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Particularities of the Antenatal Diagnosis of Duodenal Atresia: A Case Report and Literature Review

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Abstract

Introduction: Duodenal atresia is a rare malformation with an incidence of one case per 6000. It represents the typical form of small bowel obstructions. It is also associated with trisomy 21 in 30% of cases. The aim of this study is to specify the contribution of imaging in prenatal diagnosis and the outcome of duodenal atresia.

Materials and Methods: This is a descriptive retrospective study of five cases. Data were collected between 2015 and 2023 in the Obstetrics and Gynecology ward of Kairouan. Prenatal diagnosis was made by obstetric ultrasound in all cases. A fetal Magnetic Resonance Imagery (MRI) was also performed in two cases.

Results: The incidence of duodenal atresia in our study was one case per 6350 live births with a sex ratio of 4. The indicative feature was gastric dilation with duodenal involvement, creating the classic "double bubble" image with associated polyhydramnios. Three types of associated malformations were identified: trisomy 21 (in three cases), annular pancreas (in two cases), and Apple peel syndrome (in one case). All the new-borns were operated, 24 hours after birth. A duodeno-duodenostomy was typically performed. The outcome was favorable in all cases.

Keywords: Duodenal atresia, prenatal diagnosis, ultrasound, MRI, surgery.

INTRODUCTION

Duodenal atresia constitutes the most frequent cause of small bowel obstructions that can be diagnosed prenatally. It occurs in one case out of 6000 to 10,000 births [1] and it can be isolated or associated with other malformations. Approximately 30% of infants with atresia also have trisomy 21 [2]. Diagnosis is prenatal in 80 to 90% of cases. After birth, it is usually suspected in the presence of vomiting, often bilious, with or without anomalies in meconium elimination. Its management is through surgery. Nevertheless, neonatal resuscitation playing a crucial role in improving the prognosis of this condition, hence the importance of prenatal diagnosis for better neonatal management.

We report a series of five cases of duodenal atresia diagnosed prenatally in the Obstetrics and Gynecology ward of Kairouan. The objective of our study is to specify the contribution of imaging in prenatal diagnosis and the outcome of duodenal atresia.

Materials and Methods

This is a descriptive retrospective study. Data were collected from medical records and radiology reports

(MRI, ultrasound) over an extended period from 2015 to 2023. Initially, nine cases were included in the study. However, four were excluded due to incomplete medical records and one patient delivered a stillborn (due to lack of autopsy data).

RESULTS

The incidence of duodenal atresia is estimated at one case per 6350 live births. The average maternal age is 32 years, ranging from 27 to 43 years. Consanguinity was noted in one of the 5 cases. Multiparity was noted in two cases. In others cases, it was the first pregnancy. Regarding prenatal care, all pregnancies had at least two prenatal consultations, with three having nine prenatal consultations. A well-controlled gestational diabetes under dietary management complicated one pregnancy. Prenatal obstetric ultrasound allowed suspicion of the diagnosis in all five cases, with an average gestational age of 28 weeks and 6 days. In three cases, the diagnosis was suspected on the morphological ultrasound in the second trimester. However, in the other two cases, it was made during the third-trimester ultrasound. Ultrasound revealed a typical appearance with gastric and duodenal dilatation, forming the "double-bubble" sign. An absence of visualization of fluid in the downstream digestive loops was noted, suggesting complete duodenal obstruction (Figure 1). In two cases, the downstream loops were visible containing fluid. This could be explained either by incomplete obstruction or by the late onset of duodenal obstruction. The appearance of the downstream loops was not specified in two cases. In only one case, the digestive wall showed thickening with the presence of fluid outside the intestinal walls.

