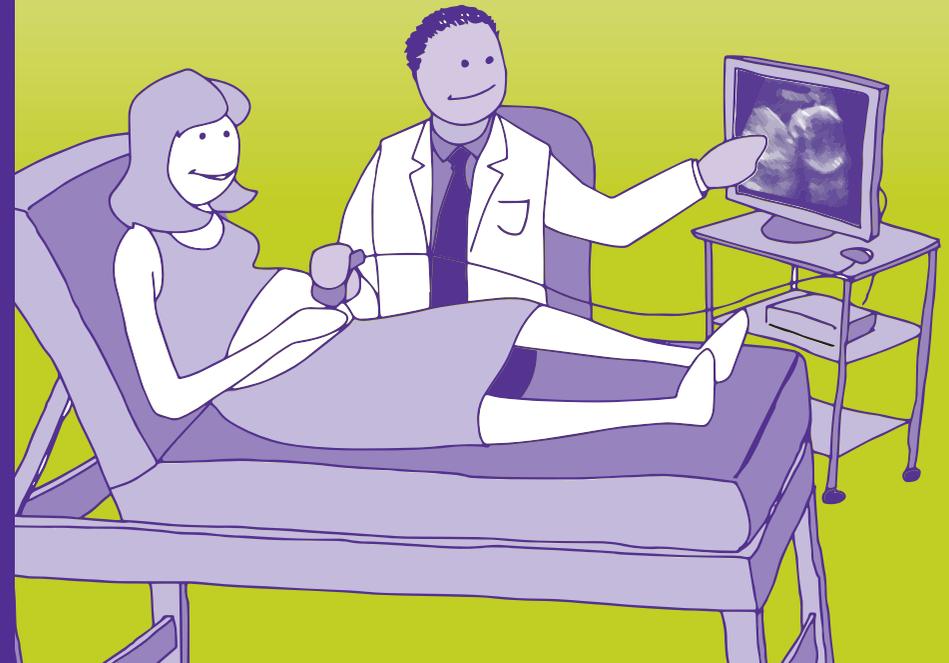




Prenatal Diagnosis of Cleft Lip and Cleft Palate



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Prenatal Diagnosis of Cleft Lip and Cleft Palate



First Edition
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Foreword

We hope this booklet will help support and educate your family. We hope it will help you:

- better understand what a prenatal diagnosis is;
- make informed decisions during your pregnancy;
- prepare for the birth of your baby;
- understand why working with a cleft palate treatment team is so important.

More than anything, we hope this booklet helps you and your family manage the enormous range of thoughts and feelings that often accompany a prenatal diagnosis of cleft. Looking back on their own prenatal diagnosis or birth of a child with a cleft, families with older children tell us that eventually, when the tide of emotions began to calm, they were ready to move on to learn about baby's needs, cleft care and most importantly, reclaim the excitement of welcoming the family's newest member.

Please note that many technical and medical terms appear in **bold** and are defined in the glossary at the end of this booklet.

Prenatal Diagnosis

What is prenatal diagnosis?

A prenatal diagnosis is information about the health and condition of your unborn baby. To learn more about your baby, your doctor or hospital may use a variety of tools and methods including:

- blood tests;
- listening to fetal heart beat;
- physical examination of the mother;
- ultrasound.

Why choose prenatal diagnosis?

Expectant parents might choose to have prenatal diagnosis to:

- learn baby's gender;
- confirm baby's due date;
- make sure baby is healthy;
- identify conditions that may affect baby before and after birth;
- help plan for labor, delivery, and post-delivery care if baby has special health needs.

Learning about baby's health before baby is born helps families and doctors prepare for any special needs that baby may have. You may be reading this booklet because your family is expecting a baby with a cleft lip and/or palate. Information from a prenatal diagnosis can help you prepare for the birth of your family's newest member.

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What is a cleft lip and/or palate?

A **cleft** is a gap in the lip and/or **palate** (roof of the mouth). It may include the **alveolus**, (bones of the upper jaw and/or upper gum). Clefts occur when the two sides of the lip or palate do not grow together during baby's development very early in pregnancy. Babies may be born with a cleft lip, a cleft palate, or both.

