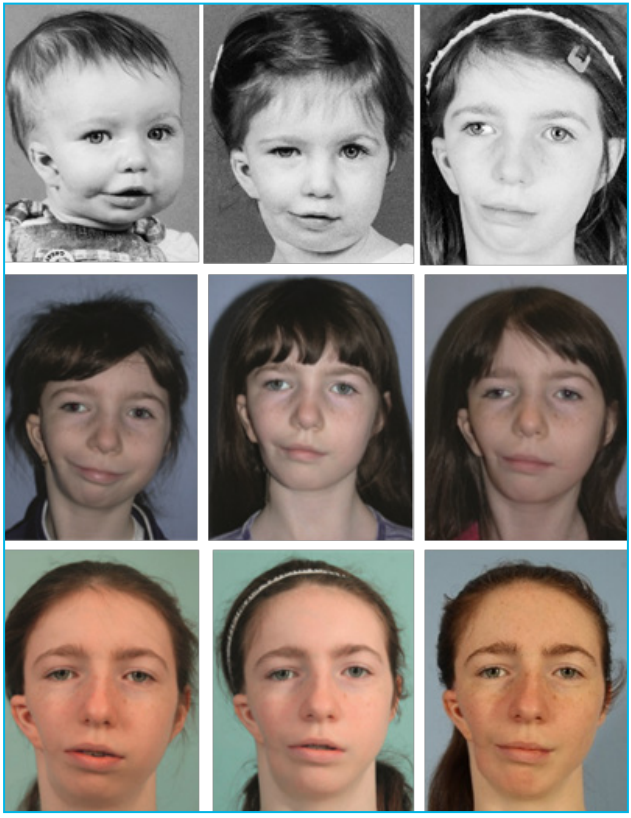
Hemifacial microsomia occurs with a frequency of 1 in 5,600 births, making it one of the most common facial differences in infants. Approximately 50% of babies born with hemifacial microsomia will have other anomalies of the spine, heart, kidneys and bladder, brain, lungs, and intestines. Genetic evaluation is recommended.

Children with hemifacial microsomia call for advanced care from a collaborative team that typically includes plastic and oral surgeons, dentists and orthodontists, and other specialists. At Boston Children's Hospital, our craniofacial team includes world-renowned specialists who are experts in treating patients with HFM.

## Diagnosis

Some babies with hemifacial microsomia will be identified in utero by prenatal testing such as fetal magnetic resonance imaging (MRI). The diagnosis is made or confirmed by clinical examination immediately after birth. An infant with bilateral or severe HFM may have difficulty breathing and feeding, though this is rare in HFM. Occasionally, babies are admitted to the Neonatal Intensive Care Unit (NICU) to be monitored for oxygen desaturations and to provide assistance with feeding.

The evaluation and management of babies with HFM requires a coordinated team approach. Additional tests, including x-rays of the spine, hearing tests, and ultrasound of the heart and kidneys, will be performed at the appropriate time to evaluate for other associated abnormalities

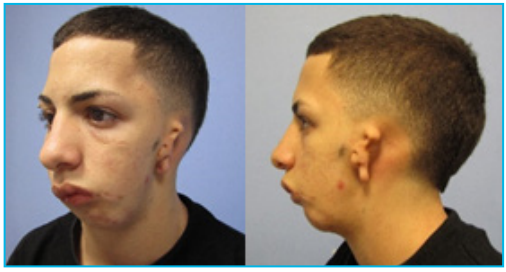


# Clinical Features of Hemifacial Microsomia

### Ear: Microtia

Definition: Underdevelopment of the outer ear structure, and sometimes involving the inner ear.

Short description: Many children with HFM have an abnormality of the outer ear structure. This ranges from a small, but otherwise normal outer ear to complete absence of the ear structure. Children with microtia typically have hearing loss in the affected ear, but the level of hearing loss can vary based on the severity of the microtia.



### Eye: Epibulbar dermoid

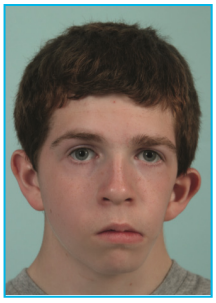
Definition: A small growth of tissue on the eye near the cornea.

