Prenatal Diagnosis and Neonatal Management of Anorectal Malformation – A Case Report

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Abstract: Intestinal dilatation and hyperechogenic bowel, some typical ultrasonographic features of anorectal malformation, were found in the 21st week of a singleton pregnancy. These findings were associated with unilateral renal agenesis and a single umbilical artery. All prenatally diagnosed anomalies were confirmed postpartum. Development of the sonographic appearance of the anorectal malformation, prenatal and neonatal management are described and discussed.

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Introduction

Anorectal malformations comprise a wide spectrum of diseases, which involve the distal anus and rectum as well as the urinary and genital tracts. Defects range from the very minor and easily treated with excellent functional prognosis, to those that are often associated with other anomalies, and are complex, difficult to manage, and have poor functional prognosis [1]**.** Anorectal malformation (ARM) was first reported by Fourniere in 1813, cited by Walker in 1948. The overall incidence of ARM in the population is estimated to 1 in 5000 newborns [2], nevertheless prenatal diagnosis of ARM remains occasional [3] (Figure 1).

Case report

A 31 year old, secundigravida, primipara, was first in contact with our Foetal Medicine Centre at 21 weeks' pregnancy during a routine scan. Her medical history revealed delivery of a female foetus with ventricular septal defect in the year 2003. Recent pregnancy was without complications. Triple test in the 16th week was negative. Ultrasound examination described hyperechogenic filling of the rectosigmoid area (Figure 2), renal agenesis (Figure 3) and single umbilical artery (Figure 4).

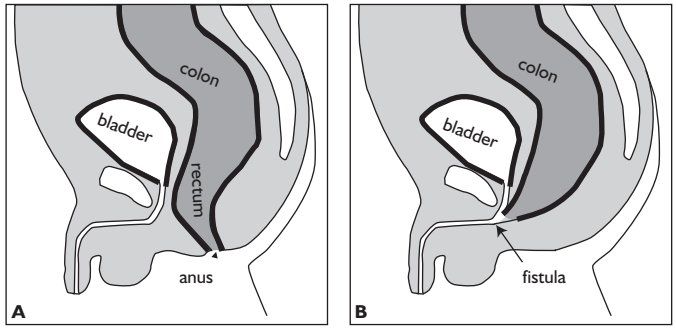

Figure 1 – Scheme of ARM: (A) normal anatomy, (B) ARM with rectourethral fistula (male).

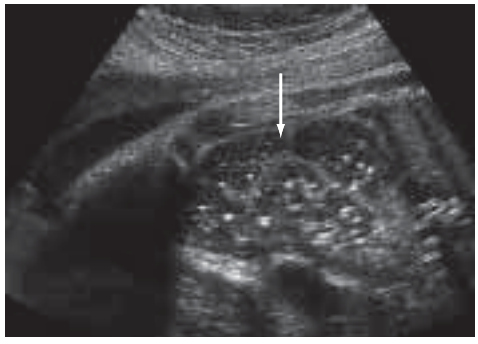

Figure 2 – Hyperechogenic filling and dilatation of the rectosigmoid area.

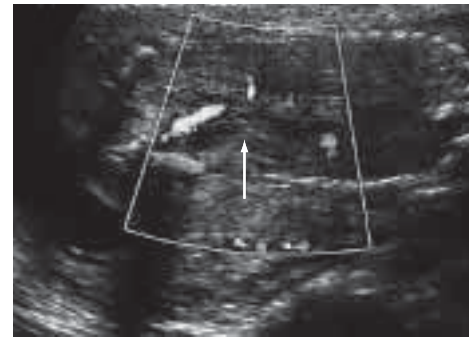

Figure 3 – Left renal artery missing due to agenesis of left kidney.

The foetal biometry was appropriate to the expected gestational age with a normal amount of amniotic fluid. Amniocentesis confirmed normal male karyotype (46, XY). We followed the foetus every 3 weeks and found the dilatation of the intestine and the hyperechogenic content more apparent (Figure 5).

These findings were suspicious for ARM. The parents were consulted about the prognosis and possible neonatal management of ARM with the neonatologist and paediatric surgeon.

