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GIANT OMPHALOCELE COMPLICATED BY RUPTURE IN A 32-SA PREGNANCY

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Abstract*: Omphalocele, also known as exomphalos, is one of the most common congenital malformations of the abdominal wall. The size of the defect and the severity of associated anomalies are determinants of overall morbidity and mortality. Prenatal screening and diagnosis of associated anomalies play a crucial role in appropriate management. The aim of this article is to review antenatal diagnostic strategies and treatment options for omphalocele, using a case report from our Department of Obstetrics and Gynaecology.*

Keywords: Omphalocele, Fetus, Abdominal Wall, Pregnancy, Genetic Abnormalities

Introduction

Omphalocele, also known as exomphalos, has its origins in the Greek word "omphalos" meaning umbilicus, the suffix "kele" evoking swelling, and the prefix "ex" denoting exterior. This condition is characterized by a hernia through the base of the umbilical cord, covered by an intact or ruptured membrane [1]. Although the precise mechanism leading to omphalocele is controversial, developmental failure can occur at various times during embryonic or fetal life. Consequently, the size of the defect can vary considerably.

One study reported an incidence of around 1 in 4000 to 7000 live births (2). In the USA, one study estimated a prevalence of 1.86 (95% confidence intervals [CI], 1.73 to 1.99) per 10,000 births (3).

Exomphalos is mainly linked to genetic alterations and chromosomal abnormalities such as trisomy 18, 13 and 21. In vitro fertilization presents a significant additional risk. Autosomal dominant familial inheritance is also recognized [4]. This congenital malformation, which is often upsetting for parents, requires the most appropriate prenatal counseling, particularly with regard to frequently associated anomalies, ideally as early as the first trimester [5]. Despite advances in surgical and neonatal care that have improved the survival of newborns with omphalocele, risks remain such as respiratory failure, prolonged stay in intensive care, feeding and growth problems, and neurodevelopmental delays [6-7].

Case report

We report the case of a 29-year-old female patient with 1st-degree consanguinity, no notable history, second gesture second pare, a live child by caesarean section for primipare on breech presentation, pregnancy poorly followed. An obstetrical ultrasound was performed in the 2nd trimester, showing a large omphalocele. The rest of the morphological ultrasound revealed no abnormalities. At 32 weeks' gestation, she presented to the emergency department in the active phase of labor. The ultrasound revealed a large omphalocele measuring 109 mm by 102 mm (Figure 1). After delivery by caesarean section for rhythm abnormality, we detected an abnormal closure of the abdominal wall, with externalization of the intestines and liver surrounded by a ruptured hernia sac (Figure 2). The malformative work-up was negative. The newborn was rapidly admitted to the paediatric ward, where a full workup was performed. The evolution after surgery was favorable, and the baby was returned to its mother after 2 months.

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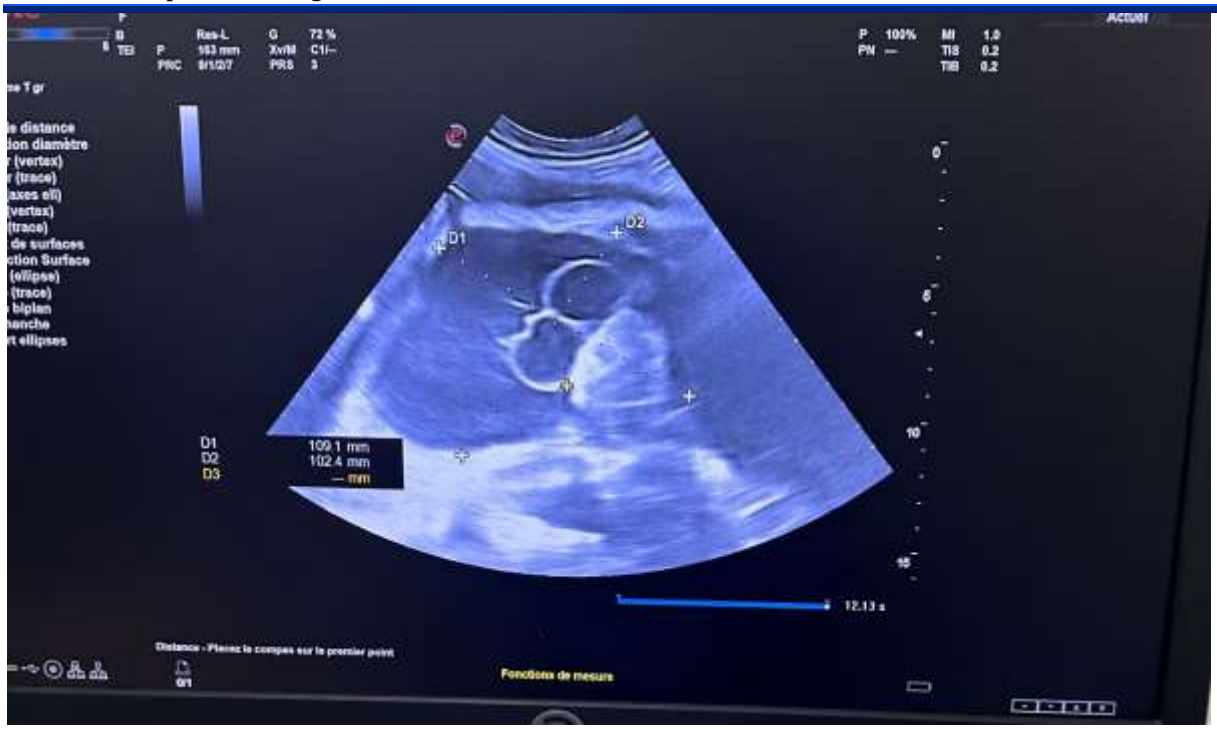

