Genetic Support Foundation

Prenatal Ultrasound

Prenatal ultrasound (also known as sonogram) examines the developing baby by using sound waves that reflect off the baby to generate a picture.

Ultrasound in the First Trimester

In the first trimester the detail that can be seen in ultrasound is limited due to the small size of the baby. Sometimes ultrasound is done early in the pregnancy to confirm the due date, or to tell if it is a twin pregnancy. At around 10-14 weeks an optional ultrasound to measure the nuchal translucency (NT)[1] can be done. This measurement is used as a screening tool for Down syndrome and other genetic conditions. It may be done in combination with a blood draw on the mother to help estimate the chances for these conditions.

Ultrasound in the Second Trimester

Most women will undergo ultrasound in the second trimester of pregnancy, usually between 16-20 weeks. Many parents look forward to this as an opportunity to find out the sex of the baby, but there is a lot more to it than that. This ultrasound is looking at the developing baby for signs of health and genetic concerns from head to toe.

What does an ultrasound test for?

It is important to keep in mind that *no* prenatal test, including ultrasound in the first or second trimester, can look for or detect all genetic conditions or birth defects. Most of the time women will

[1] The NT is the fluid filled space at the back of the baby's neck.

have a normal ultrasound, but sometimes there is a finding on ultrasound that can raise concern. Findings on ultrasound can be put into two categories: structural abnormalities and minor markers.

The severity of structural abnormalities is variable from minor and treatable to severe. Some examples of structural abnormalities are heart defects, cleft lip and palate, clubfoot, spina bifida, or extra fingers or toes. A minor marker is a finding on ultrasound that may suggest an increased chance for a certain genetic condition, but in and of itself does not cause any health concerns. Many times a minor marker is just a variation in normal development.

What if the ultrasound identifies a concern?

If a structural abnormality is seen on ultrasound, referral to specialists can be helpful to learn more about what to expect and plan next steps. For instance, if a heart defect is seen on ultrasound you may be referred to a doctor that specializes in heart concerns in babies and children to discuss the finding and make a plan for the birth of your baby.

Structural abnormalities and certain minor markers may indicate an increased chance for a genetic condition, and you should be offered genetic counseling and further options such as blood tests or amniocentesis that may provide you with more information as to whether the baby has a specific genetic condition or not. All further testing is optional.

What are the risks associated with prenatal ultrasound?

Currently, there is no evidence that ultrasound, when used appropriately, is harmful to a developing baby. However, ultrasound should be done only for medical reasons by qualified health care providers.

You can find more information and videos about prenatal genetic testing options at: www.geneticsupportfoundation.org and www.doh.wa.gov.