A rare malformation: Double duodenal atresia associated with malrotation in a patient with “Cri du Chat” syndrome

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ABSTRACT

Duodenal atresia is a common cause of neonatal obstruction. It is frequently associated with other malformations such as Down syndrome, cardiac anomalies, malrotation or annular pancreas. Double duodenal atresia is an exceptional malformation. There are only few publications on this subject and none are in association with “Cri du Chat” (Cat Cry) syndrome. We present a newborn, prenatally diagnosed with duodenal atresia and with “Cri du Chat” syndrome. The double duodenal atresia was actually of two different types (type I and type II), associated with malrotation. The second atresia was a peroperative finding at reintervention, five days later. We wish to share our experience in order to avoid unnecessary surgery and co-morbidities.

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Duodenal atresia is a frequent bowel malformation and is often diagnosed on prenatal ultrasounds. The incidence is between 1/6000 and 1/10,000 cases, with an approximately equal male to female ratio [1]. It is classified into 3 types. In type I a duodenal mucosal diaphragm obstructs the lumen. In type II a fibrous cord connects the proximal and distal blind ends of the duodenum. Finally, type III corresponds to complete separation of the two duodenal segments. Duodenal atresia is often associated with Down syndrome, cardiac anomalies, malrotation, and annular pancreas. To our knowledge, it has never been described in a patient with “Cri du Chat” (“Cat Cry”) syndrome. This is a genetic disease resulting from the deletion of different sizes of the short arm of chromosome 5p(-). The frequency is about 1 in 45,000 live born infants. The main clinical features are a high pitched mono-chromatic cry, microcephaly, broad nasal bridge, epicanthal folds, micrognathia and severe psychomotor and mental retardation. It is sometimes associated with cardiac, neurological and renal anomalies. About 10% of patients suffering of severe cardiac malformation dye within the first year of life. Beyond this age, mostly have a normal life expectancy, like in general population.

Double duodenal atresia is a very rare condition and there are only few case reports in the English literature. We present the case of a prenatally diagnosed duodenal obstruction, which turned out to be a double malformation, not noticed during the first surgery, in a patient with “Cri du Chat” syndrome.

1. Case report

The patient was the first-born of a non consanguineous couple (the mother was 34 years old). The prenatal ultrasound (US) at 21 weeks of gestation showed an intraabdominal double-bubble sign, suggestive of duodenal atresia. Progressively, a significant poly-hydramnios had developed, requiring two amnio drains at 35 and 38 weeks of gestation. No other morphological anomaly was suspected. The amniocentesis performed at 35 weeks revealed a 46XX karyotype and detected a deletion on chromosome 5p15.33p15.2 corresponding to “Cri du Chat” syndrome. The parents refused the interruption of pregnancy. The infant was delivered vaginally at 39 weeks of gestation. The birth weight was 3020 g. She was in good general condition. Initial examination showed micrognathia and slight general hypotonia. The abdominal X-ray confirmed the double-bubble sign (Fig. 1). US investigations ruled out any cardiac anomaly but found the left kidney with duplex collecting system and an inferior pelvis dilatation.

A naso-gastric tube was inserted. The patient was electively operated on the next day through a transverse laparotomy. A very dilated proximal part of the duodenum was found and a fibrous cord connected the second to the third part (type II atresia). The rest of the bowel and colon were malrotated. No other bowel anomalies
were noticed. A transverse incision was done distally on the proximal duodenum and the Vater papilla was identified. A longitudinal incision was performed on the distal duodenum. Through this, helped by a naso-gastric tube, physiological serum was carefully injected and the permeability of the bowel was checked. The continuity of the duodenum was restored by a “diamond shaped” duodeno-duodenal anastomosis. The surgery was completed with a Ladd procedure and appendicectomy. The newborn then returned to the Neonatal Unit.

In post-operative period, gastric aspirates remained non-bilious but abundant. A few days after surgery, several attempts to feed the child failed. Vomiting occurred shortly after every meal. The abdomen was non-distended and stools were few and glairous. The control abdominal X-ray on day 3 showed again a double-bubble sign. The barium meal confirmed no passage of the contrast substance farther than proximal duodenum and found a gastro-esophageal reflux (Fig. 2).

