PedsCases Podcast Scripts

This podcast can be accessed at www.pedscases.com, Apple Podcasting, Spotify, or your favourite podcasting app.

Orofacial Clefts

Developed by Dr. Julie Le and Dr. Regan Guilfoyle for PedsCases.com.
DATE

Introduction:

Hi everyone! My name is Julie Le and I'm a first year Family Medicine resident at Queen's University. Today's podcast was developed with the help of Dr. Regan Guilfoyle, a pediatric plastic surgeon at the University of Alberta. Today, we'll be discussing the topic of cleft lip and palate, and how it's managed.

In this podcast, we'll cover the following objectives:

1. Identification of the different types of cleft malformations
2. Outline the management of orofacial clefts and timing of surgical repair
3. Discuss the genetic syndromes, sequences and associations linked with cleft lip and palate

Let's jump into a case so we can get started on our journey!

Case Presentation:

You are an excited medical student on your pediatric rotation and you're at the hospital for a neonatal assessment. Upon arrival, you are presented with a 2722g female infant who was delivered via spontaneous vaginal delivery at 39 weeks gestation. You are told that the pregnancy was unremarkable with appropriate prenatal care. There were no complications or devices used during the delivery. No resuscitation was required after delivery, and the infant had APGAR scores of 9 at 1 and 9 at 5 minutes.

On general inspection, the baby is sleeping with no signs of distress. Mum states that she attempted to breastfeed immediately after delivery, but her baby wasn't latching appropriately to produce a good suck. She's concerned that there is something wrong.

You perform a physical exam including a very thorough mouth and oropharynx exam. During this examination, you discover something abnormal in the posterior aspect of the palate. You describe to your preceptor that you felt an opening on the palate.

Your preceptor turns to you and says “Well, it looks like this cleft palate was missed during the prenatal ultrasounds. What type of orofacial cleft is presenting in this case and what should we do next?”

Epidemiology:

Let’s start with epidemiology to put this case into perspective. Worldwide, the incidence of orofacial clefts is 1.7 per 1000 babies (Kawalec *et al.,* 2015). Ethnic and geographic variations also play a large role in the epidemiology of orofacial clefts. Racial groups such as Asian, Alaska Native and Indigenous North Americans have higher incidences than white or black racial groups. (Kawalec *et al.,* 2015; Lewis *et al.,* 2017). There are also differences in incidences between genders. Males have a higher incidence of having a cleft lip with or without a cleft palate compared to females (Lewis *et al.,* 2017). Females, however, have a greater incidence of having a cleft palate alone, which is typically located in the midline. Children who have a cleft palate alone have a 50% risk of an underlying associated syndrome, whereas children with a cleft lip with or without palate involvement have a 30% risk.

Definition and Categories:

So, what exactly is a cleft lip and palate? It is a craniofacial malformation where there is incomplete formation of the cleft lip, hard and/or soft palates (UptoDate, 2021). There are three categories that we typically use to define these orofacial malformations. The first is a cleft lip alone, the second is cleft palate alone, and the last is a combined cleft lip with cleft palate (CL/P). Cleft lips with or without palate involvement can present unilaterally or bilaterally.

Etiology:

The etiology of orofacial clefts seems to be related to a variety of factors that can include genetic predisposition (either alone or as part of a syndrome), environmental triggers- such as exposure to tobacco smoking, teratogens in the home and workplace, alcohol consumption, poor nutrition or viral illnesses during pregnancy, geographic location, and possibly maternal obesity and diabetes (Kawalec *et al.,* 2015).

Embryology:

To get a better understanding of this topic, let’s briefly discuss the embryology behind these malformations. There are two sets of dates I want you to remember: 4-6 weeks and 8-12 weeks of gestation.

At 4-6 weeks of gestation, fusion of the medial frontonasal process with the maxillary process of the first pharyngeal arch occurs (Abramson *et al.,* 2015). When this process fails, it results in a cleft lip. Depending on the severity of failure, the result is cleft lips of

different extents- incomplete or complete. With a complete fusion failure, the cleft will extend past the vermillion border of the lip to the ipsilateral alar base and include the alveolar process as well. The alveolar process is also known as the primary palate or premaxilla. This is the segment of the medial nasofrontal process that becomes the medial upper lip and alveolus containing the four maxillary incisors and it extends posteromedial to the incisive foramen. An incomplete cleft lip will involve the lip but will not extend to the alar base.

