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Fetal Choroid Plexus Cysts : A Practical Approach in the Perspective of Available Data

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Summary

Choroid plexus cysts are not an uncommon finding in a routine antenatal ultrasound scan. Associations between choroid plexus cysts and Trisomy 18 have been reported. Approximately one third of fetuses with Trisomy 18 have been found to have choriod plexus cysts. The present study analysed, in retrospect, the records of 42 fetuses with sonographically delineated choroid plexus cysts to determine whether it is justified to carry out an assessment for an abnormal fetal karyotype. Thirty-eight of these fetuses had no abnormal findings other than choroid plexus cysts. None of these sonographically, normal appearing fetuses had any abnormality at amniocentesis or at birth. Four fetuses had major anomalies detected sonographically, in addition to choroid plexus cysts. All 4 fetuses showed an abnormal karyotype. The data suggests that the yield of an abnormal karyotype in fetuses with isolated choroid plexus cysts is too low to justify the risk of amniocentesis.

Introduction

Choroid plexus cysts were first described by Chudleigh et al in 1984 and soon, thereafter, by Fakhrey et al (1985) and Friday et al (1985) as possible normal sonographic variants. Benacerraf and Laboon in 1989 reported a relatively high incidence of about 1 in 3 of the presence of chroid plexus cysts in fetuses with Trisomy 18. This sparked off a series of controversies on the need for an amniocentesis in fetuses found to have choroid plexus cysts on second-trimester sonography. Twenty four reports between 1985 and 1996 found no association of aneuploidy and choroid plexus cysts, whereas, 6 found two or more fetuses with choroid plexus cysts and chromosomal abnormalities. Since currently available sonographic equipment and operator expertise permit a high level of accuracy in the detection of stigmata of chromosomal abnormalities, the present study was undertaken as a retrospective analysis to assess the need and justification of karyotyping of fetuses found to have choroid plexus cysts.

Material and Methods

Clinical records and follow-up data of 1438 singleton pregnancies with gestational ages ranging between 16-26 weeks were analysed. The patients had been scanned on a Toshiba Ecocee 340 color Doppler ultrasound scanner over a 12 – month study period.

A detailed anomaly scan had been done on all these fetuses. Forty two fetuses showed choroid plexus cysts. A detailed sonography to delineate stigmata of chromosomal abnormalities was done. The stigmata specifically looked for are enlisted in Table I. Owing to the reported high association between choroid plexus cysts and Trisomy 18, parents of fetuses with choroid plexus cysts but no other sonographic anomaly were counselled about the risk of chromosomal defects. The need for a karyotype study was explained to parents of all fetuses who showed an associated dysmorphic anomaly. In parents wanting a karyotype analysis, an amniocentesis/cordocentesis was done. After

counselling, all patients opted for elective termination of fetuses with an abnormal karyotype. All the pregnancies with isolated choroid plexus cysts were followed up with another detailed sonography at 22-26 weeks. All pregnancies were followed till term and all neonates were assessed for developmental anomalies.

Table I

Sonographic Stigmata ofTrisomy 18
Polyhydramnios
Intra-uterine growth retardation
Congenital heart disease
Diaphragmatic hernia
Omphalocele
Esophageal atresia
Hydronephrosis
Club foot deformity
Clenched hands
Generalised arthogryposis
Micrognathia
Microphthalmos
Hypertelorism
Dolicocephaly
Prominent occiput
Posterior cranial fossa abnormalities
Agenesis of corpus callosum
Single umbilical artery
Thickened nuchal translucency

Observation and Results

Forty two fetses out of 1438 showed choroid plexus cysts. The cysts measured 2-10 mm across in maximum dimension. In 14 cases the cysts were unilateral (Fig. 1). In 28 cases these cysts were bilateral (Fig. 2). In 10 cases the cysts were solitary and in 32 cases the cysts were multiple. No dilatation of the lateral ventricle was seen in any fetus. A detailed sonography was done in all these cases. Thirty eight cases showed no other dysmorphic anomaly. In all these cases the cysts resolved by 22-24 weeks of gestation. Four cases showed dysmorphic anomalies. These were a thickened nuchal skin fold (Fig 3) in one case, absent stomach bubble in one case, diaphragmatic hernia (Fig 4) and moderate bilateral hydronephrosis in one case, and intra-uterine growth retardation, polyhydramnios and club feet in one case. On amniocentesis and karyotying these showed a Trisomy 21, 18, 18 and 18 respectively. These pregnancies were terminated on the patients' request. All 38 fetuses that showed resolution of cysts (Fig 5a and 5B) had been followed up serially and in the neonatal period. Normalcy was confirmed. There was no relationship between size and number of choroid plexus cysts and an abnormal karyotype.

