Introduction

Approximately 15 types of orofacial clefting exist, with cleft lip and palate being the most common. Orofacial clefts are the second most common congenital malformation of the body, and the most common congenital malformation of the head and neck. They occur in approximately 1/700 live births with a rate of about 1/1000 live births in the United States. There are approximately 7500 babies born in the United States with orofacial clefts per year with an average lifetime medical cost of $100,000 per baby or 750 million dollar lifetime cost for all children per year. Cleft lip and palate have many associated medical problems, which include otological disease, speech impairment, dental deformities, and psychosocial issues. These problems are best managed with a multidisciplinary approach requiring a coordinated medical and surgical care plan to provide optimal outcomes.

Both ethnic and gender differences occur in the birth prevalence of cleft lip and palate. Among ethnic groups, the highest rate occurs within Native American and Asian populations with approximately 2/1000 live births. There is an intermediate rate among those of European descent with approximately 1/1000 live births. The lowest rate of cleft lip and palate occurs among African populations with 1/2500 live births. There is no difference between ethnic groups for cleft palate only, with approximately 1/2000 live births. A gender difference does exist when it comes to cleft lip and palate formation with a 2:1 male to female ratio in clefts involving the lip with or without cleft palate. Consequently, a 1:2 male to female ratio exists for clefts involving the palate only. It is thought that the reason the ratio changes for cleft palate only is due to the fact that the palatine shelves close one week later in females than males.

Many speculations to the causes of cleft lip and palate have been made throughout human history including many folklore explanations. The Aztecs believed eclipses occurred because a bite had been taken out of the moon and exposure to an eclipse during pregnancy could lead to a bite out of the baby’s mouth. To prevent clefts, pregnant women were required to place obsidian knives over their abdomen before going out at night. This belief has now extended to modern day Mexico and during eclipses a metal key or safety pin is placed over the abdomen for protection. An early Chinese belief was that eating rabbit during pregnancy could lead to a “hare lip,” and others believed that bad karma or wrongdoings were the cause. In modern day Filipinos, a common belief is that force to the fetal face when fingers are in the mouth is the cause of clefts. Many other cultures believed that clefts were familial or in the blood. Most of the early
thoughts on the causes of cleft have been disproved but the belief that there is a familial component still stands today.

It has long been known that familial inheritance plays a role in cleft formation and the following data supports this theory. In two unaffected parents with one child affected, the risk for future children is 4.4% for cleft lip with or without cleft palate and 2.5% for cleft palate only. If only one parent is affected, the risk for having a child with cleft lip with or without palate is 3.2% and the risk for cleft palate only is 6.8%. If one parent and one child are affected, the risk for future children increases to 15.8% for cleft lip with or without cleft palate and 14.9% for cleft palate only. These exact percentages vary from study to study, but the common theme among studies is that as the number of family members with cleft deformities increase, the risk for future children increases as well.

The majority of clefts are considered nonsyndromic or isolated anomalies. Approximately 70% of cleft lip with or without palate and 50% of cleft palate only are nonsyndromic. No exact mechanism has been discovered for the cause of nonsyndromic cleft, and the cause is believed to be multifactorial. Clefts tend to cluster in families but are not exactly Mendelian in inheritance. It is known that normal palate development is a complex process with several proteins, growth factors, and transcription factors involved. Any disturbance in the developmental sequence can result in cleft formation.

Syndromic clefts are less common than nonsyndromic clefts and by definition are associated with other malformations or syndromes. Over 300 syndromes have been identified that are associated with orofacial clefting. The most common syndrome associated with cleft lip and palate is Van der Woude syndrome. This syndrome is autosomal dominant and has characteristic lower lip pits with orofacial clefts. Teratogen exposure and environmental factors have also been linked to cleft formation.

