LATE DIAGNOSIS OF RIGHT CONGENITAL DIAPHRAGMATIC HERNIA

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Abstract

Congenital diaphragmatic hernia is one of the most challenging malformations associated with high mortality. It is defined by the presence of an orifice in the diaphragm that allows passage of the abdominal viscera into the thorax.

Right side diaphragmatic hernia is rare (12-15%) as compared to left side (85-88%) with overall incidence of 1 in 5000 live births. Risk factors associated with poor outcome include low birth weight, prematurity, associated structural anomalies, and chromosomal defects. Treatment is a topic which depends on size and location of the defect. This is a case of isolated CDH diagnosed at 8 month postnatally. The patient was diagnosed after a respiratory infection occured and and a X- ray of the chest was taken.

The successful management of this case is presented.

Keywords: Right congenital diaphragmatic hernia (CDH), late diagnosis, infant

Introduction

Congenital diaphragmatic hernia (CDH) is a defect that occurs in 1 in 3.000-5.000 live births, of which approximately 60% occur in isolation without other congenital anomalies. This is a rare congenital anomaly of the diaphragm that occurs due to poor embryogenesis with atrophy of the diaphragm muscle fibers and loss of muscle tone. It is more frequent on the left side (85-88%), and bilateral cases have been reported, male predominance is also recognized (2:1 ratio)

Neonatal CDH is a well-recognized entity, but its presentation beyond the neonatal period varies, giving rise to erroneous clinical and radiological diagnoses. In contrast to the high neonatal mortality and morbidity rates for CDH, the prognosis for late CDH hosts if diagnosed early is generally favorable [1,2]. CDH is the result of incomplete closure of the normal pleuroperitoneal canal during fetal development. Most cases are diagnosed before birth or in the neonatal period. However, 5% to 45.5% of the CDH may appear asymptomatic during the neonatal

period, to manifest itself in childhood and adulthood. The congenital defect is identical in neonates and older patients, but the approaching and complicating symptoms of CDH in older patients differ considerably from those found in newborn patients. Although the exact etiology of most cases of congenital diaphragmatic hernia remains unknown, there is increasing evidence that genetic factors play an important role in the development of CDH. Chromosomal abnormalities have been identified as an important etiology for non-isolated CDH. In most published cases, chromosomal abnormalities were identified using a combination of anal chromosome bands-GYsis and/or fluorescence in situ hybridization(FISH). The use of new technologies, such as comparative genomic hybridization based matrices (arrayCGH) - is likely to increase the number of chromosomes identified in individuals with CDH and may aid in the identification of CDH-related genes. Trisomies 13, 18, and 21 and 45, are the commons aneuploidías described in association with CDH [1,3]. Embryologically, the etiology of CDH is postulated as the abnormal migration of myoblasts from the superior cervical somites in two of the four embryological structures that contribute to the development of the diaphragm as septum transversum beginning at week 4 of gestation and the pleuroperitoneal membrane at 8-12 weeks of gestation. Thomas hypothesized the involvement of altered myoblast growth in the pleuroperitoneal membrane, when the abdominal viscera return to the peritoneal cavity prematurely [2,4].

Case presentation

A 8 month old female infant was transferred from another clinic with the suspicion of a diaphragmatic hernia. The infant had a pulmonary infection and the parents brought her to the hospital because of respiratory distress. A X-ray of the chest was taken and a diaphragmatic right hernia was suspected (figure 1 and 2).

At arrival in our clinic the child had no respiratory distress syndrome with spO_2 =99%, with no dyspnoea or other clinical signs suggestive for a diaphragmatic hernia.

The patient was admitted in our Pediatrics department, where it received treatment for the respiratory infection. The nasal exudate reveiled an infection with Streptococcus pneumoniae for which the patient received local treatment with Ciprofloxacin 0,3%.

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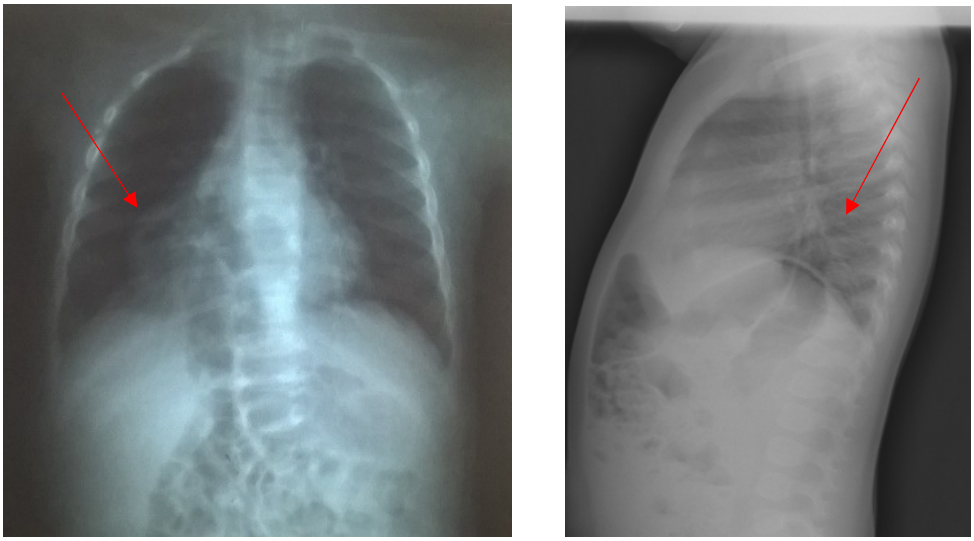

Figure 1, 2 - X-ray of the chest with colon loop in the right hemithorax.

