

OROFACIAL CLEFTING IN CHARGE SYNDROME: FOR THE PHYSICIAN

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CRANIOFACIAL ANOMALIES SEEN IN CHARGE

Orofacial clefting occurs in about 20% of children with CHARGE syndrome. These children may have cleft lip with or without cleft palate or isolated cleft palate, especially sub-mucous cleft palate.

DIAGNOSTIC CONSIDERATIONS:

In general, patients with CHARGE are more likely to have cleft lip, while those with velo-cardio-facial syndrome (VCFS) are more likely to have a cleft palate. When orofacial clefting is present in patients with CHARGE, the choanae are usually patent, so this finding (clefting) may substitute for choanal atresia in the diagnostic criteria, particularly if the remaining findings are otherwise characteristic of this condition.

Other anomalies common in CHARGE which may affect treatment and/or management of orofacial clefts:

- TE fistula or esophageal atresia
- Cranial nerve IX/X palsies
 - Laryngotracheomalacia
 - Velopharyngeal incompetency
 - Reflux
- Facial palsy, unilateral or bilateral
- Heart defects
- Ossicular malformations with or without Mondini anomaly
- Choanal atresia or stenosis

DIAGNOSTIC TESTS:

While cleft lip is obvious to the casual observer, the diagnosis of submucous cleft palate in CHARGE can sometimes be delayed. Many cases have been confirmed only when tonsils or adenoids are removed. **An evaluation looking for cranial nerve IX/X involvement is crucial prior to beginning a feeding program in order to avoid multiple aspiration pneumonias.**

CONSEQUENCES OF OROFACIAL CLEFTING IN CHARGE:

FEEDING:

Orofacial clefting can interfere with feeding in any individual. Children with CHARGE often have additional feeding problems due to neurologically-based velopharyngeal incompetency and/or reflux. Cleft lip or cleft palate can exacerbate this situation. Children with CHARGE (with or without orofacial clefting) often require g-tube feeding for significantly longer than children with other clefting syndromes.

EAR INFECTIONS/HEARING LOSS:

Children with facial clefts are prone to ear infections and possible hearing loss. Children with CHARGE typically have more ear infections and require PE tubes more often and for a longer period of time than other children with clefts. Children with CHARGE often have sensorineural hearing loss and/or conductive hearing loss due to malformed ossicles. Close follow-up by ENT and audiology is critical to maximizing the hearing in these children.

SPEECH:

An undiagnosed submucous cleft palate can interfere with speech development, already complicated in these children due to hearing loss and facial palsy.

MEDICAL MANAGEMENT AND CAVEATS

Team management approach: It is the recommendation of the American Society of Maxillofacial Surgery as well as the American Society of Plastic and Reconstructive Surgeons that management of the patient with facial clefting be provided by an interdisciplinary team of specialists offering a coordinated and consistent philosophy and a continuum of care. The cleft team may include a plastic surgeon, oral surgeon, orthodontist, otolaryngologist, ophthalmologist, social worker, nurse, audiologist, speech pathologist, and geneticist. When following a child with CHARGE, it is important to communicate with other specialists who are following the child. The feeding team members should understand the neurologic basis for incoordinated swallow and reflux. The members of the team and the approach to the child will vary from one institution to another. The specific management plan for orofacial clefting in a child with CHARGE will vary depending on the specific problems of that child as well as the protocols for a particular institution.

Early simple surgical repair maximizes optimal speech, aids in feeding, assists in reducing middle ear infections, and re-establishes normal separation of the oral and nasal cavity while minimizing growth disturbances of the upper jaw.

Post-surgical complications such as infection, dehiscence, oro-nasal fistula, and malocclusion may be more frequent in children with CHARGE. Therefore, a 23-hour stay planned for children with isolated clefts may need to be extended to an inpatient stay for children with CHARGE.

Consider insertion of PE tubes and the taking of dental impressions at the time of surgery if indicated.

Bilateral cleft lip and palate is rare in CHARGE syndrome. Although late treatable complications such as dental malocclusion, velopharyngeal incompetence, oro-nasal fistulae, and distortion of normal anatomy can often occur, initial planning during the first twelve months of life mirrors that of unilateral cleft lip and palate.