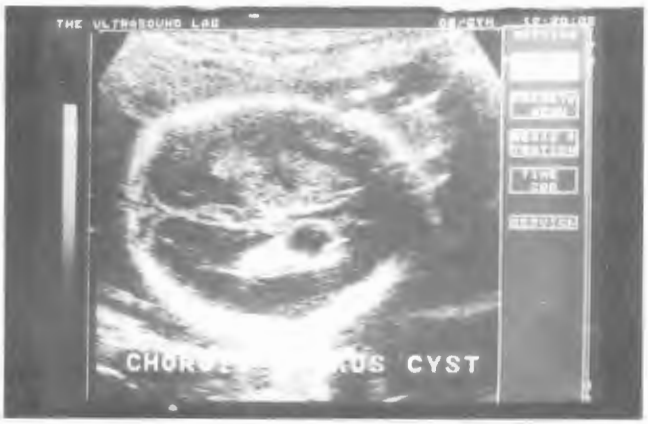

Fig.1. Unilateral large choroid plexus cyst in a 19 week fetus.

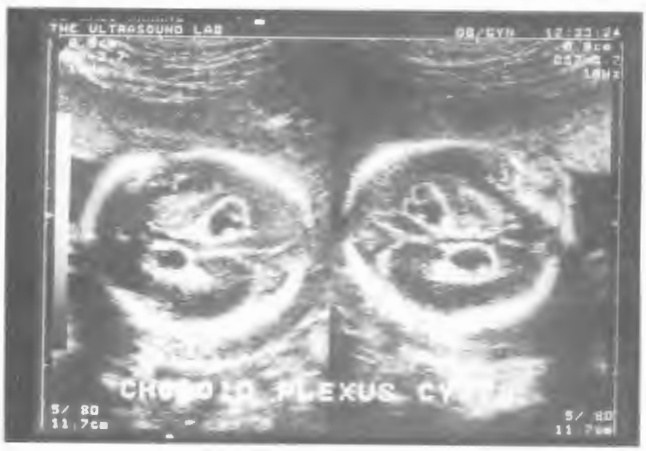

Fig 2: Multiple bilateral choroid plexus cysts in a 17 week fetus.

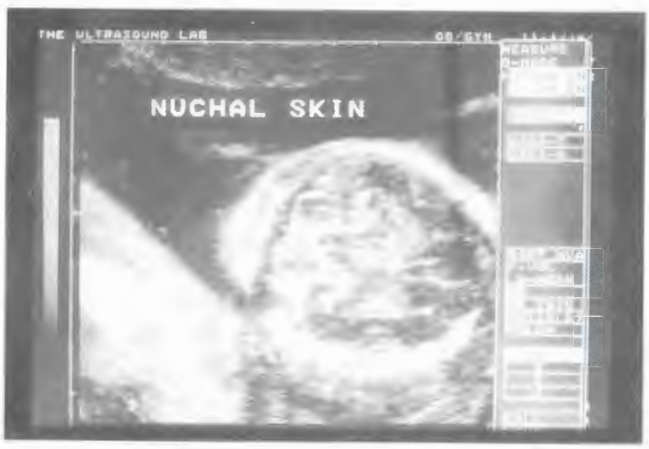

Fig. 3: Thickened Nuchal Skin Fold in a fetus with choroid plexus cyst. Karyotyping showed Trisomy 21.

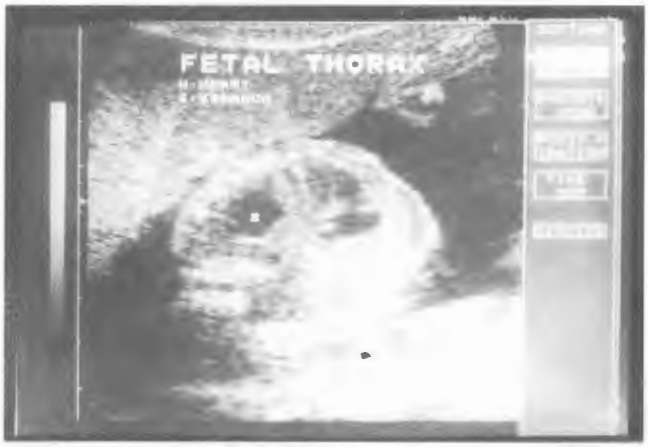

Fig. 4: Diaphragmatic herniation of the stomach in a fetus with choroid plexus cyst. Amniocentesis yielded a Trisomy 18 karyotype.

