Death of an Infant due to an Undiagnosed Congenital Diaphragmatic Hernia

Tod eines Kleinkindes infolge einer nicht-diagnostizierten kongenitalen Zwerchfellhernie

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- congenital diaphragmatic hernia
- late presentation
- unexpected death in childhood
- acute intestinal obstruction
- Bochdalek hernia

Schlüsselwörter
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- späte Manifestation
- unerwarter Tod im Kindesalter
- intestinale Obstruktion
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Background

The presented case of a 14-month-old boy who died from the effects of an undiagnosed Bochdalek hernia demonstrates the significant clinical relevance of both prenatal and postnatal screening methods, despite the known limitations of the procedures regarding CDH, and that especially in children with recurrent gastrointestinal and/or respiratory symptoms in infancy one has to consider a late manifestation of CDH as a differential diagnosis.

Case report

A 14-month-old boy became apathetic at home after 4 days of recurrent vomiting and abdominal pain and died in the hospital. Clinically, the cause of death was unclear. The autopsy showed a 2 cm postero-lateral, non-irritating diaphragmatic defect on the left side. There was a herniation of the terminal ileum, the cecum, almost the entire (very extended) colon and parts of the greater omentum (**Fig. 1**). There was a slight mediastinal displacement to the right and the left lung was significantly compressed by the herniation. The child had no other malformations or other competing causes of death.

The boy was born spontaneously after an uneventful pregnancy with multiple prenatal ultrasound scans in the 42nd week of pregnancy. No irregularities in the ultrasound examinations were apparent either pre- or postnatally. The nutritional and developmental statuses of the child were always appropriate for the child's age. From the 4th to the 13th month of life the boy suffered months of recurring changes in the stools (sometimes slimy, sometimes diarrhea, sometimes stool retention), abdominal pain and vomiting. He also suffered recurrent respiratory infections from the age of 8 ½ months until shortly before his death. The pediatrician repeatedly considered a gastrointestinal infection as the probable cause, but microbiological stool examinations could find no evidence of any pathogens. Diagnostic imaging was not carried out. The parents were recommended several times in vain that the patient's symptoms be further investigated in a hospital setting. An outpatient examination of the children in its 8th month of life in a hospital, with tests including a general medical examination and sonography of the abdomen, revealed no cause for the symptoms. After a few hours the parents prevented any further diagnostic measures by taking the child home against explicit medical advice. Documentation of such further diagnostic measures and differential diagnosis procedures were not carried out.

Discussion

After the autopsy results the leading cause of death was considered to be a respiratory insufficiency due to compression of the left lung by the herniated colon segments. The position of the herniated intestinal components undoubtedly prevented an adequate lung ventilation on the left side, and in this way oxygenation became acutely impaired. This raises the question retrospectively, whether the child's condition could have been diagnosed before its death. Upon suspicion of a CDH there is a chance of prenatal diagnosis by the combination of prenatal ultrasound/echocardiography and the MR tomographic measurement of fetal lung volume [4,8]. However, with the prenatal ultrasound diagnostics that are designed to help diagnose abnormalities of the fetus, heart failure usually remains the focus of attention due to the frequency and severity of this disorder. For this reason the areas relevant for CDH receive less scrutiny than areas such as the heart, urinary tract or the nape of the neck. It

