Congenital duodenal atresia with ‘apple-peel configuration’ of the small intestines and absent superior mesenteric artery: A case report and review of literature

Ashraf A. Alnosair, Mohamed Ibrahim Naga, Mohamed Ramadan Abdulla, Ahmed H. Al-Salem*

Department of Pediatric Surgery, Maternity and Children Hospital, Dammam, Saudi Arabia

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A B S T R A C T

Embryo logically, congenital duodenal atresia is a primary malformation that results from errors in recanalization of the duodenum in early gestation. This is in contrast to other intestinal atresias which result from vascular accidents. We report an unusual and rare case of congenital duodenal atresia, loss of the third and fourth parts of the duodenum with apple-peel configuration of remaining small bowel and absent superior mesenteric artery in a preterm child. The possibility of Strømme syndrome must also be kept in mind. The literature on the subject is also reviewed.

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Congenital duodenal obstruction (CDO) is a relatively common anomaly with an estimated incidence of 1:3000 to 1:5000 live births [1,2]. In the majority the site of obstruction is postampullary and there is a high association with Down’s syndrome [3]. Over the years, the prognosis of infants and children with CDO has improved markedly. Several factors, however, still affect the overall outcome including prematurity, and a high incidence of associated anomalies [4,5]. The causes of CDO are divided into intrinsic and extrinsic and intrinsic duodenal obstruction is thought to result from failure of recanalization of the duodenum [6]. This differentiates CDO from atresia in the rest of the intestines, which result from intrauterine vascular accident [7].

Apple peel intestinal atresia, or Christmas tree atresia, is a rare atresia in which the proximal jejunum ends blindly and the distal small bowel wraps around its blood supply in a spiral manner resembling an apple peel. It accounts for approximately 5% of all intestinal atresia and results from intrauterine vascular accident [7].

The concomitant occurrence of CDO and apple peel atresia is extremely rare and only four cases reported so far in the literature [9–12]. The possibility of Strømme syndrome which is rare consisting of apple peel intestinal atresia, ocular anomalies, microcephaly and developmental delay must be kept in mind also [13–17]. This report describes an unusual case of CDO associated with apple peel atresia, microcephaly and ocular anomalies. The literature on the subject is also reviewed and aspects of pathogenesis are discussed.

1. Case report

A baby girl, a product of premature gestation at 31 weeks and a birth weight of 1.4 kg was noticed to have bile stained vomiting. Antenatal abdominal ultrasound showed polyhydramnios and double bubble sign suggestive of congenital duodenal obstruction. Clinically, there was no abdominal distension and the oro gastric tube was draining bile-stained aspirate. There were also bilateral corneal opacities and microcephaly. Abdominal x-ray showed dilated stomach with double bubble sign with no air distally and

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* Corresponding author. P.O. Box 61015, Qatif 31911, Saudi Arabia. Fax: +966 3 8630009.
E-mail addresses: ahsaalsalem@hotmail.com, ahsaalsalem@gmail.com (A.H. Al-Salem).

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upper contrast study confirmed the diagnosis of duodenal atresia (Fig. 1). The patient underwent laparotomy which confirmed the diagnosis of congenital duodenal atresia just distal to the insertion of the biliary and pancreatic ducts. Distal to this there was absence of the remaining parts of the duodenum and superior mesenteric artery. The pancreas was normal and the bile duct was seen entering the duodenum (Fig. 2). There was also apple-peel deformity and jejunal end was found freely mobile in the abdomen (Fig. 3). The small intestines were shortened and supplied by a single vessel in a retrograde fashion. An end to end duodeno-jejunal anastomosis was done as well as appendectomy. Post-operatively, the patient did well and started on feeds gradually. She tolerated the feeds and currently on full feeds.

2. Discussion

Congenital intestinal atresia and stenosis are relatively common and among these congenital duodenal obstruction is the commonest accounting for more than 50% of all cases with a reported incidence of 1 in 3000 to 1 in 5000 [1,2,4,5]. It is also not uncommon for congenital duodenal atresia to be associated with other anomalies mainly congenital heart disease and Down’s syndrome [3,5].

The association of congenital duodenal obstruction with other intestinal atresia is extremely rare. One reason for this is that congenital duodenal atresia and intestinal atresia have different embryological etiologies.

Embryologically, the gastrointestinal tract is well developed in a five-week-old embryo. The foregut goes through a complete solid stage due to epithelial proliferation and subsequently undergoes vacuolization and recanalization to form the foregut lumen. Tandler in 1900 proposed that duodenal atresia and stenosis results from a failure of recanalization of the solid stage of embryonic duodenum [18]. This is in contrast to atresia of the rest of the small bowel which results from a vascular accident leading to interruption of the vascular supply to the atretic bowel segment [6]. This makes the association of duodenal atresia with other small bowel atresia and apple-peel atresia extremely rare.

Fig. 1. Plain abdominal x-ray showing double-bubble sign indicative of duodenal obstruction with no gas distally and contrast study confirming the diagnosis of complete duodenal obstruction.

The apple peel type of intestinal atresia is very rare and constitutes 5%–10% of all small bowel atresia [7,8]. It was first described in 1961 by Santulli and Blanc [7]. Apple peel atresia is a term used to describe a well-defined variant of high jejunal atresia near the ligament of Treitz where the bowel is shortened and assumes a helical configuration, the mesenteric defect is large and the bowel distal to the jejunal atresia is precariously supplied in a retrograde fashion by a single blood vessel from the right ileocolic or inferior mesenteric artery. The most accepted theory is that apple-peel deformity results from an intrauterine vascular accident in late gestation [6,7].

