

Correlation between isolated sonographic finding of umbilical cord cyst and fetal anomalies*

Correlação entre o achado ultra-sonográfico isolado de cisto de cordão umbilical e anomalias fetais

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Abstract **OBJECTIVE:** To correlate the isolated sonographic finding of umbilical cord cyst with fetal anomalies such as chromosomopathies and structural changes. According to the medical literature, the clinical implications of the sonographic finding of umbilical cord cyst in the second and third trimesters of pregnancy are well established; however, the meaning of this finding in the first trimester still remains controversial. **MATERIALS AND METHODS:** A retrospective study was developed with consecutive, pregnant women with single living fetuses presenting with umbilical cord cyst as an isolated finding, over a 10-year period (1996–2006). Ultrasound studies were performed in all cases for screening of fetal anomalies after the diagnosis of umbilical cord cyst. Neonates and umbilical cords were evaluated after delivery for the presence of abnormalities. **RESULTS:** Nine cases presenting umbilical cord cyst as a sole finding with no other sonographic marker for fetal abnormality were evaluated. Two cases were detected in the first pregnancy trimester and seven cases in the second and third trimesters. Fetal cytogenetic study was done by means of amniocentesis in two cases. No newborn presented with structural anomalies or aneuploidy. **CONCLUSION:** Isolated sonographic finding of umbilical cord cyst did not imply increased risk for fetal structural anomalies or aneuploidies. **Keywords:** Fetus; Umbilical cord; Cysts; Ultrasonography; Chromosome aberrations.

Resumo **OBJETIVO:** Correlacionar o achado ultra-sonográfico isolado de cisto de cordão umbilical com anomalias fetais, como cromossomopatias e alterações estruturais. Segundo a literatura médica, as implicações clínicas do achado ultra-sonográfico de cisto de cordão nos segundo e terceiro trimestres de gestação estão bem estabelecidas, entretanto, quando no primeiro trimestre, o significado ainda permanece controverso. **MATERIAIS E MÉTODOS:** Foi realizado estudo retrospectivo de gestantes da população geral, consecutivas, com fetos únicos e vivos, que apresentavam apenas o achado de cisto de cordão umbilical, num período de dez anos (1996–2006). Em todos os casos foram realizados exames ultra-sonográficos para o rastreamento de anomalias fetais após o diagnóstico de cisto de cordão. Os recém-nascidos e o cordão umbilical foram examinados após o parto para se verificar a presença de anomalias. **RESULTADOS:** Foram estudados nove casos que apresentavam cisto de cordão umbilical como único achado, sem outros marcadores ultra-sonográficos de anomalias fetais. Detectaram-se dois casos no primeiro trimestre de gestação e sete nos segundo e terceiro trimestres. Dois casos foram submetidos a estudo citogenético fetal, por meio de amniocentese. Nenhum recém-nascido apresentou anomalias estruturais ou aneuploidia. **CONCLUSÃO:** O achado ultra-sonográfico isolado de cisto de cordão umbilical não significou aumento de risco para anomalias estruturais ou aneuploidias.

Unitermos: Feto; Cordão umbilical; Cistos; Ultra-sonografia; Aberrações cromossômicas.

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INTRODUCTION

Ultrasonography has been acknowledged as the modality of choice in routine prenatal examination, and has been utilized for more than three decades for this purpose in the obstetric practice. Previously

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utilized for detecting the presence of pregnancy, fetal vitality and singleton or twin gestation, obstetric ultrasonography currently is useful in the diagnosis of small anatomical abnormalities or even to suggest the presence of chromosomal anomalies. Therefore, this method is a useful tool in pregnancy prediction and management, considering that the majority of pregnant women with malformed fetuses do not present any risk factor⁽¹⁾.

Among sonographic malformation markers, umbilical cord anomalies involving vascular, structural findings, cysts and masses may be found.

The umbilical cord develops around the seventh gestational week, and can be sonographically visualized at the eighth week^(1,2).

Umbilical cord cysts are divided into true and pseudo-cysts, the first ones presenting an epithelial lining and subdivided into omphalomesenteric duct cysts and allantoic duct cysts.

Generally, true umbilical cord cysts develop in the fetal end of the umbilical cord, and reports in the literature describe their association with gastrointestinal and genitourinary tracts abnormalities (omphalocele, persistence of the urachus and obstructive uropathy)^(1,3,4).

On the other hand, pseudo-cysts do not present an epithelial lining, and originate from a focal edema of the Wharton's jelly or from its absence because of degenerative alterations. Pseudo-cysts are more frequent than the true ones and have been found in cases of omphalocele and 18 trisomy⁽⁵⁾.

The exact differentiation between true and pseudo-cysts cannot be achieved by means of ultrasonography; this can be done only by means of a histopathologic study.

Differential diagnosis of umbilical cord cysts is umbilical cord masses including

tumors, hematomas, varices and aneurysms (Figure 1).

The present study is focused on umbilical cord cyst which has been associated to fetal anomalies and aneuploidies since 1988 in a study developed by Jauniaux et al.⁽⁶⁾. Correlation between umbilical cord cysts and anomalies was found in up to 50% of cases, particularly in findings occurred at the second and third trimesters of gestation. It is believed that the finding of umbilical cord cysts in such conditions constitute an indication for a karyotype study during the pregnancy.

