Patient Information Sheet
Intrauterine Growth Restriction

You have recently learned that your baby has intrauterine growth restriction, also known as IUGR. 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 IUGR. Currently, there are no tests available to detect all health problems.

What is IUGR?

IUGR means that baby appears to be smaller than expected when measured by ultrasound. Specifically, the baby's weight appears to be less than 10th percentile based on how far along you are in pregnancy (your gestation). This means that 90 percent of all babies who are at the same gestation as your baby weigh more than your baby. IUGR is sometimes called fetal growth restriction (FGR) or small for gestational age (SGA). It is important to have the correct due date based on an early pregnancy ultrasound to establish your correct gestation before baby can be said to have IUGR.

How many babies are found with IUGR?

In the United States and similarly developed countries, approximately 4-7% of all babies are described as having IUGR.

What causes IUGR?

There can be many reasons why your baby appears to be small on ultrasound. Some babies who appear to have IUGR just happen to be small (possibly like one or both of the baby's parents or siblings) and some babies who appear to have IUGR on the ultrasound are born healthy with no other problems. For some babies with IUGR this is the only problem seen on ultrasound (isolated). However, some babies with IUGR may have a chromosomal or genetic syndrome or a birth defect that prevents them from growing and developing normally.

In order to understand a possible chromosomal or genetic syndrome as a cause for IUGR, 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 IUGR.

IUGR can also be caused by an exposure in the baby's environment like viral infections, maternal medical problems like high blood pressure or diabetes, problems in the placenta or umbilical cord, medications, drugs and smoking, or a combination of genetic and environment (multifactorial). For many babies with IUGR the exact cause is not known.

What problems could be associated with IUGR for my baby?

Babies with IUGR have a higher chances of complications both during and after pregnancy compared to babies that are of an appropriate size and weight. Complications during pregnancy may include continued delayed growth, abnormal blood flow through different critical veins or arteries in the umbilical cord or baby's body (detected through ultrasound), or even fetal death or stillbirth. Babies who have isolated IUGR may have many types of concerns after birth, such as seizures, low blood sugar (hypoglycemia), or cerebral palsy. However, some babies with IUGR do very well after delivery and have no lifelong concerns. There are many different factors that predict how well a baby with IUGR may do once they are born. Majority of babies with IUGR have to be delivered early (preterm) to avoid them passing away inside the mother's womb. Babies born preterm may have additional complications.

For babies with syndromic IUGR, the chance for additional health problems both during and after pregnancy would depend on what the cause of the IUGR is. Some birth defects may require surgery after birth. Some chromosomal or genetic conditions can have mild features, whereas others may cause more serious health problems and developmental delays and intellectual disabilities. In general, there is no treatment that can make baby grow faster or bigger. Rarely some infections that cause IUGR can be treated. Additionally, quitting smoking or drug abuse, and controlling maternal health issues, like high blood pressure and diabetes, is important for the baby's wellbeing.

Finding the cause of the IUGR in your baby can help to provide more information about how your baby will develop and what type of testing and/or treatment is needed. However, because the cause of the IUGR cannot always be determined, your healthcare provider may not be able to know exactly what other problems your baby may have until after the baby is born. Some parents may choose to end a pregnancy that has been diagnosed with severe IUGR early in pregnancy.

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

Depending on how far along you are in pregnancy, a number of additional tests may be offered to you. If IUGR is suspected during a routine ultrasound, your doctor will first make sure that your due date is correct and a more detailed ultrasound will usually be recommended to measure the baby and look more carefully at the body parts. Typically your doctor will repeat an ultrasound every 3-4 weeks to continue and watch the baby's growth. They may also do a Doppler flow test on ultrasound, which measures the amount of blood flowing through the veins or arteries in the umbilical cord or baby's body.

In the last trimester of your pregnancy your doctor may order additional tests that may include a non-stress test (NST), which measures the baby's heart rate and movements via a belt attached to mother's belly, and a biophysical profile (BPP), which combines the NST and an ultrasound to check on the baby's wellbeing. These tests may be done once every two weeks or even more often, like once or twice a week, depending on your baby. In some cases a fetal MRI and fetal echocardiogram (an ultrasound of the baby's heart) may also be recommended. You may be referred to a high risk maternal fetal medicine (MFM) clinic or a fetal center for further evaluation.

You may also be offered different types of genetic tests. There are two types of prenatal tests, diagnostic and screening:

<u>Diagnostic Tests</u>

Depending on how far along you are in pregnancy an amniocentesis 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. This fluid is obtained by inserting a thin needle through your belly and into the amniotic sac. 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 an 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. In addition to CMA, you may also be offered testing for specific genetic syndromes to look for changes in one or more genes as well as to test for different viral infections in the fluid.

<u>Screening Tests</u>

There are multiple screening tests to determine the chances for a baby 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.

Cell-free DNA (cfDNA), also referred to as Non-Invasive Prenatal Testing (NIPT) or Non-Invasive Prenatal Screening (NIPS), is a screening test for certain chromosome conditions. During pregnancy, some of your baby's placenta's chromosome material is in your blood, along with your own chromosome material. Cell-free DNA screen is a blood test that measures the amount of chromosome material in your blood to determine if your baby could have an extra chromosome. This test screens for Down syndrome, trisomy 18, trisomy 13 and may also screen for some less severe conditions that are caused by different numbers of the X or Y chromosome. Cell-free DNA screen may also screen for several conditions caused by small missing pieces of chromosomes. While cell-free DNA screen is a good screen, it is not a diagnostic test, like the amniocentesis. 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) and blood screens for infections may also be offered.

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.

It is important to remember that prenatal screening and testing cannot detect or rule out all genetic conditions and infections. After the baby is born, if additional birth defects or developmental delays are noted, an examination by a pediatric geneticist may be recommended. The geneticist may recommend that a small amount of blood be taken from the baby to evaluate for a chromosome or genetic 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 possible cause of IUGR, the risks to future children and that of other family members, as well as to determine appropriate tests to be offered to you in future pregnancies.

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 the chromosome conditions before delivery. Other people feel that they want more information as soon as possible. After gathering all of the information you need about IUGR and testing you should make the decision that is right for you and your family.

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 IUGR. 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 find it useful to have a consultation with a maternal-fetal medicine physician (physician that specializes in high-risk obstetrical care), neonatologists, and/or a geneticist prior to delivery to discuss the specifics about the IUGR. However, depending on your specific case and what hospital you are seen at, your health care provider may recommend additional consultations and tests that are not discussed here. Support groups, either online or in person, can provide comfort for families facing this diagnosis. One-on-one therapy or family therapy may also be useful, both during and after the pregnancy.