CASE REPORT

A Rare Anterior Abdominal Wall Defect: Omphalocele - A Case Report

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Abstract:

Two most common anterior abdominal wall defects are gastroschisis and omphalocoele or exomphalos. Gastroschisis means 'stomach cleft' which is a congenital defect of the abdominal wall, usually to the right of the umbilical cord insertion and abdominal contents herniate into the amniotic sac. Exomphalos is literally translated from the Greek, means 'outside the navel'. It is also called an Omphalocele. It is a congenital abnormality in which the contents of the abdomen herniate into the umbilical cord through the umbilical ring. Textbooks grouped them together but these are different entities. These congenital malformations have a high mortality rate. Only about 60 % of children with such type of malformations survive until the end of first year of age. A male foetus of 32 weeks gestational age was sent from Dr. Ulhas Patil Medical College and Hospital, Jalgaon (Khurd) to the Department of Anatomy to examine the fetus for congenital anomalies. A case report of an Omphalocele was presented. Occurrence of such cases is very rare about 2.17 per 10000 live births as reported in literature.

Keywords: Anterior abdominal wall defect, Gastroschisis, Omphalocoele, Exomphalos

Introduction:

Anterior abdominal wall defects like omphalocele and gastroschisis remain source of significant morbidity and mortality in developing countries like India. These conditions still pose a challenge of diagnosis and management among health care givers.

In India today also most deliveries occur in remote, peripheral health centers, mostly by semiskilled and unskilled workers. If they come across such congenital anomalies, it should be immediately referred to concerned specialist for diagnosis and according management. Otherwise undue mortality can occur. So we are presenting a rare case of Omphalocele from Dr Ulhas Patil Medical College and Hospital, Jalgaon (Khurd).

Case Report:

A 31 year old third gravida mother delivered a premature, stillborn male baby at Dr Ulhas Patil Medical College and Hospital, Jalgaon (Khurd). This was sent to the Department of Anatomy for examination of congenital anomalies. On complete evaluation it was noted that baby had midline anterior abdominal wall defect with evisceration of developing bowel loops and liver covered with membranes, with overlying insertion of umbilical cord on it.

No other associated anomaly was present. Finally it was concluded that this was a rare case of omphalocele.

Discussion:

Gastroschisis and omphalocele are commonly described anterior abdominal wall defects. Gastroschisis is characterized by an intact umbilical cord and evisceration of bowel through a defect in abdominal wall, generally to the right of cord with no membrane covering. Omphalocele is characterized by herniation of bowel, liver, other organs into intact umbilical cord, the tissues

Fig. 1: Large Omphalocele Showing Attachment of Umbilical Cord over Membranes

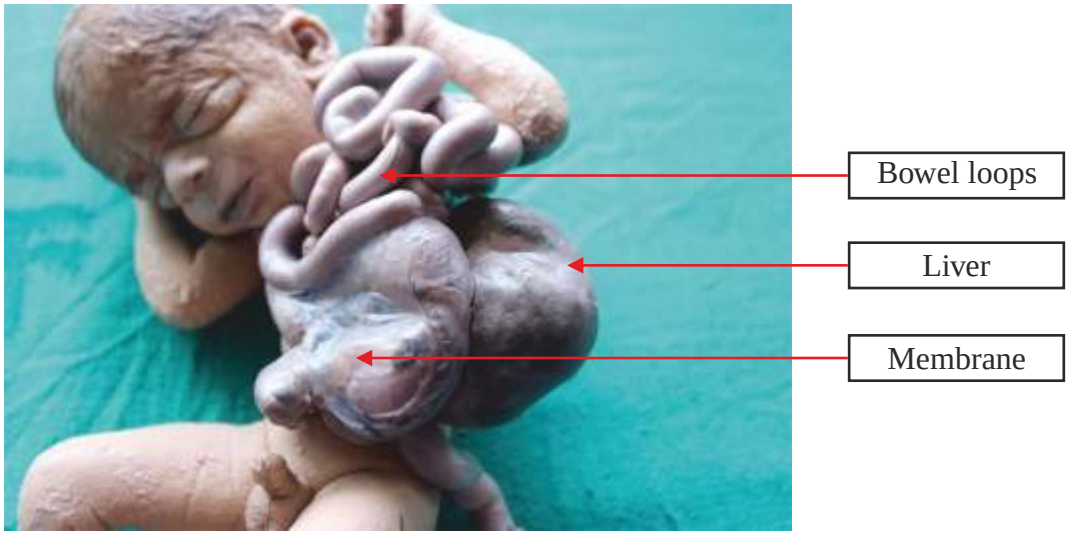

Fig. 2: Large Omphalocele Showing Bowel Loops and Liver

being covered by membranes unless later are ruptured [1]. Line of demarcation between these two entities is very fade.

