Thai Journal of Obstetrics and Gynaecology
September 1997, Vol. 9, pp. 169-173

OBSTETRICS

Clinical Significance of Sonographic Demonstration of Fetal Choroid Plexus Cysts

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ABSTRACT

Objective To evaluate the correlation of prenatally diagnosed choroid plexus cysts and neonatal outcomes.

Design Prospective descriptive analysis.

Setting Department of Obstetrics and Gynaecology, Faculty of Medicine, Maharaj Nakorn Chiang Mai Hospital, Chiang Mai University.

Subjects A total of 22 fetuses with prenatal detection of choroid plexus cyst were evaluated and followed.

Results Twenty-two cases of fetal choroid plexus cysts in second trimester were detected between estimated 4,000 scans of second-trimester obtained over 7 year period. They consisted of 18 healthy newborns, 3 newborns with trisomy 18 and 1newborn with mosaic trisomy 21. Additional sonographic abnormalities were also detected in 4 of the neonates with trisomy. None of the fetuses with apparently isolated cysts had aneuploidy.

Conclusion There is an association between fetal choroid plexus cyst with trisomy, however, chromosome study should be reserved for the cases with additional sonographic abnormalities.

Key words : choroid plexus cyst, Trisomy 18, ultrasound

With increased experience of sonographers and improvement of ultrasound technology, the prenatal diagnosis of anomalies of the fetal brain has been made with greater frequency. The choroid plexus cyst is one of the abnormalities now being recognized and reported but its clinical significance remains unclear. It is not harmful in itself,[1] but may be associated with serious chromosomal abnormalities, especially when other anomalies have also been demonstrated. The

incidence of fetal choroid plexus cysts has varied among investigators, ranging from 0.1% to 2.5%,[2-6] having an average of approximately 1% in all second-trimester fetuses. It is generally agreed that the majority of choroid plexus cysts are benign ; however, several reports have associated cysts with chromosomal abnormalities. It is difficult to determine whether choroid plexus cysts do indeed have a relationship with aneuploidy. The management of isolated choroid plexus cysts detected at 16-20 weeks' gestation remains one of the most controversial areas in obstetric ultrasound. To better clarify this controversy, we present our experience of fetal choroid plexus cysts in Thai population to evaluate the correlation of prenatally diagnosed choroid plexus cysts and neonatal outcomes.

Materials and Methods

During a 5-year peroid ranging from June 1992 to June 1997, 4,000 women in second-trimester were scheduled to undergo ultrasonographic evaluation by the authors at Chaing Mai University Hospital. Indications for these examinations included advanced maternal age (with or without amniocentesis), gestational age determinations, second-opinion sonographic evaluation, evaluation of fetal growth, and anomaly screening. All examinations were performed on the Aloka 680 EX or 650, 3.5-MHz curvilinear array. All examinations included careful inspection of the choroid plexuses. Evaluations included morphometric measurement to determine gestational age, examination of the choroid plexus for the identification and measurement of a cyst, and extensive examination to detect any structural malformations. When choroid plexus cysts were detected (Fig. 1), the proper counseling was given. Follow-up scans were arranged at approximately four weeks interval. Pregnancy outcomes were finally evaluated. Chromosomal analysis was done by either amniocentesis, cordocentesis or neonatal evaluation.

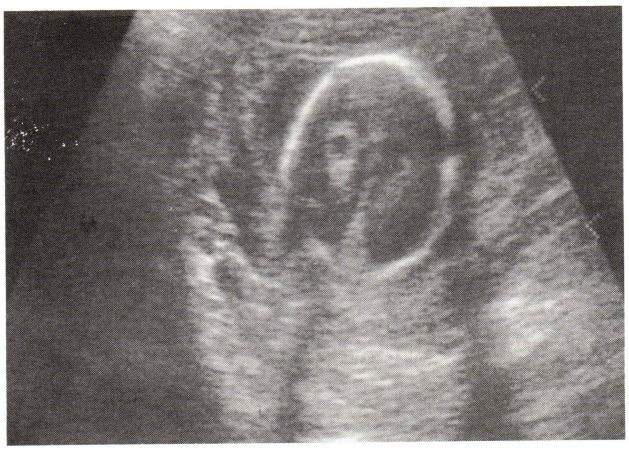

Fig. 1. Unilateral small choroid plexus cyst seen in lateral ventricle.

Results

During the study period, fetal choroid plexus cysts were diagnosed and followed-up 22 pregnancies. All were simple cysts, no complex type at all. The cases were classified into two main groups (Table 1) ; those with apparently isolated choroid plexus cysts (17 cases) and the other group which other anomalies were found on antenatal sonographic evaluation (5 cases). All of the first group had normal karyotypes, whereas 4 of 5 fetuses in the second group were aneuploidy (Table 2). Three of them were trisomy 18 and the other was mosaic trisomy 21(46, XX/47, XX 21+). One of the second group had normal karyotype but had bilateral hydronephrosis (renal pelvis diameter 14 mm on average) diagnosed at 17 weeks, without progressive change throughout pregnancy.

The prevalence of aneuploidy in all fetuses with choroid plexus cysts was 18% (4/22). However, the prevalence in the group of isolated

Table 1. Classification of pregnancies with fetal choroid plexus cysts

Group	Apparently isolated choroid plexus cyst (n = 17)	Choroid plexus cyst with associated anomalies (n =5)
Chromosomes		
Normal	17	1
Aneuploidy	-	4
Trisomy 18	-	3
Trisomy 21	-	1
Bilaterality		
Unilateral	12	3
Bilateral	5	1
Cyst Size		
2-10 mm.	16	5
> 10 mm.	1	-

Table 2. Ultrasonographic findings associated with choroid plexus cysts in patients with abnormal karyotype

No.	Age	Karyotype diagnosis	Weeks of gestation	Additional sonographic abnormalities
1	36	trisomy 18	19	Diaphragmatic hermia, rocker bottom feet
2	28	trisomy 18	20	Omphalocele, VSD, Cleft lip, brachycephaly
3	31	trisomy 18	14	Micrognathia, dilated cystern magna
4	28	mosaic	18	Small-for-date, ASD

cysts and the group with additional abnormalities were 0% (0/17) and 80% (4/5), respectively.

