CHAPTER

1

INTRODUCTION TO CLEFT LIP AND PALATE AND ASSOCIATED SYNDROMES

If you recently learned that your baby has (or will be born with) a cleft lip and palate, you may be searching for information about what this diagnosis will mean for your child and your family. Can the cleft be repaired, and if so, when? How will the cleft affect your baby's feeding? Will the cleft have any impact on other areas of development? How will others react when they see your baby?

Unless clefting runs in your family, you probably did not expect your baby to be born with a cleft, and this unexpected development can be very tough to deal with early on. Like most parents who have just been told that their child has a problem at birth, you may have struggled (and perhaps still are struggling) with feelings of shock, grief, helplessness, and anger. It may take some time for you to adjust to this unexpected diagnosis. But as you begin this journey, you should know that there are many professionals and parents of children with cleft lip and palate you can call upon for advice and support—and we will provide you with many of those resources in this book.

Because cleft lip and palate is one of the more common problems seen in newborns, it has received a lot of attention over the years. It is a treatable condition, but one that typically does require surgery. In the early months, your child may experience some cleft-related problems with feeding and ear infections that need attention. Later, problems with speech and language development may become a priority. Fortunately, all these problems can be treated.

We believe that parents who are well informed are best able to help their child through the treatment process. This book will therefore describe the early problems that children with cleft palate may encounter as well as the early treatment that they frequently receive, and we will focus on how you, the parent, can support your child through this process.

YOUR BABY'S COMMUNICATION SKILLS

The first year of life is a time of remarkable growth and discovery for all babies. Although they begin life totally reliant on their parents, babies rapidly learn to control the movements of their body and become increasingly independent. They learn to hold their head up, manipulate objects, crawl, and walk—all important accomplishments that allow them to explore their world. Of all the achievements that parents observe their baby make during this time, though, there is none more thrilling than the appearance of the first word.

Communication does not begin with the first word. It is a skill that is refined throughout the first year of life and beyond. Before that long-awaited first word ever appears, babies have laid a great deal of groundwork:

- They have practiced the sounds of their language through babbling (and thus have a group of consonants and vowels they can use to form their first words).
- They have learned the meanings of words that are used repeatedly in their homes (and so have begun to relate spoken words to objects and people around them).
- They understand that they can use sounds and gestures to convey information and to obtain what they want (and so have begun to learn the power of communication).

A cleft of the lip and palate does not prevent a baby from learning to talk, but it can influence how quickly speech develops and will probably influence the types of sounds your baby says as well. We know that some babies with cleft palate are slower to say their first word than other babies and may be slower to add words to their vocabulary even after the palate is repaired. We also know that a large percentage of children with repaired cleft palate (up to roughly 70%) ultimately require speech therapy at some point during their toddler, preschool, or school-age years (Hardin-Jones & Jones, 2005).[1] Some may need therapy to address some of the same developmental speech-language problems that are commonly seen in children without clefts, and others may need intervention to treat cleft-related problems.

In this book, we provide practical suggestions for ways that you can enhance your child's *early* speech and language development. Research has shown that the experiences parents give their babies during the first several years of development serve as an important foundation for later speech and language development. The information provided here is intended to augment, not replace, the advice you receive from the speech-language pathologist and other professionals on your child's cleft palate team. We also provide a brief overview of surgical and dental management because the presence of a cleft requires treatment from multiple specialists (including, but not limited to, a plastic surgeon, oral surgeon, ear, nose, and throat [ENT] physician, and pediatric dentist/orthodontist) and because the treatment provided by these specialists can influence speech-language development.

CLEFT LIP AND PALATE: A COMMON PROBLEM

According to the Centers for Disease Control and Prevention (CDC, 2024a), an estimated 1 in every 33 babies born in the United States each year has a birth defect. Orofacial clefts, including clefts of the lip and/or palate, are among the most common birth defects seen in newborns, occurring in approximately 1 in every 700 to 1,500 births worldwide (Mai et al., 2019; Marazita, 2023; Stallings et al., 2024). In the United States, it is estimated that 1 in 1,050 babies are born with cleft lip and palate while 1 in 1,600 babies are born with cleft palate only (CDC, 2024c).

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[1] 1. The percentage of children needing speech-language therapy differs quite a bit across studies. The difference is probably related to the age of children studied (toddler, preschool, or school-age) and whether children enrolled in early intervention are included.