Essentially, in more layman's terms, a complete cleft lip will involve part of the palate where the upper four front teeth would grow and extend past the lip to the ipsilateral nostril. An incomplete cleft lip on the other hand, would spare the nose and only involve the lip and sometimes the alveolus. In even rarer circumstances an incomplete cleft lip will also be paired with a cleft of the palate.

At 8-12 weeks of gestation, fusion of the palatal shelves of the maxillary processes occurs. This forms the secondary palate, which is composed of the soft and hard palates (Kosowski, *et. al.;* 2012). When there is inappropriate fusion of the palatal shelves, the secondary palate develops a cleft. Cleft palates can be further divided into incomplete and complete categories. A complete cleft extends from the uvula to the alveolar ridge and includes both the primary and secondary palates. An incomplete cleft involves only the secondary palate.

There is also another type of incomplete cleft palate called a submucous cleft palate. This is where the palate appears structurally intact but is compromised by both muscular and bony deficits that can be recognized by a bony notch in the hard palate and a bluish line at the zona pellucida, which is the midline of the soft palate. This indicates clefting of the submucosal palatal musculature and a bifid uvula. The muscular deficit creates abnormal insertions of the soft palate muscles into the hard palate. This is significant because it can cause velopharyngeal incompetence, which may result in speech distortions. In summary, a submucous cleft palate is a cleft of the palate musculature only, and is significant for causing speech distortions.

Classification:

The Veau System for cleft palates is used to classify orofacial clefting by which palates are affected and by laterality. It has four classes.
Class 1 is incomplete cleft with soft palate involvement only with no uni or bilateral designation.
Class 2 includes both the soft and hard palates, again with no uni or bilateral designation
Class 3 is a complete unilateral cleft of the secondary and primary palate, and includes the lip
And finally, **Class 4** is complete bilateral clefts

Diagnosis:

With improving technology and increased access to prenatal imaging, most cleft lips and palates can now be prenatally diagnosed via ultrasound or MRI (Abramson *et al.,* 2015; Marginean *et al.,* 2018). As previously discussed, the secondary palate completes development at 8-12 weeks of gestation. This means that diagnosis of defects can start from 11-13 weeks of gestation. But, as seen in our case, prenatal diagnoses can be missed and therefore, diagnosed postnatally. If a thorough oral exam is not done postnatally, a palate cleft could come to light when the infant has difficulties with feeding, such as through struggling to latch, regurgitation, being easily exhausted during feeding, or reduced volume intake.

Management:

Management and treatment of orofacial clefts tend to be a long-term process and include long-term follow up.

If untreated, the infant can have a variety of direct and associated problems such as facial deformation, increased feeding issues, airway concerns, recurrent middle ear otitis media as the eustachian tube is often affected, and speech difficulties. As the infant grows, there can be defects in tooth development and malocclusion as well (Kohli *et. al.;* 2012). Hence, upon birth, a multidisciplinary team is essential for treatment and management of the malformation, itself, as well as the associated problems. The degree of intervention is of course dependent on the degree of the malformation.

Once the malformation is identified, the first step is a referral to the cleft lip and palate team. This provides access to the team of health care workers which typically includes a pediatrician, otolaryngologist, dentist, orthodontist, oral surgeon, plastic surgeon, social worker, speech language pathologist, audiologist, geneticist, psychologist, nutritionist/dietician and respirologist (Thorne *et. al*., 2007). From there, the team assesses what the neonate's current and future needs will be, attempting to coordinate in order to minimize surgeries while maximizing the benefit to the patient. One essential component is performing a genetic evaluation to assess for syndromes or other more subtle associated anomalies (Bentz *et. al.*, 2016). The team can vary to include other disciplines depending on the child's needs.

Feeding

An immediate concern for babies born with orofacial malformations is feeding. Studies have shown that children with orofacial malformations have more delays in growth compared to children who do not (Hooper *et. al.;* 2011). Infants with cleft lips only, do not have large concerns with feeding but need specific positioning during feeds. In all modified feeding positions, the babies are held upright so that the milk can flow

downwards and prevent choking. Choking occurs when the milk enters the respiratory system through the cleft (Kumar *et. al.,* 2013).

