



European Guideline on Craniofacial Microsomia: A Version for Patients and Families

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Abstract: A European guideline on craniofacial microsomia was developed within the European Reference Network for rare and/or complex craniofacial anomalies and ear, nose, and throat disorders and published in 2020. The guideline provides an overview of optimal care provisions for patients with craniofacial microsomia and recommendations for the improvement of care. This document seeks to provide a tailored overview of this guideline for patients and their families.

Key Words: Craniofacial microsomia, Goldenhar syndrome, guideline, hemifacial microsomia, oculo-auriculo-vertebral spectrum, patient information

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This document contains information about craniofacial microsomia (CFM) and is based on the European CFM guideline

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Figure 1 is produced by Bart Mijster, The Netherlands.

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developed by The European Reference Network for rare and/or complex craniofacial anomalies and ear, nose, and throat (ENT) disorders (ERN CRANIO).¹

The European guideline on CFM was developed by ERN CRANIO to provide healthcare professionals, patients, and their families with an overview of optimal care provisions and recommendations for the improvement of care. The aim of this document is to provide a tailored overview for patients and families.

European Reference Networks are virtual networks of healthcare providers from across Europe. The networks aim to pool together expertise on complex and rare diseases and concentrate knowledge and resources. There are 24 ERNs, each focusing on a specific disease area. European Reference Network CRANIO is a network of multidisciplinary healthcare professionals from specialized healthcare providers across Europe, focusing on rare and/or complex craniofacial anomalies and ENT disorders. The patient view is considered in ERN activities via appointed patient representatives. More information can be found on the ERN CRANIO website: <https://ern-cranio.eu/>

WHAT IS CRANIOFACIAL MICROSOMIA?

Craniofacial microsomia is a term used to describe different abnormalities that affect the development of the skull and face before birth. The word “craniofacial” derives from the words cranium (skull) and face and “microsomia” means abnormally small body structures. Craniofacial microsomia occurs in 1 in every 3000 to 5000 live births and is the second most common congenital disorder of the face after cleft lip/palate.

Craniofacial microsomia can affect the growth and development of different parts of the body, such as:

- The skull and face
- The ear, jaw, cheekbone, and eye socket
- Nerves in the face, muscles, and skin
- The spine, heart, and kidneys.

The impact of CFM on different parts of the body and its severity can differ for each patient. One patient may have minor problems with the growth and development of their ear and jaw, whereas another's CFM might have a severe impact on different parts of their face. This makes it impossible to establish 1 type of treatment for all patients. Each patient is unique and each patient needs their own type of treatment.

TERMINOLOGY

Craniofacial microsomia is a clinical diagnosis, meaning that there are no specific tests or criteria available. There are many ways to describe CFM. This can be confusing for patients, families, and healthcare professionals. The different terms for CFM include:

- Hemifacial microsomia
- Goldenhar syndrome

- Oculo-auriculo-vertebral spectrum or dysplasia
- First and second branchial arch syndrome
- Facio-auriculo-vertebral syndrome or sequence.

Scientific literature has shown that for example, Goldenhar syndrome is part of the CFM “spectrum.” Therefore, it is advised to use the term “craniofacial microsomia” instead.

WHICH HEALTH PROBLEMS ARE RELATED TO CRANIOFACIAL MICROSOMIA?

A patient with CFM might experience a range of health difficulties. These difficulties vary a lot between patients and depend on the type and severity of the CFM.

Regardless of the clinical care required, most patients with CFM retain normal intelligence and can go to mainstream school. Unfortunately, these children may be regarded by others as being different. They may experience bullying, especially in childhood and may lack self-esteem. All patients and families should, therefore, have access to clinical psychology services with appropriate professional expertise and knowledge of CFM. Parents or caregivers may also experience difficulties. Raising a child with a craniofacial anomaly may be challenging and lead to parental stress as a result of various factors, such as having to attend multiple medical appointments and witnessing their child being teased and/or experiencing psychosocial difficulties.

