Patient Information Sheet
Clubfoot

You have recently learned that your baby has a clubfoot. 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 a clubfoot?
Clubfoot is an abnormal position of the foot where it is turned inward. There are several different forms of clubfoot and it may affect one or both feet.

What causes a clubfoot?
Clubfoot can be the result of an abnormal position of the baby, lack of amniotic fluid, a genetic syndrome, a chromosome problem, a neurological problem, or other unknown causes. If the clubfoot is isolated (no other problems found), then it is typically thought to be caused by multifactorial inheritance, meaning that some environmental effects of unknown origin combined with a certain genetic predisposition caused the clubfoot. In many cases the cause of clubfoot cannot be determined.

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 clubfoot.

How many babies are found with a clubfoot?
About 1 in 1000 (0.1%) babies are born with a clubfoot and it is seen more often in boys than girls.

What problems could be associated with a clubfoot for my baby?
The extent of problems that a clubfoot 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 an isolated clubfoot, the prognosis is quite good. Treatment options vary and may range from no treatment necessary to physical therapy, casts, surgery or a combination of each. It may be beneficial to discuss these options with an orthopedic surgeon.

What further testing may be offered and what will it tell me?
If the club foot is found during a routine ultrasound, then a more detailed ultrasound will usually be performed to look carefully at the baby's anatomy. An ultrasound will only modify your chance of having a baby with a chromosome problem or genetic syndrome. An ultrasound <u>cannot</u> diagnose or rule out the presence of a chromosomal problem or genetic syndrome.

Depending on your gestational age, two diagnostic testing options may be available: chorionic villus sampling (CVS) or amniocentesis. These are tests in which a small sample of placental tissue or amniotic fluid is obtained to examine the baby's chromosomes. Because these procedures are invasive, there is a risk, likely less than 1%, for complications that can lead to miscarriage.

If you choose to pursue diagnostic testing you may be offered a test called a chromosome microarray (CMA). CMA is used to identify small missing or extra pieces of the baby's chromosomes that may be associated with genetic conditions. CMA cannot detect all genetic conditions and may detect variations in the chromosomes that have uncertain clinical significance (VUS). Parental blood samples may help to clarify the meaning of a variation, but effects of these changes may not be known until after the baby is born. CMA may also detect information such as non-paternity and close relationships between parents.

If diagnostic testing is declined, there are several screening tests available in the first and second trimester. Screening tests can modify your risk for a chromosome abnormality, such as Down syndrome, and involve no risk of complications to your pregnancy. A screening test will not determine if your baby has Down syndrome, but it may help you decide whether or not you would like to pursue a diagnostic test.

Non-invasive prenatal testing, or NIPT, is a 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. 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.

The risks, benefits and limitations of screening and diagnostic testing options should be discussed with you by your genetic counselor or other health care provider. Follow-up counseling and referrals for support can be made if a chromosome condition or other genetic condition is detected prenatally. As with all situations in which prenatal testing is discussed, it is your decision whether or not this test is done.

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 a clubfoot. 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.

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 a clubfoot, you should make the decision that is right for you and your family. Please keep in mind that not all babies with a clubfoot will have the complications described in this information sheet. Though it is never possible to predict the long-term prognosis for a child with even an isolated clubfoot, most babies are born without additional medical complications.