LATE-PRESENTING CONGENITAL DIAPHRAGMATIC HERNIA IN CHILDREN

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Congenital diaphragmatic hernia (CDH) is a typical neonatal emergency. There is, however, a subset of patients who manifest this congenital anomaly in infancy and early childhood. Late-presenting diaphragmatic hernia is characterized by a variety of gastrointestinal and respiratory symptoms with acute or chronic modes of manifestation.

The aim of the study. Analysis of the clinical course, diagnosis, and surgical management of late-presenting CDH in children.

Material and methods. A retrospective study of medical charts and diagnostic studies was performed for children older than 1 month of age with CDH, treated in the University Department of Pediatric Surgery and Urology in Wrocław between 1975 and 2005.

Results. In a 30-year period, 23 children with late-presenting CDH were treated. In one case, paraoesophageal hernia was diagnosed; Morgagni hernia was diagnosed in 2 cases. Among 20 children with postero-lateral CDH, 12 were younger than 1 year of age at diagnosis. In this group, left-sided CHD was noted in 17 children, while a right-sided defect was noted in 3. Nine infants with left CDH presented mainly respiratory symptoms; in 3 cases gastrointestinal symptoms prevailed. In the group of children beyond 1 year of age, 6 manifested chronic respiratory symptoms and 2 had mainly acute gastrointestinal complaints. In all children, CDH was diagnosed preoperatively. The most accurate diagnostic modality in 17 cases was a contrast upper GI study. Intraoperative findings revealed the presence of a hernial sac in only 3 patients. A complicated course of CHD with torsion of the stomach was noted in 2 children; intestinal necrosis was seen in one patient. Small and large bowel were the organs most commonly displaced into the thoracic cavity (noted in 12 and 11 children, respectively). In all children, surgical management of CDH was successful.

Conclusions. Lack of typical clinical signs and symptoms of late-presenting CDH seems to be a factor responsible for delayed diagnosis of this anomaly in older children. Late presentation of CDH should be taken into differential diagnosis in all children presenting with gastrointestinal or respiratory symptoms. Late-presenting diaphragmatic hernia is an unequivocal indication for surgical management as soon as its diagnosis has been established.

Key words: congenital diaphragmatic hernia, late presentation, children
genital diaphragmatic hernia. It has been a subject of a considerable number of papers, but most authors focused on the presentation of its unusual clinical and diagnostic features (3, 4). Only a few papers reported a larger series of children with late-manifesting diaphragmatic defects (2, 5, 6, 7).

This fact prompted the authors of the presented paper to undertake a retrospective analysis of their experience in the management of diaphragmatic hernia with clinical presentation beyond infancy.

MATERIAL AND METHODS

Clinical notes of all patients with CDH treated at the University Department of Pediatric Surgery and Urology in Wroclaw during the period of 1975-2005 were retrospectively studied. Only patients with manifestations of diaphragmatic anomalies beyond 1 month of life were included into the group of late-presenting CDH. The patients with traumatic hernia and diaphragmatic eventeration were excluded from analysis.

Clinical symptoms, the type of presentation, as well as diagnostic and surgical management were studied in each case; the results of imaging studies and their impact on final diagnosis were also investigated. The data from operative notes allowed us to assess the anatomical features of the diaphragmatic defect, its dimensions and topography, hernial content and the presence of a hernial sac, and details of the surgical treatment. Finally, an analysis of postoperative complications and outcome was carried out.

RESULTS

Among 97 children with CDH treated during the 30-year period, there were 74 newborns (76.2%) with manifestations within the first few days of life. In 23 children, diagnosis of CDH was established after 1 month of age and these patients constituted the study group. Postero-lateral diaphragmatic defect was noted in 20 of them, while 2 patients had an antero-lateral Morgagni hernia and one had paraoesophageal hernia. The age of the patients in this group ranged from 1 month to 6.5 years. There was a predominance of infants up to 6 months of age among those having postero-lateral CDH (tab. 1). Morgagni hernia was diagnosed in two boys aged 4 months and 3.5 years. Paraoesophageal hernia was noted in a 14-month-old girl presenting with recurrent respiratory tract infections, frequent episodes of vomiting and failure to thrive. Right sided postero-lateral hernia was found in 3 patients, while 17 had a left-sided defect. Two children were affected by infantile cerebral paralysis. The associated anomalies were noted in only one child with multiple ipsilateral skeletal deformities of the ribs, spine and metacarpal bone. In one child, the lower lobe of the lung was resected due to adenomatoid malformation which was confirmed upon postoperative histological study.

