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A case of Apple-peel atresia with familial duodenal atresia

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Objective

We aimed to present a case of prenatal diagnosis of an apple-peel atresia. It is considered that the small bowel atresia (SBA) is a congenital obstruction of the lumen of the duodenum, jejunum or ileum. Its incidence is estimated from 1.3 to 2.8 out of 10.000 live births (Virgone et al. 2015). Since the research of the anatomist J. Tadler, carried out in the beginnings of the twentieth century, it has been thought that the duodenal atresia could be explained by the failure of the solid cord (that could represent the embryological origin of the duodenum) to re-canalize in the second month of pregnancy. On the contrary, it is widely accepted that the atresia of the jejunum or ileum is caused by vascular accidents leading to an obstruction of the blood supply to the small intestine (Best et al. 2012). However, this different ethiopathology is being called into question since the last works concerning the role of FGF-10 in the small intestine genesis (Jones et al. 2020). Nowadays, the most accepted classification for jejunoileal atresia (JIA) was suggested by Grossfeld et al. in 1979, and it considers four types. The III-B type, also known as apple-peel atresia, occurs in less than 10 % of all JIA, and consists of proximal jejunal atresia and a short segment of ileum spiraling around a single mesenteric vessel (Dao et al. 2019). Reports of prenatally diagnosed JIA consist of case reports and small series, with sonographic detection typically occurring late in the third trimester (Wax et al. 2006). As of today, prenatal diagnosis of JIA represents a challenge, considering that its best-reported markers (polyhydramnios and bowel dilatation) are not specific because they can be found in other congenital gastrointestinal anomalies (Chen et al. 2021). In a recent metaanalysis, the detection rate of JIA has been estimated in 50,6%. Recently, new sonographic signs have been proposed to improve the rate of prenatal diagnosis of JIA (C-sign; Chen et al. 2021) and apple-peel atresia (Barber pole sign; Lu et al. 2022). While the duodenal atresia is associated with a certain frequency with other congenital abnormalities, the JIA is relatively rare and not commonly associated with other anomalies (Kirtane et al. 2019). A recent work found only twelve published cases of coexistence of duodenal and apple-peel atresia in the same patient (Molino Gahete et al. 2022). The incidence of the presence of these congenital defects in the same family is even more rare (Gross et al. 1996). The majority of the published works found the familial coincidence of these abnormalities in the same generations of siblings, pointing the possibility of an autosomal recessive inheritance. We only found one published work that includes one case of the familial aggregation of these defects in consecutive generations (mother and son; Tatekawa et al. 2007).

Methods

This is a case report.

Results

We present the case of a 38-year-old woman, gravida 1, with a BMI pregestational of 23.8. In her medical history, we highlight that she had a congenital duodenal atresia (descendent segment), operated thirteen days after the birth, and an inferior vena cava agenesis (diagnosed in adulthood). No consanguinity was referred. This pregnancy was conceived spontaneously and was followed in our high-risk unit because of the mother medical history. The first trimester ultrasound examination was performed at 13 weeks and 1 day and disclosed a CRL according to gestational age, with a low-risk combined screening and no other abnormalities. The mid-trimester ultrasound was performed at 20 weeks and 5 days and no abnormalities were found. An intermediate scan was demanded from the high-risk pregnancy unit, and it was performed at 30 weeks and 1 day. This scan showed a bowel dilation (transverse diameter of 18mm) with echogenic walls. The content of the bowel loops was anechoic and a normal peristalsis was observed. There was no evidence of ascites, abdominal cysts or calcifications. The stomach presented normal size. The estimated fetal weight was 1300g (22^{th} percentile). No polyhydramnios was observed. A new scan was scheduled in a month. The patient was admitted in our hospital at 33 weeks and 4 days with the diagnosis of premature preterm rupture of membranes. In the hours following her admission, the patient began to have regular contractions and cervical shortening. Given that the fetus was in a breech position, we decided to perform a cesarean, that was carried out without any incidence. A girl was born, with 1750g and 7/8/9 APGAR score. A few hours after the birth, a x-scan was performed, revealing a dilatation of the bowel loops in the mesogastrium and absence of distal gas. An abdominal ultrasound scan was also carried out: there was an important distension of the duodenum and proximal jejunum, while the rest of the small bowel was collapsed. The colon itself presented a small width. Eventually, a surgery was performed two days after the birth. During the surgery, an apple-peel atresia was diagnosed and a corrective surgery with end-to-end anastomosis was carried out without any incidence. The newborn did her first stool four days after the surgery; at that moment, the enteral nutrition with breast milk was started. The parenteral nutrition was retired eleven days after the surgery and the newborn was released from the hospital at the age of 34 days. Three months after the surgery, the clinical evolution is satisfactory, with no evidence of surgical complications.

Conclusion

As the previous works have pointed out, the prenatal diagnosis of this specific condition represents a challenge, given that the most frequent ultrasound signs are very unspecific, they are not present in the 100% of the cases and they can appear in other different intestinal abnormalities. Although it is very infrequent, this defect can appear with familial aggregation of other intestinal abnormalities. Bibliography Best, Kate E, Peter W G Tennant, Marie-Claude Addor, Fabrizio Bianchi, Patricia Boyd, Elisa Calzolari, Carlos Matias Dias, et al. 2012. «Epidemiology of Small Intestinal Atresia in Europe: A Register-Based Study». Archives of Disease in Childhood – Fetal and Neonatal Edition 97 (5): F353-58. https: //doi. org/10.1136/fetalneonatal-2011- 300631. • Chen, Dan, Kwong Ho Tam, Yiwei Xiao, Juan Geng, Yu Tan, Xiaochun Zhu, Wuping Ge, Jialiang Zhou, Shangjie Xiao, y Jiaxin Chen. 2021. «New Sonographic Feature (C-sign) to Improve the Prenatal Accuracy of Jejunal Atresia». Journal of Obstetrics and Gynaecology Research 47 (12): 4196-4202. https: //doi. org/10.1111/jog.15029. • Dao, Duy T. , Farokh R. Demehri, Carol E. Barnewolt, y Terry L. Buchmiller. 2019. «A New Variant of Type III Jejunoileal Atresia». Journal of Pediatric Surgery 54 (6): 1257-60. https: //doi. org/10.1016/j. jpedsurg.2019.02.003. • Gross, E, Y Armon, K Abu-Dalu, R Gale, y M Schiller. 1996. «Familial Combined Duodenal and Jejunal Atresia». Journal of Pediatric Surgery 31 (11): 1573. https: //doi. org/10.1016/S0022-3468(96)90182-7. • Jones, Matthew L. M. , Gulcan Sarila, Pierre Chapuis, John M. Hutson, Sebastian K. King, and Warwick J. Teague. 2020. “The Role of Fibroblast Growth Factor 10 Signaling in Duodenal Atresia. ” Frontiers in

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