Breastfeeding is still encouraged for infants with orofacial malformations. However, infants with cleft palates tend to struggle with feeding and, in most cases, require specially designed bottles and/or nipples that increase milk transfer and suction pressure to allow the baby to feed more successfully. Therefore, these bottles can be used to provide supplementary feedings with breastmilk or formula (Kumar *et. al.,* 2013). Originally, bottles were changed in design to be squeezable with a NUK orthodontic nipple or hard cross-cut nipple to aid in feeding. Then came the development of nipples with one-way valves that use compression (positive pressure) versus suction (negative pressure) that help the infant actively obtain milk from bottles when they cannot create the suction pressure themselves (Nahai *et. al.,* 2005).

Post-surgical care and feeding can vary depending on the medical center. In general, infants can return to regular feeding with a bottle and re-introduction of thickened fluids or pureed solids. Hard, dry, or non-pureed solids are to be avoided as are pacifiers for up to 3 weeks post-op to allow adequate healing of the repair site.

Surgical Repair

Now we'll move onto the surgical repair portion of management.

Cleft Lip Repair

Let's start with cleft lip repair, which is generally the first surgical procedure that the patient undergoes. This procedure occurs between 3 and 6 months of age, with the caveat that the patient meets the "rule of tens". This is where the child is at least 10 weeks of age, weighs 10 pounds, and has a hemoglobin level of 10 g/dl (Nahai *et. al.,* 2005; Bentz *et. al.,* 2016). Of course, the rule falls apart in Canada as our SI units change the hemoglobin level to 100 g/L and the weight to 4.5 kg, but it's useful none the less.

The mainstay of presurgical management of infants born with complete unilateral or bilateral cleft lip and palate is the nasoalveolar molding appliance (also referred to by it's acronym NAM). The purpose of this appliance is to decrease the alveolar gap and corresponding upper lip elements and improve the nasal position of the cleft side. It is particularly useful in bilateral clefts as it can reduce the prominence and protrusion of the premaxilla prior to surgery and reduce tension on the lip repair. The appliance consists of an intraoral acrylic molding plate and intranasal stents. This appliance is then combined with taping of the upper lip and is created and managed by an orthodontist in coordination with the surgical team *(Kapadia Hitesh et. al., 2020,* Bentz *et. al.,* 2016*).*

Alternatives to nasoalveolar moldings include creating external pressure using tape or elastic devices to narrow the alveolar gap, or lip adhesion, a surgery that sutures subcutaneous flaps together to convert a complete cleft lip into an incomplete one. (Bentz *et. al.,* 2016).

Surgery can then be performed to repair the cleft lip with a goal of creating a normal appearance and achieving symmetry of the lip and nose (Bentz *et. al.,* 2016). All techniques of cleft lip repair include some element of rotation or advancement of skin flaps to limit scar contracture that will shorten the lip and contribute to new deformities. For complete bilateral cleft lips, it is advantageous to repair both clefts simultaneously for symmetry. Additionally, the upper gingivolabial sulcus requires reconstruction in a patient with complete bilateral cleft lip, as the sulcus is deficient or nonexistent with this condition (Fisher *et.al.,* 2011).

Once lip and mucosal repairs are complete, partial nasal correction is done (Bentz *et. al.,* 2016). Completely correcting the nasal deformity is unrealistic, as skeletal correction needs to be done before complete nasal correction. Since presurgical molding can only partially improve the skeletal deformity, complete nasal correction cannot be achieved during this primary surgery. Complete nasal repair is performed after skeletal maturity and any orthognathic surgery.

Following cleft lip repair, patients are discharged the same day if they tolerate the procedure and are doing well. Some surgeons chose to use bilateral arm restraints post-operatively but there is no evidence to suggest that this decreases complication rates. Additionally, there are no feeding restrictions. Scar hypertrophy occurs 3-4 weeks postoperatively and tends to improve with time. Scar massage is encouraged to help soften and lengthen the scar. If there are significant deformities that persist after healing is complete, revisions can be done before the child is of school age for psychological benefit.

