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Right-sided congenital diaphragmatic hernia and myelomeningocele: a rare association

Syed Rehan Ali
Aga Khan University

Shakeel Ahmed
Agha Khan University, shakeel.ahmed@aku.edu

CASE REPORT

Right-Sided Congenital Diaphragmatic Hernia and Myelomeningocele: A Rare Association

Syed Rehan Ali and Shakeel Ahmed

ABSTRACT

Congenital diaphragmatic hernia (CDH) is a rare birth defect with a prevalence of < 0.5 per 1,000 live births. Majority of these defects are left-sided as most studies suggest that frequency of right-sided CDH was 10% of the total. The association of CDH with myelomeningocele (MMC) is extremely rare; as in Sweed's study of 116 consecutive cases of CDH, the incidence of associated MMC with CDH was stated as 4.3%. There has been one previous case report of left-sided CDH, MMC and hydrocephalus prenatally diagnosed; but to the best of authors' knowledge, this is the first reported case of the above constellation with a right-sided CDH diagnosed prenatally.

Key Words: *Diaphragmatic hernia. Myelomeningocele. Birth defect. Hydrocephalus. Lung hypoplasia.*

INTRODUCTION

Right congenital diaphragmatic hernia (CDH) is uncommon and occurs in only 10% of cases with CDH; whereas left CDH occurs in 85% of cases, and bilateral CDH occurs in < 5% of cases.[1] About 50% - 60% of affected individuals have isolated CDH; the remainder have complex CDH, i.e. CDH occurring with additional malformations or as part of a single gene disorder or chromosome abnormality.[2] In the English language medical literature since 1975, the combination of CDH and myelomeningocele (MMC) has been reported in only 18 patients.[1] Prenatal diagnosis of the constellation of a right-sided CDH, MMC and hydrocephalus has, to our knowledge, not been reported in the medical literature as identified by Medline.

CASE REPORT

A 22-year Asian, healthy mother, was referred in her first pregnancy. She had no past medical history and was taking iron supplements antenatally. Family history was non-contributory and routine antenatal blood tests, including hepatitis B, rubella, TORCH screen, were all negative.

A routine ultrasound examination performed at gestational week 19 showed a single fetus with hydrocephalus, spina bifida, a lemon-shaped skull and small cerebellum consistent with the Arnold Chiari malformation. The fetal measurements were all on the 10th centile for gestation.

Department of Paediatrics and Child Health, The Aga Khan University Hospital, Karachi.

Correspondence: Dr. Shakeel Ahmed, Consultant, Department of Paediatrics and Child Health, The Aga Khan University Hospital, Karachi.
E-mail: shakeel.ahmed@aku.edu

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Follow-up ultrasound scan, 2 weeks later, showed, in addition, mediastinal shift of the heart to the left and the appearance of a small cystic lesion on the right side of the chest; thought to be either a small diaphragmatic hernia or congenital adenomatoid malformation. Amniocentesis was offered, but declined.

Mother delivered a 2.6 kg at term, live female infant by vaginal delivery, with Apgar scores of 1^{1} and 4^{5}. On examination, there was an obvious 7 x 5 cm MMC in the thoracolumbar region with, otherwise, no gross dysmorphic features. Arterial cord blood pH was 7.27. The neonate was intubated with a size 3.0 endotracheal tube and transferred to the neonatal intensive care unit where continuous mechanical ventilation was commenced. No other anomalies were identified.

A portable chest X-ray revealed a sizeable diaphragmatic hernia with multiple loops of bowel in the right side of the thorax, a hypoplastic right lung, and deviation of the mediastinum to the left. Despite maximum ventilatory support, the neonate began to clinically deteriorate and 12 hours after birth, she died. Autopsy was denied by the parents and subsequent chromosomal analysis showed a normal karyotype, 46XX female.

DISCUSSION

CDH is a lethal birth defect with a prevalence recently reported to be in the range of 0.17 - 0.57 per 1,000 live births.[2-4] From a meta-analysis of 12 studies compiled by Skari *et al*., the pooled frequency of right-sided CDH was 8% of the total.[5] In Sweed's study of 116 consecutive cases of CDH, the incidence of associated MMC with CDH was stated as 4.3%.[6]

There has been one previous case report of left-sided CDH, MMC and hydrocephalus prenatally diagnosed; but to the best of our knowledge, this is the first reported case of the above constellation with a right-sided CDH diagnosed prenatally.

The presence of associated major malformations is

universally seen as a negative prognostic factor. Likewise, an increased mortality rate has been shown in right-sided versus left-sided CDH.[3] Associated chromosomal anomalies have a reported prevalence ranging from 9.5% to 21%.[4,5]

It has often been stated that *in-utero* transport of prenatally diagnosed CDH and a planned delivery at a tertiary centre is an advantage for mother and baby. However, Skari's meta-analysis was unable to show this.[5]

Traditionally, lung hypoplasia has been viewed as a secondary consequence of *in-utero* compression of the fetal lung. Experimental evidence is emerging for a primary defect in lung development itself in CDH.[6] Further advances in postnatal therapy, including permissive hypercapnia and liquid ventilation along with prenatal MRI to facilitate fetal surgical planning, may hold the key to future developments in CDH care.[7]

REFERENCES

1. Dott MM, Wong LYC, Rasmussen SA. Population-based study of congenital diaphragmatic hernia: Risk factors and survival in Metropolitan Atlanta, 1968-1999. *Birth Defects Res A Clin Mol Teratol*, **67**:261-7.
2. Pagon RA, Adam MP, Ardinger HH, Bird TD, Dolan CR, Fong CT (eds). Congenital diaphragmatic hernia overview. Gene reviews [Internet], University of Washington, Seattle, Seattle (1993-2014); Updated 2010 Apr 16.
3. Barseghyan K, Jackson HA, Miller DA. Prenatal diagnosis of intraventricular hemorrhage in a fetus with congenital diaphragmatic hernia using magnetic resonance imaging. *Minerva Ginecol* **67**:383-4.
4. Benachi A, Cordier AG, Cannie M, Jani J. Advances in prenatal diagnosis of congenital diaphragmatic hernia. *Semin Fetal Neonatal Med* **19**:331-7.
5. Skari H, Bjornland K, Haugen G, Egeland T, Emblem R. Congenital diaphragmatic hernia: A meta-analysis of mortality factors. *J Pediatr Surg* 2000; **35**:1187-97.
6. Yechiel Sweed, Prem Puri. Congenital diaphragmatic hernia: Influence of associated malformations on survival. *Arch Dis Child* 1993; **69**:68-70.
7. Tudorache S, Chiutu LC, Iliescu DG, Georgescu R, Stoica GA, Simionescu CE, *et al*. Prenatal diagnosis and perinatal outcome in congenital diaphragmatic hernia. Single tertiary center report. *Rom J Morphol Embryol* **55**:823-33.