In cases of isolated choroid plexus cysts, the size of the cysts varied from 2-11 mm. and did not alter the geometry of the lateral ventricles in all cases. There were two cases of the isolated group and none of the other having a large cyst (10 mm or more). Six cases, 5 of the isolated group and 1 of the other, had bilateral choroid plexus cysts.

All of the aneuploidic fetuses were electively terminated after proper counseling. The fetus with bilateral hydronephrosis was followed-up with serial ultrasound and was delivered at 39 gestational weeks. The fetus was healthy without other associated anomaly and normal chromosomes. All women who continued their pregnancies were rescanned at 24-26 weeks'

gestation. Persistent cystic changes were visible in only 3 of 18 women who had euploidic fetuses. However, all pregnancies had no any serious obstetric complication and resulted in the deliveries of the healthy newborns at 35-40 gestational weeks. No differences in fetal and neonatal outcome were identified between unilateral and bilateral cysts. Additionally, no differences in outcome were found between persistent or resolved cysts.

Discussion

Fetal choroid plexus cyst is a common finding at autopsy in all age groups including the fetus. The pathologic characteristics have been shown to be a simple cyst surrounded by loose stromal tissue of normal choroid plexus, filled with clear fluid and without an epithelial lining.[7,8] It has become generally accepted that the majority of the findings are entirely benign, however, there are a growing number of studies indicating an association with aneuploidy, especially trisomy 18 and possibly trisomy 21.[8-12] Therefore, when such cysts are detected in utero, karyotypic and detailed sonographic examination would provide crucial information regarding the fetal prognosis.

It is noteworthy that in this study, when a choroid plexus cyst was associated with an abnormal karyotype, other structural abnormalities were always visualized on ultrasonography. It remains to be determined whether the sonographic finding of an isolated choroid plexus cyst also carries an increased risk of chromosomal anomaly. However, this series and some previous reports,[9-12] suggest that the probability of aneuploidy is high only when choroid plexus cysts are associated with any other sonographic fetal abnormalities, indicating a clear need of invasive genetic testing. The predictive value is much lower when no other anomalies are detected. In such cases, it is probably advisable to regard choroid plexus cysts as an indication for detailed ultrasound assessment, rather than invasive testing.

We agree that the most important differentiating factor between normal and aneuploidic fetuses is the detection of other structural abnormalities. Although it is not certain how sensitive these other anomalies are finding fetal choroid plexus cysts warrants a detailed ultrasonographic assessment for other anomalies. If the cyst appears to be an isolated anomaly, comprehensive counseling regarding risk for aneuploidy especially trisomy 18 and the risk for fetal loss after an invasive procedure is warranted. Whether an invasive procedure is performed ultimately should reflect the couple's own decision after genetic counseling.

In the series presented here, we were unable to evaluate the persistence of the cysts of the aneuploidic fetuses because all of them were electively terminated. In cases of normal karyotype, however, only 3 of 19 had the cysts beyond 26 weeks and all had normal outcome. Persistence of cyst may not be a useful sign in predicting fetal prognosis.

We documented a choroid plexus cyst in a mosaic trisomy 21 fetus. Whether this is a true finding or merely coincidental cannot yet to be determined.

In this study, the size and bilaterality of the cyst seemed not to be predictors of aneuploidy. Surprisingly, we found bilateral cyst only 27.3%, much less than other studies.[10] Nearly all cases of bilateral choroid plexus cysts in this series were not associated with any anomalies or aneuploidy. Although some authors[12] suggested the cysts of larger than 1 cm be related to trisomy 18 and confirmatory invasive test might be appropriate, we found that the size of the cysts

was a poor predictor of abnormal karyotype. All aneuploid fetuses had cysts < 1 cm and two cases of large cysts had normal karyotype. Thus our concern should not be limited to large cysts. It should be appreciated that since choroid plexus cysts nearly always resolve spontaneously, the size of cysts will be influenced by the gestational age of the fetus at time of scanning.

Choroid plexus cysts are easily seen in the standard biparietal view which is obtained for all routine anomaly scans. Alternative lesions detected by ultrasound screening that may potentially have higher sensitivities for chromosomal abnormalities are not easy to be detected by a routine examination. For example, ventricular septal defect and micrognathia are found in more than 50% of fetuses with trisomy 18.[13] However, these defects may be impossible to be detected even in the second trimester by expert sonographers.

If isolated choroid plexus cysts are found in early gestational age, before 18 weeks, the repeated ultrasound should be done at 20 weeks because it is possible that some of the subtle abnormalities associated with aneuploidy are not apparent in the early second-trimester. It seems that the fetus at more than 18 gestational weeks without other anomalies identified on detailed ultrasound should not be necessary to perform amniocentesis.

In conclusion, there is a strong association between aneuploidy and fetal choroid plexus cyst with additional abnormalities, whereas apparently isolated choroid plexus cyst is unlikely to increase the risk. Neither bilaterality nor cyst size was associated with aneuploidy. This small series suggests that fetal choroid plexus cysts warrant a detailed sonographic assessment and chromosome study should be reserved for the cases with additional abnormalities.

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