Cleft Palate Repair

Next, we'll explore cleft palate repair. Cleft palate repair has the goal of separating the oral and nasal cavities (Bentz *et. al.,* 2016). This contributes to normalizing feeding, reducing regurgitation and nasal irritation, establishing normal speech, and minimizing growth restrictions of the maxilla. Palatoplasty is performed between 9 to 12 months of age and is most often combined with placement of myringotomy tubes. Patients with Pierre Robin sequence or other idiopathic micrognathia typically have the repair done at 12 to 13 months of age to allow for maximal growth of the mandible. Cleft palate repairs aim to restore continuity of the hard palate and reconstruct normal soft palate anatomy and function. Hard palate repair comprises utilizing flaps to achieve closure. Soft palate repair includes intravelar veloplasty and z-plasty to lengthen the palate. Z-plasty is a technique that is commonly used in plastic surgery for scar revision to change the direction of the scar. This procedure involves two equal and opposing triangular flaps that are transposed along a shared axis (Zito *et. al.*, 2021). In this

case, it is used for closure of the soft palate. Postsurgically, patients remain in hospital for 24-48 hours with arm splints, continuous oximetry monitoring, and intravenous fluids for hydration. Oral feeding can be reimplemented in the evening and thickened fluids and purees can also be re-introduced. The patient's family is advised to avoid the use of pacifiers and to only feed pureed foods for 3 weeks post-operatively. Additionally, bilateral arm splints are to be worn for 3 weeks to avoid unintentional trauma to the palate and allow complete healing of the surgical site.

Velopharyngeal Insufficiency

The single most important outcome of cleft palate repair is adequate speech production, which cannot be adequately assessed before 3 years of age. By 3 to 4 years of age a speech-language pathologist can assess for velopharyngeal insufficiency, or VPI. To better understand velopharyngeal insufficiency, let me tell you about the velopharyngeal valve. This valve controls communication between the oral and nasal cavities and affects speech production. The valve is controlled by the pharyngeal muscles and the levator sling, which is restored during cleft palate surgery. If there is incompetency of these muscles in opening and closing the valve, this can result in nasal air emissions, hypernasality, decreased vocal intensity, short phrases, and difficulties with consonant pronunciation. In most cases velopharyngeal insufficiency can be improved with speech therapy alone. However, approximately 20% of patients will require pharyngoplasty surgery to help improve speech (Thorne *et. al.,* 2007).

Alveolar Bone Grafting and Cleft/Lip Rhinoplasty

Secondary alveolar bone grafting occurs before the growth of the adult cleft-side canine (Bentz *et. al.,* 2016) before 10 years of age. This procedure reconstructs the alveolar gap using bone graft, which is generally harvested from the iliac crest. Mucosal flaps are then created to cover the bone graft and close the defect (Wang *et. al.,* 2016*,* Power *et.al.,* 2009).

Secondary rhinoplasty and septoplasty are typically performed when patients are teenagers (Bentz *et. al.,* 2016). There is no specific age of repair so surgeons rely on the following guidelines:

1. the patient is at least one year post-menarcheal
2. growth has not been significant over the past 1-2 years, as documented by a pediatrician
3. the patient's parents are ready for the patient to proceed with the surgery

Syndromes, Sequences, Associations and Anomalies

Orofacial cleft malformations often have associated congenital anomalies, whether it is part of a syndrome or separate. Although most clefts are not associated with specific

syndromes, syndromes play a significant role in the discussion of orofacial clefts (Venkatesh, 2009).

Syndromes are split into two categories: monogenic and chromosomal syndromes. As indicated by the name, monogenic syndrome is the term for several associated anomalies that are due to a single gene. Van der Woude syndrome is one monogenic syndrome that is often linked to an anomaly with Chromosome 1q32-q41. In addition to orofacial clefts, patients with this disorder typically have depressions near the centre of their lower lip, abnormal salivary gland morphology, and missing teeth (Deshmukh *et. al.,* 2014). Another well-known monogenic syndrome is Treacher Collins syndrome. This is autosomal dominant disorder that includes orofacial clefts, downward slanting palpebral fissures, external and middle ear anomalies, and underdevelopment of the zygomatic complex, cheekbones, jaws, palate and mouth. These malformations can lead to breathing and feeding difficulties for patients (Prachi *et. al.,* 2014).