Cleft lip usually occurs around the fifth week of pregnancy, before many women even know they are pregnant. A **cleft palate** usually develops around the ninth week of pregnancy. Once the cleft has developed, it will remain throughout pregnancy and will be part of baby's first smiles. A cleft lip, with or without cleft palate, will change the appearance of baby's face. Most clefts can be repaired so that baby may eat and speak without major difficulties. Many clefts can be repaired with little scarring.

What causes a cleft?

Clefts occur because something in the environment and/or **genetics** (a family trait) has affected facial development. It is often difficult to determine the exact cause of a cleft. Clefts occur in all racial and ethnic groups. They occur slightly more often in people of Asian descent than among Caucasian and least often in people of African descent.

How common are clefts?

Facial clefts occur in about 1 of every 600 births in the United States, making them one of the most common birth differences. Left-sided clefts are more common than right-sided clefts. Some clefts are **bilateral**, affecting both sides of the mouth. About one-third of clefts involve the lip and **alveolar ridge** (upper gum). Two-thirds of all clefts extend through the entire palate. Some clefts may involve the lip and soft palate but not the hard palate.

Facial clefts occur in about 1 of every 600 births in the United States, making them one of the most common birth differences.

How will my doctor determine if my unborn baby has a cleft?

Your doctor may use **ultrasound** (also called a sonogram) to determine if your unborn baby has a cleft. An ultrasound uses sound waves to create a picture of your growing baby. The pictures often appear in shades of gray. Darker gray areas are liquids, like amniotic fluid. Lighter gray areas are solids, like bones and teeth. Most ultrasounds are two-dimensional (2D), but your doctor may use 3D or 4D ultrasounds, which show even greater detail. Baby's size and position are ideal for ultrasound between week 18 and week 26 of pregnancy.

Ultrasound quality is not always as clear as we would like due to a number of factors, including:

- the type of machine used;
- the skill and experience of the ultrasound professional;
- the weight and health of the mother;
- the stage of the pregnancy;
- the position of the baby at the time of ultrasound.

Can clefts be accurately identified through prenatal diagnosis?

In the best conditions, ultrasound can return pictures of cleft lip as early as the 20th week of pregnancy. Sometimes, ultrasound can also help parents and doctors see clefts of the alveolar ridge.

It is more difficult to get clear pictures of baby's palate. Doctors miss more cleft palates on ultrasound than they find. As technology improves, identifying clefts of the palate will become more common. Larger hospitals and university medical centers are good resources for the most advanced ultrasound technology and prenatal diagnosis expertise.

How often clefts are prenatally diagnosed?

The number of accurate prenatal diagnoses continues to increase. The National Birth Defects Prevention Study (1998-2004) reported that about 23% of babies born with a cleft had a prenatal diagnosis. The last year of the study, 2004, that percentage rose to 34%.

What does prenatal diagnosis of a cleft mean for my baby?

Once a prenatal diagnosis of a cleft is made, your doctor will determine if the cleft is your baby's only expected birth difference. In this case, the cleft would be called an **isolated or non-syndromic cleft**. If your doctor notes

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additional areas of development that may be affected, the cleft is called a **syndromic cleft**, meaning it is part of a syndrome. Most clefts of the lip and palate are isolated. Your doctor will need to gather more information to determine whether a cleft is syndromic or non-syndromic, including:

1. Family history of clefts.

Your doctor will ask questions about your family's general health history. Questions will include those about relatives who have clefts or other related birth differences. If your baby's mother, father, or grandparent had an isolated cleft lip and/or palate, baby may also have an isolated or non-syndromic cleft. If no one in baby's family has had a cleft of the lip and/or palate, (called a negative family history), more information will be needed to determine if the cleft is isolated or part of a syndrome. This is the case in most pregnancies.

2. Maternal health issues.

Clefts are more likely to occur when mothers:

- *Use alcohol and tobacco during the first 10 weeks of pregnancy.*
- *Take certain medications during the first 10 weeks of pregnancy.* For example, medicines prescribed for epileptic seizures have been linked to an increased risk of clefts. Talk with your doctor about medications and pregnancy. A good source of information about medication risks and pregnancy is MotherToBaby, a service of the non-profit Organization of Teratology Information Specialists, www.otispregnancy.org, or 866-626-6847.

