Gastrointestinal

Complete dorsal pancreatic agenesis and unilateral renal agenesis

Adriana Moreira MD*, André Carvalho MD, Inês Portugal MD, José Miguel Jesus MD

Department of Radiology, Centro Hospitalar de São João, Alameda Prof. Hêrnani Monteiro, 4200-319 Porto, Portugal

ARTICLE INFO

Article history:
Received 9 September 2017
Received in revised form 12 October 2017
Accepted 30 October 2017
Available online 1 December 2017

Abstract

Dorsal pancreatic agenesis is a very rare congenital anomaly. Unilateral renal agenesis, on the other hand, is a relatively common congenital anomaly, although its etiology is not fully understood. Renal and pancreatic embryologic development appears to be nonrelated. We report a case of a 34-year-old man who was referred to our hospital for evaluation of cholestasis and microalbuminuria. Ultrasound and magnetic resonance imaging examinations showed empty right renal fossa and absence of the pancreatic neck, body, and tail. Our case report is the second case of a dorsal pancreatic agenesis and unilateral renal agenesis in a young male patient.

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Introduction

Congenital pancreatic anomalies are not frequently encountered in radiological examinations. In fact, only around 100 cases of dorsal pancreatic agenesis, probably the least common of all, have been reported in the literature.

Unilateral renal agenesis (URA) is the most common congenital anomaly of the urinary system. To the best of our knowledge, there is only one previously published case report of dorsal pancreatic agenesis and URA [1].

Case report

A 34-year-old man was referred for a nephrology and gastroenterology appointment at our hospital because of microalbuminuria and cholestasis. The fasting blood glucose level was normal on repeated examinations.

The patient was asymptomatic, with no relevant past medical history, and physical examination showed no abdominal abnormalities besides a syndromic facies. A sonographic study revealed a solitary left kidney; no right kidney was found in the abdominopelvic cavity and only the pancreatic head was clearly visualized by ultrasound. The patient underwent abdominal and pelvic magnetic resonance imaging, which confirmed an empty right renal fossa. No ectopic kidney was found. The pancreatic neck, body, and tail were missing, corresponding to a medical condition of complete dorsal pancreatic agenesis. The pancreatic bed was occupied by bowel loops. The pancreatic head and uncinate process were normal and no parenchymal calcifications were noted. The ventral pancreatic duct of Wirsung and the common bile duct were not dilated. The rest of the magnetic resonance imaging examination of the abdomen and pelvis was normal (Figs. 1-4).
Imaging diagnosis was one of complete dorsal pancreatic and right renal agenesis.

Discussion

Dorsal pancreatic agenesis is an extremely rare congenital anomaly; the first case was reported in 1911 [2] and approximately 100 cases have been reported so far [3].

The pancreas is a retroperitoneal organ composed of a head, neck, body, and tail. During embryogenesis, it develops around the fourth week of gestation from the ventral and dorsal buds, arising from the second part of the duodenum.

The pancreatic neck, body, and tail develop from the dorsal bud and drain through the accessory duct of Santorini and the minor duodenal papilla. The ventral bud forms the major part of the head and uncinate process. During the seventh week of gestation, the ventral bud rotates dorsally around the duodenum to fuse with the dorsal bud.

Dorsal pancreatic agenesis may be either complete or partial. In complete agenesis, the neck, body, tail, duct of Santorini, and minor duodenal papilla are absent; in partial agenesis, the body, the duct of Santorini, and the minor duodenal papilla are preserved.

Abnormal embryogenesis can lead to a failure in the normal development of the dorsal pancreas, resulting in complete dorsal pancreatic agenesis [4].

Other congenital anomalies have been associated with dorsal pancreatic agenesis, such as coarctation of the aorta, Fallot tetralogy, ventricular septal defects, heterotaxy, polysplenia...
syndrome, ectopic spleen, and bowel malrotation and horseshoe kidney [5,6]. Polysplenia occurs with various congenital anomalies of visceral organs, and the spleen may play a role in normal embryonic development of visceral organs from the dorsal mesogastrium [2].

The genes underlying pancreatic agenesis in humans are not known. However, familial transmission has also been reported in the literature [7].

