Denouement and Discussion

Congenital Diaphragmatic Hernia

The patient's chest radiograph shows a congenital diaphragmatic hernia (CDH) with herniation of bowel loops (Figure [arrow]) into the right hemithorax, shifting the heart and mediastinum to the left side. A relatively gasless abdomen is also observed. To rule out diaphragmatic eventration, an ultrasonogram was obtained that confirmed the presence of a posterior right-sided CDH. The right lobe of the liver was not herniated. There was no associated hydrothorax, nor were there ascites.

A CDH affects 1 in every 2000 to 4000 live births.[1] Most of the cases occur sporadically, and usually no familial link can be identified. However, there have been reports of familial cases with autosomal recessive, autosomal dominant, and X-linked inheritance patterns; a karyotype may also show chromosomal anomalies.[2-4] This well-known structural anomaly is diagnosed prenatally in about 50% of cases. A CDH occurred on the left side in 88% of cases, on the right side in 10% of cases, and bilaterally in 2% of cases. Although a CDH is usually a disorder that occurs in newborns, up to 10% of cases have occurred after this newborn period and may even occur during adulthood. The classic presentation of patients with CDH is the development of respiratory distress in the first few hours or days of birth; this respiratory distress is due to a defect in the muscular portion or the tendon of the diaphragm, to pulmonary hypoplasia, and to pulmonary hypertension.[5] A late presenting CDH may manifest as dyspnea, chest pain, abdominal pain, nausea, vomiting, and dysphagia. The diagnosis is occasionally noted incidentally during chest radiography performed for symptoms unrelated to the CDH.[6,7] With the surgical repair of a diaphragmatic defect, whether during the first 24 hours or later, nearly half of the patients with a prenatally or postnatally diagnosed CDH can survive.[8] On the other hand, the outcome for patients with a late presentation of CDH is extremely good, with low or no mortality.[1]

An association with other organ malformations is seen in nearly 40% of cases. Cardiac anomalies are the most common organ malformations associated with CDH[9,10]; in the present case, a patent foramen ovale was observed during echocardiography. In addition, spinal, gastrointestinal, chromosomal, and pulmonary malformations are also associated with CDH.[9,11,12] In our Figure, scoliosis (double-headed arrow), hemivertebrae (asterisks), and diaphragmatic hernia (arrow) are readily seen on the chest radiograph. The abnormal positioning of the stomach may be helpful in differentiating a CDH from those few cases of congenital pulmonary airway malformation, previously known as congenital cystic adenomatoid malformation, in which the cysts are large enough to mimic the air-filled intestinal loops. In congenital pulmonary airway malformation of the lungs, the stomach and bowel are in normal position and have a normal appearance.[13] Although most CDHs (90%) occur on the left side, there is no relation between the side of a CDH and the presence of a major anomaly.[9]

At the time of hospital admission, the patient was in good general condition with no respiratory distress, cardiac failure, or developmental difficulty. The patient underwent surgical repair and had an uneventful postoperative recovery.

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