Clinical Significance and Genetic Counseling for Common Ultrasound Findings

Patient Information Sheet
Duodenal Atresia

You have recently learned that your baby has duodenal atresia. 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 duodenal atresia?

Duodenal atresia is a birth defect where first part of a baby's small intestine (the duodenum) doesn't develop properly. In typically developing babies, the duodenum is a hollow tube that allows contents from the stomach to pass into the small intestine for digestion. In babies that have duodenal atresia, there is an obstruction, or a blockage, in the duodenum that prevents the contents of the stomach from passing through. Babies born with duodenal atresia can have swelling of their abdomens and excessive vomiting. Surgery needs to be performed once a baby is born in order to correct the blockage and allow for normal digestion.

How many babies are found with duodenal atresia?

Approximately 1 in 10,000 babies are born with duodenal atresia.

What causes duodenal atresia?

Most cases of duodenal atresia are random and the cause is unknown. However, approximately one-third of babies of duodenal atresia have Down syndrome. Down syndrome is the most common chromosome disorder seen at live birth.

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. Down syndrome, specifically, is a chromosome condition caused by the presence of an extra copy of chromosome 21. 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. Duodenal atresia can be seen in some genetic syndromes, but most babies with duodenal atresia do *not* have a genetic syndrome.

What problems could be associated with duodenal atresia for my baby and how are they treated?

In approximately one-third to one-half of cases, duodenal atresia is an isolated finding and occurs in babies that otherwise healthy. In these cases, the long-term outcome after surgery is very good. In some cases, however, babies with duodenal atresia have other birth defects. These birth defects may involve the heart, the gastrointestinal system, the kidneys, the skeletal system, or the genitourinary system. The long-term outcome for babies that have multiple birth defects is variable and depends on many factors, including the severity of the other defects and the presence of a genetic syndrome. Your healthcare provider may arrange for a consult with a pediatric surgeon or a neonatologist so that you may learn more about what will be done to care for you baby after he or she is born.

Pregnancies with duodenal atresia will often have abnormally high amounts of amniotic fluid. High amniotic fluid is referred to as "polyhydramnios." Polyhydramnios may increase your risk of preterm labor. Because of this the doctors may recommend a procedure called an amnioreduction to remove the excess fluid around the baby. This fluid is removed by inserting a thin needle through the belly and into the amniotic sac and pulling out the extra fluid. Babies who are born prematurely may have more complications then those born at term.

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 duodenal atresia is suspected during a routine ultrasound, a more detailed ultrasound will usually be performed to look carefully at the baby's body. A fetal echocardiogram (a specialized 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:

Diagnostic Test:

Depending on how far along you are in pregnancy 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. 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, or if you have an amnioreduction performed, 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.

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.

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) 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. 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 the duodenal atresia, 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 duodenal atresia 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 duodenal atresia. 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, a pediatric surgeon, and a geneticist prior to delivery to discuss specifics about duodenal atresia, prognosis, and available treatments. You may also find it useful to have a consultation and an evaluation performed at a fetal center experienced at taking care of babies with duodenal atresia. 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.