Craniofacial Microsomia

What is craniofacial microsomia?
Children with craniofacial microsomia (CFM) have a small or underdeveloped part of the face, usually the ear and jaw. The eye, cheek and neck may also be affected. CFM can occur on one side or both sides of the face. This is the second most common facial birth defect after cleft lip and palate. Other names for CFM include:

- Hemifacial microsomia
- First and second branchial arch syndrome
- Otomandibular dysostosis
- Oculo-auriculovertebral sequence
- Facio-auriculovertebral syndrome
- Goldenhar syndrome
- Lateral facial dysplasia

What are the features of craniofacial microsomia?
Some children have only a slightly smaller jaw with a small, raised piece of skin (skin tag) in front of an otherwise normal-appearing ear. Some children can have a more noticeable difference, with one side of the face looking much smaller than the other. And some children have a very small, abnormally shaped ear. Some of the features that are often seen in CFM include:

**Ears:**
- Microtia (small ear)
- Aural atresia (no ear canal)
- Hearing loss
- Preauricular tags or facial tags (tags of skin in front of the ear or on the face)
- Other ear differences

**Face:**
- Facial palsy (difficulty with muscle movement)
- Small cheekbones
- Epibulbar dermoid (pinkish-white growth on the eye)
- Macrostomia (wide mouth, caused by a cleft along the corner of the mouth)
- Cleft lip and/or palate
Craniofacial Microsomia

Jaw:

• Trismus (limited opening of the mouth)
• Shortness of lower jaw
• Crooked lower jaw
• Malocclusion (teeth do not fit together well)

Although most children with CFM have differences in the face and do not have anything else wrong, other parts of the body can be affected.

Kidneys: There is a 10 percent to 15 percent chance that children with CFM will also have kidney problems. We suggest that your child have a kidney (renal) ultrasound to check the shape and function of the kidneys. This test is easy and takes about 20 minutes.

Cervical spine: Ten percent to 15 percent of children with CFM will have differences in the way the bones of the upper spine fit together. When these bones are well formed, at about 2 to 3 years of age, we will get X-rays to look at this. If there are abnormalities on the X-ray, we will refer your child to a bone (orthopedic) doctor.

Heart: It is rare for children with CFM to have problems with their hearts. If there are concerns about your child’s heart, the doctor may suggest that your child have an ultrasound of the heart (echocardiogram) to check for problems.

What causes CFM?

We don’t know why children are born with CFM. We know that something affects the development of the face during early pregnancy (around the fifth to sixth week). We do not think that CFM is caused by anything that the mother did or did not do during the pregnancy.

For most children, CFM is not inherited and will not be passed on from a parent to a child. Once two parents have had a child with craniofacial microsomia, the chance that they will have another child with CFM is slightly higher (about 2 percent to 3 percent). In rare cases, there are families that have an even higher chance of having another child with CFM. For children who have relatives with similar facial differences, we suggest a referral to a geneticist and/or genetic counselor. The counselor will talk about the chances of having another child with similar problems.

The craniofacial team has experience treating children with CFM, and will help you and your child through the different stages of treatment for CFM. As your child grows, different types of specialists will be involved in your child’s care. The craniofacial team will work together to develop a plan that will include medical tests and possibly surgery.
A craniofacial pediatrician and nurse will work closely with your family to help coordinate your child’s care and to ensure that all of your questions are answered. Your child’s treatment plan will be based on their needs. The following team members may also be involved in your child’s care:

- Audiologist (hearing specialist)
- Dietitian
- Genetic counselor
- Geneticist (specializes in possible genetic or inherited causes of conditions)
- Ophthalmologist (eye doctor)
- Oral and maxillofacial surgeon (surgeon specializing in the mouth and jaw)
- Orthodontist (dental professional who prevents or corrects misaligned teeth and jaws)
- Otolaryngologist/head and neck surgeon (ear, nose and throat doctor or ENT)
- Pediatric dentist
- Plastic and reconstructive surgeon
- Psychosocial professionals (psychologist, social worker)
- Speech pathologist

**How is CFM treated?**

The goals of treatment for CFM are to improve facial symmetry, to help the jaws and teeth fit together normally, and to maximize hearing. Treatment depends on your child’s age and how much your child is affected. As your child grows, the face grows, too. Timing the treatment carefully ensures the best results.

The following is a list of medical issues that your child may have. Not every child with CFM will have problems in each of these areas.