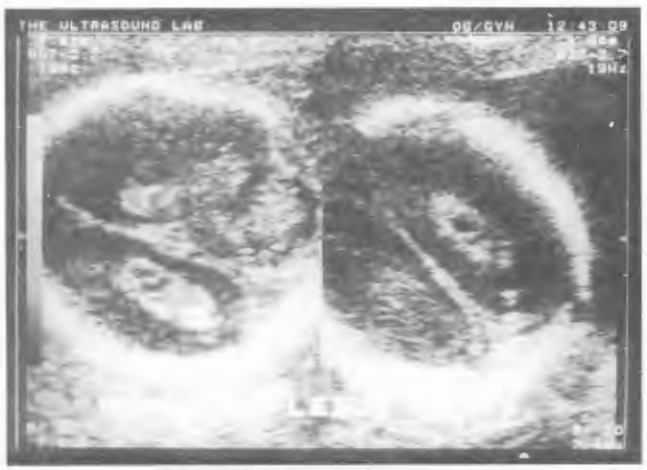

Fig. 5a. 18 week fetus with bilateral choroid plexus cysts. There was no dysmorphic developmental anomaly on sonography. Normalcy was confirmed after birth.

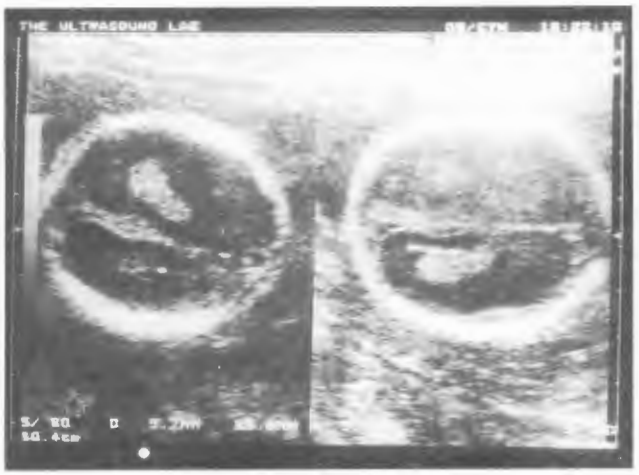

Fig. 5b. Same fetus as in Figure 5a at 22 weeks. The choroid plexus cysts have resolved.

Discussion and Conclusion

Choroid plexus cysts have attracted a great deal of attention because of an association with chromosomal abnormalities, particularly Trisomy 18, and the need for aggressive antenatal diagnosis. Choroid plexus cysts are seen in approximately 2-3% of normal fetuses during the second trimester compared with 30% of fetuses with Trisomy 18. However, Trisomy 18 is relatively rare, and most affected fetuses have multiple dysmorphic anomalies that can be detected by sonography. The present study suggests that the presence of choroid plexus cysts should initiate a search for associated dysmorphic anomalies, but, in the absence of additional abnormalities, is probably not an indication for karyotyping by itself.

This is in concurrence with meta-analysis available in recent literature. Gross et al in 1995 showed that among 13 of the larger studies addressing the issue of a karyotyping in fetuses detected to have choroid plexus cysts, the risk of aneuploidy in isolated choroid plexus cysts was 1 in 374. Gupta et al (1995) in 1995 also evaluated multiple studies and showed a risk of 1 in 150 of aneuploidy in isolated choroid plexus cysts. When other dysmorphic developmental anomalies were delineated the risk rose to 1 in 3. Benacerraf and Labodh (1989) put the association of isolated choroid plexus cysts and chromosomal abnormalities at 1 in 477. All these studies show a risk factor of an anomaly considerably lower than the usually accepted cutoff of amniocentesis related pregnancy loss (1 in 250).

Although one case in our series showed a Trisomy 21 karyotype this is not statistically significant. This is in agreement with the reports of Gupta et al (1995) who found a risk of 1 in 880 of Down's Syndrome in fetuses with choroid plexus cysts.

Since the incidence of Trisomy 18 is increased in patients with advanced maternal age and low levels of maternal serum alpha-fetoprotein (AFP), unconjugated estriol (Free E_3) and human chorionic gonadotropin (HCG) it is reasonable to, in the future, work out a fresh at-risk factor in patients who have isolated choroid plexus cysts, advanced maternal age and low triple markers. This needs evaluation in a larger planned study.

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