The children with late-presenting CDH had various clinical symptoms. In a collective analysis, predominant gastrointestinal complaints (nausea, vomiting, poor feeding, constipation, abdominal pain) and predominant respiratory symptoms (respiratory distress, cough, tachypnoea, recurrent respiratory tract infections) were noted. Children without a predominance of symptoms of either group, had a mixed clinical presentation. With regard to the mode of presentation, the patients were divided into groups with either acute or chronic manifestations. The latter refers to patients in whom clinical data indicated the occurrence of symp-

<table>
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<th>Mode of presentation</th>
<th>Clinical symptoms</th>
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Late-presenting congenital diaphragmatic hernia in children

Symptoms within days or weeks prior to the diagnosis of CDH. The data regarding the clinical presentation of CDH for all patients are presented in tab. 1. Although the small number of patients in each subgroup does not render an objective statistical analysis possible, it should be emphasized that late-presenting CDH in infants is usually characterized by an acute onset of symptoms, while in older children it manifests frequently with chronic complaints. A 5-month-old infant without previous medical problems presented with sudden cardiorespiratory arrest and had CDH diagnosed on an emergency chest X-ray. In two children with recurrent upper and lower respiratory tract infections, pleural drainage was applied in the course of their previous diseases. One of them had a pleural empyema diagnosed at the age of 13 months and required subsequent hospitalizations in a pediatric unit before his CDH was diagnosed when he was 2.5-years old. In the second patient, 6.5-year-old boy, a chest tube was inserted because of suspicion of lower lobe abscess with pleural effusion. His long-term treatment of recurrent pneumonia did not result in clinical improvement. Final diagnosis was made via a contrast upper GI study.

A 4.5-month-old infant with Morgagni hernia presented with acute respiratory infection and associated circulatory problems. A presumptive diagnosis of myocarditis was made and the child was referred to a pediatric cardiology unit. On chest CT scan a suspicion of anterior mediastinal tumor was raised. Contrast upper GI study did not reveal the presence of bowel loops in the chest. Intraoperative findings were consistent with right anterolateral hernia with intrathoracic displacement of a portion of the liver. The second patient with this type of CDH presented with mild non-specific abdominal and respiratory symptoms. Suspicion of the diaphragmatic defect was raised incidentally when he was admitted to the regional hospital for diagnostic studies of the liver due to a positive Hbs antigen test; the hernia was finally confirmed on a contrast study. At surgical exploration, a large right retrosternal defect was found with the presence of a portion of the right hepatic lobe as well as a portion of the stomach and large bowel in the chest within a hernial sac.

Among 20 children with postero-lateral defects, 17 of them had a definite diagnosis of CDH made on upper GI contrast study which showed the presence of hollow viscera within the thoracic cavity. In 3 patients, plain abdominal and chest X-rays were sufficient to diagnose a diaphragmatic hernia. Two of these children manifested with acute gastrointestinal symptoms suggesting mechanical ileus. In a boy who suffered from a sudden collapse with cardiorespiratory arrest, CDH was diagnosed on chest X-ray. In two boys with thoracic ectopia of the left kidney, preoperative imaging studies included intravenous urography and ultrasound scan. One patient with recurrent episodes of pneumonia had a bronchoscopy performed in the local pediatric hospital.

All children were subjected to an operative treatment. Nineteen children had postero-lateral CDH; in one patient with a Morgagni hernia and in a girl with a paraoesophageal hernia, the abdominal approach was used. Thoracotomy was performed in two patients – a boy with a Morgagni hernia manifesting as a mediastinal tumor and a patient treated previously with pleural drainage. A hernial sac was noted in both cases of Morgagni hernia and in 3 patients with a left postero-lateral defect.