However, other studies⁽⁷⁻¹⁴⁾ have evaluated the presence of cord cysts at the first gestational trimester, observing the transitory nature of this finding and birth of normal children in most of cases, so fetal karyotyping would not be justified.

In the literature, the prevalence of umbilical cord cysts sonographically found at different gestational ages ranges between 0.4% and 3.4%^(7,8).

The present study was aimed at correlating the isolated sonographic finding of umbilical cord cyst and fetal anomalies such as chromosomopathies and structural alterations.

MATERIALS AND METHODS

During the period from June 1996 to June 2006, nine cases of prenatal sono-

graphic studies whose sole finding had been the presence of umbilical cord cyst. These cases were collected and recorded in two units of obstetric ultrasonography utilizing equipment Logic 500, Voluson 730 and Logic 9 (GE Medical Systems; Wisconsin, EUA) with convex multifrequency (3-5 MHz) and transvaginal (5-8 MHz) transducers.

The cases identified involved non-selected, consecutive, pregnant women from the general population, with single living fetuses.

Umbilical cord cyst has been defined as an anechoic, thin-walled structure occurring along the umbilical cord and within the amniotic cavity, separated from the fetal pole and physiological herniation. Cord cyst is distinguished from the vitelline sac that has more echogenic walls and extra-amniotic location. Color Doppler was utilized for excluding differential diagnoses (Figure 2). The parameters considered in the present study are the same adopted in previous studies in the literature, and are aimed at reducing the incidence of diagnostic failure, as well as optimizing the ultrasonographic method^(9,10).

Some of the cases were offered a cytogenetic study by means of amniocentesis conducted by an obstetrician, with no interference from the investigator. Additionally, data regarding postnatal physical health outcomes for the neonates and presence, or not, of structural anomalies were collected by means of a review of the patients' records and contact with involved obstetricians and pediatricians.

RESULTS

In the present study, nine prenatal ultrasonographic images presenting umbilical cord cyst as the sole abnormality finding with no other marker for fetal abnormality were evaluated, two of them at the first gestational trimester (Figure 3) and seven at the second and third gestational trimesters (Figure 4). Table 1 demonstrates the results of this evaluation.

It is important to note that in all of the cases the cord cyst was medially located in relation to the length of the umbilical cord, and eccentrically located (para-axial) in relation to its diameter.

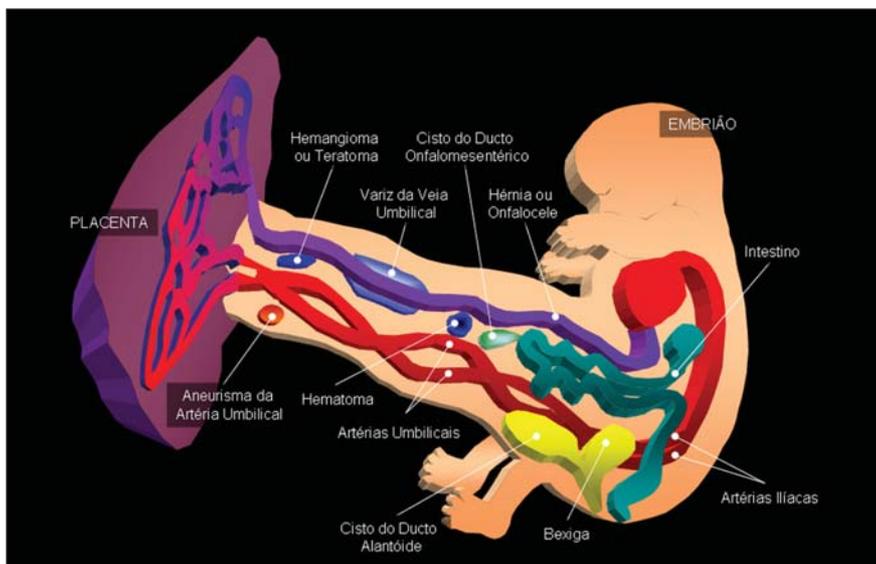


Figure 1. Umbilical cord masses. (Modified from: Callen PW, editor. *Ultra-sonografia em obstetrícia e ginecologia*. 4ª ed. Rio de Janeiro: Guanabara Koogan; 2002. p. 587).

Table 1 Description of cases.

