CME

An overview of cleft lip and palate

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ABSTRACT

Cleft lip and palate are types of craniofacial birth defects that affect thousands of children worldwide each year. These conditions are sensitive topics of conversations, often affected by the stigma of physical birth deformities and cultural myths. This article reviews the pathophysiology of cleft lip and palate, and describes the traditional management of patients with oral-facial clefts, including the extensive supportive care and an interprofessional team or *cleft team* approach that extends beyond the surgical correction.

Keywords: cleft lip, cleft palate, oral-facial cleft, birth defect, cleft repair, lip repair

Learning objectives

- Describe the pathophysiology, causes, and clinical presentation of cleft lip and palate.
- Identify possible risk factors for influencing the development of orofacial clefts.
- Discuss the perioperative, surgical, and postsurgical management of patients with orofacial clefts.

FIGURE 1. Child with a bilateral complete cleft lip and palate

Cleft lip and palate are types of craniofacial birth defects that affect about 7,000 babies born in the United States each year (**Figure 1**).[1] The CDC estimates that each year in the United States, 2,650 babies are born with a cleft palate only (CPO), and 4,440 babies are born with a cleft lip.[1] Worldwide, 1 in 700 babies is born with a cleft lip, CPO, or cleft lip and palate each year.[2,3]

Researchers find that a bilateral cleft lip is more common in males (male-to-female ratio 2:1) and CPO is more common in females.[4,5] Unilateral cleft lip is reported to be more predominant on the left side (2:1 ratio).[5] According to the World Health Organization (WHO), the diagnosis rates of cleft lip and palate vary geographically and among ethnic groups.[2,5,6] Higher prevalence of cleft defects is well documented in Asian populations and Latinos, followed by certain groups of Native Americans, with the lowest rate reported in Blacks.[2] Most cases of cleft lip and palate are diagnosed in utero by ultrasound after 20 weeks' gestation, but some are undetected and only discovered at delivery. Although rare, a small percentage of clefts are diagnosed during childhood.[2,7]

EMBRYOLOGY

In cleft lip, the lip does not completely fuse during fetal development. Cleft lip and palate, similar to neural tube defects, are birth defects that develop during pregnancy. The same gene mutation is involved in both.[8] The degree of the deformity can vary from mild to severe (**Figure 2**). CPO occurs when the roof of the mouth does not completely fuse during fetal development; this opening in the palate can extend into the nasal cavity. Because embryologic development occurs in sequential stages, cleft lip and CPO may occur independently.

RISK FACTORS

Extensive, ongoing research is dedicated to cleft lip and palate embryologic development; however, no consensus

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DOI:10.1097/01.JAA.0000721644.06681.06

Key points

- Cleft lips and cleft palates are common craniofacial birth defects worldwide.
- These orofacial clefts can range from a small notch to large splits extending into the nose.
- Multiple factors have been linked to orofacial clefts, including maternal factors, environmental factors, and genetics.
- Surgical repair can transform children's lives.
- An interprofessional or cleft team provides supportive care to the patient for years after surgery.

exists as to their cause. Researchers generally agree that genetic, nutritional, and environmental factors contribute to their formation.

Risk factors for development of an orofacial cleft include:

- Genetics
- Environmental agents such as secondhand exposure to smoke
- Medications such as phenytoin, sodium valproate, topiramate, and methotrexate
- Smoking and using tobacco products
- Alcohol
- Folate deficiency
- Maternal obesity
- Diabetes diagnosed before pregnancy is a more significant risk factor than gestational diabetes.[1,2,5]

The risk of having another offspring with a cleft increases if the parents or children have a cleft.[2] In most cases, there is no family history of cleft lip or palate, nor is there a history of complicated pregnancy.

CLINICAL CHARACTERISTICS

Children with cleft lip or palate require long-term complex and coordinated care. In 2017, the American Academy of Pediatrics (AAP) published a report dedicated to educating pediatricians on the care of patients with cleft lip and palate.[2]

Examine the oral palate, looking for a translucent appearance, an elevation of the palate during phonation, and possibly a bifid uvula. The child's speech also should be assessed for articulation and phonology especially hypernasality.

