International Journal of Contemporary Pediatrics
Meshram RM et al. Int J Contemp Pediatr. 2018 May;5(3):1142-1144
http://www.ijpediatrics.com
pISSN 2349-3283 | eISSN 2349-3291

DOI: http://dx.doi.org/10.18203/2349-3291.ijcp20181560

Case Report

Giant Omphalocele with skeletal deformities: a case report

Rajkumar M. Meshram*, Amruta Phatak, Arvind Bhurke, Chetan Chaudhari

Department of Paediatrics, Government Medical College, Nagpur, Maharashtra, India

Received: 22 January 2017
Accepted: 28 Febrary 2017

***Correspondence:**
Dr. Rajkumar M. Meshram,
E-mail: dr_rajmeshram@rediffmail.com

ABSTRACT

Omphalocele is an anterior abdominal wall defect at the base of the umbilical cord, with herniation of the abdominal contents which are covered by a membrane and associated with a very high incidence of structural and chromosomal anomalies including heart defect, vesical, genital or diaphragmatic malformations. A 1000 gm preterm neonate was born vaginally, to a non-consanguineously married primi mother with uneventful pregnancy. On examination baby had a large anterior abdominal wall defect covered by a thin membrane with widely open and bulging anterior fontanellae. Eyes, ears and lips were well developed but eyelids were fused. Liver, spleen and intestine were visible under a thin transparent membrane. Rib cage was formed, and ribs were palpable but lower limbs were directed posteriorly and genitalia was ambiguous. Baby was in gasping condition and died within 30 minutes after birth. Prenatal accurate diagnosis by providing adequate antenatal care and ultrasonogram would permit an opportunity to counsel the family and to prepare for optimal postnatal care.

Keywords: Abdominal wall defect, Giant omphalocele, Skeletal deformities

INTRODUCTION

Omphalocele is an anterior abdominal wall defect at the base of the umbilical cord, with herniation of the abdominal contents which are covered by a membrane consisting of peritoneum on the inner surface, amnion on the outer surface, and Wharton's jelly between the layers. It is a rare congenital anomaly affecting approximately 1 in 5000 live births.[1,2] The first reported case of omphalocele was described in 1634 by Pare A.[3] The exact etiology of omphalocele is not known but various theories have been postulated; these include failure of the bowel to return into the abdomen by 10-12 weeks, failure of lateral mesodermal body folds to migrate centrally, and persistence of the body stalk beyond 12 weeks gestation.[4] Omphalocele is associated with a significantly higher (50-70%) incidence of structural and chromosomal anomalies include heart defect; cleft lip and palate; vesical, genital or diaphragmatic malformations; and chromosomal anomaly.[5] Postnatal managements include protection of herniated viscera, maintenance of fluids and electrolytes, prevention of hypothermia, gastric decompression, prevention of sepsis and maintenance of cardiovascular stability. The prognosis of omphalocele is depends on size and/or contents and associated anomalies. Here, we present a case of giant omphalocele with skeletal abnormalities and ambiguous genitalia.

CASE REPORT

A 1000 gm preterm neonate was born by spontaneous vaginal delivery, to a 21 year old primigravida, non-consanguineously married mother. She had a normal, uneventful pregnancy with no history of hypertension, pre-gestational or gestational diabetes, no history of smoking, and no exposure to X-ray irradiation, no history

of drug use other than supplemental iron, vitamins and mineral. There was no history of abortion and there was no family history of fetal anomalies. Antenatal ultrasound was suggestive of anomalous baby. On examination baby had large anterior abdominal wall defect from xiphisternum to pubic symphysis with covering of thin membrane.

Figure 1: Neonate with giant omphalocele with skeletal defect.

The head was cyanosed with widely open and bulging anterior fontanellae. Eyes, ears and lips were well developed but eyelids were fused. Liver, spleen and intestine were visible under a thin transparent membrane. Rib cage was formed, and ribs were palpable but lower limbs were directed posteriorly (Figure 1). Baby had ambiguous genitalia. Baby was in gasping condition and died within 30 minutes after birth.

DISCUSSION

In fetal life, the foregut and hindgut are first seen at about three to four weeks. The intestine become elongated and progress out into the umbilical cord (physiologic herination) in between five to seven weeks of life. The cephalic portions of the intestine are drawn back into the abdominal cavity first, followed by caudal intestine; this occurs around the eleventh week of gestation. The failure of the abdominal wall to close during this process and if it involves the whole layer of abdominal wall, it presents as omphalocele.[1-3] At birth contents of omphalocele are covered with amnion, parietal peritoneum and a thin layer of connective tissue.

Omphalocele can be differentiated from gastroschisis, which is a small defect in the anterior abdominal wall typically located to the right of the umbilical ring and resulting in the herination of the abdominal contents, without a surrounding membrane that results from ischemic insult to the developing abdominal wall.