Labour was induced with prostaglandins at 41+6 weeks gestation, followed by spontaneous delivery without complications. Anorectal malformation, renal agenesis and single umbilical artery were confirmed in the eutrophic male newborn (3950 g/50 cm) with Apgar score 9-10-10. Renal agenesis was compensated by hypertrophy of the right kidney. Sigmoideostomy for faecal diversion was created by the paediatric surgeon (FN Motol) on the first day of life. The postoperative course was uneventful and the newborn was dismissed on the ninth day of life. Definitive repair of the anorectal malformation was performed with posterior sagittal anorectoplasty sec. Pena (PSARP) at the age of 7 months. A rectourethoprostatic fistula (previously undiagnosed on micturition urethrography

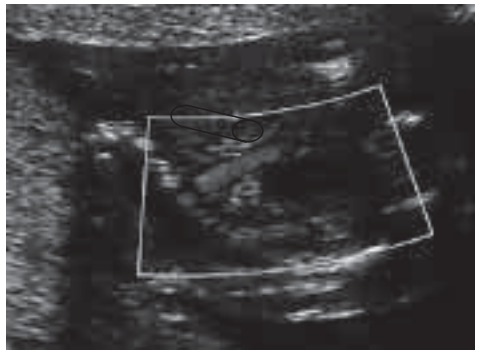

Figure 4 – Single arteria umbilicalis.

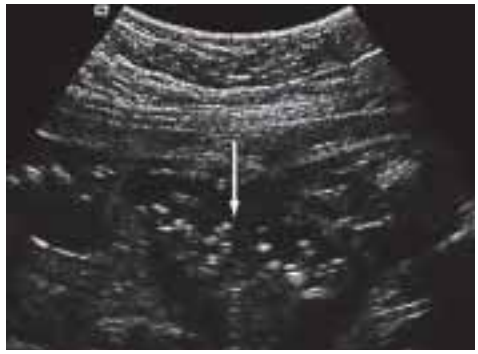

Figure 5 – Intestinal coprolites.

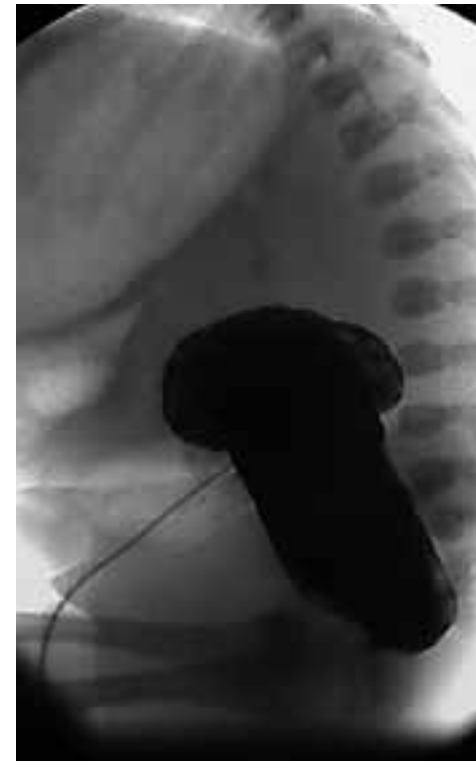

Figure 6 – Distal colostogram.

and colostogram (Figure 6) was found and closed during precise surgical dissection. The infant had an uncomplicated recovery and was discharged on the tenth postoperative day. Protective colostomy was closed two months later and the boy was doing well at the age of one year.

Discussion

Prenatal diagnosis of anorectal malformation (ARM) remains occasional, even though findings such as colon dilatation, pelvic mass, hydrocolpos, hydronephrosis, hemisacrum, hemivagina, absent radius, or absent kidney all may suggest ARM.

In a series of 69 cases published by Brantberg et al. in 2006 [2], only 15.9% of ARM cases were diagnosed prenatally. Associated anomalies were present in 85.5% (59/69), urogenital malformation in 53.6% and abnormal karyotype in 13% (mostly trisomy 21). Isolated ARM without other anomalies was found in 14.5% (10/69). Male foetuses were affected more often (82%). Harris et al. evaluated retrospectively twelve cases of prenatally undiagnosed ARM [4]. Ultrasound video recordings were reviewed, and in only 2 cases were the findings evaluated as discernible. The majority of findings (92%) were "hidden" as a complex malformation of type VATER/VACTERL (Vertebral anomaly, Anal atresia, Cardiac defect, Tracheo-Oesophageal fistula + oesophageal atresia, Renal dysplasia, Limb abnormalities) and/or caudal regression syndrome. The highest rate of detection of colonic dilatation in foetuses with ARM was in the 27th week of pregnancy. In 75% of cases ARM is diagnosed as part of VACTERL/VATER syndrome [2, 3, 5] ARM can be easily missed in the presence of other, serious and more prominent anomalies. In our case, renal agenesis and single umbilical artery were present.