Identifying an exact occurrence rate is difficult because clefting varies across both geographic location and ethnic groups. It is higher in American Indian/Alaskan Native and Asian/Pacific Islander populations than in White non-Hispanic populations, and low in individuals of Hispanic and African descent. The overwhelming majority of babies who are born with a cleft (approximately 70% to 80% of cleft lip with or without cleft palate and 50% of cleft palate) are born with an isolated cleft—meaning there are no other birth defects (physical or developmental) present.

TYPES OF CLEFTS

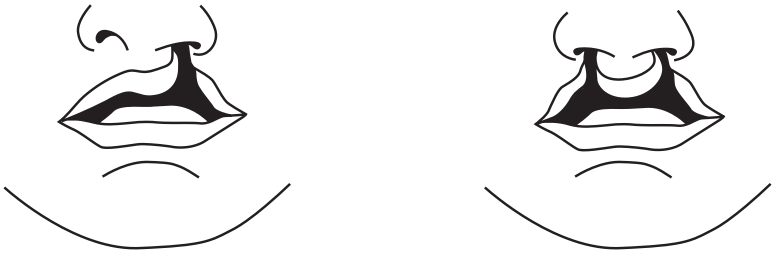
A baby's face and mouth form in the first 10 weeks after conception. During development, both sides of the face and mouth come together and fuse. A ***cleft*** (opening or gap) occurs when the tissues that form portions of the lip and palate fail to come together and fuse before the baby's birth. Clefting commonly involves some combination of

- the lip,
- the ***alveolar ridge*** (the upper gum ridge that houses teeth),
- the ***hard palate*** (roof of the mouth in front consisting of bone covered by soft tissue), and
- the ***soft palate*** (or ***velum***, the roof of the mouth in back consisting of soft tissue and muscle).

The three most common types of clefts that occur are cleft lip (CL), cleft palate (CP), and cleft lip and palate (CLP). A CL can occur on one (***unilateral***) or both (***bilateral***) sides of the lip (Figure 1.1) and can be either complete or incomplete. When a complete CL occurs, the cleft extends through the lip into the floor of the nose. In an incomplete CL, a notch appears in the lip, but the cleft does not extend all the way into the nose. The majority of cleft lips also involve a cleft of the gum ridge.

A CP can involve all or a portion of the palate. That is, the cleft may be only in the soft palate, or it may involve both the hard and soft palate. In a complete CLP, the cleft extends through the lip, gum ridge, hard palate, and soft palate. More biological male children are born with CL and CLP whereas more biological females are born with isolated CP.

Although a CL does not usually have much of an impact on anything other than a child's appearance, an unrepaired CP can influence a new-



Unilateral Cleft Lip

Bilateral Cleft Lip

Figure 1.1 Cleft Lip: Unilateral and Bilateral

born's feeding and result in an increase in ear infections. It can also influence early speech and language development.

A different type of cleft, known as a ***submucous cleft*** palate, occurs when a palatal cleft is concealed by mucosa (a thin layer of tissue). This type of cleft is not immediately obvious when looking in a child's mouth. In fact, it is often not diagnosed until a child is older and demonstrates speech that sounds excessively nasal.

To understand the impact that a submucous CP has on speech, one must first understand the role of the soft palate during speech production. Unlike the hard palate, which is made up of bone, the soft palate is made up of muscle and other tissues. When a key muscle of the soft palate (known as the ***levator veli palatini*** muscle) contracts during speech production, the soft palate is lifted up and moved backward against the back wall of the throat. This action, known as ***velopharyngeal closure***, seals the nose off from the mouth during production of the majority of speech sounds (all except the nasal sounds—the beginning sound in words like "mommy" and "no" and the ending sound in "sing").

When a child has a submucous cleft, the muscle in the soft palate has been disrupted (split) and does not function efficiently. For some children, contraction of this muscle may result in enough palatal elevation to seal off the nose if the throat is shallow (because the soft palate does not need to move a large distance). For other children, the palate may elevate but not move backward as much as needed to contact the back wall of the throat. This problem, known as ***velopharyngeal inadequacy*** (VPI) or velopharyngeal dysfunction, results in air passing from the throat into the nose (instead of the mouth) and speech that sounds excessively nasal (***hypernasal***).

Some children with a submucous cleft have normal speech and thus do not need treatment of any kind. Children with significantly hypernasal speech, however, typically need surgery. We will talk more about the problem of velopharyngeal closure when we discuss the topics of surgery and speech later in this book.