Chromosomal syndromes are anomalies that are due to chromosomal defects. Trisomies 13 and 18 are common chromosomal syndromes that include orofacial clefts. Trisomy 13, or Patau's syndrome, is caused by an extra copy of Chromosome 13. Patients with this disorder can present with malformations including microcephaly, polydactyly, and microphthalmia. Trisomy 18, or Edwards' Syndrome, is also caused by an additional chromosome. Most neonates diagnosed with this condition die before they reach the age of one. They can present with low set ears, broad foreheads, narrowed face, omphalocele, and short stature. Alongside these physical impacts, Trisomy 18 and 13 patients are impacted with cognitive impairment and global developmental delay (Cereda *et. al.,* 2012; Noriega *et. al.,* 2011).

Outside of syndromes, there are also sequences and associations.

A sequence is when the associated anomalies arise from one structural defect. A well-known sequence is the Pierre Robin Sequence. In this case, a cleft palate may develop embryologically because of a micrognathic jaw.

When there is a non-random development of a collection of morphological defects, and there is no link to a syndrome or sequence, it is called an association. For example, orofacial clefts are often associated with congenital heart defects, and it is unknown why (Venkatesh, 2009).

Case Conclusion

Alright, now that you've learned so much about cleft lip and palates, let's revisit the case!

So, your staff was just asking you "What type of orofacial cleft does this look like and what should we do next?"

With your new knowledge, you repeat your thorough intraoral exam and correctly identify the malformation as an incomplete cleft palate. As there was no indication or cause for this malformation, you also add in that it is likely a sporadic cleft palate.

You turn to the patient's parents and reassure them that orofacial clefts are one of the most common congenital defects and share that the baby will be well taken care of by a dedicated multidisciplinary team, whom you have just referred the infant to. You explain to the family that the speech pathologist and nutritionist will be here soon to do a feeding assessment and provide specialized bottles and nipples to aid in feeding. The team will also arrive to complete a medical and genetic assessment to rule out any syndromes or associated anomalies and provide additional treatment information. You let the family know that there are various stages and aspects to treatment, including various surgeries, and that the team will support the family through it all.

So now that you've identified the orofacial cleft and communicated the next steps to the family, let's review some take home points:

1. Orofacial clefts can be differentiated into 3 categories: cleft lip only, cleft palate only, or a combined cleft lip and palate. Both cleft lips only and cleft palates only can further be broken down into incomplete and complete categories. Additionally, laterality is used to denote if the cleft is present on the left, right or both sides.
2. Management of orofacial clefts is a long-term process that requires a multidisciplinary team and occurs in stages. This is important because the cleft itself needs to be addressed, as well as any associated problems that the malformation may have created, such as recurrent otitis media or speech difficulties. A major component of orofacial cleft treatment is surgery. The child will undergo various surgeries starting from 3 months to when the child reaches skeletal maturity.
3. Orofacial clefts can have associated anomalies and be linked to syndromes or sequences. Syndromes can be categorized into monogenic or chromosomal syndromes, depending on whether a single gene or an entire chromosome is affected.

Thanks for listening and we hope that you enjoyed this podcast on orofacial clefts!

References:

Abramson, Z.R., Peacock, Z.S., Cohen, H.L. & Choudhri, A.F. (2015). Radiology of Cleft Lip and Palate: Imaging for the Prenatal Period and throughout Life. *Radiographics,* 35(7), 2053-2063.

Bentz, M.L. Bauer, B.S. & Zuker, R.M. (2016). Principles & Practice of Pediatric Plastic Surgery (Second Edition) New York, NY: 10001 USA

Developed by Dr. Julie Le and Dr. Regan Guilfoyle for PedsCases.com.
May 26, 2022

Bessel, A., Hooper, L., Shaw, W.C., Reilly, S., Reid, J., & Glenny, A.M. (2011). Feeding interventions for growth and development in infants with cleft lip, cleft palate or cleft lip and palate. *Cochrane Database Syst Rev,* 2011(2), CD003315

Buchanan, E.P. & Hyman, C.H. (2013) LeFort I Osteotomy. *Semin Plast Surg*, 27(3), 149-154.

