Unusual association of diseases/symptoms

Late-presenting diaphragmatic hernia associated with intrathoracic kidney: tachypnoea as unique clinical sign

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Correspondence to Dr Sergio Sesia, sergio.sesia@ukbb.ch**Summary**

Intrathoracic renal ectopia associated with a congenital diaphragmatic hernia represents a very rare congenital malformation. The incidence of intrathoracic renal ectopia with congenital diaphragmatic hernia is reported to be less than 0.25%. The authors report on a 5-month-old female infant with late-presenting congenital diaphragmatic hernia associated with a left-sided intrathoracic ectopic kidney and adrenal gland. While prenatal ultrasound showed no pathology, the postnatal ultrasound assumed a left-sided renal agenesis. After recurrent episodes of tachypnoea, a chest x-ray confirmed the diagnosis of congenital diaphragmatic hernia which was repaired without complications. Intraoperatively, intrathoracic ectopic kidney was diagnosed. The authors discuss embryogenesis, the diagnostic algorithm and therapy as well as follow-up of this rare malformation.

BACKGROUND

The complex embryogenesis of the kidney may give rise to several renal anomalies, such as ectopic kidney. Renal ectopia is commonly occurring as lower lumbar or pelvic ectopia, secondary to failure to ascend during fetal development. With a prevalence of less than 0.01%, intrathoracic renal ectopia (IRE) represents the rarest form of all renal ectopias.¹ The incidence of IRE as a result of congenital diaphragmatic hernia was reported to be less than 0.25%.¹ We report on a case of late-diagnosed intrathoracic kidney associated to a left-sided congenital diaphragmatic hernia.

CASE PRESENTATION

Our female infant was born at full-term by uneventful vaginal delivery with a birth weight of 3400 g. The prenatal ultrasound showed no pathological result. However, the newborn attracted attention by failure to thrive and by a palpable mass in the left upper abdomen since the second month of life. Abdominal ultrasound scan performed by an experienced paediatric radiologist, could not display the left kidney. Agenesis or aplasia of the left kidney was supposed. At the age of 5 months, the girl presented with recurrent episodes of tachypnoea, but without any history of respiratory infection. Clinical examination revealed suspected diagnosis of dextrocardia. Conventional chest x-ray showed mediastinal shift to the right side and a raised left dome of the diaphragm with gas shadows in the left lung field, suggestive of congenital diaphragmatic hernia (CDH) (figure 1). Immediately performed surgery demonstrated a true posterolateral CDH (Bochdalek type) without hernial sac. The left kidney and adrenal gland, the majority of small intestine and colon up to the colon descendens were found to be protruding into the left chest cavity. Intestinal malrotation was excluded. After replacement of the organs into the abdominal cavity, the diaphragmatic defect was closed by primary repair. The left

kidney was mobilised without difficulty in a near to normal site. Opening the renal fascia, the kidney showed no deformed shape and no abnormal rotation, as well as a normal age-related size and a normal abdominal origin of renal vessels. Since the renal pelvis showed no dilatation,

Figure 1 Conventional anteroposterior chest x-ray demonstrating air-filled loops of the bowel in the left hemithorax and a paucity of gas in the abdomen. The left diaphragm is not discernable and there is an extensive mediastinal shift to the right side. The heart and mediastinum are pushed on the right side. The diagnosis is compatible with a diaphragmatic hernia over the left diaphragm. Evaluating the location of the stomach bubble on the anteroposterior chest x-ray may facilitate the diagnosis of congenital diaphragmatic hernia.¹⁰

Figure 2 Postoperative ultrasound shows correct position of the left kidney, a normal renal perfusion without any signs of urinary obstruction

dissection of the left ureter was not performed. After primary closure of the abdominal wall, the postoperative course was uneventful, and the patient was discharged on day 10 with full enteral feeding. Postoperative ultrasound demonstrated persistent correct position of the left kidney and a normal renal perfusion without any signs of urinary obstruction (figure 2). In addition, there were no signs of pulmonary hypertension during a follow-up of 30 months.

INVESTIGATIONS

Conventional anteroposterior chest x-ray and ultrasound (figures 1 and 2).

DIFFERENTIAL DIAGNOSIS

Recurrent respiratory infections (for more detailed information, please see chapter discussion).

TREATMENT

We performed primary surgical repair. For more detailed information, please see above (chapter case presentation).

OUTCOME AND FOLLOW-UP

After primary closure of the abdominal wall, the postoperative course was uneventful, and the patient was discharged on day 10 with full enteral feeding. Postoperative ultrasound demonstrated persistent correct position of the left kidney and a normal renal perfusion without any signs of urinary obstruction (figure 2). In addition, there were no signs of pulmonary hypertension during a follow-up of 30 months.