The CDC estimates that 30% of children born with cleft lip or cleft lip and palate and 50% born with CPO are associated with a syndrome.

Orofacial clefts are categorized by their location, degree of lip involvement, and the presence or absence of a cleft palate.

Cleft lips are described as unilateral, bilateral, or midline, and as complete or incomplete depending on the extent of involvement of the nose. The most common type of cleft is a unilateral cleft with the characteristic defect affecting one side of the lip, with or without a cleft palate.[2]

Submucous cleft palate, a more minor cleft palate, is caused by a malposition of the levator muscles. The diagnosis of submucous cleft palate is more difficult to make on examination, and can be missed and not discovered until a child is old enough to speak.

Clefts can be divided into two categories: syndromic and nonsyndromic.[4] Unlike cleft lip or cleft lip and palate, CPO is more likely to be associated with a syndrome or genetic defect.[2] However, most cases of cleft lip and palate do not occur with other birth defects. Common syndromes associated with cleft palate include Van der Woude syndrome, Stickler syndrome, and velocardiofacial syndrome.[4,9] Stickler and Van der Woude syndromes are autosomal dominant disorders. Children affected with Stickler syndrome often have a cleft palate, a small jaw, and collagenopathy. Children with Van der Woude syndrome often present with lower lip pits, cleft lip, and genitourinary anomalies. Van der Woude is the syndrome most commonly linked to cleft lip.[5,10] Velocardiofacial syndrome is a genetic condition, most commonly associated with cleft palate. Though symptoms may vary from child to child, many also suffer from congenital heart disease, speech problems, and immune problems.[11]

Infants born with CPO can have trouble with feedings, and are at increased risk for developing speech difficulties in early childhood.[2] They also may have recurrent otitis

FIGURE 2. Types of cleft lip: A, unilateral; B, bilateral; C, oblique facial cleft; D, median cleft lip and nasal defect; E, median mandibular cleft; and F, unilateral macrostomia

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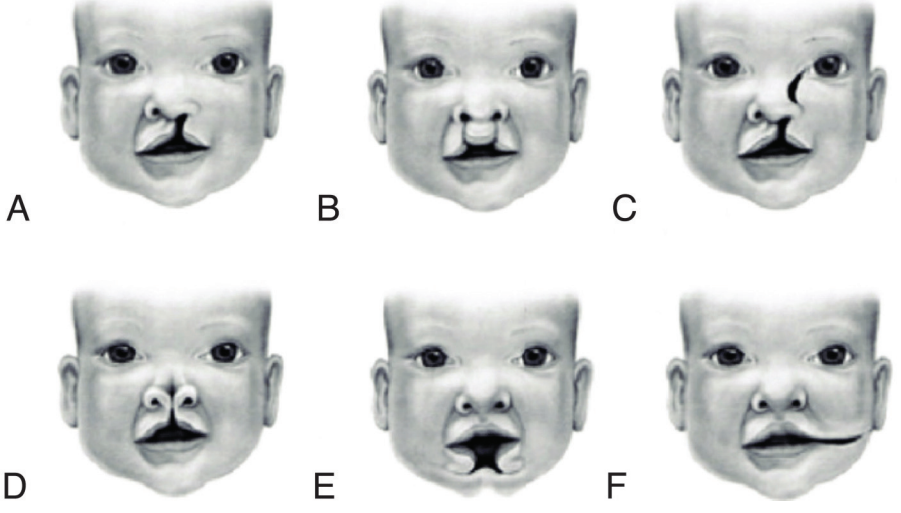

media and dental issues.[2] Children with all cleft anomalies are subject to social stigmas and may experience low self-esteem.[1,2,4]

PREOPERATIVE MANAGEMENT

The multidisciplinary team caring for a child with an orofacial cleft may include specialists in plastic surgery, oral and maxillofacial surgery, dentistry, orthodontics, otolaryngology, neurosurgery, genetics, nutrition, speech therapy, and child development. Most cleft teams are affiliated with the American Cleft Palate-Craniofacial Association (ACPA), which reports the standards and latest research in orofacial cleft, and also provides families with support and resources. Parents whose babies are diagnosed with cleft lip and palate in utero or at delivery can expect initial consultations by the cleft team to start shortly after birth, with treatments continuing into adulthood. Families and patients develop a special bond with their pediatric cleft team clinicians over the years, which makes transitioning to clinicians who only treat adults bittersweet.