CAUSES OF CLEFTING

Clefts of the lip and palate are believed to be caused by both genetic and environmental factors. That is, ***genetic*** (inherited) factors may increase a child's susceptibility to a cleft. But clefting only occurs when the effects of multiple ***genes*** interact with each other or with certain environmental factors such as drugs (medications) that can act on the developing embryo.

Research is ongoing to isolate the genes associated with isolated clefts, and a large number of potential genes/regions (approximately

FOLIC ACID FORTIFICATION

Folic acid is a B complex vitamin. Vitamins in this complex boost metabolism, enhance immune and nervous systems, and keep skin and muscles healthy. The Food and Drug Administration (FDA) issued a mandate in 1996 that folic acid be added to all "enriched" grain products by January 1998 to help reduce the risk of spina bifida and other congenital conditions in newborns. Many popular breakfast cereals now contain the daily amount of folic acid recommended for women by the CDC.

50) have been identified to date (Babai & Irving, 2023; Marazita, 2023). A major goal of such research is to identify the risk imposed by these genes as well as gene–environment interactions. Marazita (2023) reported that "cumulatively, these ~50 genes/regions explain about 20–25% of the heritability" of nonsyndromic clefts (p. 173). Although a number of suspicious environmental factors also have been identified, few have actually been documented as increasing the risk of clefting.

Factors that have been associated with an increased risk of cleft include maternal smoking (active and passive exposure) during pregnancy, maternal pregestational diabetes, and the use of some medications for acne and epilepsy (Bernheim et al., 2006; Marazita, 2023; Merritt, 2005; Shkoukani et al., 2013). Excessive alcohol consumption during pregnancy also appears to increase the risk of CLP, but the exact association is not yet clear. Other factors such as maternal obesity and maternal nutrition have been linked to an increased risk of other birth defects, so their association with CLP continues to be questioned. It is important to recognize, however, that while these environmental factors may influence the risk of clefting to varying degrees, the interaction with an individual's genetic background may result in "different outcomes for the same environmental exposure" (Marazita, 2023).

According to the CDC, there are steps a woman can take during pregnancy to lower the risk of giving birth to a baby with a birth defect. Those steps include seeking ***prenatal*** care; getting 400 micrograms of folic acid through a multivitamin, eating fortified foods, or a combination of both; avoiding products that contain nicotine (such as cigarettes and vape cartridges); and not drinking alcohol or using "street" drugs. In addition, the

CDC cautions that women should avoid environments that could result in overheating (such as hot tubs) and seek treatment for fevers higher than 101 degrees Fahrenheit. They also advise talking to your physician before taking any over-the-counter or prescription drugs and ensuring that any preexisting medical conditions (such as obesity and diabetes) have been managed.

PRENATAL DIAGNOSIS OF CLEFT LIP AND PALATE

Prenatal diagnosis of CLP by a physician has become increasingly common in recent years. Although routine ultrasound will not identify all babies with CLP prior to birth, CL can be identified at some point around the twentieth week of pregnancy. Sometimes transvaginal sonography can detect a CL several weeks earlier. A CP that occurs without a CL is much harder to identify by ultrasound (because it is difficult to look inside the mouth with this technology).

If you have been told that you are at increased risk of giving birth to a child with a cleft, you can certainly request that a detailed ultrasound be performed to check for the problem. In contrast to the limited ultrasound that takes only a few minutes and is performed to answer a specific question (for example, checking for a fetal heart rate or amniotic fluid levels), a more extensive ultrasound can be performed at 18 to 20 weeks to answer a larger number of questions and check for specific malformations. Be aware, however, that the actual identification of CLP using ultrasound depends on many factors—including, but not limited to, the position of the fetus (whether the face is hidden), the amount of amniotic fluid surrounding the fetus, and the skill of the individual performing the test.

Prenatal diagnosis of CLP is believed by some to be a mixed blessing. On the one hand, receiving the diagnosis before your baby is born gives you the advantage of preparing for your child's birth by becoming familiar with the problem, identifying a cleft palate team, and beginning to explore treatment options. On the other hand, a prenatal diagnosis can undoubtedly increase anxiety for some parents. Surveys of parents who received a prenatal diagnosis have revealed that the overwhelming majority (>80%) found it helpful to learn about the diagnosis (Sasson et al., 2021) and meet with a surgeon/cleft palate team before their child's birth (Greives et al., 2017).

