LETTER TO EDITOR (VIEWERS CHOICE)

CONGENITAL DIAPHRAGMATIC HERNIA IN SIBLINGS

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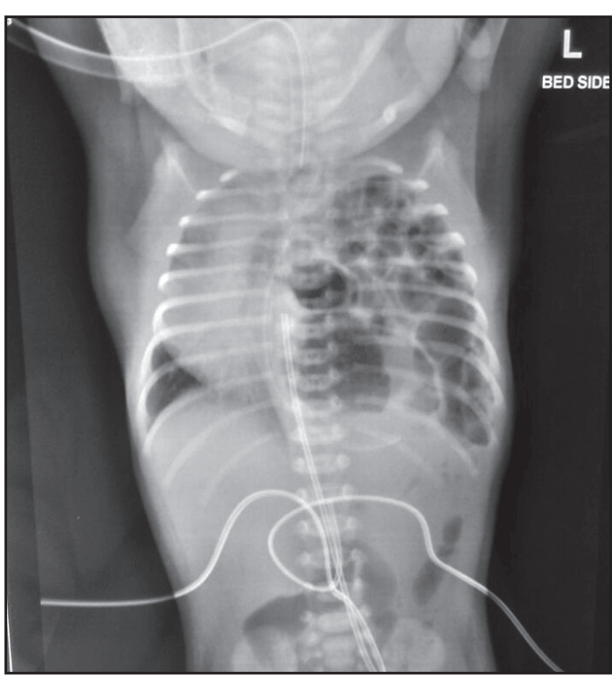
Congenital diaphragmatic hernia (CDH) occurs in one per 2500–4000 births and has a mortality rate between 30–60%. (1-3) CDH has predominantly sporadic occurrence, though up to 2% cases may be familial. (4) We present two siblings with CDH encountered at our centre.

The first child was born at term with a birth weight of 3.5 kgs and did not have any congenital abnormalities or dysmorphic features. She developed respiratory distress within the first 12 hours. Based on clinical examination and radiological investigations, a left sided diaphragmatic hernia was diagnosed. Posterolateral defect was repaired on the third day of life. Presently, she is 7 years old and has satisfactory developmental progress. The second sibling with CDH was born seven years later. He was second of monozygotic twin male babies. He was diagnosed to have left sided CDH during an antenatal scan at 20 weeks of gestation. He was born at 35 weeks gestation, with a birth weight of 2.3 kg and no other abnormalities. His postnatal X-ray showed left sided CDH (Figure 1). Posterolateral defect repair was done on seventh day of life. Post operative problems included pulmonary hypertension but eventually he had good recovery. Based on the operative description, there was no difference in the size of the hernia between the two siblings. Presently, he is 5 months old and has normal development. Parents were advised for genetic tests; however they refused for the same.

Figure 1: Chest X-ray showing congenital diaphragmatic hernia



Published reports describe more than 50 families containing two or more relatives affected with CDH. The causative chromosome abnormality could not be identified in most cases. (5) A multifactorial model of inheritance has been supported. Our patients were siblings of either sex, born seven years apart who presented with posterolateral defect and CDH. Though genetic tests could not be done as the parents were unwilling, the cases highlights the need for genetic workup in patients with familial CDH to determine risk in future pregnancies.

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