Patient Information Sheet
Cleft Lip with or without Cleft Palate

You have recently learned that your baby has a cleft lip with or without cleft palate. We expect that you may have questions about what this could mean for your developing baby. Hopefully, this information will address some of your concerns; however, we encourage you to speak to your genetic counselor or healthcare provider for more information. It is important to remember that anyone can have a baby with a birth defect. Birth defects occur in 2-5% of all births and are rarely caused by something that the parents did or did not do before or during pregnancy. Currently, there are no tests available to detect all health problems

What is cleft lip with or without cleft palate?

A cleft lip is an opening of the lip that may or may not extend to the roof of the mouth (palate). These openings are normally found in all babies during early development. However, these areas should close around the 5th or 6th week of pregnancy. If they do not close, this results in either a cleft lip alone or a cleft lip with a cleft palate. A cleft lip may be on one side of the lip (unilateral), both sides of the lip (bilateral) or in the center of the lip (midline). Since clefts of the palate are very difficult to detect on prenatal ultrasound examination, it may not be known at this time if your baby only has a cleft lip or has a cleft lip with cleft palate.

What causes cleft lip with or without a cleft palate?

Although many babies only have cleft lip with or without cleft palate and no other problems, it is thought that between 35% and 66% of children with cleft lip with or without cleft palate may have additional problems. If your baby has additional problems, it could be because your baby has a chromosome problem or a genetic syndrome. If the cleft is isolated (no other problems found), then it could have been caused by multifactorial (complex) inheritance, meaning that some environmental effects of unknown origin combined with a certain genetic predisposition caused the cleft.

In order to understand a possible chromosomal cause or genetic syndrome cause, it is helpful to know a little about chromosomes. Chromosomes are packages of genetic information that carry the instructions (genes) necessary for our growth and development. Typically, there are 23 pairs of chromosomes in each cell of our bodies. One copy of each pair is from our mother and one copy of each is from our father. The first 22 pairs of chromosomes are the same in males and females. The last pair is known as the sex chromosomes and they determine our gender. Females typically have two X chromosomes and males usually have one X and one Y. It is important to have the correct amount of genetic material to have typical development. Sometimes there is an imbalance of genetic material that may cause birth defects. The imbalance may be caused by an extra or missing whole chromosome or sometimes only by extra or missing pieces of chromosomes. Some genetic conditions are caused by changes in one or more genes in our genetic make-up that result in a particular set of characteristics or features (also called a syndrome). A genetic imbalance or genetic syndrome can produce birth defects and physical changes such as cleft lip. Rarely, certain medications or other exposures during early pregnancy may cause clefting.

How many babies are found to have cleft lip with or without cleft palate?

An average of 1 in 1,000 (0.1%) babies is born with a cleft lip. This number can be higher or lower, depending upon ethnic background. Cleft lip with or without cleft palate occurs almost twice as frequently in males than in females.

What problems could be associated with cleft lip for my baby?

The extent of problems that a cleft lip with or without palate could cause for your baby depends upon the underlying cause and whether or not your baby has any additional birth defects. Unfortunately, even with additional prenatal testing, physicians may not be completely sure if your baby has any additional problems until he or she is examined after delivery. In babies with only a cleft lip, the prognosis is quite good. Surgery will usually be performed to close the cleft in the lip early in life. However, some babies may have difficulty with feeding prior to surgery. Some children will require more than one operation to repair the cleft. Some children with cleft lip may also have speech and/or dental/orthodontic problems later in childhood. Children with a cleft palate may have recurrent ear infections. Some children may also benefit from psychological counseling.

What further testing will be offered and what will these tests tell me?

Depending on your current gestational age, a number of additional tests may be offered to you. If the cleft lip with or without cleft palate is found during a routine ultrasound, a more detailed ultrasound will usually be performed to look carefully at the baby's anatomy. A specialized ultrasound of the heart, called a fetal echocardiogram, may also be recommended.

