Patient Information Sheet
Choroid Plexus Cyst (CPC)

You have recently learned that your baby has a choroid plexus cyst (CPC). 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 a choroid plexus cyst?

The choroid plexus are normal parts of both sides of the brain. The choroid plexus make cerebrospinal fluid that bathes the spinal cord and brain. Sometimes a fluid filled cyst forms inside the choroid plexus – this is a choroid plexus cyst (CPC).

How many babies are found with choroid plexus cysts?

About 1% of second trimester prenatal ultrasounds reveal at least one choroid plexus cyst. It does not matter if there is one cyst or several, or what size a cyst is.

What problems could be associated with choroid plexus cysts for my baby?

Choroid plexus cysts do <u>not</u> cause problems. Most disappear by the last three months of pregnancy. Choroid plexus cysts are sometimes present in adults – and cause no problems. If only a choroid plexus cyst is seen, this is called an isolated choroid plexus cyst.

Rarely, babies with choroid plexus cysts have chromosome problems. However, because of this association, it is important for you to be informed about the presence of the cysts and of your options for prenatal testing. 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 choroid plexus cysts.

The specific chromosome problem most associated with choroid plexus cysts is called trisomy 18 (Edward syndrome), which is caused by an extra chromosome 18. There is an increased chance for trisomy 18 when there is a choroid plexus cyst. This chance is highest when there are multiple problems detected by the ultrasound and least likely when only choroid plexus cysts are seen. Babies with trisomy 18 usually have severe medical problems and profound intellectual disability. Only a small number of babies with trisomy 18 survive the pregnancy and the first year of life. Often people are more familiar with Down syndrome, which is caused by an extra chromosome #21. While Down syndrome has been found when there are choroid plexus cysts, it is not common. Individuals with Down syndrome often have characteristic facial appearance, low muscle tone, and an increased risk for heart and bowel defects. Individuals with Down syndrome typically have mild to moderate intellectual disability. Despite their challenges, children with Down syndrome are more like other children than different.

The chance that the baby would have a chromosome problem based on finding a choroid plexus cyst on the ultrasound depends on additional risk factors including maternal age, presence/absence of additional ultrasound findings, and the results of other screening tests. The risk to have a child with a chromosome abnormality increases as women grow older. The risk for a chromosome abnormality also increases if there are additional ultrasound findings, such as a heart defect, slow growth or other birth defects.

When there are no other risk factors, nearly all babies with isolated choroid plexus cysts are normal. When there is an isolated choroid plexus cyst, the risk for trisomy 18 is small (less than 1%) and the risk for Down syndrome is even less.

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

If the CPC is found during a routine ultrasound, a more detailed ultrasound will usually be performed to look carefully at the baby's anatomy. There are two additional types of prenatal tests: screening tests and diagnostic tests. Screening tests can modify your risk

for a chromosome abnormality such as trisomy 18 and involve no risk of complications to your pregnancy. Diagnostic tests are the most accurate tests to diagnose a genetic condition during a pregnancy; however, these tests do have a small risk of complications, including the potential for pregnancy loss.

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.

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:
Amniocentesis is a type of diagnostic test that samples a small amount of the amniotic fluid to examine the baby's chromosomes. This is the most accurate test that can be offered during a pregnancy to test for a chromosome abnormality. Because this procedure is considered an invasive procedure, there is a small risk (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.

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.

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 choroid plexus cysts. 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 any of these conditions before delivery. Other people feel that they want more information as soon as possible. After gathering all of the information you need about choroid plexus cysts, you should make the decision that is right for you and your family. Please keep in mind that not all babies with choroid plexus cysts will have the complications described in this information sheet. Though it is never possible to predict the long-term prognosis for a child with even isolated choroid plexus cysts, most babies will be born without significant medical complications.