

## Choroid Plexus Cysts Val A. Catanzarite, MD, PhD



What are choroid plexus cysts? The choroid plexus is a structure within the brain ventricles that has the function of producing cerebrospinal fluid. The choroid plexus is usually easy to see by means of sonography between week

14 and 22. Early in pregnancy, the choroid plexus has a butterfly shaped appearance and seems to almost fill the brain's ventricles. As the baby grows, the choroid plexus is visit e as a tear drop shaped gray appearing structure within the right and left ventricles of the baby's brain. The two arrows mark the choroid plexus in this image.

## **How often are choroid cysts seen on ultrasound?** In about 1:100 pregnancies, a small



clear space or "cyst" will be seen within the choroid plexus. Choroid plexus cysts can be seen within the choroid plexus of the right ventricle, of the left ventricle, or in both ventricles. They can range in size from very tiny (which is just a bit

larger than the period at the end of this line) up to nearly ½ inch in diameter. Two small choroid plexus cysts are shown in the image to the left.



A much larger choroid plexus cyst is seen in this image.

Very tiny choroid plexus cysts may be even more common in the newborn.



From experience, we know that choroid plexus cysts virtually always shrink down in size and disappear as the pregnancy progresses. They are usually gone by 26

weeks, although we have seen one pregnancy where the cyst was still visible until almost 32 weeks.

Babies with choroid plexus cysts seen in utero are at increased risk to have a chromosome problem known as Trisomy 18—which is a serious, usually lethal condition.

If choroid plexus cysts are seen on ultrasound, the first step is to arrange a comprehensive ultrasound. This is very different from the standard ultrasound scan performed by the Radiologist or Obstetrician. The sonograms at our centers are performed by experienced Perinatal Sonographers and Obstetricians with subspecialty training in Maternal and Fetal Medicine. The program performs more than 9000 sonograms per year, has experience with the diagnosis of over 1000 congenital abnormalities, and has achieved accreditation by the AIUM and ceritification as a Prenatal Diagnostic Center by the State of California, and we use high resolution ultrasound machines. We have seen many hundreds of babies with choroid plexus cysts

When a comprehensive ultrasound examination has been done and the results are normal except for choroid plexus cyst(s), the chance for Trisomy 18 is increased by a factor of about 9 relative to risk calculated based on Mom's age, serum screening, or integrated screeing. When abnormalities of the baby, such as a heart defect or abnormal hand position are seem, the risk is much, much higher.

The only way to determine whether the baby does or does not have a chromosome defect such as Trisomy 18 is by means of chorionic villus sampling or amniocentesis. What is the risk of miscarriage due to amniocentesis? In the past, the risk was quoted as approximately 1:200. This calculation was based on studies performed in the 1970's and 1980's. More recent data shows that the risk of miscarriage due to amniocentesis performed at a center such as ours is less than 1 per 500, and it may be as low as 1:1600 (see reference by Eddlemann).

There are many unanswered questions regarding choroid plexus cysts. The risk of undetected trisomy 18 is reduced when detailed examination of the fetal heart (which can typically be performed after 20-21 weeks of pregnancy) is normal, and there is as yet very limited data regarding adjustment of trisomy 18 risk determined by first trimester screening.

We have reviewed our own center's data (over 300 choroid plexus cysts; see reference in *italics* below) and are satisfied with using a 9x risk adjustment for isolated choroid plexus cysts. It is

our policy at the Perinatal Diagnosis Program to offer genetic counseling so these issues can be discussed thoroughly.

It is important to realize that choroid plexus cysts are not an abnormality of the baby. They are seen in about 1% of all pregnancies, and the vast majority of these babies are perfectly normal in every way. DiGiovanni and co workers (see references below) found that as long as chromosome testing was normal, follow up from 1-6 years of age revealed all 76 to be developmentally normal. They concluded that choroid plexus cysts do not impact infant and early childhood development.

In summary: Isolated choroid plexus cysts are seen in 1% of pregnancies; they carry an increased risk for trisomy 18. If the chromosomes are normal, choroid plexus cysts are considered a variant of normal anatomy and have no known implications for brain development or neurologic outcome of the newborn.

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