**Ears**

There are several options for treating abnormalities involving the shape of the ear. The surgeons will discuss them with you and your child. Some families choose to do nothing; others choose to have surgery. There is more information about this in the handout ‘Microtia’ (PE752) available at www.seattlechildrens.org/pdf/PE752.pdf.

Most surgeries to improve ear shape require at least three operations over a period of time. By the time your child is 6 to 8 years old, his ears are almost their adult size, so the first surgery is most often done after the age of 6 years. Your doctor can show you pictures of other children who have had ear reconstruction. The ear looks better after the surgeries, but it will not look completely normal.

Another way of treating an abnormally shaped ear is to make an artificial or prosthetic ear. This also requires several surgical procedures, but is a way to make a normal-looking ear that matches the other ear. Although prosthetic ears look normal, they need to be removed and cleaned once a day.
**Hearing**

All newborns should have their hearing checked at the birth hospital or within a few days of birth. Further testing will be done if problems are found. A hearing specialist (audiologist) will perform different types of hearing tests as your child grows.

Hearing loss related to an absent ear canal (aural atresia) may be treated with hearing aids. Sometimes, the bones of the middle ear are poorly formed or not present at all; sometimes, these bones are nearly normal.

At about 5 years of age, when growth of the ear is nearly complete, your child may need a computerized tomography (CT) scan to look at the middle and inner ear structures. CT scans are used to find out whether surgery to restore hearing is likely to be successful.

Our ear, nose and throat doctor (otolaryngologist) and hearing specialist (audiologist) will talk with you and your child about hearing issues. This includes items such as the best place for your child to sit in the school classroom, whether hearing aids are needed, and how to prevent further hearing loss.

**Eyes**

Babies with a pinkish-white growth on the eye (epibulbar dermoid) will be referred to the eye doctor (ophthalmologist). This doctor will check that the epibulbar dermoid is not interfering with your child’s ability to see. Epibulbar dermoids that are large or getting in the way of vision may be surgically removed.

**Face**

Skin tags are small pieces of extra skin that may be connected by a small stalk to the surface of the skin on the face. They are painless and do not grow or change. If your child has skin tags, talk to your child’s doctor about whether or not to have them removed.

For babies who have a cleft lip, a surgical repair is often done at 3 to 5 months of age. If your child has a cleft palate, this is repaired at about 1 year of age.

If your child has concerns about how her face looks as an adolescent, discuss this with the team. Some remolding operations on the face can be done after your child has stopped growing.

**Feeding/breathing**

Babies with CFM may have a small jaw or a cleft palate. Some babies may have trouble with breast-feeding and bottle-feeding, so we check to be sure your baby is feeding well and gaining weight. A dietician may talk with you during a clinic visit to help you provide enough calories for your baby’s growth.

If breathing is a problem, your doctors will discuss how to improve it. Some babies will need positioning devices, tubes or jaw advancement surgery to help them breathe comfortably. In some cases, a tracheostomy (a breathing tube placed in the windpipe) is needed.
Jaw and teeth

Taking care of your child’s teeth (oral hygiene) is important for all children, but especially for those with CFM. A dentist and/or orthodontist will check your child to assess their dental health, to see if there is crowding of the teeth, and to check how well the upper and lower jaws fit together. Some children with CFM have teeth missing, and we will discuss treatment for this with you. Some children may need a dental appliance or braces to provide the best dental appearance and function.

If your child has a small jawbone, your doctor may recommend an extra piece of bone (bone graft) to lengthen the jaw or make a new jaw joint. We first consider this option when your child is between the ages of 4 to 7 years. In some cases, your surgeon may recommend using a technique called distraction osteogenesis to lengthen the jawbone. This technique uses a small metal device to slowly lengthen the bone without using a bone graft.

When facial and jaw growth is nearly complete (age 13 to 16 years), most children with CFM will require orthodontics, and many will benefit from jaw repositioning (orthognathic) surgery.

Speech

Some children with CFM have differences in the muscles in the back of the mouth, and this can lead to differences in the way your child speaks (called velopharyngeal insufficiency or VPI). If you or your doctors have concerns about your child’s speech, your child will be checked by the speech pathologist.

Family and social support

Our team is sensitive to the emotional and social issues of families and patients with CFM. Social workers help patients and families access community resources and provide support and guidance to families. They can help when making decisions about surgery and can support families as they adjust to having a child with a facial difference. A social worker may also talk with your child about how his craniofacial condition is affecting his life.

Child life specialists work with children as they prepare for surgery and a hospital stay. The child life specialist uses techniques with children of all ages to reduce stress and help them learn about their condition and treatment.