- *Have diabetes.* If you have poorly managed diabetes, your baby may have an increased risk for several birth differences. If you manage your diabetes well, your baby will have the same risk of birth differences as babies born to mothers without diabetes. Some cases of gestational diabetes are actually Type II Diabetes first recognized in pregnancy. An HgA1C test may be helpful in assessing diabetic management. Ask your doctor if you might need this test.
- *Have a folic acid deficiency.* A deficiency in folic acid may result in increased risk of clefts. Even if you are getting enough folate from your diet, your body may not absorb the vitamin. This is sometimes related the use of other medications (such as anti-seizure medications). Some Latina women (8-10%) cannot absorb folic acid, so may be at higher risk of giving birth to a baby with a cleft. For more information about folic acid, visit www.cleftline.org and click on the Links tab at the top of the page.

3. **Associated abnormalities.**

If prenatal tests show birth differences in addition to the cleft, your baby may be diagnosed with a **syndrome**. A syndrome is a group of signs or symptoms that often go together. Your doctor may be able to explain how these symptoms affect your baby's growth and development. Many times, causes of birth differences are unclear. Your doctor may suggest additional tests to gather more information.

4. **Chromosomal abnormalities.**

Chromosomes are the packages that carry genetic information. If prenatal tests show more than one birth difference, your doctor might suspect a **chromosomal abnormality**. A normal human cell has 46 chromosomes. Some babies with multiple birth differences have extra chromosomes (duplication), a missing piece of a chromosome (deletion), or a rearranged chromosome. Children with chromosomal abnormalities often have developmental delays or learning problems. *Isolated clefts are not associated with chromosomal abnormalities.*

Factors that increase the risk for a chromosomal abnormality include:

- advanced age of the mother;
- abnormal first or second trimester screening results; and
- the presence of ultrasound **soft markers**, which are small differences that your doctor may see during an ultrasound examination that suggest an increased risk for problems such as chromosomal abnormalities.

How often are babies with cleft lip and palate also diagnosed with a syndrome or chromosomal abnormality?

Of all babies with cleft lip and palate, 10% to 15% are diagnosed with a syndrome or chromosomal abnormalities.

Of all babies with cleft lip and palate, 10% to 15% are diagnosed with a syndrome or chromosomal abnormalities. If tests report that chromosomes are normal, the risk for a syndrome drops to 8% to 10%.

Should I have additional prenatal testing?

Cleft lips and palates can be repaired. *Chromosomal abnormalities cannot be repaired.* The decision about additional testing for chromosomal abnormalities depends on how you might use the information you learn from it. Some questions to consider might include:

- If additional prenatal testing reports that baby may not survive long after birth or will have significant physical and/or developmental issues, will I continue the pregnancy?
- Will additional prenatal testing allow my family and me to prepare and educate ourselves about caring for a baby with special needs?
- If additional prenatal testing includes those that carry risk of miscarriage (an *amniocentesis* is one example), how do I the balance of need for more information with the risk of miscarriage?

Discuss your questions and concerns with your doctor. Your doctor has knowledge of your specific situation and is a good source for information, guidance, and support in your decision-making process.

Your doctor may recommend **amniocentesis** (taking a small amount of amniotic fluid from the sac around the baby) or other tests to learn more about your baby's chromosomes. Some of these tests have a small but real risk of causing a miscarriage.

Planning Care for a Child with a Cleft Lip or Cleft Palate

What is a treatment team?