Case	Cyst localization	Gestational age (DLM)	Indication for the study	Cyst diameter (mm)	Postnatal outcome
1	Median* / Eccentric†	8 weeks and 3 days	Routine	2.5	Normal
2	Median / Eccentric	27 weeks and 7 days	Routine	30.7	Normal
3	Median / Eccentric	9 weeks	Routine	3.0	Normal
4	Median / Eccentric	22 weeks and 4 days	Routine	32.0	Normal‡
5	Median / Eccentric	30 weeks and 2 days	Routine	17.2	Normal
6	Median / Eccentric	23 weeks	Routine	27.3	Normal
7	Median / Eccentric	21 weeks and 1 day	Routine	25.7	Normal‡
8	Median / Eccentric	22 weeks and 3 days	Routine	13.8	Normal
9	Median / Eccentric	26 weeks and 4 days	Routine	30.0	Normal

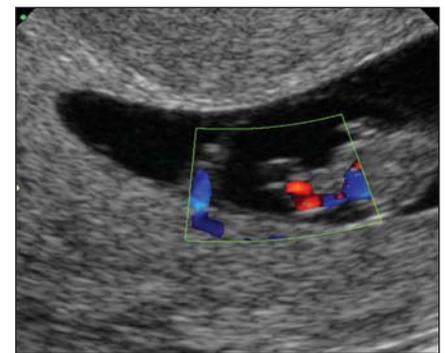
* Location of the cyst in relation to the cord length; † Location of the cyst in relation to the cord circumference; ‡ Cases with normal karyotyping. DLM, date of the last menstruation.



Figure 2. Case 3. Transverse, cystic image eccentrically located in the umbilical cord, within the amniotic membrane. The vitelline vesicle is shown at left.



A

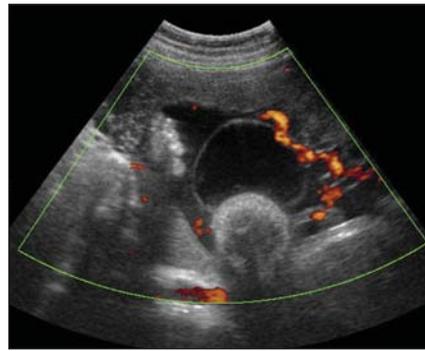


B

Figure 3. Case 1. **A:** Cystic image at the first gestational trimester, within the amniotic membrane. **B:** Utilization of Color Doppler allows the correlation between the image and the umbilical cord.



A



B

Figure 4. Case 4. **A:** Sonographic image of a thin-walled cyst proximal to the placenta and the fetus. It is possible to observe that the cystic content is anechogenic, even in relation to the amniotic fluid that presents fine debris. **B:** The utilization of Power Doppler demonstrates the umbilical cord vessels.

Postnatal outcome was normal in all of the cases. Karyotyping by amniocentesis also resulted normal.

DISCUSSION

Umbilical cord cyst etiology is still not completely known, but it is believed to be

related to an increase in the hydrostatic pressure inside the umbilical cord vessels and extravasation of fluid content into the involved region^(10,11). At the first gestational trimester, the increase in the hydrostatic pressure may be associated with the presence of a physiological herniation of the primitive bowel, whose presence and

disappearance are compatible with reports of transitory umbilical cord cysts in this gestational period. On the other hand, at the second and third gestational trimesters, the increase in the hydrostatic pressure could be a result from fetal growth restriction, omphalocele and chromosomopathies⁽¹⁰⁾.

As previously mentioned, the prevalence of umbilical cord cysts ranged between 0.4% and 3.4%^(8,9). The high prevalence reported by Ross et al.⁽⁹⁾ can be attributed to the high-risk population involved in the evaluation. In other studies, the prevalence, in general, were lower⁽¹¹⁾. Additionally, the mentioned study has demonstrated that, amongst the first-trimester cysts with spontaneous resolution, i.e., the transitory cases, association with fetal abnormalities has occurred in 13% of cases, achieving 26% including cases of the cyst persistence up to the third gestational trimester. An increase was observed in the risk for fetal abnormality in cases of cord cysts located near the cord placental or fe-

tal insertion, eccentric location in relation to the longitudinal axis of the cord, and in cases of cysts with larger diameter.

Other two studies^(8,10) have evaluated a total of 18 cases of umbilical cord cysts at the first gestational trimester, with normal postnatal outcomes in all of them. A study developed by Sepulveda et al.⁽¹⁰⁾ has concluded that the incidental detection of umbilical cord cysts at the first gestational trimester is not associated with a unfavorable prognosis for the gestation, as far as a low-risk population is concerned. On the other hand, Skibo et al.⁽⁸⁾ indicate the first-trimester umbilical cord cyst as a new, many times transitory, sonographic finding, not associated with fetal abnormalities, and that may be clinically non-significant.

The mentioned authors⁽⁸⁻¹⁰⁾ consider that the persistence of the finding exceeding the first trimester should be differently approached and even indicate fetal karyotyping, considering the higher risk for aneuploidies and malformations.

So far, the approach suggested by studies in the literature is that a fetal karyotyping is performed in cases of umbilical cord cyst persistence at the second and third gestational trimesters, and detailed ultrasonographic studies are performed by an experienced sonographer for evaluating the fetal anatomy looking for other markers for

fetal abnormalities in cases of umbilical cord cysts at the first gestational trimester. It is important to note that, unlike in the present study, in the majority of above mentioned studies the finding of umbilical cord cyst was not isolated.

Therefore, despite the small casuistic in the present study, the isolated sonographic finding of umbilical cord cyst did not represented an increase in the risk for structural or aneuploidic anomalies. Based on these findings, there is no indication for fetal karyotyping in cases of isolated sonographic finding of umbilical cord cyst at any gestational age.

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