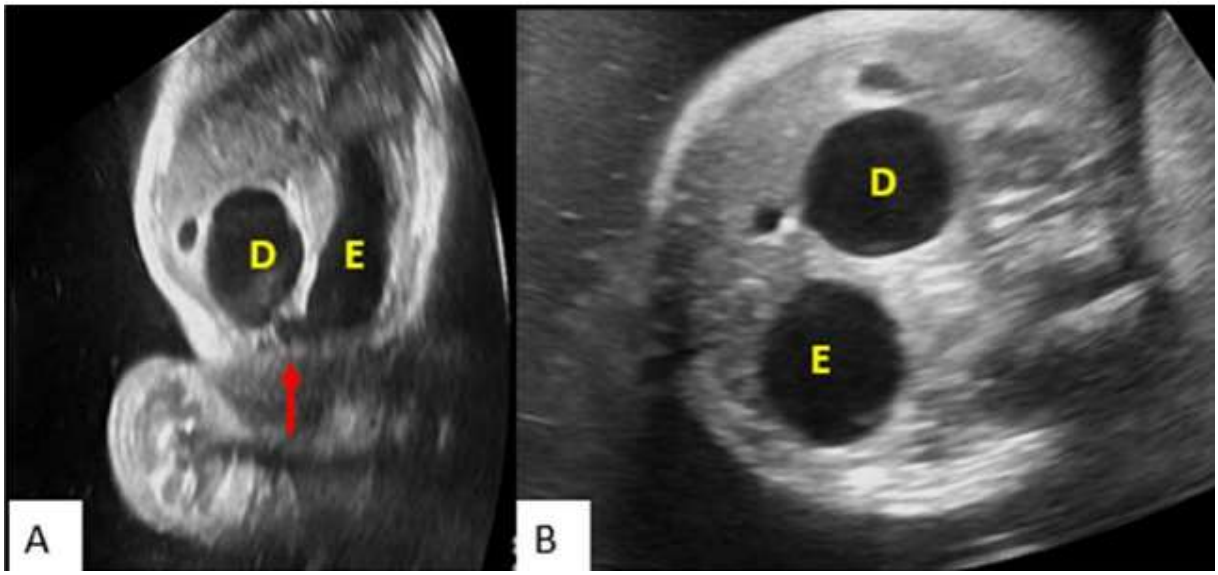

Figure 1: Duodenal atresia in a fetus at 26 weeks gestational age: axial (B) and parasagittal (A) fetal abdominal ultrasound sections showing a typical appearance of double bubble (B) with communication (A, red arrow) (E: stomach, D: duodenum) with polyhydramnios.

Fetal MRI was subsequently performed in two cases where the diagnosis of complete duodenal obstruction was uncertain:

In the first case, the diagnosis was suspected during morphological ultrasound at 25 weeks of amenorrhea due to a double-bubble appearance. However, the downstream digestive loops were visible suggesting incomplete duodenal obstruction, such as duodenal diaphragm or annular pancreas.

In the second case, obstetric ultrasound performed at 29 weeks of amenorrhea revealed distension of the digestive loops, suggesting a malformation mimicking jejunal atresia. In both cases, a fetal MRI performed at 37 weeks and 31 weeks of amenorrhea respectively, showed significant swelling of the stomach and duodenum, with a tapered appearance of the distal end of the duodenal segment and flattened downstream digestive loops. This indicated a complete duodenal obstruction (Figure 2).