SPECIAL CONSIDERATIONS IN TREATMENT OF FACIAL CLEFTS IN CHARGE:

- 1) Heart defects (present in 2/3 of children with CHARGE) may be complex. These may affect timing of surgery and overall stability of the patient.
- 2) Possible unexpected reaction to anesthesia. Some children with CHARGE are resistant to sedation, while others are slow to recover from anesthesia. Because of risks of anesthesia, it may be appropriate to combine surgical procedures in these children.
- 3) Laryngomalacia or tracheomalacia is common in CHARGE and can result in surgical or anesthetic complications.
- 4) Cranial nerve palsies complicate post-operative feeding in a major way. Most importantly, apparent abnormalities of cranial nerves IX and X cause secretions and food to pool in the pharynx. Esophageal peristalsis is uncoordinated. Aspiration and reflux are common. This improves in weeks, months or years, with a few children having permanent problems. In addition, facial palsy is usually unilateral and may affect lip closure and may compromise ultimate cosmetic outcome of cleft lip.

NON-MEDICAL MANAGEMENT

If a patient has CHARGE, orofacial clefting is likely to be only one of a number of serious medical problems for that child. The cleft may or may not be a high-level concern to the parents. It is critical to talk to the parents about the whole child, and not just about the cleft. Parents are often especially concerned with how the cleft correction fits into other surgical schedules.

The orofacial team, which includes ENT, audiology, and feeding specialists, can be a great help to parents (even if the child does not have a cleft) because they have experience with feeding difficulties, PE tubes, and ear infections, all of which are common in CHARGE. Communication with other specialists following the child is critical to effective care of these children. These children often have multiple medical appointments every month or even every week, so coordination of appointments is appreciated by the parents.

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CLEFT LIP AND PALATE (CL/P) IN CHARGE: PARENT INFORMATION

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NORMAL STRUCTURE AND FUNCTION:

You know what the lips are! The **palate** is the roof of the mouth: the bony and muscular structure between the oral (mouth) and nasal (nose) cavities.

EMBRYOLOGY:

The lips and palate develop between 5 and 7 weeks of gestation (pregnancy). They result from the growth, merging, and fusion of five "processes:" two maxillary (upper jaw) processes, two mandibular (lower jaw) processes, and one frontonasal (nose) process (see diagram). The lower lip is formed when the mandibular processes unite. The upper lip is formed from the two maxillary processes and the frontonasal process. They come together (two from the side, and one from above) to form the upper lip. The "cupids bow" in the center of the upper lip shows the borders where the processes meet.

The primary (hard) palate, the nasal cavity and the choanae (passages from the back of the nose to the throat) result from merging of the medial nasal processes at 5 weeks. At 6 weeks, the secondary (soft) palate processes develop and fuse. This creates the soft palate, the nasal cavity and the choanae. The lips, palate and nasal cavity should be complete by about 7 weeks gestation. If any of these processes fail to fuse or merge, a gap, or cleft, results.

FREQUENCY OF OROFACIAL CLEFTING IN CHARGE:

Orofacial clefting (cleft lip or cleft palate) occurs in about 20% of children with CHARGE syndrome. Submucous cleft palate is often not diagnosed in the newborn period, so the frequency of clefting may be even higher. It is possible to have both choanal atresia and cleft palate.

Diagnosis of Clefts in CHARGE

Cleft lip is obvious at birth. The newborn exam performed in the delivery room usually includes an evaluation of the palate. A cleft of the hard palate should be apparent as part of this exam. A submucous cleft is more difficult to diagnose and is sometimes not recognized until much later.

CLEFT LIP (with or without cleft palate, CL/P):

Cleft lip can be **unilateral** (UL, one-sided) or **bilateral** (BL, two-sided). Cleft lip typically occurs between the side and center (cupids bow) portions of the upper lip. It may involve only the lip, or extend into the gum and even into the primary (hard) palate.

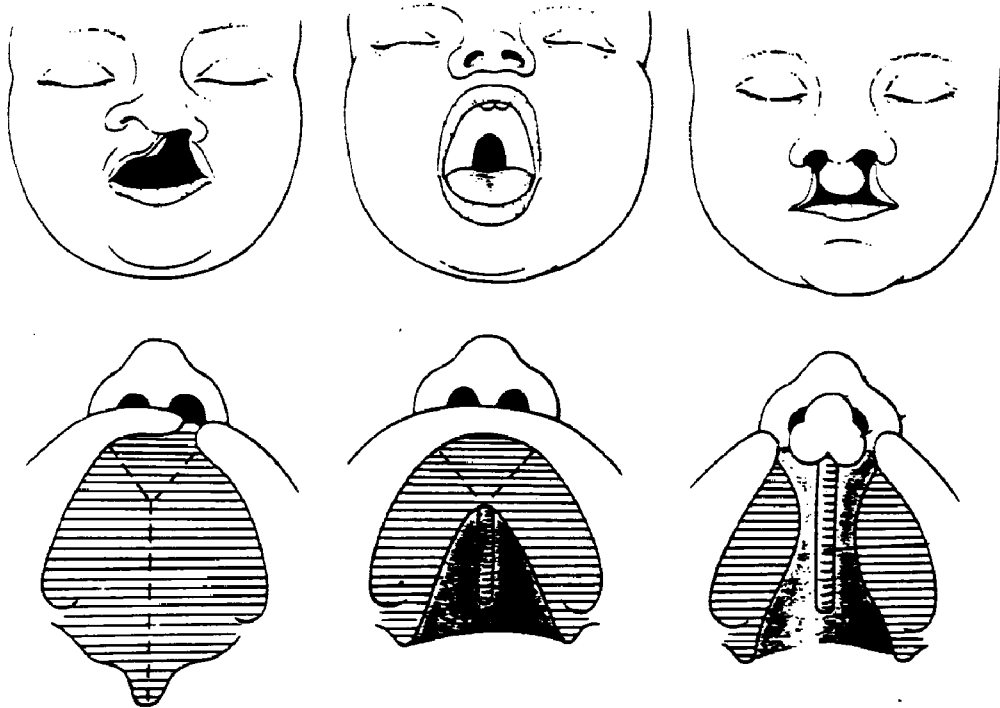
CLEFT PALATE (CP):