Although neonates with CPO can have difficulty with feedings and are prone to aspiration, breastfeeding is encouraged for these infants, and support from a certified lactation consultant should be offered to the mother. Breastfeeding can resume immediately following cleft lip repair procedure.[3] Special bottles and nipples are available to facilitate feeding.[4] Encourage parents and caregivers to feed newborns in an upright position to avoid the risks of reflux. Infants born with orofacial clefts should acquire similar growth and weight gains as infants born without orofacial clefts.[2] Failure to thrive or gain weight may postpone a surgical correction and increase the risk of postoperative complications.[2,12]

In preparation for surgery on infants with cleft lip or cleft lip and palate, cleft teams may offer nasoalveolar molding (NAM) therapy. The flexibility of the skin and cartilage of the roof of the mouth in a newborn lets this therapy reduce the width of the cleft lip defect by repositioning the distorted nasal cartilage and realigning the gums before surgical repair. NAM therapy may be started at 1 week of age; the average treatment time is 3 to 6 months. NAM molding includes a custom acrylic orthodontic plate to cover the infant's palate that usually is evaluated every 2 weeks.

Audiologic and otolaryngology evaluations are essential components of the cleft team care. As with all newborns, those with cleft lip and palate undergo a newborn hearing screening before discharge. Newborns with CPO often fail this screening. Many neonates with clefts fail the newborn hearing screening. These infants are diagnosed with middle ear effusions.[2] Eustachian tube dysfunction contributes to a higher frequency of recurrent episodes of acute otitis media and conductive hearing loss.[4] According to Tsao and colleagues, patients who have tympanostomy tubes placed for middle ear effusion have better speech and language development.[13] Tympanostomy tube insertion procedures often are coordinated with the cleft palate repair.

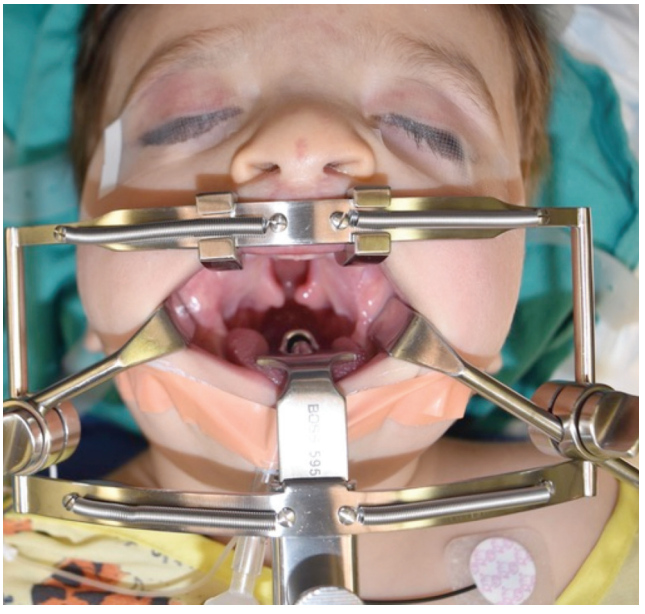

FIGURE 3. Intraoperative photograph during cleft palate repair procedure

Children with orofacial clefts can struggle with speech and language difficulties that can affect them for the rest of their lives. All children born with a cleft palate begin speech therapy at an early age. Speech services are provided through early intervention before age 3 years, and then through the school district. Children are discharged from speech therapy once their speech is proven to be age-appropriate. Even after corrective surgery, some children with a cleft palate will continue to struggle with a speech difficulty, velopharyngeal insufficiency (VPI). VPI occurs when the soft palate fails to properly close against the posterior pharyngeal wall.[2] A child with VPI has hypernasal speech.[2] Surgical management for VPI may be indicated if the child fails speech therapy.[2]