A treatment team is a group of cleft specialists who work together with families and patients to design a treatment plan for a child's cleft care. The best care for a child with a cleft or other craniofacial birth difference happens when a family works with a cleft palate or craniofacial treatment team. The treatment team includes the child, the child's family, several doctors, and other cleft-care specialists. Team care begins when the prenatal diagnosis is made and is vital through the first days learning to feed a baby with a cleft. Your family's relationship with a treatment team will support you and your child throughout childhood, the teenage years and beyond. Some of the cleft-specialists that contribute to team care may include:

- a *surgeon* (such as a plastic surgeon, an oral/maxillofacial surgeon, a craniofacial surgeon, an otolaryngologist, or a neurosurgeon);
- a *speech-language pathologist* who assesses speech and feeding problems
- a *pediatric dentist* or other dental specialist
- an *orthodontist* who straightens the teeth and aligns the jaws;
- a *geneticist* who screens patients for craniofacial syndromes and helps parents and adult patients understand the chances of having more children with these conditions;
- a *nurse* who helps with feeding education and monitors child's health;
- a *psychologist* or *other mental health professional* who supports your child and family's emotional needs;
- an *audiologist* who assesses hearing;
- a *pediatrician* who monitors overall health and development; and
- other specialists.

Coordination between the family, child, and treatment team about goals for care and treatment lead to the best outcomes for the child. As your child grows, your child will be assessed by the treatment team every 1-2 years through young adulthood.

How do I sort through my feelings?

While each parent and family member will experience his or her own personal emotions and reactions to the prenatal diagnosis, the recovery process for each may have lots in common:

- *Disbelief*, confusion, fear or similar responses may be the first feelings to flood in after a prenatal diagnosis.

Consult with a cleft palate treatment team as soon as you know your baby has a cleft. Find a team by contacting us: **The Cleft Palate Foundation**
1.800.242.5338
www.cleftline.org

- *As the reality* of the diagnosis becomes clear, many experience a sense of loss and sadness. Viewing ultrasound images may be helpful at this stage.
- *Naming the loss* helps family members actively mourn whatever the loss(es) may be. Recognizing that baby's face may look different than imagined (naming the loss) and recognizing the feelings associated with that difference (actively mourn) is one example.
- *Recovery* moves families into problem-solving. As emotions peak then settle, families may be then be ready to educate themselves about clefts and cleft care.

We encourage parents and families to treat themselves and their loved ones with extra care and patience during this recovery process. Families who have had similar experiences tell us that once the tide of emotions began to calm, they were ready to learn more about their baby's needs and cleft care and reclaim the excitement of welcoming baby into their family.

What should I do in the meantime?

- 1) *Take time to think about this new information.* You knew you were expecting a baby. Now you know something specific about him or her. Give yourself time to digest this information.
- 2) *Pay attention to how you are feeling.* Notice your feelings and how they may change over time. You may feel a range of emotions including disappointment, anger, grief, guilt, or even relief over your child's diagnosis. It is okay to talk to your loved ones about these feelings, and it's also okay to take some time alone to process them yourself.
- 3) *Adjust to the new information.* Though some things may feel very different now that you know your baby will be born with a cleft, many things will remain the same. You will continue to prepare for your baby's arrival with excitement, but may need to make some adjustments to your plans. For instance, your baby will likely need to be fed by bottle because of his/her cleft, rather than fed by breast.
- 4) *When you're ready, begin to educate yourself.* As you adjust to the new information, your emotions may require less attention, allowing you time and energy to address some of the questions you may have. As your expertise in clefts and cleft care grows, we hope that your confidence in planning for the future will grow as well.
- 5) *Expect that others will have as many questions as you have.* Think about helpful ways to respond to curious questions from both strangers and friends. Brief responses to questions may help others quickly learn

about your baby's condition and allow you more time to show off your beautiful new family member.

- Keep your responses short by providing a very brief explanation. For example, "My baby was born with a cleft, it will be repaired when he/she is a little bigger." Practice until you come up with a response that feels right to you.
- Try to focus on the positive in your explanation; your child hears and experiences your responses far more than he or she notices questions from others!
- Remember that a cleft is only one small feature of your child.
- Your child's cleft will not stop him or her from experiencing all the ups and downs of childhood.
- Your dreams for your child's future do not need to change because of the prenatal diagnosis of a cleft.

6) *Begin to learn what you can do with this new information.* This may be the best part! When you are ready to begin problem-solving, your craniofacial team will give you the information you need to help your child. In fact, you may eventually feel like a cleft-care expert yourself.