Anatomy

The orbicularis oris is the muscle that surrounds the lips and acts as a sphincter for the mouth. The vermilion is the red portion of the lips and an important landmark is the wet/dry border. Along the upper vermilion is the vermilion-skin border or white roll. Two midline elevations arise from the vermilion and form the Cupid’s bow. Two raised vertical columns of tissue extending from the apices of the bow are known as the philtral columns. The area of depression between the columns is known as the philtral dimple. The philtrum extends from the cupid’s bow to the columella, which are both important structures when reconstructing the lip. The tubercle is the midline protuberance of the upper lip, which is also important in reconstruction. The hard palate is made up of the palatine processes of the maxilla and the horizontal plates of the palatine bone. It is bound anteriorly and laterally by the alveolar arches and is continuous with the soft palate posteriorly. An important structure here is the incisive foramen, which marks the division between the primary and secondary palate during development. Greater palatine and lesser palatine foramen are important landmarks from which the greater palatine and lesser palatine nerve/artery enters the palate. This is important because the flaps for two-flap palatoplasty receive their blood supply from the greater palatine artery. The soft palate consists of two parts. The anterior portion is the palatine aponeurosis, which attaches to the posterior margin of the hard palate and is formed by the expanded tendon of the tensor veli palatini. The muscles of the soft palate attach to the aponeurosis. The muscular posterior portion of the soft palate consists of five muscles: tensor veli palatini, levator veli palatini, palatoglossus, palatopharyngeus, and the uvular muscle.
The blood supply to the palate is mainly from the greater palatine artery, which arises from the maxillary artery, but it receives some supply from the lesser palatine and ascending palatine arteries. The greater palatine and the nasopalatine nerves innervate the hard palate. The greater palatine nerve supplies the gingivae, mucous membranes, and glands of most of the hard palate. The nasopalatine nerve supplies the mucosa of the anterior portion of the palate. The soft palate receives sensory innervation from the lesser palatine nerves and receives its motor innervation from two cranial nerves. The tensor veli palatini is supplied by cranial nerve V and the four other muscles are supplied by cranial nerve X.

Embryology

Normal embryological development of the lip and palate is essential in understanding how clefts form. The development of the lip and palate begins around the 4th week of embryological development and is completed by the end of the 12th week. By the end of the 4th week, the five facial prominences have formed which include the frontonasal process, the paired maxillary processes, and the paired mandibular processes. These five facial prominences surround the developing oral cavity.

During the 5th week of development the nasal placodes form from the frontonasal process. The nasal placodes invaginate to form the nasal pits, along with the lateral and medial nasal prominences or swellings. By the end of the 6th week, the paired maxillary processes have grown medially and pushed the medial nasal prominences together. Fusion of the medial nasal prominences form the philtrum, middle upper lip, nasal tip, and columella. Fusion of the paired maxillary prominences with the paired medial nasal prominences forms the complete upper lip with the maxillary prominences forming the lateral lip. The lateral nasal prominences form the bilateral nasal alae. Palate formation begins at the end of the 5th week of development and is completed by the 12th week of development. The completed palate is formed by the primary palate and the secondary palate, which are separated by the incisive foramen. As the maxillary processes grow, the medial nasal prominences are pushed together and fuse to form not only superficial structures of the face but also a deeper structure referred to as the primary palate. The primary palate consists of the central maxillary alveolar arch with four incisors and the hard palate anterior to the incisive foramen. The primary palate’s development is completed before the secondary palate begins development. During the 6th week, shelf-like outgrowths from the bilateral maxillary processes grow vertically down on both sides of the tongue. During the 7th week, the tongue moves inferiorly and the palatal shelves migrate to a horizontal position above the tongue. Palatal fusion occurs in an anterior to posterior direction and completes with uvular fusion by the end of the 12th week. This fusion occurs one week later in females.

Cleft Formation

Disruptions at any stage of the developmental process can result in clefts of the lip and palate. The process is complex and requires many steps to form a complete lip and palate. Any disturbance in timing of a particular step can result in formation of a cleft. Obstructions can result in abnormal positioning of the developing palate, which can prevent proper fusion of the palate. Pierre Robin Sequence is an example of when positioning prevents closure of the palate. The micrognathia does not allow the tongue to move inferiorly which prevents the palatal shelves from migrating into a more horizontal position for fusion resulting in a wide U shaped palate. Several stages in the development process require fusion of facial prominences. Failure of fusion of the maxillary and nasal prominences on one side result in cleft lip with or without primary palate and failure of fusion bilaterally results in bilateral cleft lip with or without primary palate. The failure of fusion of the palatal shelves will result in clefts of the secondary palate.
Classification

Several classification systems have been developed for cleft lip and palate, with no one universally recognized classification system. The various classification systems are all based on the fact that clefts can be unilateral or bilateral and complete or incomplete. The Veau classification is the simplest and most utilized of the classification systems. Class I is incomplete cleft involving only the soft palate. Class II is a cleft involving the hard and soft palate. Class III is complete unilateral cleft involving the lip and palate. Class IV is complete bilateral clefts involving the lip and palate. Most surgeons use modified versions of the available classification systems or simply describe the defect that is present.

