CASE REPORT

Unrecognised diaphragmatic hernia in a refugee child: an incidental diagnosis

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SUMMARY

A 4-year-old boy from Syria was evaluated at the emergency department because of an upper airway viral illness. His physical examination showed a significant intensity reduction of all heart sounds in the absence of any other pathological signs. As the child was affected with Down's syndrome and had suffered thoracic and abdominal trauma because of bombardments, a diaphragmatic hernia was immediately suspected and was confirmed through a simple chest X-ray. A careful clinical examination is crucial in refugee children and adolescents, as several medical and surgical disorders could have escaped previously.

BACKGROUND

Diaphragmatic hernia (DH) is defined as the presence of abdominal viscera in the chest because of a defect in the diaphragm. In children, DH can be congenital or acquired. Congenital diaphragmatic hernia (CDH) occurs in 1 in 3000 live births, accounting for 8% of all birth defects and 1%–2% of infant mortality and, thus, represents one of the most prevalent and lethal congenital anomalies.

Anatomically, CDH develops through a posterolateral defect ('Bochdalek hernia') in 70%–75% of cases or through an anterior defect ('Morgagni hernia') in 23%–28%, whereas central defects are very unusual, accounting for 2%–7% of cases. Importantly, CDH can be associated with additional anatomical defects and be part of genetic syndromes; for instance, Morgagni hernia resulted to be associated with chromosomal anomalies, such as Down's syndrome. At least half of all CDHs are diagnosed during prenatal ultrasound screening, and the remaining cases are usually diagnosed shortly after birth because of the onset of respiratory symptoms and distress.^{1,2} However, diagnosis of CDH in infants or older children has been reported, too. Late-presenting CDH is characterised by a great variety of clinical symptoms, as this may be diagnosed in completely asymptomatic children, as well as in patients with chronic, acute or even sudden symptoms.^{3,4} In children, acquired DH (ADH) is observed less frequently than in adults, where the traumatic aetiology is more likely. Blunt thoracic/abdominal trauma, as those occurring in traffic accidents, fall from height or assaults, can cause diaphragmatic lesions leading to ADH. However, in politically unstable world areas afflicted by civilian wars and terrorist attacks, blunt traumas due to

bomb blasting can involve the paediatric population as well as adults.⁵

CASE PRESENTATION

A 4-year-old refugee boy from Syria was brought to the paediatric emergency department by the local social services in order to get a medical evaluation because of mild episodic cough and transient respiratory discomfort during the previous night. No fever or other remarkable symptoms were reported and, actually, at his arrival to the hospital, the child was in very good clinical condition, was very lively and, particularly, did not complain of cough or any other respiratory clinical manifestation, except for runny nose. Indeed, his vital signs were normal (non-invasive oxygen saturation: 97%, normal values (n.v.): >95%; heart rate: 96 beats per minute, n.v.: 90–120; respiratory frequency: 22 breaths per minute, n.v.: 20–30; arterial blood pressure: 101/58 mm Hg, n.v.: >80/>50 mm Hg; capillary refilling <1 s, n.v.: <2 s). The child was affected with Down's syndrome that was diagnosed at birth (based on his physical stigmata) and confirmed by the analysis of the karyotype. Anyway, no major malformation was reported and, particularly, parents provided a normal medical report concerning heart ultrasound performed during the first year of life. Importantly, the child presented several scars localised both at the limbs and at the trunk, as he suffered traumatic injuries during the bombing in Syria.

The patient resulted to be acutely affected by an upper airway respiratory infection, but the physical examination revealed a significant reduction of all heart sounds. Actually, this finding was not consistent with the patient's slim body, neither with his good clinical condition, as no symptoms or signs of cardiorespiratory instability (as in patients with significant pericardial effusion or cardiomyopathies) were recorded at all. Importantly, both his chromosomal disease and his traumatic personal history were highlighted; therefore, such a finding of softened heart sounds addressed the clinical suspicion towards the possibility of an asymptomatic or mildly symptomatic diaphragmatic hernia.

INVESTIGATIONS

Through a simple double-projection chest X-ray (figure 1), a previously unrecognised large bowel herniation in the anterior mediastinum, between the heart and the sternum, could be evidenced. ECG showed normal findings, except for a reduction of

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Figure 1 (A) Anteroposterior chest X-ray demonstrating abnormal content in the mediastinum, whose appearance is consistent with large intestine 'haustra'; no signs of pleural effusion and pneumothorax are evident. (B) Lateral chest X-ray showing the interposition of large intestine herniation between the sternum and the heart, displaced posteriorly.

the signal amplitude in all the derivations. Blood cell count and biochemistry were all in the normal range for age. A complete radiological assessment through CT was completed in the tertiary hospital centre where the child was referred.