Most patients with dorsal pancreatic agenesis are asymptomatic.

As to symptomatic patients, the majority suffer from epigastric pain, typically aggravated by meals [3,8]. Abdominal pain is more common in partial agenesis. About half of the symptomatic patients have diabetes mellitus, which occurs more often in cases of complete agenesis. The majority of these patients develop recurrent or intermittent pancreatitis and some exhibited signs of pancreatic exocrine failure [9].

The differential diagnosis of dorsal pancreatic agenesis includes pseudoagenesis, pancreas divisum, pancreatic head carcinoma, pancreatic pseudolipodystrophy, and pancreatic lipomatosis [8,10].

The absence of the dorsal duct of Santorini helps differentiate dorsal pancreatic agenesis from pseudoagenesis and lipomatosis.

Congenital renal agenesis is defined in terms of a complete absence of renal tissue at birth. This condition includes bilateral renal agenesis, which is not compatible with extrauterine life, or URA, the most common congenital abnormality of the urinary system, with an incidence between 1 of 500 to 1 of 3200 births. Men are more commonly affected, with a ratio of 1.2-2.3:1. The left kidney is more commonly absent in URA than the right kidney [11].

Renal development involves fusion of the metanephric blastema and the ureteric bud.

The metanephric blastema, the undifferentiated mesenchyma in the nephrogenic ridge, is the proximal component that originates from the glomeruli and tubules up to the distal collecting tubules. The collecting ducts, calyces, and pelvis originate from the ureteric bud, which is the distal component.

During the fifth week of gestation, the Wolffian duct gives rise to the ureteric bud and the latter penetrates the metanephric blastema. Nephrogenesis begins at the seventh week of gestation under the influence of the ureteric bud. By the 20th week, the ureteric bud branches into 15 generations to form the collecting duct system. At this time, nephrogenesis is only 30% complete and will proceed until the 36th week [12].

Despite the fetal kidney’s minor role in homeostasis in utero, it is, nevertheless, an important source of amniotic fluid. Hence, during pregnancy, oligohydramnios may be the first indication that the fetus has bilateral renal agenesis or URA [13].

Renal agenesis is caused by the absence of the nephrogenic ridge or the failure of the ureteric bud to induce development of the metanephric blastema.

In true agenesis, the ureter and ipsilateral bladder hemitrigone are absent. The contralateral kidney undergoes compensatory hypertrophy, which occurs primarily after birth [12].

Vesicoureteral reflux is the most common abnormality of the contralateral kidney, occurring in about 15% of the cases [14]. Other reported abnormalities in the remaining kidney include renal malrotation or ectopia, ureteropelvic junction obstruction, ureterovesical junction obstruction, ectopic ureter with partial obstruction of collecting system, dysplasia, and hypoplasia.

Other anomalies associated with renal agenesis are skeletal abnormalities, anorectal malformations, cryptorchism, and cardiovascular abnormalities.

VATER is an acronym to describe a nonrandom constellation of congenital anomalies that may go from vertebral and ventricular septal anomalies to anorectal atresia, tracheal and esophageal lesions, and radial bone and renal abnormalities [15].

Lal et al. [16] have reported a case of dorsal pancreatic agenesis with associated pancake kidney and bicornuate uterus, and Bagul and Traipathi [1] reported the case of a young man with a complete dorsal pancreatic agenesis and URA. Worth mentioning is also the case presented by Lee et al., referring to a 30-year-old woman with complete agenesis of the dorsal pancreas and unilateral kidney agenesis associated with a mutation of the pancreatic and duodenal homeobox 1 [17].

Pancreatic and duodenal homeobox 1 is a homeodomain transcription factor that plays a critical role in early pancreatic formation [18] and is vital for the growth of the pancreatic buds [19].

However, as referred to previously, the pancreas has an endodermal origin, unlike the genitourinary system, whose origin is mesodermal, and the embryologic development of the pancreas and kidney appears to be nonrelated.

Conclusion

In view of its rarity, our case report adds up to the existing knowledge and makes room for a better understanding of this rare congenital anomaly.

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