

Isolated anorectal atresia diagnosed in the first trimester – case report

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Abstract

Anorectal atresia (ARA) is a congenital anomaly with an incidence of 1:1500 to 1:5000 in newborns. This malformation can be isolated, but is often associated with multiple congenital anomalies. Until recently, the diagnosis was often made in the third trimester of pregnancy or even after birth but, according to a review, it was established that prenatal diagnosis can be made even in the first trimester. Early diagnosis is essential for the management and planning of birth and postpartum treatment. We present the case of a fetus diagnosed with ARA at 12+0 weeks at the first-trimester ultrasound anomalies scan. The karyotype was performed and the result was normal. ARA was diagnosed and considered isolated since further detailed examinations performed in the second and third trimesters found no other anomalies. After birth, at 36 weeks, the diagnosis was confirmed as low anorectal atresia with perineal fistula. The newborn was scheduled for surgery at 3 months of life.

Keywords: anorectal atresia, prenatal diagnosis, isolated anomaly

Rezumat

Atrezia anorectală (ARA) este o anomalie congenitală cu o incidență de 1:1500 la 1:5000 de nou-născuți. Această malformație poate fi izolată, dar este adesea asociată cu multiple anomalii congenitale. Până de curând, diagnosticul se făcea adesea în al treilea trimestru de sarcină sau chiar după naștere, dar în urma unui review s-a stabilit că diagnosticul prenatal se poate face chiar din primul trimestru. Diagnosticul precoce este esențial pentru managementul și planificarea nașterii și a conduitei postnatale. Prezentăm cazul unui făt diagnosticat cu ARA la vârsta gestațională de 12 săptămâni, în cadrul morfologiei de trimestrul întâi. S-a efectuat cariotipul fetal, iar rezultatul a fost normal. ARA a fost diagnosticată și considerată izolată, deoarece în urma evaluărilor ecografice efectuate în al doilea și al treilea trimestru de sarcină nu s-au constatat alte anomalii. După naștere, care a avut loc la 36 de săptămâni de gestație, s-a stabilit diagnosticul de atrezie anorectală joasă cu fistulă perineală. Nou-născutul a fost programat pentru intervenție chirurgicală la vârsta de 3 luni de viață.

Cuvinte-cheie: atrezie anorectală, diagnostic prenatal, anomalie izolată

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Atrezia anorectală, malformație izolată diagnosticată în primul trimestru de sarcină – prezentare de caz

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Introduction

Anorectal atresia (ARA) is a congenital anomaly, with a significant incidence, of 1:1500 to 1:5000 in newborns^(1,2). This malformation can be isolated, but is often associated with multiple congenital anomalies as part of the VACTERL syndrome (vertebral, anal, cardiovascular, tracheoesophageal, renal and limb)⁽³⁻⁵⁾, complex genitourinary defects, caudal regression syndrome, or omphalocele-exstrophy-imperforate anus-spinal defects syndrome (OEIS)^(3,4,6). ARA is the result of a deficient development of the urogenital septum, a mechanism where the distal rectal pouch is prevented from reaching the perineum. As a result of this mechanism, abnormal development of the perianal muscular complex also occurs⁽⁷⁾. Until recently, the diagnosis was often made in the third trimester of pregnancy or even after birth⁽²⁾ but, after a review, it was established that prenatal diagnosis can be made even in the first trimester⁽⁸⁾. Early diagnosis is essential for the management and planning

of birth and for the postpartum course of action. We present a case of a fetus diagnosed with ARA at 12+0 weeks, within the first-trimester morphology.

Case report

A 46-year-old primipara was referred to our center for a first-trimester anomaly scan at 12+0 weeks. The pregnancy was obtained after *in vitro* fertilization (IVF) with egg donation at the fourth attempt. No pre-implantation genetic diagnosis (PGD) was performed and the egg donor was 23 years old. The patient reported no drug consumption, non-smoking, antiplatelet medication, and vaginal progesterone substitution.

The scan revealed a slighter increased NT (3.2 mm), but no other soft markers for aneuploidy. At the lower abdominal level, mostly in the left quadrant, we noticed an anechoic oblong structure with well-demarcated smooth walls, without a Doppler signal containing a pediculated hyperechogenic image that moved inside the anechoic one

(Figures 1, 2, 3 and 4). The complex image was labeled as distal bowel dilatation after checking the complete separation from the neighboring structures (urinary bladder). Fetal sex was confirmed as male, and a thorough evaluation of the fetal anatomy was performed. No other anomalies were found. Anorectal atresia diagnosis was advanced. Considering the possible association with chromosomal anomalies, free fetal DNA test was recommended and it was repeated because of the inconclusive first result. The second result indicated a high risk (6/100) for trisomy 21. Amniocentesis with MCA was performed with a normal result (arr (1-22)x2,(XY)x1). Close follow-up of the fetus was conducted monthly. At the mid-trimester anomaly scan, the anechoic image disappeared, and a hyperechoic rimmed structure of 13 mm was found instead (Figure 5). No other anomalies were obvious. Later in pregnancy, even this image vanished. Considering the initial diagnosis, we evaluated the perianal muscular complex (PAMC). The examination was difficult due to the fetal position, but at 29 weeks the PAMC aspect was abnormal and the typical “target sign” was not obvious (Figure 6). From this consideration, we counseled the couple as having a very

high likelihood of ARA, and pediatric surgery evaluation was scheduled.

