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Giant Isolated Omphalocele Discovered at Birth in a Term Neonate: A Case Report

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Abstract:

- ***Introduction:***

Omphalocele is a congenital malformation of the anterior abdominal wall characterized by herniation of abdominal contents through the umbilical ring, covered by a membranous sac. Its incidence is estimated at 1 per 4,000 to 7,000 live births. Although it is often detected on prenatal ultrasound, it may exceptionally be discovered at birth.

- ***Case Report:***

We report the case of a male newborn weighing 3,560 g, born at 38 weeks and 6 days of amenorrhea to a 37-year-old multigravid woman (G4P3) with no notable personal or family history and no known consanguinity. The pregnancy had been monitored by a general practitioner, with normal reported fetal biometry. Delivery was vaginal, with an Apgar score of 10. Clinical examination at birth revealed a large omphalocele with an intact sac containing bowel loops.

- ***Conclusion:***

This case illustrates the importance of rigorous prenatal ultrasound surveillance and immediate multidisciplinary management in the delivery room. The incidental discovery of an omphalocele at birth, without antenatal diagnosis, highlights the limitations of obstetric follow-up performed exclusively in primary care.

Keywords: *Omphalocele; Abdominal Wall Defect; Term Neonate; Missed Antenatal Diagnosis; Pediatric Surgery Mots-Clés: Omphalocèle; Malformation De La Paroi Abdominale; Nouveau-Né À Terme; Diagnostic Anténatal Manqué; Chirurgie Pédiatrique.*

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I. INTRODUCTION

Omphalocele (or exomphalos) is a rare congenital malformation of the anterior abdominal wall resulting from failure of closure of the lateral embryonic folds between the third and fourth week of development. It is characterized by the persistence of a herniation of abdominal viscera — which may include bowel loops, the liver, or other organs — through the umbilical ring, covered by a sac composed of peritoneum and Wharton's jelly into which the umbilical cord inserts.

Its incidence varies across studies between 1/4,000 and 1/7,000 live births. Omphalocele is associated in 30 to 70% of cases with other malformations or chromosomal anomalies (trisomy 13, trisomy 18, Beckwith-Wiedemann syndrome), which makes its antenatal diagnosis of major importance for genetic counseling and therapeutic planning.

Prenatal diagnosis by ultrasound is possible as early as the first trimester of pregnancy and allows for planned multidisciplinary management. Nevertheless, cases of postnatal diagnosis do exist, particularly in contexts of insufficient medical follow-up or when the malformation is isolated with an intact, poorly visible sac. We report here a

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case of neonatal omphalocele discovered at birth in a context of prenatal follow-up by a general practitioner without specialized ultrasound assessment.

II. CASE REPORT

This is a 37-year-old woman, gravida 4 para 3 (G4P3), who had carried three previous pregnancies to term, all delivered vaginally with living, healthy children. She had no notable medical or surgical history. There was no consanguinity and no exposure to teratogenic drugs or to medicinal herbs during this pregnancy.

The current pregnancy, estimated at 38 weeks and 6 days of amenorrhea based on the last menstrual period (LMP), had been followed by a general practitioner. The reported prenatal biometry was normal, with no anomalies signaled.

The patient was admitted to the delivery room in advanced labor: cervix 100% effaced, dilated to 4 cm. Delivery was vaginal and uncomplicated. The newborn was male, with a birth weight of 3,560 g and an Apgar score of 10 at both the first and fifth minutes of life, indicating excellent immediate neonatal adaptation.

On systematic clinical examination of the newborn in the delivery room, a large omphalocele was incidentally discovered. The sac was intact, globular in shape, with a smooth and shiny surface and a grey-greenish membranous appearance; the umbilical cord, clamped, inserted into its upper portion. The sac most likely contained bowel loops, with no signs of rupture or immediate vascular compromise. The abdomen outside the defect appeared flat. No other malformations were visible apart from the omphalocele.

The newborn was immediately transferred to the neonatal intensive care unit for specialized management (protection of the sac, central venous access, gastric decompression by nasogastric tube, infectious workup, and scheduling of surgical correction).

Fig 1 Giant Omphalocele with Intact Membranous Sac Discovered at Birth. Intrapartum View of tthe Abdomen of a Term Newborn (38 Weeks + 6 Days, 3,560 G, Apgar 10). A Large Globular Umbilical Mass is Noted, with an Intact and Shiny Grey-Greenish Membranous Wall; The Clamped Umbilical Cord Inserts Into its Upper Portion. The Contents of the Sac Most Likely Include Bowel Loops. No Signs of Rupture or Visible Necrosis.

III. DISCUSSION

This case presents several remarkable features that warrant discussion.

- *Failure of Prenatal Diagnosis*

The most striking aspect of this observation is the absence of prenatal diagnosis despite a pregnancy carried to term. An omphalocele with hepatic contents — likely in this case given the volume and appearance of the sac — usually has a diameter greater than 4 cm and should be detectable as early as the first-trimester ultrasound (increased nuchal translucency, persistent umbilical herniation after 12 weeks) and systematically confirmed at the second-trimester scan. Follow-up exclusively by a general practitioner, without recourse to specialized morphological ultrasound, very likely constitutes the explanatory factor for the diagnostic failure in this case. This case argues for the requirement of specialized obstetric follow-up including at least three ultrasound examinations (11–14 weeks, 20–24 weeks, 32–34 weeks) with detailed morphological assessment.

- *Giant Omphalocele with Hepatic Contents*

An omphalocele is described as “giant” when the sac measures more than 5 cm and/or contains liver. This form accounts for approximately 10 to 20% of omphaloceles and poses particular surgical challenges. Primary one-stage repair (direct closure) is often impossible due to the disproportion

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between the viscera and the abdominal cavity — the abdomen not having had the opportunity to develop normally in utero. Progressive reduction techniques (compressive "silo" dressing, application of silver sulfadiazine to promote epithelialization of the sac) may be used as alternatives.

➢ *Prognosis and Associated Malformations*

Although clinically isolated in this case, an omphalocele should systematically prompt a search for chromosomal (karyotype), cardiac (echocardiography), renal, and cerebral anomalies. Association with Beckwith-Wiedemann syndrome (macrosomia, macroglossia, hypoglycemia) must be considered. The survival rate of isolated omphaloceles at term exceeds 90% in specialized centers, conditioned by the absence of chromosomal anomaly and the quality of initial surgical management.

➢ *Immediate Management in the Delivery Room*

Initial management in the delivery room is crucial. It includes: protection of the sac by sterile wrapping with moist saline compresses (without compression), insertion of a nasogastric tube on gentle suction, maintenance of normothermia (the sac favors fluid and thermal losses), peripheral venous access, and correction of any hypoglycemia. Prophylactic antibiotic therapy is recommended. Transfer to a pediatric medical-surgical unit must be organized without delay.

IV. CONCLUSION

This case of a giant omphalocele unexpectedly discovered at birth, in a context of prenatal surveillance by a general practitioner without specialized morphological ultrasound, illustrates the stakes of structured obstetric follow-up. It also underscores the need for training delivery room teams in the recognition and initial management of abdominal wall malformations. Despite the abrupt nature of this discovery, the excellent immediate neonatal adaptation of this newborn (Apgar 10, adequate weight) constitutes a favorable prognostic factor, provided that early surgical management and a complete malformation workup are carried out.

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