Cleft Anatomy

In incomplete cleft lip, the muscle fibers of the orbicularis oris are often intact but hypoplastic. Varying degrees of clefting can be present. In complete cleft lip, the orbicularis oris inserts at the columella medially and the ala laterally on the cleft side. The columella is displaced to the normal side and the nasal ala on the cleft side is displaced laterally, inferiorly, and posteriorly. The nasal tip is deflected towards the noncleft side. The alveolus may or may not be involved with clefts of the lip.

Bilateral cleft lip is similar to the unilateral cleft lip with a few exceptions. The orbicularis oris attaches to the lateral cleft margins bilaterally at the nasal ala, with varying degrees of premaxilla protrusion possible. The nasal deformities are symmetrical with practically a doubling of the unilateral nasal deformity. The bilateral nasal alae are displaced laterally, posteriorly, and inferiorly which gives a widely flared appearance. The columella is extremely short in bilateral clefts of the lip.

Clefts of the palate vary from submucosal clefts to complete clefts extending to the maxillary alveolus. Clefts of the primary palate are anterior to the incisive foramen and those of the secondary palate are posterior to the incisive foramen. Clefts of the secondary palate develop due to failure of fusion of the palatal shelves, which results in abnormal insertion of muscle into the medial edges of the cleft. Vomer attachment is variable. With unilateral cleft lip and palate, it attaches to the noncleft side and with bilateral clefts it has no connections.

Management

Cleft lip and palate are not just surgical problems and require a multidisciplinary approach to management. Children born with clefts are faced with many problems that require long-term coordinated care. A typical cleft care team consists of physicians from multiple specialties. The management of patients with clefts begins at birth with management of feeding and airway concerns. Cleft lip and palate patients are predisposed to developing otological disease and speech disorders. Otolaryngologists play a pivotal role in management of airway concerns, speech disorders, and ear problems. Depending on the specific surgical center, reconstructive surgery may require the skills of an otolaryngologist as well.

Isolated cleft palate rarely results in airway compromise. Airway issues are usually associated with coexisting structural abnormalities. Pierre Robin Sequence is the most documented cause of airway compromise in children born with cleft palate. Pierre Robin Sequence refers to the grouping of micrognathia, glossoptosis, and U shaped cleft palate. Glossoptosis can lead to airway obstruction. Several syndromes have been associated with the development of Pierre Robin Sequence with Stickler and Velocardiofacial syndrome being the most common. The initial management of newborns with Pierre Robin Sequence is the most documented cause of airway compromise in children born with cleft palate.
Robin Sequence is sometimes as simple as placing the newborn in prone position; however, some severe cases may require tracheostomy. Mandibular distraction can be used for correction as well.

One of the most critical aspects in management of a newborn with cleft lip and palate other than airway concerns is nutrition and feeding. Before the patient leaves the hospital feeding techniques must be established. Cleft palate limits the ability to suck due to the common cavity between the mouth and the nose. Cleft lip alone usually does not result in feeding issues. Special bottles have been developed to improve the feeding in patients with cleft palate. Some patients require premaxillary orthopedics to feed appropriately.

Most children with cleft palate have some form of ear disease. The abnormal insertion of the tensor veli palatini is believed to be the cause or contribute to the development of middle ear disease. Persistent otitis media with effusion has been estimated to be between 80-95% in children with cleft palate. Paradise et al. in 1969 devised the term “universality of otitis media in cleft palate children” after demonstrating that 96% of cleft patients had middle ear effusion in a study of cleft patients at his institution. Szabo et al. in 2009 did a retrospective study looking at the presence of middle ear effusion and treatment outcomes in cleft palate patients from Connecticut Children’s Medical Center between 2000-2005. They found that 61 of 74 or 82% of patients passed their newborn hearing screen. Ninety-eight percent of the patients in the study developed middle ear fluid requiring at least one set of tubes. Interestingly, only 74% of the patients in this study required one or two sets of tubes before resolving the eustachian tube dysfunction sufficiently where the middle ear effusion did not reaccumulate.