Research has shown that overall satisfaction with the support and information provided by medical professionals does not differ between

DETERMINING YOUR RISK OF HAVING ANOTHER BABY WITH A CLEFT

Many factors are considered when determining an individual's risk of giving birth to a child with a CLP. If a cleft condition has previously occurred in your family or if you have already given birth to a child with a cleft, a genetic counselor can help you identify the approximate risk of giving birth to another child with a cleft. A number of factors will be considered in determining your risk, including family history of clefting (number of relatives affected), the race and gender of affected relatives, and the type (severity) of cleft the affected family members had.

Generally speaking, when a parent already has one child with a cleft, the risk of giving birth to another child with a cleft is approximately 2% to 5%. A parent with a cleft who is the only one in their family with a cleft has a comparable risk of giving birth to a child with a cleft. The risk increases when there are multiple individuals in the family with a cleft.

mothers who received the diagnosis before their child's birth versus after the birth (Robbins et al., 2010). One potential benefit of prenatal diagnosis is that it can help with your baby's early feeding. In their study, Robbins and colleagues found that mothers who had received a prenatal diagnosis of clefting were more satisfied with the help they received in feeding their baby. The researchers speculated, though, that the parents "may have been better prepared to ask for help" (p. 480).

Whether or not your current child was diagnosed prenatally with a cleft, you may want to pursue a prenatal diagnosis if you decide to have another baby. You may also want to talk to your child's doctor about your risk of giving birth to another child with a cleft. See the box for general information on the risk of recurrence.

CLEFTING ASSOCIATED WITH OTHER CONDITIONS

Clefts of the lip and palate can occur as isolated conditions or as part of a syndrome that includes other problems. The term ***syndrome*** refers to a combination of problems and behaviors occurring together that characterize a single condition. Clefts involving the lip and/or palate are classified as syndromic clefts when they occur with other malformations (usually two or more) in a recognizable pattern; clefts that occur as isolated problems are classified as nonsyndromic clefts.

Although clefts involving the lip and palate have been associated with almost 500 syndromes (Dixon et al., 2011), not all have been associated with specific genes, and the majority of these syndromes are very rare. It has been estimated that approximately 70% of these children are born with isolated clefts, and 30% have other anomalies as well.

Even though the majority of these children do not have a recognizable syndrome, it is important to point out that an appreciable number do have additional anomalies. In a recent study of 9,403 children with CLP or CP, Fitzsimons and colleagues (2023) found that while 39% had at least one other congenital malformation (in addition to the cleft) in their health records, only 17% had malformations across two or more body systems. The prevalence of additional malformations was greater in children with CP than in those with CLP. We should point out that some of these malformations represent significant health concerns, but others may reflect relatively minor issues. Your child's cleft palate team can help you understand the significance of other anomalies that may be present as well as advise on additional treatment (if any) that may be needed.

What are the implications for your child if they have an associated syndrome? In each syndrome, there are certain medical or developmental conditions in addition to clefting that can affect your child's development. These conditions vary widely from syndrome to syndrome and can range from mild to severe. Some syndromes are identified at birth or soon after, and some may not be identified until a child does not meet expected developmental milestones.

If there are suspicions that your child may have a syndrome, your child's cleft palate team can refer you to a geneticist, a professional who can identify genetic syndromes. A genetic evaluation consists of a detailed medical and family history, physical examination of your child, and laboratory tests (for both child and parents), if recommended by the geneticist. The results of the evaluation will provide you with information that you can use to plan for your child's early intervention needs and follow-up, as well as for planning whether you wish to have additional children.

It is beyond the scope of this book to describe in detail all of the syndromes that are associated with clefting. If you have questions about a specific syndrome, consult with members of your child's cleft palate team. There are also a number of patient and family support organizations that have websites that either provide information about or are devoted to specific conditions. For example, Ameriface (www.ameriface.org) and FACES: The National Craniofacial Association (www.faces-cranio.org) provide resources (website links, discussion groups, etc.) for numerous syndromes and other conditions.

We will now provide brief descriptions of two conditions that are frequently seen by cleft palate teams.

22q11.2 Deletion Syndrome

During the past 30 years, 22q11.2 deletion syndrome (***22q11.2DS***), a relatively common syndrome, has received a lot of attention. Individuals with this syndrome have palatal problems, heart problems, and a characteristic facial appearance, as well as learning disabilities and many other associated problems. This syndrome, which occurs in approximately 1 in 2,000–4,000 births (Cortes-Martin et al., 2022; Fung et al., 2015), is the most frequently occurring syndrome involving clefts or VPI. The term 22q11.2DS is a collective term that encompasses other conditions (such as velocardiofacial syndrome and DiGeorge syndrome) because all involve the deletion of the same genetic material on chromosome 22.