Short description: This small growth is typically present at birth but may go unrecognized until later in development. This may affect vision in some cases. A surgical procedure for removal may be needed.

### Cheek: Soft tissue deficiency

Definition: Underdevelopment of the tissues under the skin that support the cheek, such as fat and muscle, causing a lack of fullness around the cheek and jaw.

Short description: This lack of filling in the cheek can cause it to appear smaller, deflated, or sunken on one side of the face.



### Breathing: Obstructive sleep apnea

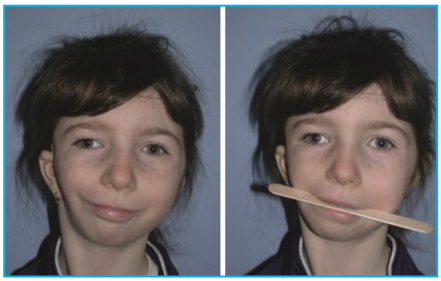
Short description:

In some patients with HFM, breathing is impacted by the abnormal jaw position. This particularly occurs during sleep and while lying on the back. This may cause snoring or more severe respiratory problems.

*Resnick CM, Kaban LB, Padwa BL. 62 - Hemifacial Microsomia: The Disorder and Its Surgical Management. In: Brennan PA, Schliephake H, Ghali GE, Cascarini L, eds. Maxillofacial Surgery (Third Edition). Churchill Livingstone; 2017:870-893. doi:10.1016/B978-0-7020-6056-4.00063-0*

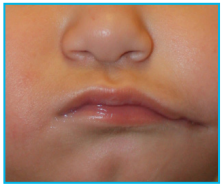
### Jaw: Mandibular asymmetry

Definition: A lower jaw (mandible) that is underdeveloped or smaller on one side of the face compared to the other.
Short description: A child with mandibular asymmetry will typically have a mouth that appears uneven or slanted upwards toward the underdeveloped side. This may affect the facial appearance and the ability to bite the teeth together effectively. The mandibular asymmetry is the most common finding in hemifacial microsomia.



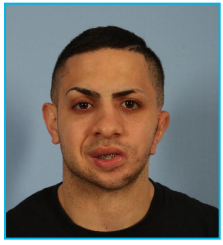
### Mouth: Macrostomia

Definition: A mouth that is abnormally wide or large for the face.
Short description: Macrostomia is caused by an abnormal formation of the muscles that attach around the corners of the lips, leading to an abnormally large appearance of the mouth.



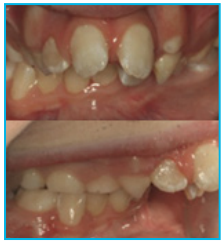
### Nerve: Cranial nerve abnormality

Definition: Major nerves of the head (cranial) can sometimes be affected, with resulting weakness of all or part of the affected side of the face.
Short description: Over 25% of patients with HFM will experience facial nerve weakness, mostly of the lower lip and sometimes of the eyebrows.

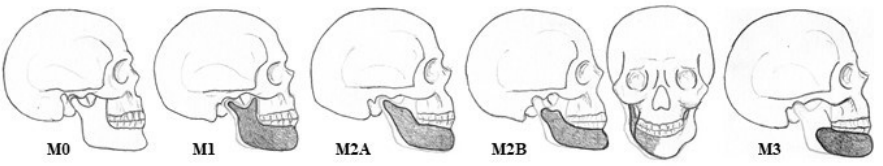


### Teeth: Overlapping and poor positioning

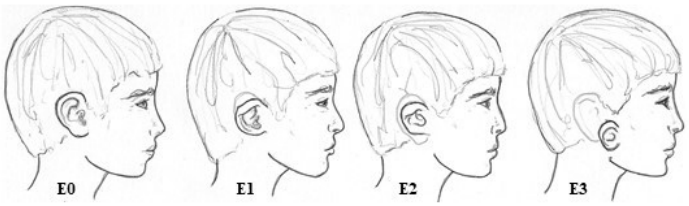
Short description: Children with HFM may have missing teeth, delayed tooth growth, or crowding of the teeth within the mouth. These problems may be worsened by the jaw asymmetry. Orthodontic treatment is used to correct and control the proper growth of the teeth.