Chromosomal problems can be diagnosed prenatally through a test called amniocentesis. Amniocentesis is a procedure in which a needle is inserted through the mother's abdominal wall, through the uterus, and into the amniotic sac under ultrasound guidance. A small amount of amniotic fluid is withdrawn. Cells that have been shed by the developing baby are found in this fluid and can be

grown (cultured) in a laboratory. The chromosomes within these cells can be analyzed for abnormalities by looking at the number and structure of the chromosomes as well as reading the chromosomes in more detail by a test called a chromosomal microarray (CMA). This test can detect imbalances in chromosomes caused by small pieces that are extra or missing. The results of an amniocentesis are generally available within about two weeks. Because amniocentesis is an invasive procedure, there is a small risk, likely less than 1%, for complications that can lead to miscarriage. The risks, benefits, and limitations of amniocentesis should be discussed with you by your genetic counselor or health care provider. Follow-up counseling and referrals for support can be made if a chromosome problem is detected prenatally. As with all situations in which amniocentesis is discussed, it is your decision whether or not this test is done.

Non-invasive prenatal testing, or NIPT, is a type of screening test for certain chromosome conditions. During pregnancy, some of your baby's chromosome material is in your blood, along with your own chromosomes. NIPT is a blood test that measures the amount of chromosome material in your blood to determine if your baby could have an extra chromosome, such as in Down syndrome where there is an extra 21st chromosome. This test also screens for two other more severe chromosome conditions, trisomy 13 and trisomy 18, and may screen for some less severe conditions that are caused by different numbers of the X or Y chromosome. NIPT is a highly accurate screen; however, it is not a diagnostic test and it can only screen for a few common chromosome conditions. The detection rate for these chromosome conditions is typically between 90-99%. Other screening blood tests (often called a triple, quad, penta, or first trimester screen) may also be offered. Your genetic counselor or health care provider can discuss these various screening options in more detail.

You will likely be asked questions about any family history of clefting or other physical changes associated with a genetic form of clefting. If you have not already done so, you may be offered an appointment to speak to a medical geneticist (a doctor specializing in genetics and birth defects). He/She may briefly examine your lips, teeth and sense of smell to determine if you or your partner has a mild manifestation of a genetic form of clefting.

After the baby is born, it is important that your baby be thoroughly examined by a geneticist. This examination will help to determine if the cleft is part of a syndrome. If a chromosome analysis was not performed by amniocentesis, the geneticist may recommend that a small amount of blood be taken from your baby to see if your baby has a chromosome problem. This examination and possible additional tests are important, since an accurate diagnosis will provide you with the best information regarding the prognosis for your baby, the cause of the cleft, the potential recurrence risks to future children and that of other family members, as well as to determine appropriate tests to be offered to you in any future pregnancies. When a cleft lip with or without cleft palate is isolated and unilateral, the recurrence risk in future pregnancies is 3-5%. The risk is higher for bilateral clefts (6-8%).

Some studies suggest that taking a specific vitamin, folic acid (also known as folate), prior to and during the first few months of pregnancy may reduce the incidence of clefting. It is unknown, however, if this prevents or reduces the chance of clefting for those who have already had a child with a cleft lip. It is suggested that you discuss folic acid supplementation further with your doctor prior to planning any future pregnancy.

What do I do now?

This information is only intended as an introduction to some of the terms and tests that you have already heard or will be hearing about from your genetic counselor and healthcare provider. We hope that this information sheet will be helpful as you begin to understand more about cleft lip. We understand that any time something of concern is found through prenatal screening and testing, parents are going to be worried. Please don't hesitate to contact your genetic counselor with any questions or concerns you have. We are here to help you and your baby.

You may also find it useful to have a consultation with a craniofacial team, pediatric surgeon, and/or neonatologist prior to delivery.

The decision to have one or more of these tests or to do no additional testing is a difficult and personal one. There is no one right decision. Some people will decide to have no further testing because they do not feel they need to know if their baby will have any of these conditions before delivery. Other people feel that they want more information as soon as possible. After gathering all of the information you need about cleft lip, you should make the decision that is right for you and your family.

Resources:

1. Cleft Palate Foundation (CPF), 1504 East Franklin Street, Suite 102, Chapel Hill, NC 27514-2820 Tel. 919-933-9044, 1-800-24-CLEFT, fax 919-933-9604, www.cleftline.org, email: info@cleftline.org
2. National Foundation for Facial Reconstruction. 317 East 34th Street, Room 901, New York, NY 10016, Tel. 212-263-6656, Fax 212-263-7534, www.nffr.org, email: info@nffr.org
3. Public Health Education Information Sheet "Cleft Lip and Palate" published by: March of Dimes Birth Defects Foundation; 1-888-MODIMES or online at www.marchofdimes.com