Chromosomes are threadlike structures located in our cells that contain genes. Ordinarily, people have 23 pairs of chromosomes in each cell, with one chromosome in each pair inherited from the father and one from the mother. Specific conditions occur when chromosomes are damaged, duplicated, or missing. With the diagnosis of 22q11.2DS, children are typically missing a small portion (microdeletion) of chromosome 22 (22q11.2 is the specific location). When this syndrome is suspected from the combination of clinical findings that are present (heart disease, VPI, facial characteristics), a special blood test (FISH: fluorescence in situ hybridization) can be performed to confirm the specific microdeletion. If FISH analysis does not identify a deletion, more sophisticated tests can be performed.

Infants with 22q11.2DS often have significant feeding problems. These difficulties may be related to a number of factors including (but not limited to) problems with low muscle tone and maintenance of the airway, weakness associated with heart disease, VPI (and the subsequent difficulty with sucking), and slow emptying of food from the stomach with chronic constipation.

Virtually all children with 22q11.2DS have speech-language, developmental, and motor problems throughout childhood. They begin talking later than other toddlers, produce fewer consonant sounds, and may not begin combining words to produce short sentences until after 2 years of age. Some children benefit from learning sign language until their speech skills improve enough that they can be understood. Although children with this syndrome initially have trouble making themselves understood due to the small number of different consonants they produce, their speech and motor skills often develop more quickly between the ages of 3 and 5 years.

In school, children with 22q11.2DS may make adequate academic progress early on, but they have increasing difficulty as learning becomes increasingly abstract. They tend to have more difficulty with arithmetic than with reading or spelling (although their performance in each of these areas tends to lag behind that of their peers). Computer work and music have been identified as areas of strength. Children with 22q11.2DS usually need ongoing intervention because of the complex nature of their problems. Because the syndrome is associated with many different problems and each child may have a different combination of these problems, the syndrome affects children differently.

There are a number of strong treatment and advocacy groups across the country that provide publications and other resources for individuals with 22q11.2DS and their parents. The names and addresses of some of these foundations and treatment centers are included in the Resources section at the back of the book.

Pierre Robin Sequence

Pierre Robin sequence (also known simply as Robin sequence) is a condition that includes ***micrognathia*** (a small lower jaw), a wide cleft palate, and ***glossoptosis*** (tongue positioned in the back of the mouth, blocking the airway). The term “sequence” refers to the sequence of events that occur as a result of poor growth of the lower jaw. When the jaw does not grow appropriately in utero, the tongue cannot drop from its position between the two palatal shelves, the palatal shelves cannot come together, and a cleft palate occurs. In addition, poor growth of the lower jaw results in the tongue being positioned further back in the mouth, which may obstruct the airway.

Although Pierre Robin sequence can occur as an isolated condition, some cases occur as part of a larger syndrome. In fact, 22q11.2DS is one of the syndromes most commonly associated with Pierre Robin sequence.

A clinical examination may be all that is needed to identify the condition when it occurs in isolation, but it is a good idea to obtain a genetic evaluation to rule out the possibility of an associated syndrome.

For babies with Pierre Robin sequence, maintaining an open airway is an early priority. Because the normal-sized tongue is large compared to the small lower jaw, it tends to fall backward into the airway and interfere with breathing. Depending on the severity of the problem, the baby's surgeon may recommend treatments that range from simple adjustments in positioning to surgeries that either anchor the tongue temporarily in the front of the mouth or lengthen the lower jaw to bring the tongue forward.

Feeding a baby with Pierre Robin sequence can be challenging. Even once the airway problem has been resolved, the small lower jaw and the cleft palate can make it difficult for the baby to suck and latch onto a nipple effectively. We will address some of the feeding problems experienced by these babies in Chapter 3.

As children with Pierre Robin sequence get older, speech problems associated with the cleft palate may become evident. In general, though, these children do not typically have other developmental problems unless the sequence is associated with a larger syndrome.

SUMMARY

CLP is a common congenital (present at birth) condition that is believed to be the result of both genetic and environmental factors. Although clefts involving the lip can be identified on ultrasound by 20 weeks, prenatal diagnosis of cleft palate is more difficult and less reliable. CLP is a complex problem that requires treatment from a number of different professionals. We will discuss these specialties and the interdisciplinary team in the next chapter.