Anterior abdominal wall develops from fusion of four ectomesodermic folds that is cranial, caudal and two lateral folds. Failure of fusion of them results in omphalocele [2]. If omphalocele is associated with caudal fold failure; it results in extrophy of bladder or cloaca. If it is associated with cranial fold failure, it results in Pentalogy of Cantrell [3]. Perturbation of retinoic acid levels during development secondary to micro-duplication of ALDH1A2 gene present at 15q21.3 could underlie the pathological basis of Pentalogy of Cantrell [4]. Gastroschisis is probably caused by disruption of right omphalomesentric artery. This results in full thickness defect in abdominal wall located just lateral and usually to the right of intact umbilical cord resulting in evisceration of abdominal contents into amniotic space [5].

Overall incidence of anterior abdominal wall defects is 2.63 per 10000 live births, while that of gastroschisis is 0.46 per 10000 live births. Incidence of omphalocele is 2.17 per 10000 live births which is lowest amongst Indian population. Younger mothers below age of 25 years carry 2.8 fold higher risk of having a baby with gastroschisis [6-9]. Older mothers above age of 35 years carry 1.8 fold higher risk of having a baby with omphalocele [6, 10]. Higher incidence of gastroschisis is noted with cigarette smokers [11, 12], use of recreational drug like cocaine, amphetamine, marijuana and alcohol [13, 14], in undernourished [15], who use over counter medication like ephedrine, pseudoephedrine etc [12, 15]. One third of omphalocele cases and more than half of gastroschisis cases are premature births [7, 16]. Both mainly affect male children of first para first gesta mothers. Both are associated with prematurity and low birth weight [17].

Omphalocele is associated with significantly higher incidence of other structural and chromosomal anomalies [18] like Trisomy [18, 13 and 21], Turner, Klinefelter and Triploidy syndromes [19, 21]. Roughly it is 54 %. Omphalocele is associated with cardiac anomalies [21-25], gastrointestinal, genitourinary [22], neural tube [24, 26] and musculoskeletal defects. Omphalocele is a part of Beckwith–Widemann syndrome [27], Pentalogy of Cantrell [28] Meckel–Gruber syndrome [29], and lethal cleft palate Omphlocele syndrome [30]. Gastroschisis is associated with intestinal atresia in 25% of cases and cryptorchidism in 31% otherwise association with other anomalies is rare.

Prenatal diagnosis of such anterior abdominal wall defects is important and they are often diagnosed during routine prenatal ultra-sonography. In normal embryogenesis, extra abdominal intestinal loops return to abdominal cavity by 11 weeks of gestation so confirmation of diagnosis of anterior abdominal wall defects should be preferably delayed till 14th week of gestation [5].

Abdominal wall defects are also diagnosed prenataly by serum markers like Maternal Serum Alfa Feto Protein (MSAFP). The median value of MSAFP in gastroschisis is 9.42 multiples of median (MOM). While in omphalocele, it is only 4.18 MOM making it little bit difficult to diagnose [5].

Mainstay of treatment of such anterior abdominal wall defects is to reduce herniated viscera into the abdomen and to close with fascia and skin to create a solid abdominal wall with relatively normal umbilicus while minimizing risk to the baby. But in comparison with gastroschisis, omphalocele doesn't require urgency in surgical management so long as the viscera are covered with membranes. A complete evaluation for associated defects can be done and other problems can be treated.

Prognosis of infant with omphalocele is usually is fatal as it is commonly associated with chromo-somal and structural anomalies like cardiac defects. Prognosis of infant with gastroschisis is determined primarily by condition of exteriorized bowel, contemporary mortality being around 8% [31]. Patients with gastroschisis have good survival rate of 90 to 95 % provided they do not have catastrophic bowel loss, sepsis and long term complications of short bowel syndrome.

Such anterior abdominal wall defect can be prevented. Taking multivitamins during pregnancy seems to influence occurrence of anterior abdominal wall defects. Use of multivitamins during pregnancy is associated with 60% reduction in risk of symptomatic Omphalocele [17].

References:

1. Weber T, Au-fliegner M, Downard C, Fishman S. Abdominal wall defects. *Curr Opin Paediatr* 2002; 14(4):491-497.
2. Vermeij-keers C, Hartwig NG, Van Der Werff JF. Embryonic development of ventral body wall and its congenital malformations. *Semin Paediatr Surg* 1996; 5(2):82-89.
3. Cantrell JR, Haller JA, Ravitch MM. A syndrome of congenital defects involving the abdominal wall, sternum, diaphragm, pericardium, and heart. *Surg Gynecol Obstet* 1958; 107(5):602–614.
4. Steiner MB, Vengoechea J, Collins RT. Duplication of the ALDH1A2 gene in association with pentalogy of Cantrell: a case report. *Journal of Medical Case Reports* 2013; 7:287-290.
5. Martin RW. Screening for fetal abdominal wall defects. *Obste Gynecol Clin North Am* 1998; 25(3):517-26.
6. Tan K BL, Tan KH, Chew SK, Yeo G S H. Gastroschisis and omphalocele in Singapore: a ten-year series from 1993 to 2002. *Singapore Med J* 2008; 49(1):31-36.
7. DI Tanna GL, Rosano A, Mastroiacovo P. Prevalence of gastroschisis at birth: Retrospective study. *BMJ* 2002; 325(7377):1389–1390.
8. Yang P, Beaty TH, Khoury MJ, Chee E, Stewart W, Gordis L. Genetic-epidemiologic study of omphalocele and gastroschisis: Evidence for heterogeneity. *Am J Med Genet* 1992; 44(5):668–675.
9. Kazaura MR, Lie RT, Irgens LM, Didriksen A, Kapstad M, Egenaes J, Bjerkedal T. Increasing risk of gastroschisis in Norway: An age-period-cohort analysis. *Am J Epidemiol* 2004; 159(4): 358–363.
10. Tan KH, Kilby MD, Whittle MJ, et al. Congenital anterior abdominal wall defects in England and Wales 1987–93: Retrospective analysis of OPCS data. *BMJ* 1996; 313(7062):903-906.
11. Hoyme HE, Higgingbottom MC, Jones KL. The vascular pathogenesis of gastroschisis: Intrauterine interruption of the omphalomesenteric artery. *J Pediatr* 1981; 98(2):228-231.
12. Werler MM, Sheehan JE, Mitchell AA. Association of vasoconstrictive exposures with risks of gastroschisis and small intestinal atresia. *Epidemiology* 2003; 14(3):349-354.
13. Hoyme HE, Jones MC, Jones KL. Gastroschisis: Abdominal wall disruption secondary to early gestational interruption of the omphalomesenteric artery. *Semin Perinatol* 1983; 7(4):294-298.
14. Torfs C, Velie EM, Oeschli FW, Bateson TF, Curry CJR. A population based study of gastroschisis: demographic, pregnancy and lifestyle risk factors. *Teratology* 1994; 50(1):44-53.
15. Torfs CP, Lam PK, Schaffer DM, Brand RJ. Association between mothers' nutrient intake and their offspring's risk of gastroschisis. *Teratology* 1998; 58(6):241-250.
16. Rankin J, Dillon E, Wright C. Congenital anterior abdominal wall defects in the north of England,1986–1996: occurrence and outcome. *Prenat Diagn* 1999; 19(7):662-668.
17. Chircor L, Mehedinti R, Hincu M. Risk factors related to omphalocele and gastroschisis. *Rom J Morph Emb* 2009; 50(4): 645-649.
18. Suita S, Okamatsu T, Yamamoto T, Handa N, Nirasawa Y, Watanabe Y, *et al*. Changing profile of abdominal wall defects in Japan: results of a national survey. *J Pediatr Surg* 2000; 35(1):66-71.
19. Nyberg DA, Fitzsimmons J, Mack LA, Hughes M, Pretorius DH, Hickok D *et al*. Chromosomal abnormalities in fetuses with omphalocele. Significance of omphalocele contents. *J Ultrasound Med* 1989; 8(6):299-308.
20. Gilbert WM, Nicolaides KH. Fetal omphalocele: Associated malformations and chromosomal defects. *Obstet Gynecol* 1987; 70(4):633-635.
21. Baird PA, MacDonald EC. An epidemiologic study of congenital malformations of the anterior abdominal wall in more than half a million consecutive live births. *Am J Hum Genet* 1981; 33(3):470-478.
22. Grosfield JL, Dawes L, Weber TR. Congenital abdominal wall defects: Current management and survival. *Surg Clin North Am* 1981; 61(5):1037-1049.
23. Mayer T, Black R, Matlack ME, Johnson DG. Gastroschisis and omphalocele: An eight year review. *Ann Surg* 1980; 192(6):783-787.
24. Mann L, Ferguson-Smith MA, Desai M, Gibson AAM, Raine PAM. Prenatal assessment of anterior abdominal wall defects and there prognosis. *Prenat Diagn* 1984; 4(6):427-435.
25. Mabogunje OA, Mahour GH. Omphalocele and gastroschisis: Trends in survival across two decades. *Am J Surg* 1984; 148(5): 679-686.
26. Hauge M, Bugge M, Nielson J. Early prenatal diagnosis of omphalocele constitutes indication for amniocentesis. *Lancet* 1983; 2 (8348): 507.

27. Boyd PA, Bhattacharjee A, Gould S *et al*. Outcome of prenatally diagnosed anterior abdominal wall defects. *Arch Dis Child Fetal Neonatal Ed* 1998; 78 (3): F 209-13.
28. Leon G, Chedraui P, San Miguel G. Prenatal diagnosis of Cantrell's pentalogy with conventional and three-dimensional sonography. *Journal of maternal-fetal and neonatal medicine* 2000; 12 (3):209-211.
29. Van Zalen-Spreock RM, Van Vugt JMG, Van Geijn HP. First-trimester sonography of physiological midgut herniation and early diagnosis of omphalocele. *Prenat Diagn* 1997; 17(6):511-518.
30. Czeizel A. New lethal omphalocele-cleft palate syndrome? *Hum Genet* 1983; 64(1): 99.
31. Driver CP, Bruce J, Bianchi A, Doig CM, Dickson AP, Bowen J. The contemporary outcome of gastroschisis. *J Pediatr Surg* 2000; 35(12):1719-1723.

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