18 days after she was transferred to our institution, the operation was scheduled.

At operation a median laparotomy was done. There was a large defect in the right diaphragm, approximately 4/3 cm. The hernial contents (colon loop) were reduced, the leaves of the diaphragm were dissected and the defect was closed in a single layer with nonabsorbable sutures. Thoracic drainage was not necessary, a small tear in the right pleura was sutured.

The postoperative period was uneventful. The patient could be extubated on the same day after the operation and was discharged on the 10th postoperative day.

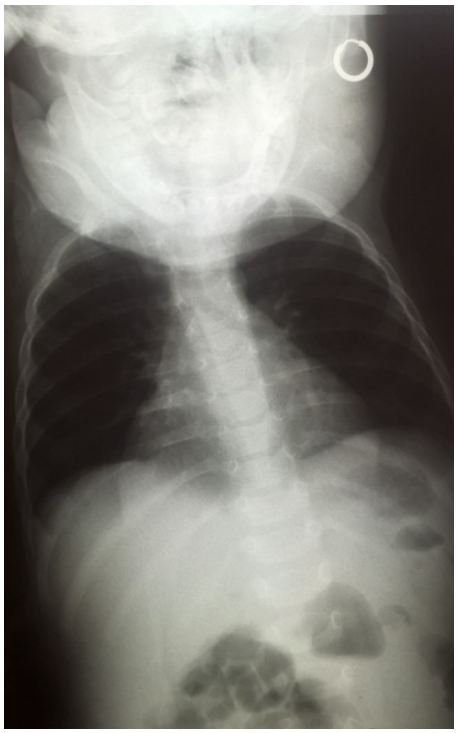

Figure 3 – postoperative chest X-ray with normal, expanded lungs and hemidiaphragms at the same level.

Discussion

Prenatal diagnosis by ultrasound detects more than 50% of CDH cases at a mean gestational age of 24 weeks. Three-dimensional ultrasound and fetal magnetic resonance (MRI) are other methods of prenatal diagnosis used in assessing the severity and outcome of the CDH. There is no prenatal diagnosis of CDH in this case. After birth the child did not show any signs of respiratory distress and had a normal development without medical problems until the age of 8 month.

Historically CDH was regarded a surgical emergency and the new born was rushed to the operating room for surgical correction. In 1987 Bohen et al. proposed a period of medical stabilization and delayed surgical repair in an attempt to improve the overall condition of the neonate.

The surgical repair can be difficult in right sided CDH because the size of the defect. The defect in the presented case was relatively large. Only a thin rim of diaphragm was present anteriorly and the posterior rim was fortunately present.

If the liver is herniated into the thorax the reduction of liver can be a difficult problem. Liver replacement in the abdomen can be complicated by kinking of hepatic veins causing profound hypotension. Potential anatomic anomalies such as possible hepatopulmonary fusion [6,7], anomalous venous drainage is uniquely associated with right sided defects.

Survival based on liver herniation alone is 43% as compared to 93% survival without liver herniation [8]. The series published by Fischer et al. [9] has shown the survival rate (right CDH 55% to left CDH 77%) ECMO requirement (right CDH 40% vs left CDH 15%) prosthetic material in right CDH vs left CDH (76% vs 41%) and abdominal wall (38% vs 19%) repairs. These data support that right side CDH carries a high mortality and morbidity. The repair of a CDH may be as variable as clinical management. The type of repair is dependent on the size of the defect. If the defect is small, a tension free primary surgical closure should be performed with non-absorbable sutures. If the defect is wide, primary closure may be attempted with a patch (Marlex, Goretex, Dacron) or a muscle flap (latissimus dorsi, serratus anterior).

Sometimes if the defect is not too large, the patient is diagnosed with delay and has only repeated pulmonary infections which eventually leads to a chest X-ray and the diagnosis of a right congenital diaphragmatic hernia.

Conclusion

Preoperative physiological stabilization and subsequent elective repair has become the corner stone of management of CDH like our case.

Presentation of right congenital diaphragmatic hernia beyond the neonatal period gives rise to erroneous clinical and radiological diagnoses. In contrast to the high neonatal mortality and morbidity rates for CDH, the prognosis for late CDH hosts if diagnosed and treated correctly is generally favorable

Success in this difficult case is optimized by the close cooperation between pediatrician, anesthetist and the pediatric surgeon.

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