At 36 weeks, the baby was delivered by caesarean section, with 2770 g and a 9 Apgar score. Postnatal examination revealed anorectal atresia with a fistula at the perineal level, allowing meconium passage (Figures 7 and 8). The newborn underwent surgical evaluation with dilatation of the fistula and was scheduled for surgery at 3 months of life.

Discussion

Anal atresia occurs as an isolated congenital anomaly, with a reported frequency of 1.11 per 10,000 births in Europe⁽¹⁾. Anal atresia is more often associated with other congenital anomalies, such as VACTERL syndrome (vertebral defects, anal atresia, tracheoesophageal fistula, esophageal atresia, renal anomalies and limb malformations) or caudal regression syndrome⁽⁹⁻¹¹⁾. Prenatal diagnosis is based on the presence of a dilated fetal bowel on ultrasound⁽¹²⁾. This diagnosis is commonly corroborated by the presence of further congenital anomalies since anal atresia rarely occurs as an isolated anomaly⁽⁹⁾.



Figure 1. The lower abdomen of the fetus presents an anechoic oblong structure with well-demarcated smooth walls and a pediculated hyperechoic image



Figure 2. The pediculated hyperechoic image moved inside the anechoic one



Figure 3. The Doppler signal is present only in pelvic vessels



Figure 4. The pediculated structure has no Doppler signal



Figure 5. Hyperechoic rimmed structure



Figure 6. The typical "target sign" was not obvious



Figure 7. The postpartum aspect of the perineal region



Figure 8. The detailed aspect of postpartum ARA

In a cohort study of patients with ultrasound consistent anal atresia, Harris et al. confirmed the diagnosis either at birth or at fetal autopsy. Among them, 11 of 12 patients (92%) with anal atresia had abnormalities associated with either VACTREL syndrome or caudal regression syndrome⁽¹³⁾. In our case, the abnormality was isolated.

It is well known that anorectal anomaly is more frequent in male fetuses⁽¹⁴⁾, and in our case the fetus was male.

The etiology of ARA appears to be multifactorial. Chromosomal aberrations have been reported in only one case of Down syndrome. Genetic mutations involved in this condition are rare⁽¹⁵⁾. In our case, it was assumed that the abnormality was related to the assisted reproductive procedure.

The diagnosis of ARA may be difficult in the first trimester when the diagnosis is based only on indirect

ultrasound signs. ARA has a low prenatal detection rate of only 16%⁽²⁾. In a 10-year study on 56 children, prenatal diagnosis of ARA by MRI versus ultrasound could not be established below 20 weeks of gestation⁽¹⁶⁾.

Fourteen cases have been reported in the literature in which the diagnosis was suspected following visualization of fetal intraabdominal or pelvic cystic structures⁽¹⁷⁻²⁰⁾. Cystic structures are represented by distended intestines filled with fluid of undetermined origin. The accumulation of fluid can be explained by the fact that the intestinal mucosa is not capable of absorption in early gestation and the rectum ends in the fundus⁽²¹⁾.

The most common form of these structures is tubular, which is determined by associated anomalies (rectovesical fistula, megacystis, complex cloacal anomalies). A tubular structure was also the defining feature in our case.

The presence of intracystic hyperechoic structures may suggest the existence of intestinal disorders due to

enterolithiasis⁽⁷⁾. In our case, the hyperechogenic structure was interpreted as an image produced by peristalsis.

The pregnancy assessment and management of a cystic abdominal tubular formation in a fetus at 12 weeks of gestation should take into consideration the differential diagnosis⁽²²⁾, including the possibility that bowel dilatation is a transient finding⁽²³⁾. Cystic transient images in pregnancy with spontaneous resolution in the second and third trimesters are reported in the literature, particularly if the right colon is affected⁽²¹⁾. In our case, the anechogenic image faded in the second trimester. It is thus recommended that serial sonographic examinations and perhaps other tests, including karyotyping, be offered to try to exclude other significant malformations. According to a review by Pleş et al., the appearance of abnormalities on later

examination was reported not only as transient findings, but also as a change in echogenicity from anechoic to hyperechoic structures in two cases⁽⁸⁾.

Conclusions

The diagnosis of anorectal atresia in the prenatal period is clinically important because prenatal diagnosis allows the planning of birth and the postnatal management. This case demonstrates the importance of maintaining a high clinical suspicion for anal atresia in a fetus with intraabdominal tubular anechogenic imaging in the first trimester and monitoring with serial ultrasound to establish a diagnosis of certainty. ■

Conflict of interests: The authors declare no conflict of interests.

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