

ISOLATED GIANT OMPHALOCELE WITH BOWEL LOOPS: AN UNCOMMON CONGENITAL ANOMALY CASE REPORT

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Abstract

An omphalocele is a congenital defect that affects the development of the abdominal wall in the umbilical region, resulting in a hernial-type sac of variable size. Here we describe a case report of a isolated omphalocele in which bowel loops were the only content.

Keywords: *bowel loops, omphalocele*

I. Introduction

An omphalocele represents an embryological defect of the umbilical ring and medial segments of the two lateral abdominal folds. The incidence is nearly 2.5 cases per 10,000 live birth.⁽¹⁾ With omphalocele, the intraabdominal viscera herniate into the base of the umbilical cord and the herniated viscera are covered by the peritoneum and amnion. Omphalocele was categorized into two groups, One group consisted with abnormal karyotype & associated structural anomalies. The second group consisted with a normal karyotype and an omphalocele as an isolated US finding.⁽²⁾ Our case belongs to the second group having an omphalocele as an isolated US finding.

II. CASE REPORT

A newborn was presented with midline abdominal wall defect into which abdominal contents are extruded into the sac-like structure at the base of the umbilical cord. [fig 1, fig 2]. The sac is covered by the thin membrane [amnion]. On ultrasonography, bowel loops were noted as a content [fig 3]. Bowel loops were dilated. On plain radiograph, dilated bowel loops are noted outside the abdominal wall [fig 4]. Rest of the organs were within normal limits with no congenital abnormality.



Figure 1



Figure 2



Figure 3



Figure 4

III. DISCUSSION

Omphalocele represents an embryological defect of the umbilical ring and medial segments of the two lateral abdominal wall folds that form the anterior abdominal wall during foetal growth.⁽³⁾ The aetiology of omphalocele is unknown, but various theories have been postulated. These include failure of the bowel to return to the abdomen by 10–12 weeks, given that physiological umbilical herniation should not persist beyond 12 weeks. After 12 weeks of gestation, one must consider omphalocele as the cause of a mass in the base of the umbilical cord and beyond the confines of the abdominal wall.⁽⁴⁾ Another possibility is that omphalocele results from the failure of the embryonic lateral folds to fuse in the midline.⁽⁵⁾ Ultrasonographic (US) examinations have revealed the completion of this process at 12 weeks gestation.⁽⁶⁾ The size of the defect is variable and can range from 4 cm to more than 10 cm.⁽⁷⁾ Omphalocoeles can be divided into two groups depending on the size of the hernial defect; minor and major (giant). A minor omphalocele occurs when the defect is 4 cm or less. A major or giant omphalocele is classified as a 5 cm or larger defect.⁽⁷⁾ The hernial sac may contain small and large bowel, stomach, liver, spleen, urinary bladder, uterus and ovaries. Our patient had a giant omphalocele containing the bowel loops as a content. Omphalocoeles can also be subdivided according to the site of the defect into epigastric (classic omphalocele) with cephalic fold defect, central type with a lateral wall defect greater than 4 cm and the hypogastric/caudal type with caudal fold defect.⁽³⁾ Chromosomal anomalies are common (accounting for 40%–60%), and these include trisomies 13, 18 and 21 as well as Turner, Klinefelter and triploidy

syndromes.^(3,8) Associated cardiovascular anomalies are seen in up to 50% of patients with omphaloceles. Diagnosis of omphalocele can be easily made by prenatal ultrasonography. A definitive diagnosis of omphalocele is possible only beyond 12 weeks of gestation, when confusion with physiologic midgut herniation is no longer possible. A number of sonographic features differentiate an omphalocele from physiologic midgut herniation. For example, a midgut herniation seldom exceeds 7 mm in diameter, whereas omphaloceles are much larger, sometimes even larger than the abdomen. In addition, midgut herniation seldom persists after 12 weeks of gestation or in a foetus with a crown-rump length measurement of more than 44 mm. MR imaging studies of the foetus may help in corroborating and refining the ultrasonographic diagnosis of complex foetal defects.^[8]

The optimal mode of delivery for foetuses with antenatally diagnosed abdominal wall defects has been subject of controversy. How et al^[10] reported that these foetuses may safely be delivered by the vaginal route and caesarean delivery should perform for obstetric indications only. In another study, emphasized that elective caesarean section improve the outcome of neonates with abdominal wall defect. In our case because of giant omphalocele, caesarean route is preferred.

In conclusion, omphalocele can be of isolate & nonisolated types.^[9] Omphalocele containing liver & spleen are more likely associated with the abnormal karyotype with subsequent poor fetal outcome while containing only bowel loops are less likely associated.^[9] In case of pregnancy continuation serial ultrasonography is recommended to detect any alteration in fetal growth.

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