Parents of newly diagnosed children with CFM should also have access to a specialist clinical psychology service with expertise and knowledge of CFM. Contact with patient support groups could help parents and families build a support network with others who have had similar experiences to them.

Most countries in Europe have their own patient support groups. These groups are often led by patients and families themselves. The treating physician should provide patients and families with more information on existing groups.

The following list outlines the difficulties that patients with CFM may encounter and lists some of the recommendations made in ERN CRANIO’s European CFM guideline.

- Breathing difficulties. Patients with CFM are more likely to suffer from breathing problems whilst sleeping, particularly obstructive sleep apnea (OSA). This is characterized by snoring and/or increased effort to breathe. About 18% of CFM patients have OSA. The severity of OSA in CFM patients varies. Patients with CFM on both sides of the face or severe underdevelopment of the lower jaw on 1 side are at increased risk for OSA. These patients should be screened for OSA with a sleep study in the first year of life. All other patients with CFM should be screened for OSA with a questionnaire once every 2 years at least up to 6 years of age.
- Feeding difficulties. Patients with CFM may suffer from an underdeveloped jaw, or weak nerves in their face or chewing muscles. In approximately 26% of the patients, this leads to chewing, swallowing, sucking, or growth problems. The pediatrician should screen and monitor all patients regularly for feeding problems with a questionnaire once every 2 years at least up to 6 years of age.
- Eye problems. Patients with CFM regularly present with epibulbar dermoids (a swelling on the eye) or other eye anomalies, such as a defect in the eyelid or iris. These anomalies can cause various difficulties such as a lazy eye, irritation, and problems with eyelid closure and they may have an aesthetic impact. All patients should be seen by an orthoptist and ophthalmologist before the age of 5 years to check for visual acuity. If eye anomalies are present, the

patient should be examined by a specialized orthoptist and ophthalmologist before the age of 5 years. If surgical treatment is considered this should be discussed in a multidisciplinary team and with the parents, carefully evaluating the harms and the benefits, especially if the child’s vision is still developing.

- Hearing difficulties. Sufficient hearing is needed for children to learn language properly. However, many CFM patients have hearing problems caused by damage to their inner or outer ear. In fact, these hearing problems can occur on both sides of the head even if CFM affects just 1 side. All patients with an underdeveloped outer ear and/or cleft palate should be regularly seen by an ENT doctor to assess for hearing difficulties. If ear reconstruction or surgical placement of a hearing aid is considered, this should always be discussed in the multidisciplinary team, because this could interfere with reconstruction of the external ear.
- Spinal problems. Approximately a third of patients with CFM have spinal problems, such as a wedge-shaped vertebrae (called hemivertebrae), fusion of 2 or more vertebrae (called block vertebrae) or a curved spine (called scoliosis). Patients with spinal problems are also at higher risk for other problems with their heart or kidneys. All CFM patients should, therefore, be checked for neck and back problems. Patients should also be examined by a child nerve specialist (pediatric neurologist) in cases where there is numbness or weakness in the back, or neck pain.
- Speech and language difficulties. Patients with CFM may have speech, language, and communication needs due to an underdeveloped jaw, teeth that are not aligned properly, or weak muscles in their face. Patients might also have an opening in their upper lip or the roof of their mouth, which is called a cleft lip/palate. Also, patients with CFM are at risk of hearing problems. All of these issues can make it hard to speak and learn language properly. Therefore, all babies and children with CFM should be screened for language skills by a speech and language therapist once every 2 years at least up to 8 years of age.
- Dental problems. Some CFM patients are more likely to have missing teeth or other problems with their teeth. All CFM patients should therefore see a dentist regularly. Also, all patients with CFM should be seen by an orthodontist from the multidisciplinary team, regularly from 5 years of age. Orthodontic treatment should be discussed and coordinated by the multidisciplinary team and not decided solely by a private orthodontist. It is encouraged for hospitals to have associated dental clinics/orthodontists for routine care to reduce travel time for patients and families.

