Clinical Case Reports International

Case Presentation
Published: 27 Feb, 2023

# Omphaloceles: Antenatal Diagnosis and Treatment

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## Abstract

The omphalocele is a rare malformation. Its frequency is estimated 1/5000 of births. This malformation results from a closing defect of the abdominal wall and it's frequently associated with other chromosomic abnormalities.

We report three new cases of omphaloceles accrued to a 27-, 43- and 39-years old patients. The diagnosis by the ultrasound exam was easy and precocious.

A fetal karyotype was performed to all the patients and was constantly abnormal. Therefore, the medical interruption of the pregnancy was practiced to the three patients. On the occasion of those three observations and a literature review, we remind the diagnostic aspects and the management modalities of this congenital malformation.

**Keywords: Omphalocele; Parietal defect; Chromosomal aberration; Trisomy 18**

## Introduction

Abdominal wall closure defects include a set of parietal malformations. Omphaloceles are the typical forms of these parietal defects. Their frequency is estimated at 1/5000 deliveries. Thanks to advances in ultrasound, the antenatal diagnosis of these malformations is made possible from 11 to 12 weeks of amenorrhea. The importance of these conditions lies in their frequent association with chromosomal or malformative abnormalities [1]. Which can therefore constitute a sign of chromosomal aberrations.

### Observation 1

Ms. M.O, 27-years-old, is a third gestity, nulliparous and having two early miscarriages. She had consulted for prenatal follow-up of a 3-month pregnancy (Unclear Last Menstrual Period). The obstetrical examination was normal.

The obstetrical ultrasound showed an evolving monofetal pregnancy. The biometrics (Biparietal diameter: 20 mm, Craniocaudal length: 49 mm) corresponded to a pregnancy of 12 weeks. We also noted a well-defined round echogenic formation 13 mm in diameter hanging from the anterior abdominal wall (Figure 1), without any other visible malformation, in particular no nuchal anomaly. A fetal karyotype on amniotic fluid, done at 15 weeks, concluded in trisomy 21. A medical termination of the pregnancy was decided. This was performed with intravaginal Cytotec (1 tablet every 6 h). The expulsion had taken place after the 3rd tablet. The foeto-pathological examination confirmed the diagnosis of omphalocele and revealed a wide cleft lip and palate.

### Observation 2

A 43-year-old patient, tenth gestity, ninth parity, she had no particular notable history or malformations in the family. She had consulted for excessive uterine height at a term of 22 weeks.

The obstetrical ultrasound had shown a one progressive pregnancy. The fetus presented a round, well-defined 06 diameter mass with echogenic content and inside a small trans-sound image (digestive structure) had been objectified. The umbilical cord was inserted on the surface of this mass (Figure 2). We also found manifest polyhydramnios and extreme shortness of the long bones (humerus, femur). The diagnosis retained was that of omphalocele associated with micromyelia. A fetal karyotype was performed and concluded with trisomy 18. Therapeutic termination of pregnancy was indicated at 24 weeks. Expulsion of a female stillborn of 800 gr. The foeto-pathological examination had concluded to polymalformative syndrome with extreme shortness of the long bones, a giant omphalocele containing small loops, the stomach and the left liver (Figure 3), as well as a diaphragmatic hernia, hypoplasia of the left heart with mitral atresia indicating a Cantrell pentalogy (Figure 4).

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**Received Date**: *09 Feb 2023*
**Accepted Date:** *23 Feb 2023*
**Published Date:** *27 Feb 2023*

***Citation:***
*Ragmoun H, Riadh M, Sarra H, Rihab Y, Montacer H. Omphaloceles: Antenatal Diagnosis and Treatment. Clin Case Rep Int. 2023; 7: 1494.*

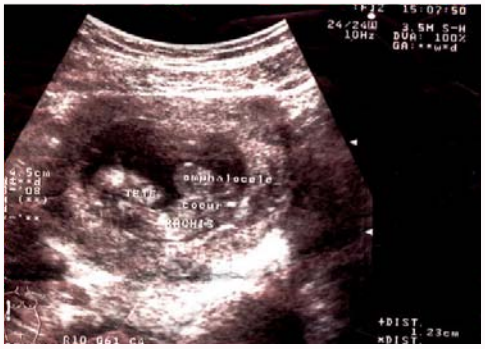


**Figure 1:** Well-limited echogenic formation hanging from the ventral wall of a 10 weeks fetus.

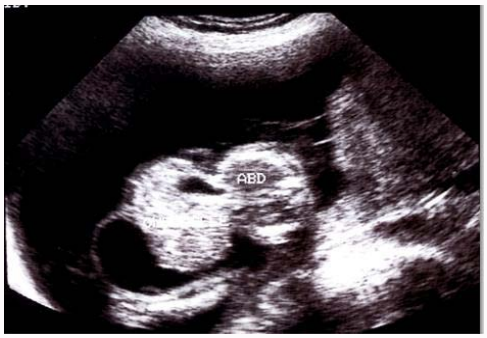


**Figure 2:** Ultrasound appearance of the omphalocele containing the gastric clarity and on the surface of which the umbilical cord is inserted.

## Discussion

Omphalocele is a defect of central seat ventral closure resulting in herniation of the abdominal viscera into the base of the umbilical cord. Its frequency varies according to the series from 1/2000 to 1/5000 births [1]. Maternal age does not appear to be a risk factor [2-4]. Some authors report a greater incidence in male fetuses [5,6]. In our work, two of our patients were over 38 years old and the omphaloceles had affected two boys and one girl.

The review of the literature reveals several family cases, whether they are recurrences of omphalocele in the same patients or sporadic family cases including 8 cases of omphalocele in the same family [1,7]. These familial cases of omphalocele suggest the possibility of a genetic origin [3]. This risk of recurrence, although low, justifies genetic counseling both in isolated omphaloceles and those associated with a polymalformative syndrome [6]. None of our patients had similar cases in the family.

