Case Report

Giant Omphalocele with skeletal deformities: a case report

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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-consangunineously married primi mother with uneventful pregnancy. On examination baby had a large anterior abdominal wall defect covered by 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. The first reported case of omphalocele was described in 1634 by Pare A. 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. 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. 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-consangunineously 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 herniation) 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. At birth contents of omphalocele are covered with amnion, parietal peritoneum and a thin layer of connective tissue.

Omphalocele can be differentiated from gastrochisis, which is a small defect in the anterior abdominal wall typically located to the right of the umbilical ring and resulting in the herniation 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 gastrochisis ranges from 0.4 and 3 per 10000 births. 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 gastrochisis occur equally in male and female. One third of omphalocele neonates are premature and low birth weight.

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. About 45% of patients of with omphalocele have comitant 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. 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.

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. Linnaus et al reported a giant omphalocele associated with liver injury in preterm labor with transverse lie. Prenatal ultrasound could potentially identify the overwhelming majority of abdominal wall defects and accurately distinguish omphalocele from gastrochisis and this identification would permit an opportunity to counsel the family and to prepare for optimal postnatal care. Four-fold rise of maternal serum alpha 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.

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.\(^6\) 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.

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