WHAT ARE POSSIBLE SURGICAL TREATMENTS THAT PATIENTS WITH CRANIOFACIAL MICROSOMIA MIGHT EXPERIENCE?

Craniofacial microsomia patients often have 1 or more surgeries before becoming an adult. There is no “one-size-fits-all” (surgical) treatment approach for patients with CFM. Below, the various options are explained. Most surgical treatments concern the face, although some patients might need treatment for problems of the body such as the spine, hands, feet, heart, or kidneys. It is important to note that most of the listed treatments will never be necessary. However, as it is possible that 1 or more of these surgical treatments are necessary, the full list has been provided.

Involvement of a psychologist before and after facial surgery is advised to support the patient and their family with acceptance,

expectation management, and the psychological impact of the surgery itself.

Jaw Surgeries

Patients with CFM often have small or underdeveloped jaws that require surgery. The requirement for and timing of treatment is based on clinical assessment by the multidisciplinary team. If surgery of the lower or upper jaw is performed for aesthetic reasons only, it is advised to postpone this until adulthood. The jaw asymmetry might re-occur after the initial treatment if this is performed during childhood. This should be discussed within the multidisciplinary team of doctors and with patients and caregivers. The various types of jaw surgeries that might be offered include:

- Mandibular distraction osteogenesis is a surgical procedure that lengthens the lower jaw, thereby pulling the tongue forward. This opens the airway and improves breathing, eating, and sleeping. Mandibular distraction is effective for patients with breathing difficulties and may prevent the need for placement of a breathing tube (tracheostomy) or lead to decannulation. The outcomes of mandibular distraction on facial asymmetry are unpredictable. Facial asymmetry often reoccurs when mandibular distraction is performed during childhood. This could lead to (unnecessary) secondary surgery to restore facial symmetry.
- Orthognathic surgery is performed to improve the functioning of the teeth and jaw and improve facial symmetry. Improved functioning will mean the patient is better able to chew.
- Mandibular reconstruction might be needed to treat patients with a severe underdevelopment of the lower jaw, which makes distraction impossible. In some patients the mandibular joint may be absent. Reconstruction of the lower jaw and joint could be performed by using bone from another part of the body, such as a rib. If this is needed, it should be performed when children are older than 6 years old.
- Temporomandibular joint implants are custom-made implants that take the place of the missing mandibular joint (see above). These implants might be needed to treat severely underdeveloped lower jaws.

Facial Nerves Surgery

Some CFM patients have undeveloped nerves on 1 or both sides of their face, called facial palsy. This may cause problems with eye closure, articulation of speech, drooling, or asymmetric facial mimics and smiles. Although patients with CFM commonly experience facial differences besides facial palsy, this problem with nerve functioning can have a big impact on both social and psychological functioning. Some recommendations for treatment have been made:

- Eye: It is important to protect the eye if full eyelid closure cannot be achieved. All patients with eyelid closure problems should be referred to an eye doctor (ophthalmologist). Surgical treatment may be needed to achieve eyelid closure. This could be done with placement of a gold weight or platinum chain in the upper eyelid, with transfer of muscle, or by using healthy nerves from the other side of the face (called cross-facial nerve grafting).
- Mouth: Patients with an asymmetric depression of the lower lip may be treated by placing botulinum toxin in the healthy muscle that causes lip depression. The effect will last for several months. Surgical removal of the muscle has a permanent effect and should only be performed if outcomes of treatment with botulinum toxin are satisfactory. If

outcomes of treatment with botulinum toxin are not satisfactory, transfer of muscle to the affected side can be used to restore lip depression.

- Aesthetics: Surgery might be needed to restore facial movement and treat an asymmetric smile. The timing of treatment should be coordinated with other surgeries. The goal of this treatment is to achieve spontaneous facial movements and a spontaneous smile. In this type of surgery, the healthy facial nerve is used and crossed to the affected side of the face. A muscle (often the gracilis muscle) is connected to this nerve, which makes movement possible. In patients without a healthy facial nerve (for instance bilaterally affected patients) a different nerve could be used. A facial physical therapist should be part of the multidisciplinary team and help the patient achieve optimal facial movement.