The patient was re-operated on day 5. By the same incision, the inspection of the abdomen was performed. It confirmed a dilated proximal part of the duodenum, an intact anastomosis which looked permeable, and a non-dilated bowel, with the same size as at the first surgery. The anastomosis was opened on the anterior side and was permeable. The dilated duodenum was incised vertically at the limit between the first and the second part. A complete duodenal diaphragm was identified at the superior duodenal flexure (Fig. 3). This explained the total duodenal obstruction, the non-bilious vomiting and the persisting of the double-bubble sign on the X-ray. The diaphragm was excised and a termino-terminal anastomosis was performed. A naso-gastric tube was advanced transanastomotically until the fourth part of the duodenum for post-operative feeding. A second naso-gastric tube was placed to decompress the stomach.

Progressively the gastric aspirates diminished and enteral nutrition was started on the third post-operative day. This was well tolerated and it was slowly increased. The intestinal transit became regular. The feeding tube was withdrawn on day 9 after the second surgery and oral feeding was started. No abdominal problem was noticed. The patient was discharged at 20 days of life. The follow-up was performed regularly, by a multidisciplinary team including a surgeon, a nephrologist, a gastroenterologist, a neurologist and a physical therapist for her multiple problems related to the “Cri du Chat” syndrome. A pH-impedancemetry was performed and confirmed the gastroesophageal reflux seen during the barium meal. A treatment by Omeprazole was started. Two years later there were no surgical problems, just a developmental delay and a slight motor retardation.

2. Discussion

Duodenal atresia is a common cause of bowel obstruction. Certain case reports present familial cases in siblings from non-consanguineous parents [1] or 3 brothers, 2 girls and 1 boy from consanguineous parents suggesting a genetic transmission [2].

Our patient has micrognathia, a feature present in the “Cri du Chat” syndrome, as well as renal duplex collecting system and inferior pelvis dilatation.

Double duodenal atresia had already been described, but in limited number. About 22 case reports exist in the literature to our knowledge [3]. Stringer et al., published 4 cases of double duodenal atresia of different forms, associated with a biliary tree malformation in 2 patients, with no genetic anomalies [4]. Grosfeld et al., in his 20-year survey, found 3 patients with double duodenal atresia [5].

More recently, Keys et al., presented a case of prenatally diagnosed duodenal atresia which turned out, at surgery, to be a double
atresia with perforation on the anterior part of the second duodenum leading to free bile in the peritoneum, which explained the greenish coloration of the scrotum at birth [6].

Another rare presentation of double duodenal atresia, published by Migita et al., is as a 3 cm diameter abdominal cyst, which prenatally mimicked a duodenal duplication on US and MRI, [3]. The biliary tree was abnormal, in a “Y” shape, and was connected to the first and second part of the duodenum.

In our case, the double duodenal atresia was not associated with a biliary tree anomaly.

The rare case of a double duodenal web, the incomplete form, was reported by Sharma et al., in an 11 month-old cachectic infant [7]. The first web was situated on second part of the duodenum and the second one was at the duodeno-jejunal junction.

Ferguson et al., published a similar case to ours. A girl operated on for duodenal atresia had to undergo a second surgery for a gastroduodenal web [8].

After our experience, we propose, peroperative, to ask the anesthetists to advance the naso-gastric tube until the proximal incision and then to withdraw it, in order to assess the duodenal permeability.

Another point we missed were the gastric aspirates. The fact that they were non-bilious previous to the first surgery should have made us reflect about a possible obstacle above the Vater ampulla. We were reassured by the presence of bile and identification of the papilla during the first surgery and we continued the anastomosis without questioning this incoherence.

We stress the importance of knowing this variant of the malformation. It is necessary to check the permeability of the whole bowel, even if the anatomy seems to be normal, in order to avoid a second intervention, prolonged hospital stay, parenteral nutrition and any other comorbidities, like in our case.

3. Conclusion

While duodenal atresia is a common intestinal malformation, a double one is exceptional, but it exists. Moreover, in our case there were combined types (I and II) of duodenal atresia. The surgeon should be aware of this rare situation and he should carefully assess the permeability of the whole duodenum and the rest of the bowel to rule out any possible associated anomalies. A simple advancement of the naso-gastric tube up to the anastomosis could avoid such situation. We wished to share our experience in order to prevent unnecessary surgery.

References