Among children with right-sided postero-lateral CDH, small and large bowel loops were found in the chest in two of them, and stomach and large bowel in the third case. In the group of left-sided defects, small bowel and large bowel were the most frequently displaced viscera and found in 12 and 11 cases, respectively. The stomach was noted in the thoracic cavity in 6 children, the spleen in 5 patients, the left kidney in 2, and greater omentum in a single case.

In 6 children, complications of the hernia were noted intraoperatively. Stomach volvulus was found in two children; furthermore, in two patients the displaced bowel was strangulated at the hernial ring. In one patient, segmental resection of a 50-cm long ischaemic portion of the small intestine was necessary. In two children, a fibro-purulent exudate covering the displaced bowel loops and lining the pleural cavity was noted.

In all patients the diaphragmatic defect was closed primarily. In a girl with paraoesophageal hernia, an additional Nissen fundoplication was performed.

Postoperative complications occurred in 3 children. In the abovementioned boy with a previous history of recurrent pneumonia and
pleural drainage, intrapleural haemorrhage was noted in the immediate postoperative period and he required an additional thoracotomy. Two other complications were minor and consisted of wound dehiscence. All patients were discharged from the surgical department in good condition.

**DISCUSSION**

During the period of the last 30 years, 94 children were treated for congenital posterolateral diaphragmatic hernia. This is without doubt the most common anatomical variant among all diaphragmatic defects. According to the arbitrarily accepted medical literature definition, late manifesting CDH is a diaphragmatic defect diagnosed outside the neonatal period (5, 7). Adhering to this criterion, children with CDH in these age groups constituted 21% of all cases. Other variants of diaphragmatic hernia were noted in single cases and confirm observations made by the other authors indicating its very rare incidence in childhood (2, 4, 8). It seems that the relative frequency of late manifesting CDH with regard to all diaphragmatic defects depends basically on the number of babies affected by CDH treated in the same pediatric surgical centre (3, 4). This fact is responsible for discrepancies between the data reported from various surgical departments.

Although in the presented paper Morgagni hernia and paraoesophageal hernia were included, a detailed analysis of their clinical aspects was carried out in the group of 20 children with posterolateral defects. The number of patients does not render an objective statistical study possible, but basic epidemiological data are concordant with those observed by the other authors indicating its very rare incidence in childhood (2, 4, 8). It seems that the relative frequency of late manifesting CDH with regard to all diaphragmatic defects depends basically on the number of babies affected by CDH treated in the same pediatric surgical centre (3, 4). This fact is responsible for discrepancies between the data reported from various surgical departments.

In the presented series of 20 children, diaphragmatic defect was diagnosed by the age of 3 years in 95% of cases. The data from the collective review based on a survey of literature dealing with this subject indicate that CDH may manifest in older children as well. Baglaj noted that 23.4% of left-sided hernias and 15% of right CDHs were recognized in children older than 5 years of age (3). It seems therefore that the patient’s age cannot be taken as unequivocal criterion excluding CDH in a child. There are also reports of CHD diagnosed in adult patients (9). It should be stressed, however, that more than 80% of cases of this clinical variant of CDH manifest clinically in infancy and early childhood (2, 4, 10).

Late-presenting CHD is characterised by variability of clinical symptoms. A child affected by this anomaly may present with symptoms from the respiratory system, gastrointestinal system, or both systems simultaneously. Such non-specific manifestations with absence of pathognomonic symptoms are responsible for diagnostic difficulties in a considerable number of patients (4, 5, 7, 11). The mode of clinical presentation in an infant or older child may be acute or chronic when the symptoms are mild or of a recurrent nature. Upon analysing 20 children with posterolateral hernia, the authors showed a significant predominance of acute presenters among infants, regardless of the side of hernia. In contrast, older children with chronic presentation of the diaphragmatic defect prevailed more than twofold. In both clinical groups, respiratory symptoms were noted more frequently than those from the gastrointestinal tract. Unfortunately, a low number of patients with right CDH preclude a precise clinical analysis.