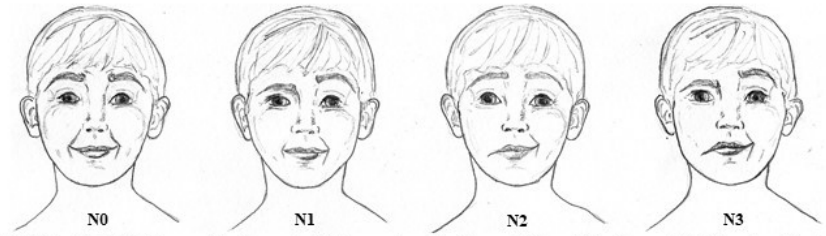

**ORBIT:** **O0** Normal orbit size and position; **O1** Abnormal orbit size; **O2↓** Inferior orbital displacement; **O2↓** Superior orbital displacement; **O3** Abnormal orbital size and position



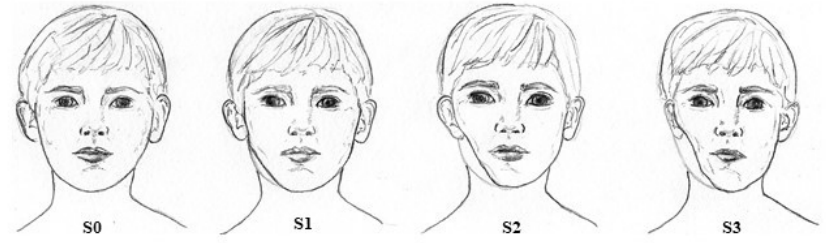

**MANDIBLE:** **M0** Normal mandible; **M1** Mandibular & temporomandibular joint components present with normal shape, smaller in size; **M2A** Joint components present with coronoid diminutive or absent; **M2B** No articulation with temporal bone, small ramus and offset; **M3** All joint components are absent with very small ramus.




**EAR:** **E0** Normal auricle; **E1** Mild hypoplasia and cupping, all structures present; **E2** Absence of external canal and concha hypoplasia; **E3** Malpositioned lobule with auricle absent, lobular typically inferior and anteriorly displaced

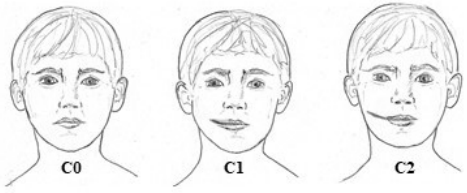



**NERVE:** **N0** No facial nerve involvement; **N1** Temporal and/or Zygomatic branch involvement; **N2** Buccal and/or Mandibular and/or Cervical branch involvement; **N3** All branches affected




**SOFT TISSUE:** **S0** No soft tissue deficiency; **S1** Minimal soft tissue deficiency; **S2** Moderate soft tissue deficiency; **S3** Severe soft tissue deficiency

**MACROSTOMIA**
**(TESSIER #7 CLEFT):**
**C0** No cleft
**C1** Cleft terminates medical to anterior border of masseter
**C2** Cleft terminates lateral to anterior border of masseter




*Redrawn from original classification system established by:*
*J Gougoutas, Alexander & J Singh, Davinder & W Low, David & P Bartlett, Scott. (2008). Hemifacial Microsomia: Clinical Features and Pictographic Representations of the OMENS Classification System. Plastic and reconstructive surgery. 120. 112e-120e. 10.1097/01.prs.0000287383.35963.5e.*

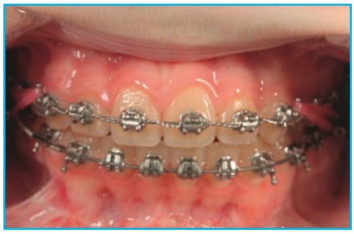
# Hemifacial Microsomia Treatment

## What treatment is available for hemifacial microsomia?

Some children with HFM will need more procedures than others. During infancy, the initial goals are to ensure your baby is breathing and feeding well. Later in childhood and adolescence, goals are to improve appearance and function of the affected areas. Our team will develop a personalized surgical treatment plan for your child. Your child's treatment team will include experts from many specialties.

### Treating Dental Abnormalities

Your child may need braces or other dental appliances to create appropriate space for and improve alignment of the teeth. Orthodontic (braces) treatment is also often necessary in preparation for reconstructive jaw surgery.

