CASE REPORT

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Congenital diaphragmatic hernia in identical twins

Accepted: 17 June 1999

Abstract The authors present a pair of identical twins with congenital diaphragmatic hernia (CDH) diagnosed prenatally, who underwent successful surgical repair. They were diagnosed as having CDH at 32 weeks' gestation and showed respiratory distress soon after cesarean section at 33 weeks' gestation. Both survived after scheduled perinatal management followed by surgery, for which the prenatal diagnosis of CDH was valuable.

Key words Congenital diaphragmatic hernia · Identical twins · Prenatal diagnosis

Introduction

The birth prevalence of congenital diaphragmatic hernia (CDH) is reported to be 3.3 per 10,000 births [14]. Although this disease is thought to be due to a developmental accident rather than genetic transmission, the possibility of a genetic factor cannot be completely excluded. So far, 52 families with CDH in siblings have been reported [1–4, 6, 9–11, 13, 15, 16]. We report a pair of identical twins who were diagnosed as having CDH prenatally and treated successfully.

Case report

A 20-year-old primigravida delivered live female twins at 33 weeks' gestation by emergency cesarean section because of acute toxemia. Both twins had been diagnosed as having CDH at 32 weeks' gestation when ultrasound (US) showed intestinal loops in the left thoracic cavity and shifting of the mediastinum to the right. In twin A, the stomach was also observed in the thoracic cavity. The lung-thorax transverse area ratio (L/T ratio) [8] by US was 0.19 and 0.23 in twins A and B, respectively (Fig. 1). Since the placenta was monochorionic and diamniotic, they were presumed to be monozygotic.

Twin A weighed 1,857 g at birth and had Apgar scores of 1 at 1 min and 3 and 5 min. Twin B weighed 1,561 g at birth and had Apgar scores of 2 at 1 min and 3 and 5 min. They both required high-frequency oscillatory ventilation just after birth. The alveolar-arterial oxygen difference was 537 mmHg and 323 mmHg in twins A and B, respectively. Plain roentgenograms confirmed the diagnosis of left CDH in both infants (Fig. 2). Twin B was operated upon on day 1 of age and her postoperative course was uneventful. Twin A was operated upon at 2 days of age after persistent pulmonary hypertension had been stabilized by inhalation of nitric oxide. The diaphragmatic defects were 2.7 × 0.8 cm and 2.0 × 1.0 cm in twins A and B, respectively; both were closed directly.

At 7 days of age, twin A deteriorated suddenly because of perforated peritonitis, and an ileostomy was performed. Mechani-
chal ventilation was necessary for a total of 28 and 13 days in twins A and B, respectively. Twin A was operated upon at 40 days of age to close the ileostomy. Laser therapy for retinopathy of prematurity was carried out at 6 weeks of age in both twins. Both were discharged after 151 and 113 days of hospitalization, respectively, in good condition. At 18 months of age, both twins showed normal physical and mental develop.

Discussion

CDH most often occurs as an isolated event, yet there have been reports of familial recurrence [1–4, 6, 9–11, 13, 15, 16]. These reports suggest that the risk for future siblings ranges from 0.9% [1] to 2.0% [11] after one infant is affected. The majority of familial cases occurred in siblings, with a few occurring in other close relatives [2, 3, 11]. Three sets of twins, excepting stillborn infants, were reported to have CDH to date, all of which were diagnosed after birth [6, 9, 15]. Ours is the first report of a pair of identical twins with CDH diagnosed before birth and treated successfully. Families with one affected child should be advised that CDH is not always a sporadic anomaly. Considering the poor outcome, careful counseling of the parents and prenatal examination in subsequent pregnancies should be recommended [10].

Accurate prenatal diagnosis enables better planning of the site and type of delivery and postnatal therapy in CDH. The severity of this disease can be predicted by measurement of the L/T ratio, which reflects the degree of lung hypoplasia. We have been using this parameter in the prenatal assessment of CDH patients [8]. We organized two independent teams for perinatal treatment of each patient in collaboration with neonatologists and anesthesiologists after prenatal assessment. Scheduled and well-organized management carried out just after birth presumably contributed to the good outcome.

Recently, prenatal administration of steroid or vitamin E has been reported to be effective in accelerating pulmonary maturation in animals [7, 12]. Harrison et al. have reported that high-risk fetuses with CDH appear to benefit from temporary tracheal occlusion, which accelerates lung growth and corrects the pulmonary hypoplasia, performed fesoscopically [5]. These prenatal medical/surgical treatments will presumably reduce mortality in compromised patients diagnosed as having CDH before birth.

References


Fig. 2A, B Chest X-ray films at birth: multiple loops of bowel filled with gas in thoracic cavity in A twin A and B twin B