First Trimester Ultrasound and Second Trimester Ultrasound (Level II Ultrasound)

To the Point- Ultrasound (also known as sonogram) is an imaging technique that is used to examine internal organs and structures. It is commonly used in pregnancy. Ultrasound uses sound waves, which reflect off body structures to generate a picture.

Overview

The Basics

Ultrasound may be used at different times during the pregnancy to evaluate different things. There are generally two types of ultrasound that are used as genetic screening tests: the nuchal translucency or NT ultrasound done in the first trimester; and the anatomical fetal survey in the second trimester, often referred to as a level II ultrasound when performed by a perinatologist or maternal-fetal medicine doctor.

How it Works

**Ultrasound in the 1st Trimester**

In the first trimester of pregnancy the fetus is still very small – at 12 weeks the baby measures only a few inches in length and there is still a lot of early development taking place. The detail that can be seen in ultrasound at this time is pretty limited, however ultrasound is often done early in pregnancy for the following reasons:

- Confirmation of the presence of an intrauterine pregnancy
- Estimate how far along you are in the pregnancy
- To see if there is more than one baby developing (i.e. twins, triplets)
- Measure the nuchal translucency which is a screening tool for Down syndrome and other genetic conditions. This ultrasound is usually done between 10-13 weeks in the pregnancy.
- To evaluate pain or bleeding
Most women will undergo ultrasound in the second trimester of pregnancy – usually between 16-20 weeks. Some women think that the most important thing about this ultrasound is determining the gender of the baby, but there is a lot more to it than that! This ultrasound is often referred to as the anatomic survey (because it is looking at the developing baby’s anatomy). In a way, the anatomic survey is the most comprehensive genetic and health screening test offered in pregnancy as it assesses the baby from head to toe. The ultrasound is a way to look for physical signs that may indicate concern about the baby, but usually it provides some reassurance that the baby is developing as usual. It also evaluates important factors like where the placenta is located and how much amniotic fluid is surrounding the baby.

Results

Most of the time women will have a normal anatomic survey ultrasound and will leave with some of the first pictures for the baby book and the ever so important news of boy or girl. But sometimes there is a finding on ultrasound that can raise concern. Having some background about what is being looked for with this ultrasound can help you be prepared for that possibility. We will discuss two different categories of ultrasound findings:

1. Structural abnormalities
2. Minor markers

**Structural abnormalities (birth defects)**

Structural abnormalities are relatively rare and are also incredibly variable. A structural abnormality can be something that is very minor and easily treated or something very severe and even life-threatening. Some examples of structural abnormalities that can be seen on ultrasound are heart defects, cleft lip and palate, club foot, spina bifida, or extra fingers or toes. A structural abnormality can be an isolated finding – meaning that there is nothing else out of the ordinary impacting the health of the baby. Or, a structural
abnormality could be an indicator of an underlying genetic condition or syndrome that may be associated with other health and developmental concerns.

## Minor Markers

A minor marker is a subtle finding on ultrasound that may suggest an increased chance for certain genetic conditions, but in and of itself a minor marker does not cause any health concerns. In fact, most of the time a minor marker is a just a variation in normal development and is not associated with a chromosome or genetic condition at all. For example, shorter than average femur (thigh) bones are considered a minor maker which increases the likelihood of Down syndrome. This is because *many* babies with Down syndrome have shorter than average thigh bones. But there are also *many* babies who do NOT have Down syndrome but DO have shorter than average thigh bones. There are also some babies with Down syndrome that have perfectly average thigh bones. It is important to take news of these minor markers with a grain of salt. While they may increase the chance that your baby has Down syndrome or another genetic condition, most of the time this turns out not to be the case.

### What are some other minor markers that may be seen in the 2\(^{nd}\) trimester?

- Shortened or Absent Nasal Bone
- Choroid Plexus Cyst
- Echogenic Intracardiac Focus
- Increased thickness of the nuchal fold
- Pyelectatis
- Short femur bones
- Short humerus bones
- Single Umbilical Artery (SUA)

### What if a minor marker or multiple markers are seen on my ultrasound?

You should be counseled by a medical professional that can provide you more information about what these findings may mean for your baby. Depending on what marker(s) is seen,
your doctor may offer diagnostic testing, such as amniocentesis to evaluate for certain genetic and chromosomal conditions like Down syndrome at this point. It is always a personal choice whether to undergo amniocentesis. If you decide not to undergo amniocentesis it may be recommended that you have a follow-up specialized ultrasound to more carefully evaluate the baby’s heart, called a fetal echocardiogram. Babies with chromosome conditions including Down syndrome have a higher chance for a heart defect and if minor markers suggest an increased risk for a chromosome condition, it may be important for planning your delivery to determine if the baby is likely to have a heart defect.

What if a structural abnormality is detected?

Next steps will depend on the specific finding or concern. Often, diagnostic testing like amniocentesis will be discussed. Sometimes an abnormally on ultrasound is associated with a higher likelihood of certain genetic or chromosomal condition in the baby and further testing may be able to help explain what is being seen on ultrasound. Regardless of whether or not you decide to do follow-up diagnostic testing and regardless of the results, it is not uncommon for more follow-up ultrasounds to take place after a concern has been found. It may be recommended that you deliver your baby at a hospital with a critical care nursery that is equipped to address any special medical needs your baby may have. Some structural abnormalities can be repaired with surgery and some cannot. In the rare situation that a baby is diagnosed with life threatening abnormalities on ultrasound, expectant parents should be counseled about their options. Some women choose to end their pregnancy if serious abnormalities are discovered. Some women decide to carry their pregnancies and may be supported through a perinatal hospice program if their baby is not expected to survive.

How to Decide

It is fairly common to have at least one ultrasound during pregnancy, typically around 16-20 weeks of the pregnancy. However, some women may have an earlier ultrasound or more ultrasounds if they have bleeding, pain or other concerns are identified on an initial ultrasound.

Currently, there is no reliable evidence that ultrasound is harmful to a developing baby. For example, there have been no associations between ultrasound and birth defects, childhood cancer or developmental problems later on in life.

However, according to the American College of Obstetricians and Gynecologists patient information on ultrasound, it is possible that harmful effects could be identified down the
road. Their recommendation is that ultrasounds be performed only for medical reasons by qualified health care providers¹.

References

1) [http://www.acog.org/Patients/FAQs/Ultrasound-Exams](http://www.acog.org/Patients/FAQs/Ultrasound-Exams)