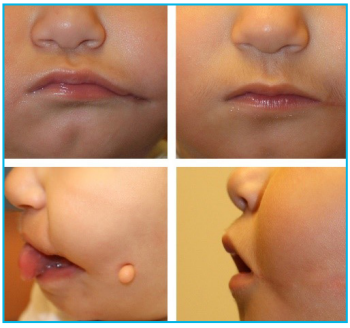


### Treating Ear Abnormalities

Ear tags are typically removed or relocated during infancy. Reconstructive surgical procedures for your child's ear, if needed, usually occur between 5 and 8 years-of age. This may involve rib cartilage or a MEDPOR® implant. Several procedures may be needed over approximately 1 to 2 years to construct a missing or severely abnormal ear. Laser hair removal is often performed before or after ear construction to correct the hairline around the ear.

### Treating Mouth Abnormalities

Macrostomia repair is typically performed between 3-6 months of age.



## Treating Breathing Abnormalities

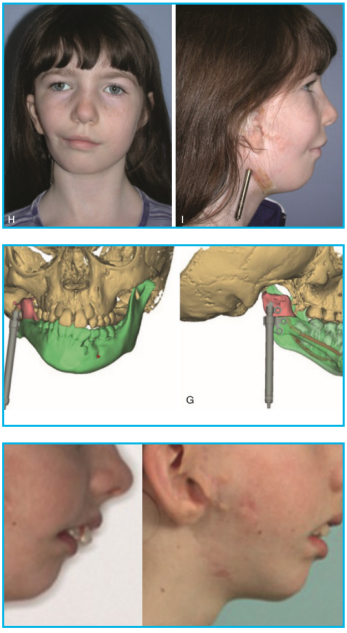
Rarely, patients with very severe or bilateral (both sides) HFM may have difficulty breathing. Sometimes a CPAP (pressurized oxygen treatment) or a tracheostomy are necessary.

## Treating Jaw Abnormalities

Most children with HFM have a significantly underdeveloped upper and/ or lower jaw. This can lead to difficulty chewing and keeping the teeth properly aligned. The treatment depends on the extent of your child's findings. Reconstructive operations may include:

- **Distraction osteogenesis:**
  This is a jaw lengthening procedure during which the surgeon will make a cut in the jawbone and insert a device that will gradually stretch and lengthen the bone over time. Depending on the extent of the deformity and your child's function, this procedure can be done during growth as the permanent teeth begin to erupt or can be delayed until growth has been completed.

- **Bone graft/flap:**
  If the jaw deformity is more extensive, surgeons may take and transfer bone and cartilage from elsewhere in the body and reconstruct the underdeveloped part of your child's jaw. Rib bone and a portion of the leg bone are common sources for bone grafts.

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After surgery, your child may still have some asymmetry (unevenness) in the appearance of the jaw. Additional surgical procedures are often needed over the course of many years to enhance appearance and function.

### Treating facial nerve paralysis

In some cases of HFM, children can have facial nerve weakness, with decreased movement in parts of their face. Your surgeon and clinical team will assess nerve function and will work with you and your child on the best approach to establish the greatest possible degree of movement. Typical treatments may include:

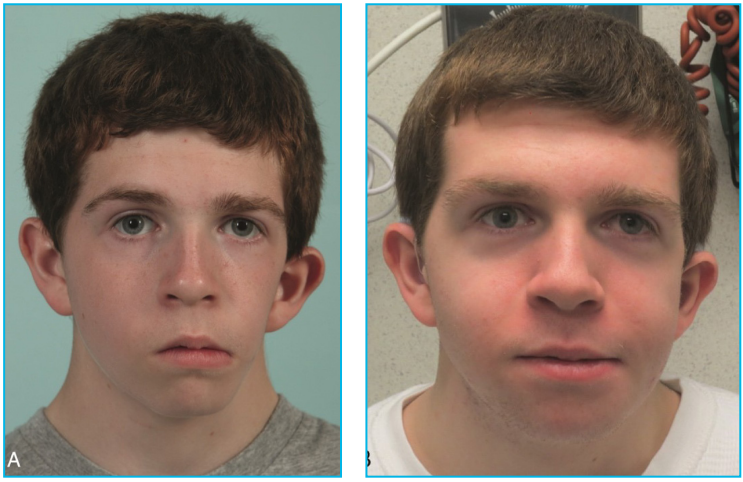
- **Botox:** Muscle imbalance can sometimes be addressed using Botox.