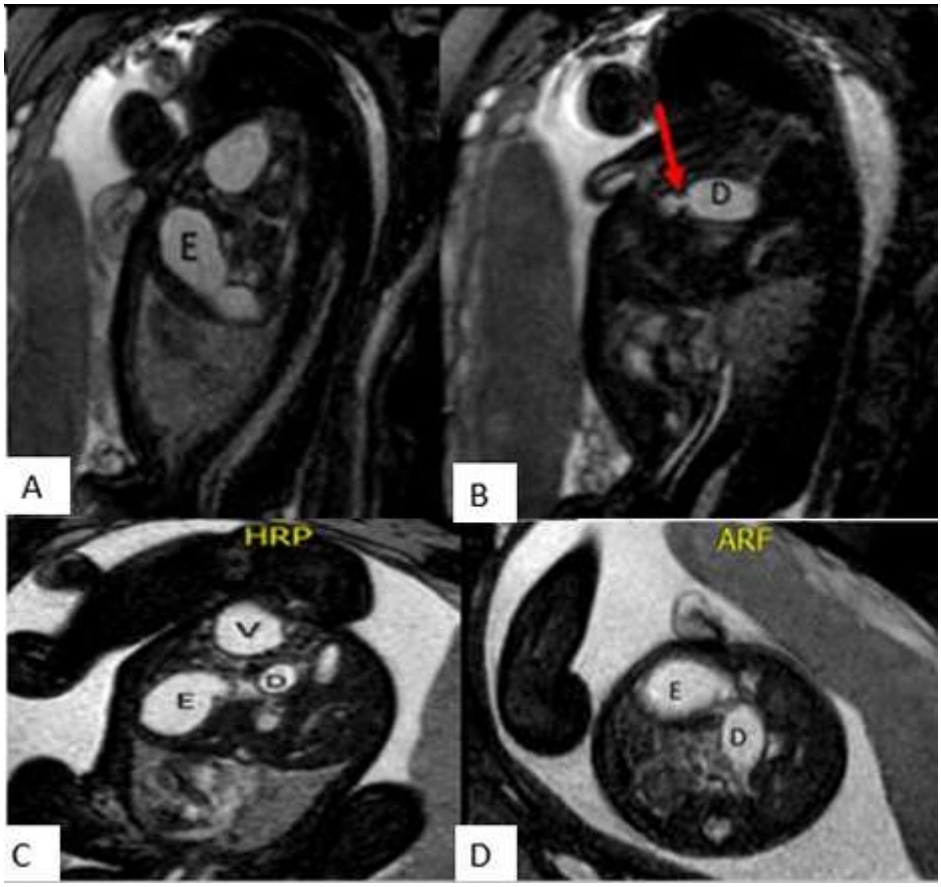

Figure 2: Fetal MRI at 31 weeks gestational age (A, B: T2 sagittal, C: T2 coronal, D: T2 axial) showing a distension of the gastro-duodenal region (C, D). A "double bubble" communication is noted (B: red arrow) with a bird's beak appearance of the duodenum without visualization of fluid signal in the downstream digestive loops. This suggests a complete duodenal obstruction (E: stomach, D: duodenum, V: bladder).

All cases in our series involved singleton pregnancies. In four cases, the patients had a vaginal birth. A caesarean section was performed in one case due to a scarred uterus. There was a male predominance with a sex ratio of 4. The average birth weight was 2850 g, ranging from 2200 g to 3750 g. Three deliveries occurred before 37 weeks of gestational age but not before 36 weeks of gestational age. In our study, duodenal atresia was associated with other malformations. Specifically, trisomy 21 was found in three cases out of the five in our series. While annular pancreas was noted in two cases, Apple Peel syndrome was found in one case. No other congenital malformations were found.

Additionally, no amniocentesis was performed. Postnatal karyotyping was requested in three cases due to facial dysmorphia observed at birth. It confirmed the trisomy 21 diagnosis.

All five newborns was operated on, 24 hours after birth. The surgery involved a duodeno-duodenostomy. The surgical exploration revealed duodenal atresia in all five cases, associated with Apple Peel syndrome in one case and annular pancreas in two cases. Postoperative evolution was uneventful in all cases, with no complications reported.

DISCUSSION

The incidence of duodenal atresia is estimated at 1 case per 6000 to 10,000 births. A male predominance with a sex ratio of 2 to 3 is commonly reported in the literature. In the majority of reported series, the average birth weight was around 2 kg. In our series, the average birth weight was 2850g, which can be explained by deliveries occurring between 36 and 37 weeks of amenorrhea.

Duodenal atresia results from a failure of recanalization. The intestinal tube fails to canalize its lumen, which normally occurs during the sixth week of gestation. This failure of recanalization may explain some forms of biliary atresia that can be associated. Based on the nature of the obstruction, duodenal atresias are classified into three types:

- Type I (92%), where an obstructing septum or membrane is formed by the mucosa without defects in the musculature or mesentery. (Fig. 3A).
- Type II, comprising only 1% of all duodenal atresias, consists of two blind ends of the duodenum connected by a short fibrous cord. (Fig.3B).
- Type III, with two blind ends of the duodenum completely disconnected (Fig.3C), occurs in 7% of cases. Eighty-five percent of duodenal atresias are located at the junction of the first and second parts of the duodenum.