Figure 1: large omphalocele measuring 109 mm by 102 mm

Figure 2: externalization of the intestines and liver surrounded by a ruptured hernia sac .

Discussion

The estimated prevalence of omphalocele in the Paris region was 6.8 cases per 10,000 births over the period 2009 to 2013. During this period, a medical termination of pregnancy (IMG) rate of 70% and a foetal death in utero (FDIU) rate of 2% were observed. The incidence of omphalocele is higher in women over 35 and under 20, and affected newborns are more likely to be male. Prenatal diagnosis of omphalocele is now generally established in the first trimester in over 90% of cases.

Because of the association of omphalocele with chromosomal abnormalities and congenital heart defects, prenatal diagnosis often leads to an investigation of the foetal karyotype.

Before the 1970s, up to 80% of infants with omphalocele died of starvation and associated complications after the operation. However, since the 1970s, the development of total parenteral nutrition and staged closure methods has led to an increase in the survival rate of infants with omphalocele.

At the embryological level, there is no clear consensus as to the precise mechanisms leading to the pathogenesis of omphalocele. MARGULIES put forward the hypothesis that omphalocele forms before the 3rd week of gestation. This could result either from the failure of the mesodermal transverse septum to unite with its amniotic covering, or from a defect in the proliferation of embryonic connective tissue in the transverse septum. These processes are normally responsible for the formation of the supra-umbilical part of the abdominal wall. [13]

Omphaloceles are generally classified as major and minor according to their dimensions, although this classification may be less relevant than obtaining a detailed description of the anomaly, with a thorough analysis of the contents and structure of the hernial sac, as well as any associated chromosomal abnormalities (11).

Prenatal ultrasound is highly sensitive in detecting omphalocele. It can also identify associated anomalies and sonographic features suggestive of chromosomal abnormalities, facilitating the decision to terminate the pregnancy. In a study of 88 cases, the ratio of omphalocele circumference to abdominal circumference was found to be a predictive indicator: a ratio greater than 0.75 was associated with staged closure, while a ratio less than 0.57 indicated predominant closure (5).

Magnetic resonance imaging is not routinely performed to evaluate omphalocele, but it may prove useful in giant omphalocele, helping to predict postnatal outcome. For infants whose observed versus predicted total lung volume is less than 50%, prolonged respiratory support and hospitalization are usually required (12).

If an omphalocele is suspected, it is recommended that a third-level diagnostic center be consulted for a thorough evaluation via ultrasound for possible structural anomalies, with particular emphasis on those associated with trisomy 18 and 13 (14). A fetal echocardiogram is essential, as 47% of cases present with cardiac anomalies (15).

Careful examination of the vertebrae and gastrointestinal tract should be performed to exclude the syndrome of omphalocele, bladder exstrophy, anal imperforation and spinal cord anomalies (OEIS) (16). Normal imaging of the fetal bladder, kidneys and caudal anterior abdominal wall is necessary to exclude bladder or loacal exstrophy (17).

In cases of renal hypertrophy, macrosomia and hydramnios associated with omphalocele on ultrasound, Beckwith-Wiedemann syndrome is suspected, and cytogenetic examinations may be specifically directed to the exclusion of 11p anomalies or uniparental disorganization.

Approximately 20% of fetuses present with intrauterine growth retardation (IUGR) (18). However, the assessment of IUGR is complex due to difficulties in measuring abdominal circumference.