The present case we are reporting and those reported in the literature have common findings as outlined in Table 1. They all had an atretic third and fourth parts of duodenum and proximal jejunum with typical apple-peel configuration of the ileum and total absence of superior mesenteric artery. This association is extremely rare and only four cases were reported before [9–12]. Ours is the fifth. All these cases raise questions regarding the embryological etiology of congenital duodenal atresia. In all these cases, the totally missing superior mesenteric artery whether congenital or acquired as a result of intrauterine vascular accident resulted in duodeno-jejunal atresia and apple-peel atresia. In these patients the pancreatico-biliary ducts drain into the proximal second part of the duodenum. We feel that the extent of duodenal atresia depends on the degree of pancreatico-duodenal arcade interruption. Arbell et al. reported a prematurity with an extensive atresia from the first part of the duodenum to the mid small bowel; malrotation of the distal part, in volvulus and in an “apple peel” configuration; no connection of the bile ducts to the bowel; and the presence of a type II choledochal cyst [19]. Pumberger et al. reported four children, including two from one family, with duodeno-jejunal atresia associated with malrotation, volvulus, and absent parietal attachment of the mesentery [20]. In all, there is absence of the mesentery and distal parts of the superior mesenteric artery and the small intestine was supplied in a retrograde fashion from the right colic artery. In all these cases and those reported in the table, the pathoanatomical findings are different from the classical apple peel atresia. We feel that apple
peel atresia is of two types, the classical apple peel atresia characterized by duodenal atresia with the biliary and pancreatic secretions draining into this part of the duodenum. There was absent 3rd and 4th parts of duodenum as well as the superior mesenteric artery and apple-peel deformity with the jejunal end freely mobile and a marginal artery supplying the small intestines retrogradely.

In 1993, Strømme et al. described a new syndrome consisting of apple peel intestinal atresia in siblings with ocular anomalies and microcephaly [14]. They reported two siblings who presented shortly after birth with signs of upper intestinal obstruction due to apple peel jejunal atresia. In addition to intestinal atresia, both siblings exhibited severe microcephaly and ocular abnormalities. Subsequently, five more cases were reported (Table 2). All had apple peel jejunal atresia, microcephaly and ocular anomalies except one who had simple jejunal atresia [13–17]. Strømme syndrome is rare consisting of apple peel intestinal atresia, ocular anomalies, microcephaly and developmental delay. Microcephaly is more frequently observed but brain scans usually reveal no neurological abnormalities and some patients exhibit developmental delays. Jejunal atresia has an apple peel appearance in most but not all patients. The ocular manifestations comprise a wide range of anterior chamber anomalies with sclerocornea/corneal leukoma being the most common. Infants generally do well following

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Table 1

Summary of the reported cases of congenital duodenal atresia with ‘apple-peel configuration’ of the small intestines and absent superior mesenteric artery.

<table>
<thead>
<tr>
<th>No.</th>
<th>Authors (year)</th>
<th>Age and gestation</th>
<th>Sex and weight</th>
<th>Anomalies</th>
<th>Outcome</th>
</tr>
</thead>
<tbody>
<tr>
<td>1.</td>
<td>Weber and Freeman (1999)</td>
<td>NB 36 weeks</td>
<td>F, 2.1 kg</td>
<td>Loss of 3rd and 4th parts of duodenum and proximal jejunum, apple-peel atresia, absent SMA, DS, CHD.</td>
<td>Survived</td>
</tr>
<tr>
<td>2.</td>
<td>Tatekawa et al. (2007)</td>
<td>NB 36 weeks</td>
<td>F, 2.104 kg</td>
<td>Duodenal atresia, apple-peel atresia and multiple intestinal atresias.</td>
<td>Survived</td>
</tr>
<tr>
<td>3.</td>
<td>Ahmed et al. (2009)</td>
<td>8 days</td>
<td>M, 1.2 kg</td>
<td>Loss of 3rd and 4th parts of duodenum, apple-peel atresia, absent SMA.</td>
<td>Died</td>
</tr>
<tr>
<td>4.</td>
<td>Patil et al. (2011)</td>
<td>6 days</td>
<td>F, 1.6 kg</td>
<td>Duodenal atresia, apple-peel atresia, malrotation.</td>
<td>Died</td>
</tr>
<tr>
<td>5.</td>
<td>Present case Alnosair et al.</td>
<td>NB</td>
<td>F, 1.4 kg</td>
<td>Loss of 3rd and 4th parts of duodenum, apple-peel atresia, absent SMA.</td>
<td>Survived</td>
</tr>
</tbody>
</table>

CHD, congenital heart disease; DS, Down’s syndrome; NB, newborn; SMA, superior mesenteric artery.
intestinal surgery but no information regarding long term prognosis is available. Our patient had a classic Stromme syndrome (microcephaly, apple peel intestinal atresia and ocular manifestations) but in addition our patient had duodenal atresia with absence of the third and fourth parts of duodenum and absent superior mesenteric artery. This association is the first to be described in Stromme syndrome. The management of these patients is complex and difficult and the extent of duodenal atresia is important in this regard. The proximal atretic jejunal end is also mobile which makes it difficult to try and anastomose to the atretic duodenal end without twisting the apple-peel segment. Every attempt should be made to anastomose the jejunal end to the duodenal end without twisting the single vessel supplying the small intestines. A central venous catheter should be placed in these patients as they may require prolonged TPN. Apple-peel as an isolated anomaly is known to be associated with a high incidence of complications (63%), and mortality (54%). This must be kept in mind and because of the possibility of familial occurrence, genetic counseling is important.

**Conflict of interest**
We declare no conflict of interest.

**References**