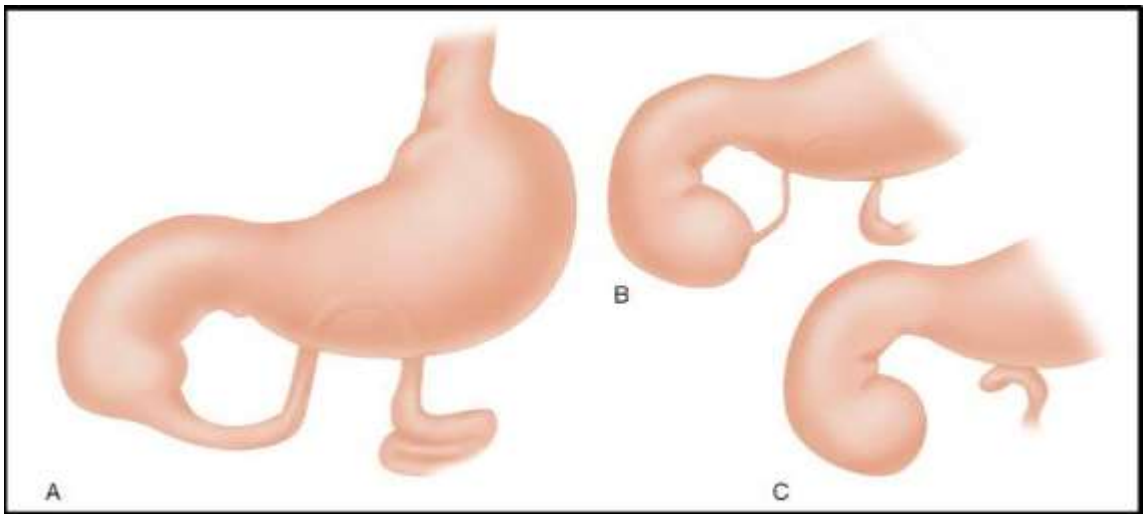

Figure 3: Different types of Duodenal atresia [16] 4A : type I, 4B : type II, 4C : type III

Prenatal diagnosis of duodenal atresia is primarily achieved through obstetric ultrasound, with a sensitivity ranging from 87% to 94% according to studies. In most series, the diagnosis could be made starting from 20 weeks of amenorrhea. However, few studies mentioned diagnosis before 20 weeks gestation. By 29 weeks of amenorrhea, the amount of amniotic fluid ingested by the fetus exceeds the absorptive capacity of the gastro-duodenal mucosa. This leads to abnormal dilation of the stomach and duodenum upstream of the obstruction, resulting in polyhydramnios. This distension forms the characteristic "double-bubble" appearance, pathognomonic for duodenal obstruction. This feature was observed in all cases in our series,

prompting suspicion of duodenal atresia. A superior transverse abdominal section shows two well-defined liquid images, corresponding to the dilated stomach and the initial portion of the duodenum. Confirming continuity between the stomach and duodenum, creating an hourglass image, is essential for ruling out other differential diagnoses such as choledochal cyst, mesenteric cyst, or digestive duplication.

These observed liquid images may contain fine moving echogenic particles (vernix or meconium). It is necessary to verify the persistence of these images during successive checks to confirm the diagnosis. In fact, transient images of gastric or even gastro-duodenal dilation can simply indicate a fetus vomiting or visualizing intestinal peristaltic movements.

Fetal MRI serves as a complementary second-line technique to ultrasound, in case of an uncertain diagnosis. Thanks to its excellent contrast resolution, MRI can precisely locate the anomaly. It is also superior in determining the nature of the obstruction as it visualizes a specific signal from the digestive tube content. This allows a more focused diagnosis regarding the duodenal obstruction and its etiology. Approximately one-third of infants with duodenal atresia also have trisomy 21. This highlights the importance of systematic fetal karyotyping in cases with a typical prenatal presentation of duodenal atresia.

In conclusion, prenatal diagnosis of digestive anomalies, particularly duodenal atresia, allows a proper care for the newborn immediately after birth. This field has seen significant development over the past two decades due to advances in fetal imaging. The complexity of this pathology arises from the high frequency of associated anomalies that significantly affect the prognosis. Hence, systematic second-trimester ultrasound screening is crucial. An analysis of downstream digestive loops usually suggests complete duodenal obstruction. However, this may sometimes require fetal MRI for better analysis and characterization of the obstruction type, along with a comprehensive assessment of associated anomalies.

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