### Diagnosis

Thanks to advances in ultrasound, antenatal diagnosis of omphaloceles made possible from the first trimester. This diagnosis is usually easy. The omphalocele presents as a new formation hanging from the anterior abdominal wall, its limits are clear and distinct from the abdominal contour. Inside this mass there are heterogeneous echoes, intestinal loops or even hypoechoic images corresponding to the liver as well as many other organs that can be highlighted. At the same time, we have an abdominal circumference reduction. Furthermore, it is easy to find the implantation of the umbilical cord at the level of this mass.

Before the 11$^{th}$ week, it is difficult to confirm the parietal defect

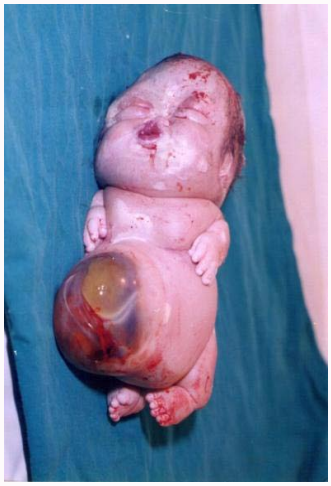

**Figure 3:** Giant omphalocele with severe micromelia at 20 weeks.

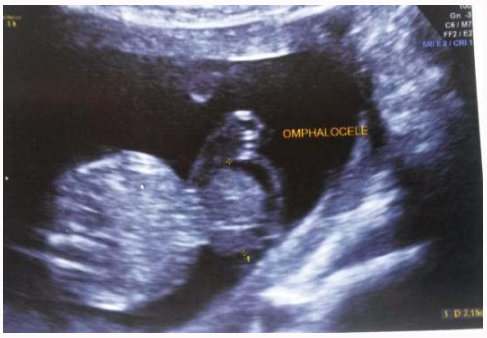


**Figure 4:** Obstetric ultrasound at 15 weeks: Transversal section of the abdomen showing a well-limited echogenic formation located at the base of the umbilicus.

[5,8]. However, the presence of an echogenic, homogeneous swelling larger than the abdominal diameter would be in favor of the diagnosis rather than a simple delay of intestinal reintegration into the abdominal cavity [2,9]. In our cases the diagnosis of omphalocele was easy, including the case revealed in the first trimester of pregnancy.

The presence of excess amniotic fluid should lead to a search for an associated congenital malformation. Isolated omphaloceles not accompanied by polyhydramnios [2]. In our observations, the only patient who had polyhydramnios actually presented an omphalocele associated with a polymalformative syndrome such as pentalogy of Cantrell.

### Differential diagnosis

Omphalocele must be differentiated from a sacrococcygeal teratoma, particularly in cases of inferior colostomy. It should not be confused with an allantoic cyst of the umbilical cord, the latter is purely liquid, projecting on the umbilical path, on the other hand the abdominal wall is intact.

### Chromosomal abnormalities

In any omphalocele, a fetal karyotype is systematically performed. Chromosomal abnormalities are consistently found in 20% to 54% of cases. It was performed in all three patients. The chromosomal formulas were constantly pathological (two cases of trisomy 18 and one trisomy 21). This risk of chromosomal aberration is increased by advanced maternal age and by the coexistence of omphalocele with

other congenital malformations [1]. In literature, the most frequently found chromosomal abnormalities are trisomy's 13, 18, 21 [4,6].

### Associated malformations

The association of omphaloceles with other malformations has also been reported in numerous series. The most commonly found anomalies are cardiac anomalies (CIV, CIA, tetralogy of Fallot...), skeletal and limb anomalies (club foot, Amelia, etc.) and the nervous system malformations (anencephaly, hydrocephalus, spina-bifida), renal malformations.

### Three syndromes deserve to be individualized

- Cantrell's pentalogy which combines a voluminous epigastric omphalocele containing the abdominal viscera, aplasia of the lower part of the sternum, anterior defect in the diaphragm, absence of the anterior pericardium and cardiac malformations.

- Inferior colostomy which includes a hypogastric omphalocele with vesicointestinal fistula and anal imperforation, or bladder exstrophy.

- Beckwith-Wiedemann syndrome: This is an autosomal transmission condition with a 27% risk of recurrence [1,9]. It is defined by the triad: Omphalocele, Macroglossia and gigantism, with a difficult ultrasound diagnosis.

### Management and treatment

The obstetric management of omphaloceles depends on whether they are isolated or associated with chromosomal or malformative abnormalities. Thus, in the event of polymalformative syndrome or associated chromosomal anomaly, therapeutic termination of pregnancy is indicated [1,3].

On the other hand, in isolated omphaloceles, continuation of the pregnancy is authorized subject to regular prenatal monitoring and delivery in a specialized center with immediate surgical management [10]. Under these conditions, recent studies report that the neonatal survival is close to 100% [3,5,6]. In our work, the demonstration of chromosomal abnormalities and/or other associated malformations justified termination of pregnancy in all cases.

## Conclusion

Omphalocele is a rare embryopathy: 1/5000 births. Thanks to ultrasound, prenatal diagnosis is made possible from 12 weeks of pregnancy. This diagnosis is usually easy to make. Before any omphalocele, a fetal karyotype is systematically performed. A chromosomal aberration is found in 20% to 54% of cases. In addition, the search for associated malformations is essential, these affect one out of two patients.

The prognosis of omphaloceles depends more on the presence of congenital malformations or associated chromosomal abnormalities than on its size. In isolated omphaloceles neonatal survival is approximately 100% of cases.

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