The authors representing the Congenital Diaphragmatic Hernia Study Group found in all children affected by right-sided defect respiratory symptoms exclusively, while patients with left CDH had gastrointestinal complaints occur-
ring two-fold over respiratory complaints (4). In the above mentioned collective review of 362 children with late-manifesting CDH, a significant predominance of acute presentation in infants and young children with left-sided anomalies was noted. In the group of children aged more than 5 years, there was a near equal number of acute and chronic presenters. However, among those with right CDH, only infants had a higher number of the acute mode of presentation (3). All these discrepancies in the literature with respect to the clinical data seem to have its origin in small numbers of patients reported by most authors. The largest series of 26 children treated in one center was presented by surgeons from Toronto (2). Including the present paper, there have been only 10 clinical papers reporting more than 10 patient with late-presenting hernia published to date (5-8, 10-14).

A relevant aspect of late-manifesting CDH refers to its potential sudden presentation with acute cardiorespiratory arrest in an apparently healthy child without a past medical history of any pathologic condition. Such was the case in a 5-week old infant noted in the presented study. Prompt resuscitation allowed to stabilize the vital functions and subsequently diaphragmatic hernia was diagnosed as a cause of his sudden clinical deterioration. The literature survey indicates that such an unusual course of late CDH was noted in 7 patients, but in all of them the diaphragmatic defect was found on autopsy. In 7 other children with acute or chronic presentation, death had occurred before surgical intervention could be undertaken (3, 15, 16). Taking this data into account, it must be stressed that late-presenting diaphragmatic hernia can not be regarded as a completely benign condition associated uniformly with a very good outcome. On the other hand, it seems that this clinical variant of CDH should be considered as an unequivocal indication for surgical management as soon as its diagnosis has been established.

Variability in the clinical presentation of CDH in older children is responsible for diagnostic pitfalls and diagnostic delay. In one child with Morgagni hernia from the presented study, a mediastinal tumor was diagnosed initially. In two patients with left-sided CDH presenting with recurrent pneumonia, pleural drainage was applied in the course of their disease. Lack of clinical improvement despite prolonged medical treatment prompted their referral to a surgical center where the diagnosis of CDH was finally made. The data from the literature indicate that erroneous diagnosis of pneumothorax or pleural effusion in a child with CDH may be responsible for the occurrence of serious iatrogenic complications resulting from injury of displaced viscera. In 9 children, gastric perforation by pleural drain was noted. Single cases of liver injury and colonic perforation were also found (2, 3, 14).

Among the children presented here, a diagnosis of CDH was made on upper gastrointestinal tract study in 17 of them. In 3 children, chest and abdominal plain films were sufficient to recognize a diaphragmatic anomaly. In one child with Morgagni hernia, computed tomography of chest was performed preoperatively; this child was the only patient in whom the liver exclusively was displaced into the chest. Bowel was present within the chest in all three children with right postero-lateral hernia. Late-manifesting CDH may be associated with a wide range of radiological features. Intrathoracic gastric or intestinal displacement gives a typical appearance of the defect in most cases. On the other hand, an absence of hollow viscer may be responsible for the diagnostic dilemma. This holds true, especially in cases of right CDH with liver protrusion which occur in most instances. In such cases, ultrasonography, computed tomography or even scintigraphy have found their application (5, 8, 17).

In 4 of the above presented children, a complicated course of hernia was noted with incarceration of displaced viscera within the hernial ring. In one of these patients, resection of a necrotic segment of the small bowel was necessary. Two patients were found to have volvulus the stomach. The data from the literature have shown that complication of late-presenting diaphragmatic hernia, noted at surgical operation or autopsy, occurred in 12.4% of patients (3). This fact again confirms that CDH may be associated with potentially serious sequels and therefore such risks should be taken into consideration whenever indications for surgical management are discussed.