With the improved quality of first-trimester ultrasound screening (19), early diagnosis is becoming feasible (20). The presence of a "physiological" intestinal hernia during this period may cause confusion. Great caution is recommended when diagnosing omphalocele before 12 weeks, to avoid confusion with physiological intestinal hernia.

If the parents decide to continue the pregnancy with a view to postnatal corrective surgery, regular assessments of fetal growth and amniotic fluid volume are recommended. In case of deficient growth, Doppler examinations of the umbilical artery and amniocentesis may be necessary.

According to a meta-analysis, the results do not support the systematic use of Caesarean section to improve neonatal outcomes. Specifically, this meta-analysis found no improvement in the development of ischemic bowel, small bowel obstruction, necrotizing enterocolitis, sepsis or mortality (21).

There appears to be little advantage in opting for Caesarean section at delivery, unless the lesion is significant and likely to interfere with labour or sustain trauma (18, 22, 23). This consideration is particularly relevant when the lesion is significant and includes the liver (24; 25).

Delivery should be carried out as quickly as possible, preferably in a center equipped for adequate surgical management. By avoiding prematurity, incubation and ventilation are rarely necessary, unless an associated cardiopulmonary anomaly compromises the situation.

Surgical treatment of a small or medium-sized omphalocele should focus on direct closure of the fascia and skin. It is possible to remove the sac or hold it in place during fascial closure (26). Although advances in screening have increased, non-fatal congenital anomalies are frequently detected after birth. Approximately 17% of infants with omphalocele have chromosomal abnormalities, 32% have congenital heart disease, 8% have central nervous system abnormalities, and 22% have abnormalities affecting all systems (27). The presence of associated anomalies has a significant impact on long-term survival. Those with isolated omphalocele have the best one-year survival rate, exceeding 90%, with improvement over time. Between 2001 and 2005, the one-year survival rate improved by 22% compared with the previous five years, suggesting potential improvements in the prenatal diagnosis or treatment of newborns with omphalocele.

In general, outcome literature indicates an overall survival rate of 75% for children born alive with an omphalocele. Children with this condition face a high burden of early medical and surgical interventions during childhood, with risks of feeding difficulties, non-prosperity, gastroesophageal reflux disease (GERD) and intestinal obstruction due to adhesions. Although intestinal volvulus is rare, it can be fatal if not identified early. Giant omphalocele is a complex subset that is often associated with more pronounced neurodevelopmental and motor delay. Despite these early challenges, careful management and follow-up can enable children with omphalocele to eventually achieve a health status and quality of life similar to that of the general population, particularly in the absence of associated anomalies (28).

Conclusion

Omphalocele affects around 3 in 10,000 births. The clinical outcome depends on the associated structural and chromosomal abnormalities, as well as the gestation period up to delivery.

The use of prenatal ultrasound diagnosis and karyotyping is of crucial importance, enabling informed decisions to be made about pre- and postnatal management. It is recommended that prenatal care and counseling be provided in a multidisciplinary manner, and that parents ideally receive information about prognosis and outcome based on prospective population data.

References

Mastroiacovo P, Lisi A, Castilla EE, et al. Gastroschisis and associateddefects: an international study. Am J Med Genet A. 2007;143A(7):660–671.

Tassin M, Descriaud C, Elie C, et al. Omphalocele in the first trimester: prediction of perinataloutcome. Prenat. Diagn. 2013; 33: 497 -501.

Parker SE, Mai CT, Canfield MA, et al: National BirthDefectsPrevention. UpdatedNationalBirthPrevalenceestimates for selectedbirthdefects in the United States, 2004 -2006. Birth. Defects. Res. A. Clin. Mol. Teratol. 2010; 88: 1008 -16.

Frolov P, Alali J, Klein MD. Clinicalriskfactors for gastroschisisand omphalocele in humans: areview of the literature. PediatrSurg Int 2010 Dec;26(12):1135–48.

Kleinrouweler CE, Kuijper CF, van Zalen-Sprock MM, Mathijssen IB, Bilardo CM, Pajkrt E. Characteristics and outcome and the omphalocelecircumference/ abdominal circumference ratio in prenatallydiagnosedfetalomphalocele. FetalDiagnTher 2011;30(1):60–9.

Danzer E, Gerdes M, D'Agostino JA, Bernbaum J, Hoffman C, Rintoul NE, et al. Patient characteristics are important determinants of neurodevelopmentaloutcomeduringinfancy in giantomphalocele. Early Hum Dev. 2015;91:187–93

Hijkoop A, Peters NCJ, Lechner RL, van Bever Y, van GilsFrijters APJM, Tibboel D, et al. Omphalocele: fromdiagnosis to growth and developmentat 2 years of age. ArchDis Child FetalNeonatal Ed. 2019;104:F18–F23.