- **Dynamic muscle transfer:** If your child has decreased facial mobility, a muscle transfer from one body part to the face can enhance facial expressions and function.

### Treating facial soft tissue bulk

Following correction of the bony profile of the face, there may still be differences in the amount of cheek and facial fat on the underdeveloped side. This can be improved by adding fat.

- **Fat transfer:** We transfer soft tissue from elsewhere in the body to increase the fullness of your child's cheek and forehead.




(A) Pre-operative photograph, (B) Post-operative photograph
*Resnick CM, Kaban LB, Padwa BL. 62 - Hemifacial Microsomia: The Disorder and Its Surgical Management. In: Brennan PA, Schliephake H, Ghali GE, Cascarini L, eds. Maxillofacial Surgery (Third Edition). Churchill Livingstone; 2017:870-893. doi:10.1016/B978-0-7020-6056-4.00063-0*

# Frequently Asked Questions

**How common is Hemifacial microsomia?**
Hemifacial microsomia is one of the most common craniofacial abnormalities and occurs in approximately one in 5,600 infants.

**Did my baby inherit HFM?**
It is important to know that nothing you (or your partner) did or did not do caused the HFM. In most cases, HFM happens by chance and did not originate from a parent. Inheritance of HFM from a parent occurs less than 3% of the time.

**Is Hemifacial microsomia painful?**
No, children will not feel any pain or discomfort from having HFM.

**What type of medical specialists will be involved in my child's care?**

- **Craniofacial Surgeons** are plastic and oral surgeons who will conduct the majority of your child's procedures.
- **Orthodontists** help to assess and adjust facial growth and development of teeth through treatment and after.
- **Otolaryngologists** investigate and address hearing problems that may occur.
- **Ophthalmologists** assess vision quality and movement of the eyes, for potential correction.
- **Geneticists** help diagnose the genetic component of HFM and provide information about the possibility of future inheritance.

**Will my baby be able to breastfeed?**
Most babies with HFM can breastfeed. If your child has a cleft palate or macrostomia and cannot create the suction necessary to express milk directly from your breast, you may still provide breast milk to your baby if you desire by pumping and giving it by specialty bottle.

Your baby's ability to feed, whether by breast or bottle, depends on the severity of jaw asymmetry and macrostomia. Right after birth, your care team and our specially trained craniofacial nurses will help you determine the type of feeding method that is best for you and your baby.

**How can I ensure that my baby is properly nurtured, as well as nourished?**
It's essential to remember that your child is a normal baby. Facial asymmetries will improve with surgery. Having a strong family and/or social support network can help. There are a number of support groups that many families find helpful in addressing issues that may arise through the child's growth and development.

**How can I ensure my baby is gaining sufficient weight?**
Weigh your baby once a week. If he or she is not gaining more than one ounce per day, you should talk to your pediatrician about increasing the calories in the milk.

Calories can easily be added by concentrating formula or adding powdered milk to breast milk. Your pediatrician or cleft nursing team can advise you on the right approach.

If your baby has still not gained enough weight even after increasing calories, your pediatrician may recommend an appointment with a gastrointestinal or nutritional specialist.

**Are there any psychological or developmental delays associated with HFM?**
While there are no direct psychological developmental delays recorded in children with HFM, the emotional stress associated with having the condition and undergoing treatments can cause children to be psychologically distressed or uneasy during childhood. Your care team will work with you and your child to address and manage these concerns. Despite this, most individuals with HFM grow up to be fully functioning adults.

# Further Resources and Support

## Resources at Boston Children's Hospital

**Cleft and Craniofacial Center** 617-355-6309

**Contact a Nurse** 617-355-4513

**Center for Families** 617-355-6279
center.families@childrens.harvard.edu

Helps families locate the information and resources they need to better understand their child's particular condition and take part in their care. All Boston Children's patients, families and health professionals are welcome to use the Center's services at no extra cost.

## Outside Resources:

www.facesofchildren.org
https://www.changingfaces.org.uk/Home
http://www.faces-cranio.org

## Notes:

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Boston Children's Hospital Department of Plastic and Oral Surgery

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Plastic and Oral Surgery