Practically all patients with unrepaired cleft palate will develop some form of speech abnormality. The primary goal of palate repair is to restore function of the velopharyngeal valve, which should restore normal speech. Even with surgical closure of the palate, 10-20% of patients have been reported to have velopharyngeal insufficiency. These patients will have varying degrees of hypernasality and articulation errors such as glottal stops and pharyngeal fricatives. The velopharyngeal insufficiency following palate surgery is first managed by speech pathology. If speech pathology alone is insufficient, it can be managed surgically with pharyngoplasty or palatoplasty.

**Surgical Correction**

Timing of surgical correction varies among individual institutions. Typically, presurgical techniques are used from 1-3 months. Cleft lip repair is performed at 3 months of age and myringotomy tubes are placed at the time of repair. The palate is usually repaired from 9-12 months of age. Initial orthodontic treatment begins at 1 year of age. Alveolar bone grafting is performed from 7-8 years of age before permanent teeth have erupted. Around 18 years of age or skeletal maturity, midface advancement is performed if necessary.

Sometimes presurgical steps must be taken to improve surgical outcomes. When a wide cleft or premaxilla protrusion is present, it is advantageous to narrow the cleft and/or mold the premaxilla before definitive surgery is performed. There are surgical and nonsurgical techniques that facilitate presurgical correction. Taping has been used effectively to reduce the width of a cleft and reduce premaxilla protrusion in a nonsurgical manner. With taping, strips of hypoallergenic tape are placed with tension across the cleft and secured to the patient’s cheek. This molds bony tissue by applying gentle pressure to protruding portions of the maxilla. Nasoalveolar molding devices are custom-made orthopedics which utilize wiring and nasal stenting to mold the nasal cartilage, premaxilla, and alveolar ridge. The nasal
stenting can be elongated and adjusted to lengthen the columella and mold the nasal cartilage. This method takes advantage of the malleability of immature nasal cartilage and its ability to maintain form once corrected. When taping and nasoalveolar molding is not effective, lip adhesion can be considered. Lip adhesion is when a complete cleft is surgically converted to an incomplete cleft. Lip adhesion is performed at 2-4 weeks of age and extends definitive repair to 4-6 months to allow proper healing. It has the same indications as the nonsurgical forms of correction, which include wide unilateral clefts or premaxilla protrusion. The disadvantage to performing lip adhesion is the development of scar tissue, which could interfere with definitive repair. Most surgeons do not perform lip adhesion because of the possibility of extensive scar tissue formation.

**Cleft Lip Correction**

Definitive cleft lip repair is performed at approximately 3 months of age. Many surgeons follow the “rule of tens” when deciding the proper timing for lip repair. The “rule of tens” states that a patient must be at least 10 weeks old, weigh at least 10 lbs., and have a hemoglobin of 10 or higher. Definitive lip repair must be delayed to 5-6 months of age if lip adhesion is performed to allow for proper healing and scar maturity.

**Milliard Rotation-Advancement Technique**

The Milliard rotation-advancement technique was first introduced in 1957. It is the most widely used procedure for cleft lip repair. In North America, 46% of physicians report using the Milliard technique and 38% report using a modified version of the technique. The advantages of this technique include: placement of the scar along the philtral columns, allows complete muscular repair, and minimizes wasting of normal tissue. Disadvantages include the requirement for extensive undermining. Nine landmarks are marked using blue dye to create the cuts necessary to achieve proper symmetry with closure. The rotation flaps are first cut on the medial edge of the cleft followed by the advancement cuts on the lateral portion of the cleft. Downward and lateral rotation of the medial segment and medial advancement of the lateral segment into the cleft allows for closure. The closure consists of 3 layers: mucosal, muscular, and cutaneous. The ultimate goals of the Milliard technique consist of tensionless closure, formation of the cupid’s bow, creation of the philtrum, and repositioning of the nasal ala to a more symmetrical position. Generally not many complications arise, but the most common is hypertrophic scarring and scar contracture, which can be partially prevented with early aggressive massage. Nostril stenosis can occur when repairing wide clefts.

