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COMMUNITY MIDWIVES

Choroid Plexus Cysts

What are Choroid Plexus Cysts?

In order to understand what choroid plexus cysts (CPC's) are, it is important to have a general understanding of the choroid plexus. The brain contains pockets or spaces called ventricles with a spongy layer of cells and blood vessels called the choroid plexus. The choroid plexus has an important function of producing a fluid called cerebrospinal fluid. The fluid produced by the cells of the choroid plexus fills the ventricles and then flows around the brain and the spinal cord to provide a cushion of fluid around these structures.

CPC's occur within the choroid plexus and are thought to arise from fluid trapped within this spongy layer of cells.

How common are CPC's? Do they impact on an individual's health?

Fetal CPC's are now recognized in 1 out of every 100 pregnancies where an ultrasound is performed between 16 and 20 weeks.

It is believed that 50% of adults in the general population have one or more tiny CPC's. CPC's have no impact on an individual's health or development.

Fetal CPC and the Association with Chromosome Problems

Sonographically detectable fetal choroid plexus cysts (CPC) were first described in 1984 and were initially regarded as a benign variant until 1986, when a link between choroid plexus cysts and Trisomy 18 was postulated and further reinforced by subsequent studies. Recently, researchers have noted a slightly increased incidence of CPC's in fetus' with an extra chromosome, including Trisomy 18, Trisomy 21 (Down syndrome), or less commonly, Trisomy 13.

What is a Trisomy?

Normally, within the nucleus of all of our cells, there are 46 chromosomes, each paired so that there are 23 pairs. One copy of each pair is received from the mother and one from the father. Babies with, for example, Trisomy 21 aka Down syndrome, have 47 chromosomes because they have received an additional chromosome 21 from either the egg or the sperm at conception. Individuals with Down syndrome have typical physical features and a high risk of developing particular health problems. Some are treatable; however, all individuals with Down syndrome have some degree of mental disability.

Other less common trisomies which can be seen in newborn babies include Trisomy 18 or Trisomy 13. Babies with these chromosome problems do poorly and are often lost during the pregnancy or die within the first year of life.

My baby has a CPC. What is the risk of a chromosome problem?

The risks are low. The risk is estimated at 1% (1/100) of delivering a baby with a chromosome problem. Additional factors that may change the risk:

- Mother's age at the expected date of delivery
- The size and number of the fetal CPC's
- Evidence of other "fetal findings" seen at the time of the ultrasound that suggest a chromosome problem. Babies with chromosome problems can quite often have other problems affecting major organs, such as the heart, limbs, stomach, etc, therefore CPC's that are NOT associated any other findings are considered very unlikely to be

significant – the estimates vary between 1 in 100 and 1 in 300 chance of having a baby with a chromosome problem.

What options are available to me?

Isolated CPC's

Given that the risk of miscarriage caused by amniocentesis may be greater than the risk of Trisomy in a fetus with isolated CPC, it is considered imprudent to do amniocentesis on all fetuses with isolated CPC's.

1) Your midwife will provide you with a referral to see a Medical Geneticist who can best provide you with the most current information about fetal CPC's, answer your questions and concerns, and outline options available to you.
2) If you are between 15 and 21 weeks of pregnancy, you can choose to have a Triple Screen test: a blood test that can give you a more accurate risk assessment of the chances of having a baby with Trisomy 18, Trisomy 21 or open spina bifida. If you have already had a Triple Screen, then the risk results will be recalculated.
    a) If this test result shows elevated risk of a chromosomal problem, then you will be offered an amniocentesis.
3) If you are older than 35, you can choose to go directly to amniocentesis without doing a Triple Screen.

CPC's + other ultrasound findings

If, in addition to the CPC's, there are other findings on ultrasound that also indicate an elevated risk of chromosomal abnormalities, you may be offered an amnio directly.

Confirmed chromosomal abnormality

If an amnio confirms a chromosomal abnormality, then your options will depend on where you are in pregnancy and how profound the fetal abnormality is.

IMPORTANT TAKE HOME MESSAGES:

It is important for you to recognize that the chances are strongly in your favor of a normal pregnancy outcome. The detection of these cysts has identified your pregnancy as at only a slightly greater chance of having a chromosome abnormality than the general population.

Fetal CPC's do not have any effect on the development of the fetus. We would expect that if the fetus has normal chromosomes, there would be no other associated health problems about which to be concerned (i.e. your baby will have normal development and intelligence). These cysts do not affect the production or circulation of the cerebrospinal fluid. No special treatment or tests are needed for the baby at delivery.