Patient Information Sheet
Intracardiac Echogenic Focus (ICEF)

You have recently learned that your baby has an intracadiac echogenic focus (ICEF). 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 an intracardiac echogenic focus (ICEF)?

An ICEF is seen on ultrasound as an echogenic area, or bright spot, in the fetal heart. The focus is most often located in the left ventricle of the heart, but can sometimes be seen in the right ventricle or other areas of the heart. The left ventricle is one of the lower chambers of the heart.

How many babies are found with ICEF?

This is not an uncommon finding on ultrasound and some studies have shown it may be found in as many as 20% of babies, depending upon your ethnic background.

What problems could be associated with an ICEF for my baby?

An ICEF most likely represents normal variation in development. In an otherwise healthy baby, the ICEF is not expected to cause any harm to the baby and does not affect the structure or function of the heart. It does not signify a heart defect in the baby.

There have been some studies that suggest there may be an increased risk for a chromosome abnormality, such as Down syndrome, when a baby is found to have an ICEF. 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 ICEF.

Down syndrome is a specific chromosomal abnormality caused by an extra copy of chromosome #21. As a result, individuals with Down syndrome often have characteristics in common including a flattened facial appearance, up-slanting eyes, a single crease in the palm of the hand, low muscle tone, and short stature. Individuals with Down syndrome typically have mild to moderate intellectual disabilities and are at an increased risk for medical problems, such as heart and bowel defects. Despite their challenges, children with Down syndrome are more like other children than different. Today, there are more opportunities than ever before for individuals with Down syndrome to develop their talents and abilities. For example, more teens and adults with Down syndrome are graduating from high school, finding employment and living independently. This is not something that can be predicted prenatally or even in the early years of life.

An ICEF is most concerning if there are other differences identified on ultrasound or if you are already at an increased chance for having a baby with a chromosome condition because of maternal age (age 35 or greater) or abnormal screening tests during your pregnancy. In an otherwise low risk pregnancy, the finding of an ICEF is not likely to significantly increase the chance for a chromosome condition.

What further testing may be offered and what will it tell me?

If the ICEF is found during a routine ultrasound, a more detailed ultrasound will usually be performed to look carefully at the baby's anatomy. Approximately 25-50% of children who have Down syndrome will not have any additional findings on a detailed ultrasound. The ultrasound will only modify your chance of having a baby with a chromosome problem such as Down syndrome. It cannot diagnose or rule out the presence of a chromosomal abnormality. If there are other concerns about the structure of the baby's heart, a specialized ultrasound of the heart can be performed, called a fetal echocardiogram. If your baby is found to have an isolated ICEF and is otherwise at low risk for chromosome conditions, this ultrasound is not necessary.

If you have had a detailed ultrasound that showed no other differences and there are no other risk factors for your pregnancy, then most likely no additional testing is necessary. It is important to remember that an ICEF is most likely a normal variant in the development of a healthy baby.

However, when additional testing is indicated, there are two types of prenatal tests: screening tests and diagnostic tests. 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. Diagnostic tests can evaluate the number and structure of your baby's chromosomes. They are 99% accurate in detecting extra or missing chromosomes. Unfortunately, all diagnostic tests have a risk of complications including the potential for pregnancy loss.

Screening Tests

There are multiple screening tests to determine the chances for a pregnancy to have a chromosome condition. These screening tests cannot diagnose or rule out the presence of a chromosome condition but may be used to 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.

Diagnostic Tests

Depending on your gestational age, an amniocentesis procedure may be offered. The amniocentesis is a diagnostic test in which a small sample of amniotic fluid is obtained to examine the baby's chromosomes. Because the procedure is considered an invasive procedure, there is a risk, likely less than 1%, for complications that can lead to miscarriage.

If you choose to pursue amniocentesis, you may be offered an additional test called a chromosome microarray (CMA.) It 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. 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.

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 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 ICEF. 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 ICEF, you should make the decision that is right for you and your family. Please keep in mind that not all babies with ICEF 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 ICEF, most babies will be born without significant medical complications.