Cleft palate results from a failure of fusion of the palatal shelves. Isolated cleft palate (without cleft lip) is in the back of the palate. It can involve the hard and soft palate or just the soft palate. Children with cleft palate typically have underdevelopment of the mid-face (flat midface) and often a small chin.

Submucous cleft palate: Submucous cleft palate involves the muscles (but not the bones) of the soft palate. Swallowing and speech are often affected.

Bifid uvula:

Bifid (split or double) uvula (little thingee that hangs down in the back of the throat) is considered a mild form of cleft palate. This alone does not usually create problems. It may be a sign to look more closely for a possible submucous cleft palate.



EFFECT OF CLEFTING IN THE CHILD:

Although cleft lip is a cosmetic problem, the more immediate concern is the effect on feeding. Special nipples may help the child suck from a bottle. Nursing is sometimes difficult or even impossible. Cleft palate makes feeding even more difficult, as the milk can pass through the palate into the nose. Children with CHARGE often have additional feeding problems due to facial palsy, reflux, and/or swallowing problems (see FEEDING section). **Nurses and other specialists in a cleft palate clinic often have extensive experience with feeding difficulties and can be a big help to parents.**

Swallowing and speech are affected by cleft palate, sometimes even after surgical correction. Children with a cleft palate are more prone to ear infections and aspiration. Children with CHARGE (with or without a cleft) typically have multiple ear infections and require multiple PE tubes (see EARS section). Children with cleft palate are prone to conductive hearing loss (see HEARING section).

Most children with CHARGE and a cleft (and many without clefts) are unable to take food by mouth for some time and require a gastrostomy tube (g-tube) for some time (see FEEDING section).

Children with cleft lip/palate often have orthodontic problems which will need to be addressed. Children with CHARGE may have particular dental problems as well, but these have not yet been well-described.

MEDICAL MANAGEMENT

TEAM APPROACH:

It is the recommendation of the American Society of Maxillofacial Surgery as well as the American Society of Plastic and Reconstructive Surgeons that management of the patient with facial clefting be provided by an interdisciplinary team of specialists offering a coordinated and consistent philosophy and a continuum of care. Members of the Craniofacial team may include a plastic surgeon, oral surgeon, orthodontist, otolaryngologist, ophthalmologist, social worker, nurse, audiologist, speech pathologist, and geneticist. Every institution will have a slightly different team and perhaps a different approach to facial clefts. Make use of these team members! Many of them will have had experience with feeding problems in other children.

TREATMENT/SURGERY:

The treatment for facial clefting involves a series of surgical procedures which may take place over many years, even through the patient's young adult life. The actual timing and type of treatment takes into consideration the child's growth, development, and other medical problems. Prior to surgery, it is often necessary to take dental impressions. Some children will need a pre-surgical orthodontic appliance and home therapy to prepare for surgery.

Surgery is not usually scheduled immediately for cleft lip and palate. Children with CHARGE often have more immediate surgical needs, such as choanal atresia or a heart defect. Many palatal clefts will be repaired between eight and twelve months of age in a single stage, although surgery on very wide clefts may be delayed until up to eighteen months of age.

OTHER TESTS/PROCEDURES:

Audiological (hearing) testing should be done early. Children with CHARGE have an even higher risk of hearing loss than do other children with clefts. Insertion of PE tubes and the taking of dental impressions may be done during the same anesthesia as cleft surgery.

Non-medical management

Most of the non-medical concerns related to facial clefts are feeding concerns. See the FEEDING section for more information.