According to the latest guidelines from the AAP and the American Academy of Pediatric Dentistry (AAPD), all children, including children with orofacial clefts, should have their first dental visit by 1 year of age.[2] Children with orofacial clefts also will require oral health and orthodontic evaluations to ensure proper occlusion and optimal oral hygiene.[2] Their upper and lower jaws often develop at different rates, which is a significant cause of dental malalignment and overcrowding. Patients with a cleft through the alveoli of the maxilla, a bony process of the alveolar bone that forms the teeth sockets, have a higher rate of missing teeth, and/or ectopic teeth in the cleft site.[2]

CORRECTIVE SURGERY

Cleft lip repair traditionally is performed at ages 3 to 4 months. Surgical repair for cleft palate generally is done later but typically is recommended to be completed between ages 7 and 12 months (**Figure 3**). The ultimate surgical goals of cleft repair are to return the lip and nose to ana-

tomical harmony, and to restore palatal function and normal speech development. Different surgical techniques are considered and the reconstructive surgical procedure is individualized to each patient's cleft type.

Cleft lip repairs aim to create a symmetric and balanced lip and nose with minimal scarring. Cleft lip repairs undergo the most change during the first year after surgery; however, they continue to remodel for years following the surgery. Most children are hospitalized overnight for observation following their cleft palate procedure. Patients are prescribed a soft diet for 1 month, and should avoid using straws, to protect the surgical site from injury and possibly infection. Postoperative evaluation may vary depending on the patient's condition, as well as the surgeon's training and prior experience treating similar cases. Most patients are evaluated at 2 weeks postoperative.

Common surgical complications include bleeding, wound dehiscence, wound infection, and respiratory concerns such as laryngospasm.[5]

Annual follow-ups are essential to evaluate lip and nose symmetry with the child's craniofacial growth. Before adulthood, many children will require another surgical procedure on their nose because asymmetries often occur as the child grows, most often during puberty. Educate parents and patients about sun safety tips to protect the scar. The patient's individual healing and outcome depends on the type and the size of the cleft defect, as well as the patient's genetics. Patients with darker skin coloring are at higher risk for developing hypertrophic scars and keloids.[14] Nutrition plays an important role in cell proliferation and protein synthesis to support wound healing. However, obesity is associated with impaired wound healing and an increase risk of postoperative infections. Disorders of the immune system, red blood cell (sickle cell anemia), and genetic conditions (such as Klinefelter syndrome) also may impair wound healing.[15]

PREVENTION

The AAP, CDC, Institute of Medicine, and US Preventive Services Task Force all recommend that women of childbearing age take 0.4 mg of folic acid daily to help prevent neural tube defects.[2] The AAP believes that folic acid supplementation before pregnancy also may protect against orofacial clefts.[2] Of course, all prospective parents should receive genetic counseling to rule out or identify possible inherited diseases or defects.

CONCLUSION

Orofacial cleft defects among children are a global problem; their repair often is associated with organizations such as Smile Train and third-world mission trips.[2] The United States is not immune to these birth anomalies, but we are fortunate to have a healthcare system with advances that offer children surgical correction at a young age. More than 8,000 cleft lip and cleft palate repair procedures are performed annually in the United States.[2] Most children lead full lives with imperceptible scars due to early treatments, compared with children in developing countries whose clefts often go unrepaired into adulthood, and who may suffer stigmatization. Clinicians must help parents understand their child's diagnosis, prognosis, and treatment expectations, and offer them reassurance and comfort. Many resources are available online to parents and caregivers, from sites such as the ACPA (www.acpa-cpf.org). Patients with cleft lip or palate require long-term postoperative follow-up to assess proper healing, speech progression, and quality of life, but they can expect to lead normal, productive lives. **JAAPA**

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