Contrary to neonatal CDH, late presenting diaphragmatic defects are reported to have a very good outcome. Accurate diagnosis allows for timely surgical management to be undertaken, consisting usually in placement of pro-
truded viscera back into the abdominal cavity and closure of the diaphragmatic defect (12, 14). In all of the analysed above children, simple closure of the hernial ring was possible without the need of application of reconstructive techniques. Other authors confirm this observation in their reports. Only in 4 children with late-presenting CDH reported in the literature, was a prosthetic patch used in order to restore continuity of the diaphragm (3). Recently, there have been a few case reports published indicating a usefulness of minimally invasive techniques in repair of diaphragmatic hernia in children (18-21). However, in every case when a thoracoscopic or laparoscopic procedure is considered, the surgeon must take into account a risk of a complicated course of hernia with the necessity of expanding a range of surgical interventions.

CONCLUSIONS

1. Lack of typical clinical features in children affected by late-presenting diaphragmatic hernia is responsible for a considerable diagnostic delay in many cases.
2. Late-presenting CDH should be considered in the differential diagnosis in infants and older children manifesting with respiratory or gastrointestinal symptoms.
3. Late-presenting diaphragmatic hernia is an unequivocal indication for surgical management as soon as its diagnosis has been established.

REFERENCES

There are several points requiring special consideration among the Authors' conclusions regarding congenital diaphragmatic defects not evident at the time of birth. These defects might lead to life threatening complications like cardiac arrest, visceral incarceration with a tumor-like picture, pneumothorax or pulmonary effusion with subsequent attempts of pleural drainage which may potentially cause further iatrogenic complications. Therefore, one has to agree with the statement that a delayed presentation of symptoms does not in reality mean a defect with an expected mild clinical course. The moment of diagnosis should automatically be an indication for an operative treatment which usually promises a very good result. My personal surgical experience fully supports the authors' observations of the atypical clinical course of these defects, even to the point of being finally diagnosed post-mortem. However, these cases should be considered as rare.

There is a surprisingly high percentage of delayed diaphragmatic defect manifestations (23.8% – 23 children out of a total number of 97 treated) reported by the Authors. It seems to be one of the largest series from a single surgical center, not only in Poland but also in the world. The frequency of occurrences of the diaphragmatic defects with a late presentation varies in the literature; it statistically ranges from 5-20% and is significantly lower in highly specialized pediatric surgical centers. However, it should be stressed that the analysis referred to children undergoing a diagnostic process before being referred to a surgical institution. The doctors taking care of these patients could have less experience in diagnosing these lesions, and this could be a reason for such a high rate of occurrence in the presented material; even more than a total number of newborns treated.

It was proven that a lack of symptoms like cyanosis and dyspnoea could easily delay reaching the final diagnosis. An isolated diaphragmatic defect without herniation of the abdominal contents is difficult to be diagnosed by imaging unless it shows a misplaced stomach or a segment of the bowel. It also applies to prenatal investigations. Based on our own material as well as published data from other centers, the Authors pointed out a different, more acute way of presentation of the defect in newborns and infants. More chronic, recurrent symptoms are typical for older children. The presence or absence of a hernia sac had no influence on the way and time of the onset of symptoms; it only could be connected to the time of defect formation during pregnancy. On the contrary, the type of diaphragmatic hernia had an influence on the time of the defect detection. Periesophageal (hiatal) and Morgagni's types of diaphragmatic hernias rarely give symptoms in newborns, and actually should not be analyzed as one group along with a Bochdalek's type of hernia. One has to agree with the authors in that a small number of patients with periesophageal (hiatal?), Morgagni's and right-sided hernias does not justify drawing any conclusions of their initial presentation. Left-sided diaphragmatic hernias had a clearer, more typical clinical picture with symptoms related more often to the respiratory, rather than digestive systems.

To summarize, I would like to state that the retrospective analysis of cases of the delayed diaphragmatic hernia symptom's presentation had proven the necessity of taking into account the possibility of CDH in any case of atypical respiratory and digestive tracts dysfunction. Also, contrast examination of the digestive tract might lead to a final diagnosis.

Prof. dr hab. Czeslaw Stoba
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