Centracare Health

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Clinical Significance and Genetic Counseling for Common Ultrasound Findings

Patient Information Sheet
Use of Second Trimester Ultrasound and Down syndrome:
What does it mean?

You have had or are considering having a second trimester ultrasound to provide a risk assessment for your baby to have Down syndrome. We expect that you may have questions about Down syndrome and what this ultrasound is able to show. 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 Down syndrome?

Down syndrome is a genetic condition that is not typically inherited in a family. 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 disability 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.

What causes Down syndrome?

Down syndrome is caused by an extra copy of chromosome #21. 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. Sometimes there is an imbalance of genetic material that may cause birth defects. In the case of Down syndrome, an extra copy of chromosome number 21 is present.

How many babies have Down syndrome?

Approximately 1 out of every 800 babies is born with Down syndrome. A couple's chance of having a baby with Down syndrome increases with the mother's age.

What is ultrasound and what can it tell me about Down syndrome?

Ultrasound is a routine procedure that is performed during pregnancy to examine a baby's anatomy and growth. Ultrasound uses sound waves, which pass safely through the mother's abdomen, to create a picture of the developing baby. In addition to evaluating the baby's growth and development, it can also look for signs that may make your doctor more concerned about certain types of birth defects. Ultrasound may detect structural birth defects, such as a heart defect, that could cause health problems for the baby. Ultrasound may also detect subtle differences (known as soft markers) that may not affect the health of the baby, but may increase the concern for Down syndrome or other health conditions.

Listed here are some of the signs that ultrasound can detect which may increase the concern about the chance for your baby to have Down syndrome. Please remember that each of the signs listed here are often seen in babies who do **not** have Down syndrome. Also, not every baby with Down syndrome will have signs detected by ultrasound.

Structural birth defects:

- Cystic hygroma (cystic area at the back of the neck)
- Duodenal atresia (a narrowing or blockage of the intestine)
- Heart defects (structural changes in the heart)

Soft markers:

- Absent or hypoplastic nasal bone (missing or small bone in the nose)
- Echogenic bowel (bright area in the intestines)
- Increased nuchal fold (thicker than usual skin at the back of the neck)
- Intracardiac echogenic focus (bright spot in the tissue of the heart)
- Mild hydronephrosis (slightly increased fluid in the kidneys)
- Shortened femur or humerus length (shorter than usual long bones in the legs/arms)
- Ventriculomegaly (slightly increased fluid measurements in the brain)

The ability to identify signs of Down syndrome by ultrasound can differ from center to center. Individual factors, like the size of the baby or the mother, can also affect the ability of ultrasound to identify these signs. Your healthcare provider should consider other information about the risk for Down syndrome in your pregnancy, including maternal age and other prenatal test results.

When these signs are detected, a woman's chance to have a baby with Down syndrome may be increased. In general, the more signs seen on the ultrasound examination, the greater the risk is for Down syndrome. The fact that these signs have been seen does not mean that your baby has Down syndrome, only that additional testing may be offered to you to diagnose or rule out Down syndrome.

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

If you have had an ultrasound that has increased your doctor's concern about Down syndrome, your doctor or genetic counselor will talk with you about the option to have additional prenatal testing. Depending on your current gestational age, a number of additional tests may be offered to you. There are also 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

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, two diagnostic testing options may be available including chorionic villus sampling (CVS) or amniocentesis. These are diagnostic tests in which a small sample of placental tissue or amniotic fluid is obtained to examine the baby's chromosomes. Because these procedures are considered invasive procedures, there is a risk, likely less than 1%, for complications that can lead to miscarriage. The risks, benefits and limitations of amniocentesis and/or CVS should be discussed with you by your genetic counselor or health care provider. Follow-up counseling and referrals for support can be made if a chromosome problem is detected prenatally. As with all situations in which diagnostic testing is discussed, it is your decision whether or not this test is done.

If you choose to pursue a CVS or 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. If you have any questions about ultrasound, Down syndrome, or prenatal testing, please speak with your doctor or genetic counselor.

What should 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 screening options for Down syndrome. 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 a certain condition before delivery. Other people feel that they want more information as soon as possible. After gathering all of the information you need about Down syndrome, you should make the decision that is right for you and your family. Please keep in mind that not all babies with Down syndrome will have the complications described in this information sheet.