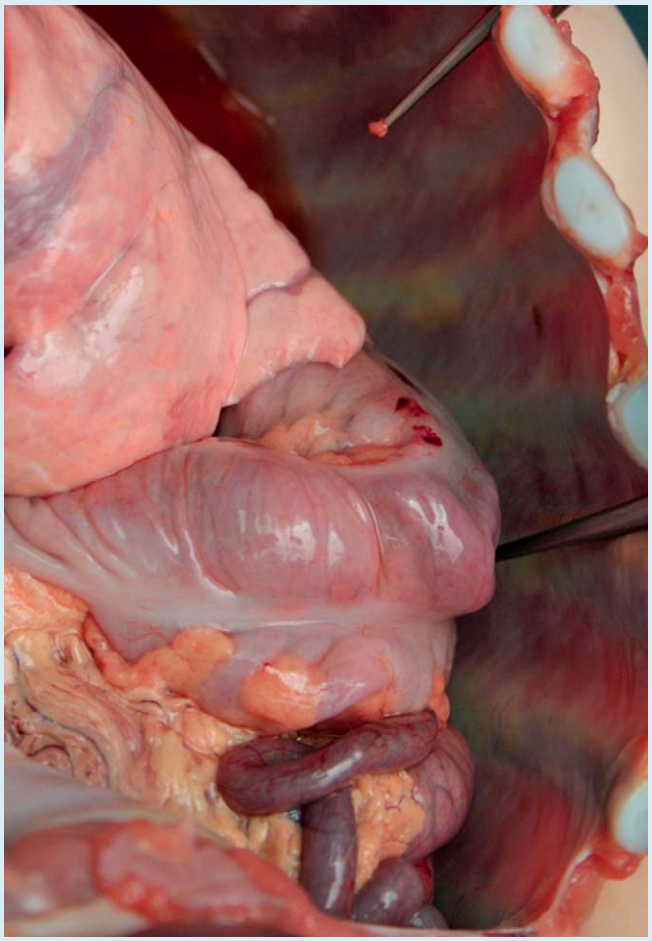

Fig. 1 Autopsy view of the left thoracic cavity-compression of the lung because of the herniated colon.

Table 1 Clinical characteristics of patients with late presenting CDH, according to literature.

respiratory symptoms (43 %) [5]
e. g. cough (without percentage) [2]
e. g. recurrent respiratory infections (14 %) [6]
e. g. dyspnea/tachypnea/cyanosis (without percentage) [2]
gastrointestinal symptoms (33 %) [5]
e. g. recurrent vomiting (18 %) [6]
e. g. intermittent abdominal pain (18 %) [5, 6]
respiratory and gastrointestinal symptoms (13 %) [5]
no symptoms (11–23 %) [5, 6]
failure to thrive (32 %) [5]

is also possible that, even where there is a predisposition for the disease, the detected herniation of the intestine only occurs at a later timepoint, so that the typical features of manifest disease may be missing at the time of investigation. In addition, all screening methods are subject to error, as has been described for the prenatal detection of congenital heart defects by obstetrical ultrasound [10].

Postpartum, the diagnosis of CDH is often made on the basis of respiratory problems in the newborn with cyanosis and dyspnea, both of which result from the pulmonary hypoplasia that is also usually present. Since the CDH seems to coincide with a systemic immune response starting immediately after parturition, the diagnosis can also be supported by significantly raised serum concentrations of cytokines [7]. In addition, pulse oximetry is also available postnatally as an adjunctive screening method for mechanical respiratory disabilities that impair gas exchange [9]. However, this method is not routinely found in maternity hospitals and neonatal units so that "error rates" from this procedure have not been reliably calculated.

Late presentation of CDH (after the neonatal period) is associated with a multitude of clinical symptoms that can occur in the most varied combinations (**Table 1**). For the diagnostic procedure with later manifestations of CDH, it is of utmost importance to take differential diagnostic considerations into account with recurring gastrointestinal or respiratory infections in childhood. As regards the necessary diagnosis, the guidelines of the German Society of Pediatric Surgery only indicate that a lung auscultation, a chest x-ray and ultrasound examinations of the abdomen and internal organs be performed [1]. However, it should be noted that in up to 25 % of all cases clinical and radiological misdiagnoses have been made and that apparently regular screening results can not ultimately rule out disease with any certainty. [2, 3].

Conclusion

From this case it can be concluded that in children with recurrent respiratory and/or gastrointestinal symptoms after exclusion of other (often infectious) causes, differential diagnoses for rarer causes, e. g. the late manifestation of CDH, should be carried out since fatal outcomes can now be prevented almost completely in a pediatric surgical setting.

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