DIFFERENTIAL DIAGNOSIS

The diagnosis of diaphragmatic hernia could be safely ascertained by chest X-ray, as described above. In this clinical setting, the main differential diagnosis is among different types of diaphragmatic hernia, based on the location of the defect of diaphragm, leading to the dislocation of some abdominal organs into the thoracic cavity. A classification of CDH based on the primitive defect is mentioned in the Background section, and CT scan is able to provide an appropriate anatomical study. In this clinical case, the definition of a congenital or acquired origin—or maybe both—of the diaphragmatic hernia was the most challenging diagnostic point, as the patient both was affected by a predisposing genetic condition and was exposed to traumatic injuries being able to lead to diaphragmatic hernia. Actually, the medical report concerning heart ultrasound performed in the first few days after birth was normal; however, a minor constitutional defect of the diaphragm that evolved eventually could not be excluded completely.

TREATMENT

The child was immediately referred to the paediatric surgery department of a tertiary hospital in order to receive further diagnostic investigations and the appropriate therapeutic intervention. According to the information received by the surgeon, the child underwent laparotomy to reduce the intestinal herniation and to repair the diaphragmatic defect. During the surgical procedure, an anterior defect of the diaphragm was observed and a mild congenital defect worsened by the previous abdominal/thoracic trauma resulted to be the most probable hypothesis of causality.

DISCUSSION

This case report evidenced how a careful medical evaluation associated to the appropriate attention to the patient's personal history can lead to the diagnosis of a previously unrecognised disease, such as diaphragmatic hernia beyond the neonatal period; this finding represents a very unusual circumstance for

paediatricians operating in developed countries. Indeed, in these geographical areas, most diaphragmatic hernias are congenital and, thus, are diagnosed in the first months of life. As mentioned in the introduction, CDH is a consequence of diaphragm malformations and, thus, are usually evidenced by prenatal ultrasonography or are recognised at birth or in the first days of life because of the occurrence of acute respiratory distress (indeed, this anomaly of the diaphragm is often associated to lung hypoplasia or compression).² However, some cases of CDH have a late presentation in infancy or result to be even asymptomatic; if so, the diagnosis is made through a chest X-ray performed for other reasons.^{6,7}

Despite these considerations, the diaphragmatic hernia in our patient could have been actually acquired after a traumatic rupture of the diaphragm, as the previous medical reports (including heart ultrasound performed at 1 year of age) showed no abnormalities. Moreover, the child was a victim of bombardments in Syria, leading to multiple injuries, including blunt thoracic/abdominal trauma.

CDH can be caused by the rupture of the diaphragm, as a result of either penetrating or blunt injury of the abdomen as well as the thorax. Large perforation of the diaphragm with herniation of abdominal contents into the chest cavity usually is associated with respiratory distress and requires urgent operative correction, but small defects of the diaphragm could be actually unnoticed for a long time if no significant injury of intra-abdominal or intra-thoracic organs is concomitant. These findings are often seen in adults because of traffic accidents, fall from height or violent acts. Thus, the diagnosis of diaphragm rupture can be missed during the early phase of the post-traumatic assessment, which can lead to a progressive and prolonged intestinal herniation, resulting in severe complications (such as intestinal obstruction, ischaemia or sepsis) or, on the contrary, to mildly symptomatic or asymptomatic conditions,^{4,5,8} as it was in our case probably.

Very recently, Er *et al* assessed the demographical data and injury characteristics of patients who were injured in the Syrian Civil War in order to define differences in injury characteristics between adult and children. They reported blunt trauma as being the most frequent mechanism of injury in children. In the acute clinical setting, the most frequently injured region of the body was the head and many presented polytraumatic injuries;

however, around 48% of children presented an abdominal and/or thoracic trauma, too.⁹ Thus, the diagnosis of diaphragm rupture may be often missed during the early phase of the post-traumatic assessment, also because of a major attention to multiple and more severe injuries as well as the lack of appropriate radiological tools in those areas. Therefore, a missed diagnosis of diaphragmatic rupture in the acute setting can lead to a progressive and prolonged intestinal herniation that can result in severe complications or, on the contrary, in only mildly symptomatic or asymptomatic conditions; the latter situation is sometimes evidenced by accident, namely through a chest X-ray performed during febrile respiratory illnesses or lung infections, for instance.^{3 10}

Actually, in our case, only an attentive physical examination along with a plausible personal history could lead to the clinical suspicion of diaphragmatic hernia in the absence of any evident lower respiratory and/or gastrointestinal symptoms. Unfortunately, refugee patients are exposed to misdiagnosis and/or underdiagnosis of several diseases, including remarkable surgical

conditions, in addition to nutritional deficiencies, infectious diseases and mental health disorders.^{11 12}

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Learning points

- ▶ Diaphragmatic hernia can be asymptomatic for a long time and only a careful clinical examination could reveal it: only mild and isolated signs, such as the reduction of intensity of heart sounds, could be present.
- ▶ Children with a personal history of important thoracic and/or abdominal trauma because of bombing should be monitored for long-term complications: diaphragmatic hernia should be considered in this clinical setting.
- ▶ Children coming from war areas must be fully examined in order to diagnose diseases being asymptomatic or mildly symptomatic, which could escape to a superficial assessment.

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