Bilateral cleft lip repair is a challenging and technical procedure. There are many surgical techniques that are currently used for repair of bilateral cleft lips. All of which include the common goals of symmetry, orbicularis oris closure, philtrum formation, formation of the median tubercle, and positioning of the ala cartilages to construct the nasal tip and columella.

**Cleft Palate Correction**

The primary goal of cleft palate repair is to restore function by separating the nasal and oral cavity. This creates a velopharyngeal valve allowing for proper swallowing and speech, and the preservation of midface growth. The timing of closure has been somewhat controversial. Speech outcomes are better with early closure but midfacial growth may be hindered. Most surgeons repair cleft palate between 8-12 months of age to minimize speech abnormalities. The Bardach two-flap palatoplasty and the Furlow
palatoplasty (or some modified version of the two) are the most common surgical techniques used in the United States for repair of cleft palate.

**Bardach Two-Flap Palatoplasty**

Margins of the cleft are cut to separate the nasal and oral mucosa. Lateral incisions are made to the bone, medial to the alveolus and carried posteriorly to the retromolar area. Mucoperiosteal flaps are elevated bilaterally and the neuromuscular bundle is isolated. Care must be taken to avoid damage to the neurovascular bundle arising from the greater palatine foramen. Damage to this vessel would compromise blood supply to the flap. Closure occurs in three layers. Closure of the nasal mucoperiosteum proceeds from anterior to posterior. The velar musculature is reapproximated, and the oral mucoperiosteum is closed from posterior to anterior along the cleft margin. The lateral oral mucoperiosteum is then reapproximated to the medial alveolus. Keys to optimizing surgical outcomes with two-flap palatoplasty include aggressive repositioning of the soft palate muscles, adequate mobilization to create a tensionless closure, and complete palatal closure in one setting. One possible complication is oronasal fistulas, which have been reported to have an incidence from 3.4% to 29%. Other complications include maxillary growth disturbance and velopharyngeal insufficiency.

**Furlow Double Opposing Z-plasty**

Furlow double opposing z-plasty is a surgical technique for closure of the soft palate and reconstruction of the levator sling. It has the advantage of repositioning the musculature into a more anatomical location, lengthening the palate, and improving speech outcomes. As with other types of palatoplasty, fistulas can form especially in the presence of wide clefts.

Four flaps are created with two anterior mucosal flaps and two posteriorly situated mucomuscular flaps. The flaps with nasal mucosa are transposed and sutured together, followed by the flaps with oral mucosa. This recreates the muscular sling and lengthens the palate. This surgical technique can be modified for clefts involving the hard palate. It combines two-flap palatoplasty with z-plasty.

**Conclusion**

Cleft lip and palate are common head and neck congenital malformations that require a multidisciplinary approach to management of the many medical and surgical needs of the patient. Otolaryngologists play a pivotal role in the cleft care team and are capable of managing many of the problems these patients face.

**Faculty Discussion: Harold Pine, MD**

That was a very good talk Dr. Smith and it's very frustrating and difficult to understand these difficult concept for one who went through a residency and fellowship where the plastic surgeons did the majority of this work and a fellowship and it's hard to understand these concepts until you've actually seen or helped with some of these procedures and it's a tough thing to do when most of this work is actually done in another department. So when opportunities arise for you do take advantage of them because our board does consider this fair game and does generally hold you responsible for at least in general concepts about all these things and they're certainly questions every year on the inservice about clefts and genetics and so forth.
From a practical standpoint and where otolaryngologists end up being involved is really the care of their ears. I think that a number of years it was just a given without much thought that if you had a cleft palate you were going to get ear tubes and there wasn’t a whole lot of testing in between that was necessary. It was just assumed that this is what you were going to need and how it happened this is how it happened. Now, and there is literature to support this not every child will need early intervention and I certainly support since we have the people available, get these kids tested and this is another one of these problems where we’re almost always consulted early on, shortly after birth. So, not a whole lot for us to do except if there’s an airway issue but really to insure that there’s good followup with us and especially with audiology. The way we do it here generally speaking is to that we will attempt to place a first set of ear tubes at the time of a cleft lip repair which is generally to be around three to five months of age.

References