7) *Make use of your craniofacial treatment team and all its resources.* If you have received a prenatal diagnosis of cleft lip or cleft palate, discuss your concerns with your craniofacial team. Your treatment team should be a primary resource throughout your baby's infancy and well into childhood. Some of the benefits of team care include:

- feeding education and support;
- connection with other parents and children affected by clefts;
- parent engagement in treatment planning;
- education about speech development and clefts;
- pre- and post-surgical support.

Your treatment team should be your first resource for questions, care, and planning!

Looking Ahead

In preparing for your baby's birth, you have done one of the most important and positive things possible: You have begun educating yourself about prenatal diagnosis and its benefits for you and your family.

At this moment, you may feel as if most of the attention is focused on the cleft rather than on the birth of your baby. Our hope is that as you continue to learn about caring for a child with a cleft, the cleft itself will become less important. This will make room for plans and dreams for life with your baby and the child and person he or she will become.

If you are expecting a baby with a cleft, other booklets in this series may be useful to you as you plan for baby's arrival:

Feeding Your Baby (booklet and video series)
Your Baby's First Year
Preparing for Surgery
Genetics and You

You'll find copies of these and other publications at our website, www.cleftline.org. To request complimentary printed copies, call 1.800.24.CLEFT or send an email to info@cleftline.org. We look forward to hearing from you and supporting you and your family in your cleft care journey.

For More Information

Las publicaciones de la Fundacion del Paladar Hendido tambien se ofrecen en espanol. Favor de llamarnos para recibir copias en espanol.

This publication and many others have been produced by:

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The Cleft Palate Foundation (CPF) maintains a growing collection of booklets and fact sheets that present an introduction to and explanation of many elements of cleft and craniofacial care and treatment. All publications are authored and regularly revised by representatives of professional disciplines serving the field of cleft and craniofacial care and treatment.

A publications order form for institutions including current pricing, bulk order rates and shipping and handling fees may be accessed at the CPF website or by calling the ***Cleftline*[™]** at 1.800.24.CLEFT. All fact sheets are available at the website as open-access, PDF documents. Families, patients, students and other individuals may request complimentary packets of publications by emailing info@cleftline.org or by calling ***Cleftline*[™]**.

To date, the Cleft Palate Foundation has shared over 10,000 custom-made ***Gund*[™]** Teddy Bears with repaired cleft lips for children and families all over the world. Please visit www.cleftline.org or call ***Cleftline*[™]** for more information about CPF's Teddy Bears.

You can help us continue our mission! Please consider a contribution to the CPF ***Cleftline*[™]** Fund. Visit www.cleftline.org or call ***Cleftline*[™]** today! Thank you.

HOPE AND HELP ARE ON THE LINE

GLOSSARY

ALVEOLUS - The bones of the upper jaw and upper gum.

AMNIOCENTESIS - Testing a small amount of amniotic fluid removed from the sac around the unborn baby.

BILATERAL CLEFT - A cleft affecting both sides of the mouth.

CLEFT - A separation of the parts of the lip or palate (roof) of the mouth. A cleft happens in the early weeks of prenatal development (between weeks 5 and 12 of pregnancy). It may include the alveolus.

CHROMOSOMAL ABNORMALITY - A normal human cell has 46 chromosomes. Some babies with multiple birth differences have extra chromosomes (duplication), a missing piece of a chromosome (deletion), or a rearranged chromosome. Children with chromosomal abnormalities often have developmental delays or learning problems. Isolated clefts are not associated with chromosomal abnormalities.

GENETICS - The study of genes, heredity and family traits.

ISOLATED or NON-SYNDROMIC CLEFT - The only expected birth difference for a child with a cleft.

ULTRASOUND (also called a sonogram) - An ultrasound uses sound waves to create a picture of your growing baby and may help determine if your unborn baby has a cleft.

SYNDROMIC CLEFT - A cleft that is one symptom among other symptoms that combine as parts of a syndrome.

SOFT MARKERS - Small differences that your doctor may see during an ultrasound that suggest an increased risk for problems such as chromosomal abnormalities.

TREATMENT TEAM - A group of healthcare professional specializing in cleft and craniofacial care. Treatment teams work together with families and patients to design a treatment plan for a child's cleft care.

UNILATERAL CLEFT - A cleft that affects one side of the mouth.