Cereda, A. & Carey, J.C. (2012) The trisome 18 syndrome. *Orphanet journal of rare diseases.* 7 (81), doi: 10.1186/1750-1172-7-81

Deshmukh, P.K., Deshmukh, K., Mangalgi, A., Patil, S., Hugar, D., & Kodangal, S.F. (2014) Van der woude syndrome with short review of literature. *Case Reports in dentistry.* 2014 (8714260) doi: 10.1155/2014/871460

Fisher, D., Sommerlad, B. (2011) Cleft Lip, Cleft Palate, and Velopharyngeal Insufficiency. *Plastic & Reconstructive Surgery*. 128 (4), 342-360.

Kapadia H, Olson D, Tse R, Susaria S. (2020) Nasoalveolar Molding for Unilateral and Bilateral Cleft Lip Repair. *Oral and Maxillofacial Surgery Clinics of North America.* 32(2), 197-204.

Kawalec, A., Nelke, K., Pawlas, K., & Gerber, H. (2015) Risk factors involved in orofacial cleft predisposition- review. *Open Med (Wars)*, 10(1), 163-175.

Kumar Jindal, M., & Khan, S.Y. (2013) How to feed cleft patient? *Int J Clin Pediatr Dent,* 6(2), 100-103.

Marginean, C., Sasarean, V., Marginean, C.O., Melit, L.E. & Marginean, M.O. (2018). Prenatal diagnosis of cleft lip and cleft lip palate-a case series. *Med Ultrason,* 20(4), 531-535.

Nadjmi, N., Amadori, S., & Van de Casteele, E. (2016). Secondary Cleft Lip Reconstruction and the Use of Pedicted, Deepithelialized Scar Tissue. *Plast Reconstr Surg Glob Open.* 4(10), e601.

Nahai, F.R., Williams, J.K., Burstein, F.D., Martin, J. & Thomas, J. (2005). The Management of Cleft Lip and Palate: Pathways for Treatment and Longitudinal Assessment. *Semin Plast Surg.* 19(4), 275-285.

Noriega, M.A. & Siddik, A.B. (2021) Trisomy 13. Retrieved from https://www.ncbi.nlm.nih.gov/books/NBK559091/

Power, S.M., Matic, D.B. (2009) Gingivoperiosteoplasty following alveolar molding with a Latham appliance versus secondary bone grafting: The effects on bone production and midfacial growth in patients with bilateral clefts. *Plast Reconstr Surg*. 12: 573-582.

Developed by Dr. Julie Le and Dr. Regan Guilfoyle for PedsCases.com.
May 26, 2022

Prachi, S., Tupkari, J.V., Benjamin, T. & Singh, A. (2011) Treacher Collins syndrome. *Journal of oral and maxillofacial pathology: JOMP.* 15(3), 348-351.

Thorne, C.H., Beasley, R.W., Aston, S.J., Bartlett, S.P., Gurtner, G.C. & Spear, S.L. (2007). Grabb & Smith's Plastic Surgery (6th Edition) Philadelphia, PA: 19106 USA.

Uptodate (2021). Etiology, prenatal diagnosis, obstetric management, and recurrence of cleft lip and/or palate. Retrieved from: https://www.uptodate.com/contents/etiology-prenatal-diagnosis-obstetric-management-and-recurrence-of-cleft-lip-and-or-palate

Venkatsh, R. (2009) Syndromes and anomalies associated with cleft. *Indian Journal of Plastic Surgery*, 42(Suppl), S51-S55.

Wang, Y, Liao, Y., Chen, P.K. (2016) Comparative Outcomes of Primary Gingivoperiosteoplasty and Secondary Alveolar Bone Grafting in Patients with Unilateral Cleft Lip and Palate. *Plast Reconstr Surg*. 137 (1), 218-227.

Zito, P., Jawad, B.A. & Mazzoni, T. (2021) Z Plasty. Retrieved from: https://pubmed.ncbi.nlm.nih.gov/29939552

Developed by Dr. Julie Le and Dr. Regan Guilfoyle for PedsCases.com.
May 26, 2022