Choroid plexus cysts and their anatomical correlations: importance in fetal prognosis. A critical literature review

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Abstract

The objective of this investigation was to carry out a literature review on the choroid plexus cysts and their importance in fetal prognosis with search on *PubMed*, *Web of Science*, *LILACS* and *MEDLINE* databases using keywords in English without time restrictions. Choroid plexus cysts, which are also termed intraventricular neuroepithelial cysts, choroid epithelial cysts or ependimary cysts, are structures localized within the interior of the lateral ventricles, comprising secretory epithelium, the principal function of which is the production of cerebrospinal fluid. Ultrasound images of the choroid plexuses within the lateral ventricles consist of hyperechogenic structures at the level of the body, trigon and inferior horn of the ventricles. Between the 16th and the 20th week of gestation, cystic structures in the interior of the ventricular plexuses may be observed at a rate of 0.3-1.0% in an ultrasound examination of the fetal head, as is shown. Most choroid plexus cysts regress spontaneously without after effects, although there is a possible association with chromosomal abnormalities, notably trisomy 18 (Edwards' syndrome) and less frequently cited, trisomy 21 (Down syndrome). The presence of this kind of cyst is considered to be a lesser marker for them, except when combined with other more frequently used and accepted ecographic markers, such as nuchal translucency, intracardiac echogenic focus and others. Its isolated presence as the sole characteristic of this aneuploidy is rare. The majority of choroid cysts are transitory and of little clinical significance, and may be identified through a computerized tomography examination. As such, the existence of isolated choroid cysts does not indicate the confirmation of chromosomal aneuploidies so much as an alarm that should trigger an investigation in greater depth in search of other more important markers, emphasizing the importance of pre-natal monitoring.

Keywords: choroid plexus, cysts, fetal prognosis.

1 Introduction

Choroid plexus cysts, which are also termed intraventricular neuroepithelial cysts, choroid epithelial cysts or ependimary cysts, are structures localized within the interior of the lateral ventricles, comprising secretory epithelium, the principal function of which is the production of cerebrospinal fluid, according to Nicolaides, Rodek and Gosden (1986) and Hertzberg, Kay and Bowie (1989). They are asymptomatic artifacts found particularly during Magnetic Resonance and Ultrasound and relatively common in autopsies and are generally less than 1 cm in size, although if larger, they produce obstructive symptoms according to Melo, Garcia, Fernandes et al. (2003).

Ultrasound images of the choroid plexuses consist of hyperechogenic structures at the level of the body, trigon and inferior horn of the ventricles (VINTZILEOS et al., 1998). Between the 16th and the 20th week of gestation, cystic structures in the interior of the ventricular plexuses may be observed at a rate of 0.3-1.0% in an ultrasound examination of the fetal head.

Various recent ultrasound findings have been attributed to trisomy 18, such as choroid plexus cysts and retardation of fetal growth (30-60%).

Most choroid plexus cysts regress spontaneously without other effects, although there is a possible association with chromosomal abnormalities, notably trisomy 18 and less frequently cited trisomy 21. The presence of this kind of cyst is considered to be a lesser marker for them, except when combined with other more frequently used and accepted ecographic markers, such as nuchal translucency, intracardiac echogenic focus and others. Its isolated presence as the sole characteristic of this aneuploidy is rare. The majority of choroid cysts are transitory and of little clinical significance, being habitually associated with other alterations in fetal anatomy identified by echography.

This study aims to highlight the importance of choroid plexus cysts in fetal prognosis.

2 Material and methods

An updated bibliographical survey from 1985 to the present was carried out, emphasizing the association between the presence of cysts of the choroid plexus and fetal aneuploidy. *PubMed*, *Web of Science*, *Scielo*, *LILACS* and *MEDLINE* databases were used as search reference using keywords in English and no time restrictions. In this systematization, papers were searched, using the keywords "cyst", "choroid plexus" and "fetal prognosis", by randomized combination until these terms were included.

3 Results

According to Paz (2001), the cyst is observed in 1% of normal fetuses and in 50-60% of fetuses with trisomy 18,

representing a significant difference. Since this is not an exclusive characteristic of Edwards' syndrome, it is also seen at lower frequency in Down, Turner's and Klinefelter's syndrome, as well as in other triploidies. 80% of cases of trisomy of chromosome 18 present cysts of the choroid plexus.

Vintzileos (1998), cited that for the majority of fetuses with choroid plexus cysts, the presence of them increases the risk of trisomy by a factor of 13.8. It was not found a report of increased risk of Down syndrome (Chromosome 21) in the presence of isolated choroid plexus cysts (GUPTA, CAVE, LILFORD et al. 1995; GUPTA, KHAN, THORNTON et al., 1997);

In the three cases of children with isolated choroid plexus cysts studied by Herini, Tsuneishi, Takada et al. (2003), two merely presented an increase in the cephalic circumference without the presence of trisomies, with one case showing the presence of sacral myelomeningocele. All three presented normal neuropsychomotor development with only one case of after-effects due to ventroculomegaly.

According to Whitlow, Lazanakis, Kadir et al. (1998), the majority of aneuploidies were detected by increased nuchal translucency and/or the presence of structural anomalies (78%; 25/32) including cyst of the choroid plexus, albeit never in isolation, but as in combination. There was no increase of the risk of aneuploidy with regard to the number of cysts, or their size or bilaterality (GROSS, SHULMAN, TOLLEY et al., 1995).

4 Conclusion

Paz (2001), is of opinion that the presence of choroid plexus cysts as a characteristic of Edwards' syndrome (trisomy of chromosome 18) is controversial within the literature, without any consensus on when amniocentesis should be recommended in the event of an isolated cyst, i.e. in the absence of other abnormalities. Various authors such as Gross, Shulman, Tolley et al. (1995) and Gratton, Hogge and Aston (1996) have highlighted the need to investigate cysts by invasive methods, examinations such as amniocentesis or cordocentesis for the study of karyotype only if other important cited markers are present.

Reinsch (1997) and Sullivan, Giudice, Vaveldis et al. (1999) highlight the need for invasive procedures such as amniocentesis for the study of karyotypes only if other risk factors are present, such as: dismorphies in ultrasound examinations, advanced maternal age, obstetric precedents of chromosomal abnormalities or positive serological markets (altered triple test). Against this, Morcos, Platt, Carlson et al. (1998) recommends amniocentesis and the study of karyotypes in the presence of choroid plexus cysts, even where these are isolated.

Gupta, Cave, Lilford et al. (1995) demonstrated that the risk of trisomy 18 in fetuses without choroid plexus cysts is 1:4,600 in 20-year-old women and 1:90 in 45-year-old women.

Gratton, Hogge and Aston (1996) demonstrated that 80% of cases of trisomy 18 which had visible cysts in ultrasonography also presented other associated ultrasound abnormalities, corroborating two other studies by Ferriman, Linton, Woods et al. (1998) (97%) and Snijders, Shawa and Nicolaides (1994) (97%) respectively. For Gupta, Cave, Lilford et al. (1995) and Gupta, Khan, Thornton et al. (1997) there are no reports of an increase in the incidence of Down syndrome in the presence of isolated choroid plexus cysts, but according to Chervenak, Isaacson and Campbell (1993) the cysts may be observed until the second quarter in normal fetuses, regressing spontaneously, but may also be associated with trisomy 18.

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