

Clinical Significance and Genetic Counseling for Common Ultrasound Findings

Patient Information Sheet Absent Cavum Septum Pellucidum

You have recently learned that your baby has an absent cavum septum pellucidum. 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 an absent cavum septum pellucidum?

The cavum septum pellucidum (CSP) is the name for a fluid-filled space in the middle of the developing brain. The CSP should be visible in your baby's brain on an ultrasound between 18 and 37 weeks of your pregnancy. Seeing the CSP during this period of pregnancy is a sign that the baby's brain is likely developing normally. When the CSP is absent it means that your doctors could not see this fluid-filled space when looking at your baby's brain on ultrasound. The CSP itself does not seem to cause the brain to work differently, but babies who have an absent CSP have a higher chance of having other brain abnormalities that may affect how the baby's brain works. Therefore, when your doctor notices that your baby's CSP is absent, he or she may recommend additional testing and imaging.

How many babies are found with absent CSP?

It is not known exactly how many babies have absent CSP during pregnancy, but it may be as high as 4 in 1000. About 2-3 per 100,000 people are found to have an absent CSP after they are born.

What causes absent CSP?

Most of the babies that have an absent CSP also have another problem in the brain's development. There are many kinds of different brain abnormalities that can cause the CSP to be absent on ultrasound. The brain abnormalities that can cause absent CSP can be associated with many different things, including chromosome conditions, genetic syndromes, brain injury, brain infection, and problems with the blood flow to the brain. In some cases, we may not know what caused a structural brain change to occur.

About half of the babies with absent CSP have a diagnosed genetic condition with may be inherited or happen by chance in the baby for the first time. In order to understand a possible chromosomal cause or genetic syndrome cause, 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 brain problems and absent CSP.

When absent CSP and/or other brain abnormalities are isolated (no other physical or genetic problems found) it could have been caused by multifactorial (complex) inheritance, meaning that multiple factors (including genetics and the environment) influence the development of the brain. While it is rare, some babies can only have an absent CSP. If your baby has truly isolated absent CSP) this may be a normal variant for your baby.

What problems could be associated with absent CSP for my baby?

The most common brain problem causing absent CSP is a difference in the development of the corpus callosum. The corpus callosum is a bundle of nerve fibers that connect the two sides of the brain. In some babies with absent CSP, the corpus callosum is missing or is small. However, some babies with absent CSP have a normal corpus callosum. Other babies with an absent CSP may have other brain problems, such as holoprosencephaly (a difference in the way the two sides of the brain form), septo-optic dysplasia (a difference in structures in the middle of the brain that can cause vision problems), schizencephaly (a cleft in the brain tissue), or hydrocephalus (too much fluid in the brain).

Generally babies that have a problem in their brain development usually have some developmental delays and may need extra time and help to learn new skills. They also have a higher chance of intellectual disability, or low IQ, as well as other problems such as seizures or problems with their vision. Sometimes, when the brain abnormality is very severe, babies may pass away after delivery. It is important to remember, however, that some babies only have an absent CSP. Babies that only have an isolated absent CSP usually do very well and have a high chance of developing normally.

Finding the cause of the absent CSP 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 absent CSP 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. There is no cure for absent CSP or other brain problems and it cannot be treated or fixed during pregnancy. Some parents may choose to end a pregnancy that has been diagnosed with absent CPS with another brain problem.

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 absent CSP is found during a routine ultrasound (after 18 weeks of your pregnancy), a more detailed ultrasound will usually be recommended to look more carefully at the baby's brain and other body parts. 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 and meet with a genetic counselor. There are two types of prenatal tests, diagnostic and screening:

Diagnostic Tests

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 or to test for different viral infections in the fluid.

Screening Tests

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. 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 absent CSP, 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 absent CSP and testing you should make the decision that is right for you and your family.

What is the risk for this to happen again?

Parents of a pregnancy with absent CSP are often worried about the risk for it to happen in another pregnancy. The chance of absent CSP happening again would depend on the cause of the absent CSP. For truly isolated absent CSP, the chance of it happening again is typically less than 1%. However, if the absent CSP is caused by another brain problem or genetic condition, the chance of it happening again may be as high as 50%.

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 absent CSP. 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), neurologist, and/or geneticist, prior to delivery to discuss the specifics about absent CSP. After your baby is born a consultation with ophthalmology and endocrinology is also recommended. 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 absent CSP and other brain abnormalities. 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.