Soft Tissue Surgery

Most CFM patients have mild to severe loss of soft tissue in their face. Usually, this means a lack of fat under the skin or weak muscles, which can make the face look uneven. If a patient desires treatment, this might be performed to restore the shape of the face and create symmetry. Various types of treatment can be used to treat the soft tissue deficiency:

- Fat grafting (fat transfer) is used to transfer fat from a different part of the body to the face to increase the facial symmetry. This type of treatment can start from childhood on. In case of a severe shortage of soft tissue, several surgical procedures might be required to achieve an optimal result.
- Pedicled flaps might be proposed to correct the facial soft tissue deficiency. A pedicled flap is a piece of soft tissue (skin, fat, and sometimes muscle), which is left attached to the original site by a narrow base of tissue and is transposed to a new location. There is minimal scientific evidence regarding the use of pedicled flaps in CFM patients, and therefore, this is strongly discouraged.
- Free flaps are detached from blood supply at the original location and then transferred to another location and connected to the blood supply at that site. Results of free tissue transfers to treat soft tissue deficiency in CFM patients are mostly considered to be satisfactory or positive in scientific literature. However, this treatment can have a significant impact on the patient. Complications occur in 27% of patients and include hematoma, flap loss, infections, or sagging. Additional surgery to make the muscle less bulky are needed in 20% of the cases. The long-term effects of reconstruction with free flaps are not well known. Therefore, a free tissue transfer should only be performed in patients with a very severe soft tissue deficiency.
- Implants such as silicone or polyethylene can be a good method to treat the soft tissue deficiency in patients with CFM, especially for the angle of the lower jaw. The long-term effects of implants for soft tissue correction are unknown. Therefore, implants for soft tissue restoration should not be used in the growing patient and placement should be postponed until the patient is at least 18 years old.

Microtia Surgery

Underdevelopment of the outer ear (called microtia) is 1 of the most common characteristics of patients with CFM. Ear reconstruction is only performed if the patient and family feel that it is required. As hearing difficulties are common in patients with an underdeveloped ear, the timing of treatment for both hearing and ear reconstruction should be coordinated. Patients should always be