Snijders RJ, Brizot ML, Faria M, Nicolaides KH. Fetalexomphalosat 11 to 14 weeks of gestation. J Ultrasound Med 1995; 14: 569–574.

Blazer S, Zimmer EZ, Gover A, Bronshtein M. Fetalomphaloceledetectedearly in pregnancy: associated anomalies and outcomes. Radiology 2004; 232: 191–195

Yazbeck S, Ndoye M, Khan AH. Omphalocele: A 25-year experience. J PediatrSurg 1986; 21 (9):761- 763.

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Danzer EVT, Bebbington MW, Siegle J, Rintoul NE, Johnson MP, Flake AW, Adzick NS, Hedrick HL. Fetal MRI-calculated total lung volumes in the prediction of short-termoutcome in giantomphalocele: preliminaryfindings. FetalDiagnTher 2012;31(4): 248–53

Margulies L. Omphalocele (amniocele): Itsanatomy and etiology in relation to hernias of umbilicus and the umbilicalcord. American Journal of Obstetrics&Gynecology. 1945 Jan 1;49(5):695-9.

Benacerraf, B.R. (1989). The use of sonography for the antenataldetection of aneuploidy, Clinic in Diagnostic Ultrasound, 215, 21–54

Crawford, D.C., Chapman, M.G., Allan, L. (1985). Echocardiography in the investigation of congenitalanterior abdominal walldefects, Br. J. Obstet. Gynaecol., 92, 1034–1037.

Chen, C.P., Shih, S.L., Jan, S., Jeng, C.J., Lau, C.C. (1997). Perinatalfeatures of omphalocele–extrophy– imperforate anus– spinal defectscomplex (OEIS), Am. J. Perinatal., 14, 275–279.

Gosden, C., Brock, D.J.M. (1981). Prenataldiagnosis of exstrophy, Am. J. Genetics, 8, 95–98.

Carpenter, M.W., Curci, M.R., Dibbins, A. (1994). Perinatal management of ventral abdominal walldefects, Obstet. Gynecol., 64, 646–650.

Nicolaides, K.H., Snijders, R., Cheng, H.H. (1992). Fetalgastrointestinal and abdominal walldefects: associatedmalformations and chromosomalabnormalities, FetalDiagn. Ther., 7, 102–115.

Schmidt, W., Yarkoni, S., Crelin, E.S. (1987). Sonographic visualisation of physiologicalherniation in the first trimester, Obstet. Gynaecol., 69, 911–915.

Sally Y. Segel, MD, Sara J. Marder, MD, Samuel Parry, MD, and George A. Macones, MD, MSCE Fetal Abdominal Wall Defects and Mode of Delivery: A SystematicReview The American College of Obstetricians and Gynecologists. NOVEMBER 2001

Lewis, D.F., Towers, C.V., Garite, T.J., Jackson, D.N., Nageotte, M.P., Major, C.A. (1990). Fetal gastroschisis and exomphalos: Is Cesarean section the best mode of delivery?, Am. J. Obstet. Gynecol., 163, 773–775

Kirk, E.P., Wah, R.M. (1983). Obstetric management of the fetuswithomphalocele and gastroschisis. A review and report of 112 cases, Am. J. Obstet. Gynecol., 146, 512–516

Barcholucci, L. (1983). Discussion. In: Kirk, E.P., Wah, R.M. Obstetric management of the fetuswithomphalocele or gastroschisis: areview and report of 112 cases, Am. J. Obstet. Gynecol., 146, 512–515

Irving, I. (1967). Exomphalos and macroglossia. A study of 11 cases, J. Pediatr. Surg., 2, 499–503.

M. D. Kilby, Division of FetalMedicine, AcademicDepartment of Obstetrics, Floor 3, Birmingham Women'sHospital, Edgbaston, Birmingham, B15 2TG, U.K.Institute of Child Health, Ladywood, Birmingham, B16 8ET, U.K. 3 Department of Ultrasound, Birmingham Women'sHospital, Birmingham, B15 2TG, U.K. EXOMPHALOS (OMPHALOCELE)Received 18 August 1998 Accepted 1 September 1998

Marshall J, Salemi JL, Tanner JP, et al. Prevalence, Correlates,and Outcomes of Omphalocele in the United States, 1995-2005. ObstetGynecol 2015; 126:284-293

Joanne E. Baerg MD Professor of Surgery , Amanda N. Munoz MD ClinicalScholarFellow , Long Term Complications and Outcomes in Omphalocele, Seminars in PediatricSurgery (2019), doi: https://doi.org/10.1053/j.sempedsurg.2019.04.004.