There are regional differences in the incidence of abdominal wall defect however the incidence of omphalocele ranges between 1.5 and 3 per 10000 births whereas incidence of gastroschisis ranges from 0.4 and 3 per 10000 births.[6,7] Older mothers above the age of 35 years carry 1.8-fold higher risk of having a baby with omphalocele and slight male preponderance is observed while gastroschisis occur equally in male and female. One third of omphalocele neonates are premature and low birth weight.[8,9]

Omphalocele is associated with significantly higher incidence of other structural and chromosomal anomalies like Trisomy 18, 13 and 21, Turner, and Klinefelter and Triploidy syndrome.[10] About 45% of patients of with omphalocele have concomitant congenital heart diseases with progression to pulmonary hypertension. Small omphalocele size is associated with fewer cardiac anomalies but has an increased prevalence of gastrointestinal anomalies.[11] Other associated anomalies include gastrointestinal, genitourinary, neural tube defect and musculoskeletal defects. Omphalocele is also a part of Beckwith-Widemann syndrome, Pentology of Cantrell, Meckel-Gruber syndrome and lethal cleft palate Omphalocele syndrome.[12,13]

The issue of mode of delivery for abdominal wall defect is highly debatable. Some reports advocate for the practice of caesarean versus vaginal delivery in giant omphalocele containing liver, however numerous studies have found no difference in outcome between the modes of delivery for smaller abdominal defect.[14] Linnaus et al reported a giant omphalocele associated with liver injury in preterm labor with transverse lie.[15] Prenatal ultrasound could potentially identify the overwhelming majority of abdominal wall defects and accurately distinguish omphalocele from gastroschisis and this identification would permit an opportunity to counsel the family and to prepare for optimal postnatal care. Four-fold rise of maternal serum alfa fetoprotein is also used as a marker for prenatal diagnosis of abdominal wall defect. Use of multivitamins during pregnancy is associated with 60% reduction in risk of symptomatic omphalocele.[9]

Postnatal management include protection of herniated viscera, maintenance of fluids and electrolytes, prevention of hypothermia, gastric decompression, prevention of sepsis and maintenance of cardiovascular stability. For small or medium-sized defects it is often appropriate to attempt primary closure after removal of the omphalocele sac while some giant omphalocele requires a skin flap or nonoperative management approach. Complications occur more frequently with giant defects. Potential short-term complications include necrotizing enterocolitis, prolonged ileus, and respiratory distress. Long term complications include parenteral nutrition dependence; gastroesophageal reflux, feeding

intolerance, and neurodevelopmental delay. Overall, advances in surgical therapies and nursing care have improved outcomes for infants with omphalocele; survival rate for those with isolated omphalocele are reported at 75 to 95 percent.[16] Infants with associated anomalies and giant omphalocele have the poorest outcome.

CONCLUSION

Giant omphalocele with multiple congenital anomalies is associated with poor outcome. Prenatal accurate diagnosis by providing adequate antenatal care and ultrasonogram would permit an opportunity to counsel the family and to prepare for optimal postnatal care.

Funding: No funding sources
Conflict of interest: None declared
Ethical approval: Not required

REFERENCES

1. Marshall J, Salemi JI, Tanneer JP, Ramakrishnan R, feldkamp ML, Marengo LK, et al. Prevalence, correlates and outcomes of omphalocele in the united states, 1995-2005. Obstet Gynecol 2015;126(2):284-93.
2. Benjamin B, Wilson GN. Anomalies associated with gastroschisis and omphalocele: analysis of 2825 cases from the Texas Birth Defects Registry. J Pediatr Sur. 2014;49(4):514-9.
3. Gratiot JH. Persistent omphalomesentric duct with omphalocele. Am J Surg. 1953;86(6):747-50.
4. Kurkchubasche AG. The fetus with an abdominal wall defect. Medicine and health, Rhode Island. 2001;84(5):159-61.
5. Stoll C, Alembik Y, Dott B, Roth MP. Omphalocele and gastroschisis and associated malformations. Am J Med Genet Part A. 2008;146A(10):1280-5.
6. Curry JI, McKinnney P, Thornton JG, Stringer. The aetiology of gastroschisis. Br J Obstet Gynaecol. 2000;107(11):1339-46.
7. Rankin J, Dillon E, Wright C. Congenital anterior abdominal wall defect in the North England, 1986-1996: occurrence and outcome. Prenat Diagn. 1999;19(7):662-8.
8. Tan KBL, Tan KH, Chew SK, Yeo GSH. Gastroschisis and omphalocele in singapore: a ten-year series from 1993 to 2002. Singapore Med J. 2008;49(1):31-6.
9. Chircor L, Mehedinnti R, Hincu M. Risk factors related to omphalocele and gastroschisis. Rom J Morph Emb. 2009;50(4):645-9.
10. Nicholas SS, Stamilio DM, Dicke JM, Gray DL, Macones GA, Odibo AO. Predicting adevse neonatal outcome in fetuses with abdominal wall defects using prenatal risk factors. Am J Obstet Gynecol. 2009;201(4):383e1-6.
11. Kumar HR, Jester AI, Ladd AP. Impact of omphalocele size on associated conditions. J Pediatr Surg. 2008;43(12):2216-9.
12. Tanriverdi S, Aycicek R, Bagci O, Karaboga B, Sencan A, Koyuncu FM. Prenatal diagnosis of omphalocele and Beckwith-Wiedemann syndrome: a case report. Perinatal J. 2011;19(3):140-4.
13. Leon G, Chedraui P, San Miguel G. Prenatal diagnosis of Cantrell's Pentology with conventional and three-dimensional sonography. J Materal-Fetal Neonatal Med. 2000;12(3):209-11.
14. Anteby EY, Yagel S. Route of delivery of fetuses with structural anomalies. Eur J Obstet Gynecol Reprod Biol. 2003;106(1):5-9.
15. Linnaus ME, Donato B, McMahon L, Chamblis L, Notrica DM. A case of traumatic rupture of a giant omphalocele and liver injury associated with transverse lie and preterm labor. J Ped Surg Case Rep. 2016;14:4-7.
16. McNair C, Hawes J, Urquhart H. Caring for the newborn with an omphalocele. Neonatal Network. 2006;25(5):319-27.

Cite this article as: Meshram RM, Phatak A, Bhurke A, Chaudhari C. Giant Omphalocele with skeletal deformities: a case report. Int J Contemp Pediatr 2018;5:1142-4.