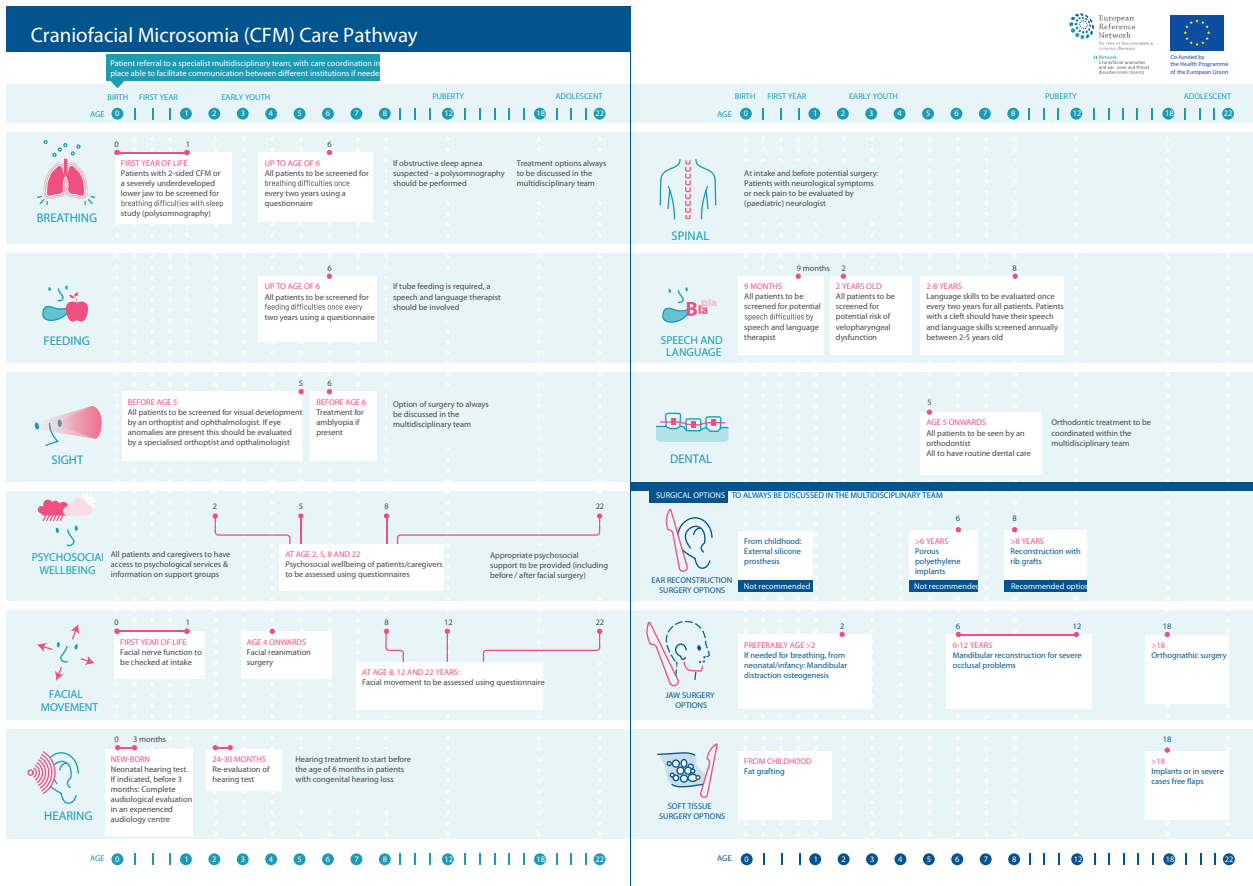


FIGURE 1. Craniofacial microsomia (CFM) care pathway. An infographic.

treated within a multidisciplinary team setting. The choice for 1 of the treatment options for ear reconstruction is based on the potential outcomes, benefits, and harms of treatment. Patients and parents should be able to make an informed choice and weigh up the advantages and disadvantages of treatment options. The default option for microtia is “no reconstruction,” however, if the patient and family would like to proceed then there are several options for treatment:

- Silicone prosthesis. An artificial ear can be attached with either glue or an implant. Treatment with silicone prosthesis attached with glue may be performed in children. However, such treatment for a young child is discouraged. Treatment of the ear deformity in young children is often initiated to comfort the parents, and this rationale is debatable. Worries about their child being bullied are taken seriously and support should be provided, but it is believed that surgery will not be the solution. Although treatment before the age of 8 is not recommended, if it is chosen it is recommended to use a silicone prosthesis attached with glue. A silicone prosthesis attached on implants is only considered appropriate in patients without other options and should only be placed in adults.
- Porous polyethylene implants are also called Medpore implants. This type of reconstruction is quicker and has a lower burden for the patients compared to ear reconstruction

with their own tissue. This type of reconstruction poses risks of infection and scarring or fractures of the implants as a result of contact sport. If the reconstruction with Medpore fails, other types of ear reconstruction are challenging. Therefore, it is advised to approach this treatment with caution. If reconstruction with Medpore is the treatment of choice, it is advised to postpone treatment until the child is aged 6 since at this age the width or the normal ear is approaching its mature size.

- Reconstruction with rib grafts is considered the optimal choice of treatment because it is the most durable treatment option with good long-term outcomes. This is especially relevant in patients with CFM as a life-long treatment solution is needed. The timing of treatment should be discussed within the multidisciplinary team and should be performed from the age of 8 onwards.

An infographic (Fig. 1) is included to accompany the text in this document.

More information on CFM and experienced craniofacial centres is available on the ERN CRANIO website: www.ern-cranio.eu.

REFERENCE

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