DISCUSSION

The embryology of congenital anomalies of the kidney is well described.² Incidents during induction of the metanephric tissue, ascent of the kidney by cephalad migration and positioning of the kidney up to 8th gestational week may affect presence, number and/or the position of the kidney. With a prevalence of less than 1 in 16 000, IRE is a rare entity. It is more commonly found on the left (62%) than on the right side (36%) and is very rarely bilateral (2%). Males are more frequently affected than females.¹ Although a thoracic kidney may be associated with a deformed shape, an abnormal rotation, an elongated ureter, a high origin of renal vessels and medial deviation of the lower renal pole, the function of the kidney is usually normal.³ A classification of IRE into four groups was proposed³: (a) closed diaphragma, (b) eventration of the diaphragma, (c) CDH or acquired hernia and (d) as consequence of traumatic rupture of the diaphragma. Renal ectopia may be associated with genetic anomalies and several malformations.¹ Thus, amniocentesis for karyotyping should be offered to the parents and a thorough exam of the newborn is mandatory. The exact cause of CDH remains still unclear. During embryogenesis, the diaphragm is said to develop out of the septum transversum, the pleuroperitoneal membranes and the esophageal mesenterium.⁴ CDH of the Bochdalek type seems to be the result of the failure of complete fusion of the pleuroperitoneal membrane with the septum transversum.⁴ An intrathoracic kidney with concomitant CDH may be caused by direct herniation through the diaphragmatic defect into the intrathoracic space or by primary location in the posterior mediastinum due to a true embryologic ectopia.⁵ The association of ectopic adrenal gland with IRE

and CDH has not been well reported in literature. Many case reports of IRE do not mention the location of adrenal glands.⁵ It was suggested that adrenal gland determines final renal position, but the causal relationship is discussed controversially. There is a debate about a causal relationship between CDH and IRE.⁶ IRE may result from the delayed closure of the diaphragm or the accelerated ascension of the kidney before normal closure. The low incidence of right-sided IRE is explained by the early fusion of the pleuroperitoneal channel on the right side and by the presence of the liver as a barrier. The association of CDH and IRE was described 40 times in literature from 1997 to 2007. Only in 12 cases a CDH was associated.⁶ Further investigations are necessary to clarify the aetiology of IRE. On prenatal ultrasound, kidneys can be seen as early as at 12 weeks of gestation.⁷ IRE may be detectable prenatally by ultrasound. Some authors suggest the superiority of MRI in contrast to ultrasound in diagnosing and characterising IRE associated with CDH.⁸ In our case, neither CDH nor IRE was diagnosed by prenatal ultrasound, but by postnatally requested anteroposterior chest x-ray. Various pathological conditions such as lung sequestration, bronchogenic cyst, congenital cystic congenital pulmonary airway malformations and congenital lobar emphysema can present with similar radiographic findings.⁹ Radiological findings can also mimic other chest problems, such as pneumonia with cavitation, pleural effusion, massive pneumothorax and pneumatocele.¹⁰ Evaluating the location of the stomach bubble and the placement of the nasogastric tube on chest x-ray may facilitate the diagnosis of CDH.¹⁰ A high index of suspicion of CDH is required if the gastric bubble is not seen under the diaphragm on the left side. Intrathoracic located kidney is usually asymptomatic and most reported cases present an incidental finding on a chest radiography.¹¹ CDH is an anomaly that usually is diagnosed prenatally or in the neonate. However, CDH may present beyond the neonatal period in 5–20%.¹² Because of non-specific clinical signs, late-presenting CDH may pose significant diagnostic problems and result in delayed diagnosis.^{10–11 13} Symptoms may cause chest problems such as respiratory distress, pneumonia, or abdominal problems such as abdominal pain, vomiting, haematemesis and feeding difficulties. Early- and late-presenting CDH may lead to different clinical presentation. Respiratory symptoms seem to be associated more with early CDH and gastrointestinal symptoms with late-presenting CDH.¹³ In our patient, recurrent tachypnoea associated with failure to thrive was reported. In the absence of additional malformation such as hydronephrosis, malrotation or megaureter, ectopic kidney requires no specific surgical treatment,¹⁴ as in our case. Regarding the uneventful follow-up, we did not perform neither a cystogram to diagnose a vesico-ureteral reflux nor a dimercaptosuccinic acid-scintigraphy to evaluate renal function.

Learning points

- Late-presenting CDH may pose a significant diagnostic problem and lead to delayed diagnosis. Its clinical manifestations are variable and include respiratory and abdominal symptoms. In the case of an abnormal location of the stomach bubble on anteroposterior chest x-ray or its absence in the abdomen a high index of suspicion of CDH is mandatory.
- When recurrent respiratory symptoms occurred, chest x-ray should be performed to exclude late-presenting CDH. In contrast, IRE without concomitant CDH is usually asymptomatic. When abdominal ultrasound shows an empty renal fossa, IRE should be considered in the differential diagnosis of chest masses.
- IRE usually requires no specific treatment, in contrast to IRE associated with congenital diaphragmatic hernia, which must be treated surgically.

Competing interests None.

Patient consent Obtained.

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