Due to the shift of the first morphologic ultrasound examination to the end of the first trimester, it is possible to diagnose ARM at the end of the first trimester [6, 7].

The picture of ARM can change its appearance during the course of pregnancy. The finding of intestinal dilatation can be transient – visible only in the 16th week, but not describable on the ultrasound examinations later in pregnancy [4]. In another case from the literature the intestinal dilatation was visible in the first and then later in the third trimester [8]. In both cases supra-levator anal atresia was postnataly confirmed. In our case, there was progression of the sonographic picture of intestinal dilatation with hyperechogenic foci as the pregnancy progressed toward term.

Enterolithiasis (intestinal coprolites) was described prenatally in 48 cases in the literature [9]. From these 48 cases, there were 35 foetuses with ARM. In 66% (23 cases), communication between the intestine and urogenital tract were described; 12 cases (34%) were without communication. Shimotake et al. used infrared spectrophotometry in analysis of coprolites obtained during creation of colostomy. They confirmed that the coprolites consist of amoniumhydrogen urate.

This finding supports the theory that coprolites are formed in the intestine in presence of urine [10]. The intestinal content together with low pH of the urine participates in the formation of coprolites. In our case we prenatally described colon dilatation with hyperechogenic foci. The intestinal coprolites were confirmed on neonatal ultrasound, and rectourethoprostatic fistula was found during definitive repair of ARM.

The adequate counselling of the parents plays important part in the management of these types of malformations [11]. Regarding ARM, the sensitivity of prenatal sonographic diagnosis does not by far reach a hundred percent. As we deal more or less with suspicion, the counselling has to reflect all these uncertainties. In most of the cases, ARM is accompanied by other malformations, seriously changing the whole prognosis. Surveillance in cases of isolated ARM is about 95%. Involuntary bowel movement, soiling and constipation are main problems in functional results. Each defect has a different prognosis. Isolated ARM with low defects usually has excellent results, while patients with abnormal sacrum (more than two vertebrae missing) and flat perineum (absent muscles) suffer from faecal incontinence**.** Pena reported voluntary bowel movement in 80% of adults [12]. Skaba reported 79% full continence in children with perineal fistula [13]. Chen et al. compared postoperative results from the point of view of the anorectal functions in 108 newborns in whom PSARP sec. Pena was performed [14]. In 32% the infralevator type was found, and 68% supralevator type, which is connected with a higher rate of complications. The main complications were constipation (17.6%), ileus 1.9% and incontinence 2.7%. Rintala et al. reported long-term results in 48 patients with supralevator type of ARM from the point of stool continence. 20 patients (42%) were completely continent, in the same number continence was satisfactory, 8 patient (16%) were incontinent with the necessity of consequent surgical treatment [15].

Conclusion

Anorectal malformation is a relatively common malformation, which could be diagnosed during prenatal ultrasound examination, already at the end of the first trimester.

Anecho/hypoechogenic dilatation of the intestine is the main sonographic feature of this malformation. Hyperechogenic foci in the intestinal lumen (coprolites) do the diagnosis more reliable and communication between the intestine and urogenital system (uretrorectal fistula) is very probable. In the majority of cases ARM are associated with other malformations, like VACTERL syndrome. Isolated ARM with low defects usually has a good prognosis.

References

1. LEVITT M. A., PENA A.: Anorectal malformations. *Orphanet J. Rare Dis.* 2(33): 1–13, 2007.
2. BRANTBERG A., BLAAS H. G., HAUGEN S. E., ISAKSEN C. V.: Imperforate anus: A relatively common anomaly rarely diagnosed prenatally. *Ultrasound Obstet. Gynecol.* 28(7): 904–910, 2006.
3. GILBERT C. E., HAMILL J., METCALFE R. F., SMITH P., TEELE R. L.: Changing antenatal sonographic appearance of anorectal atresia from first to third trimesters. *J. Ultrasound Med.* 25(6): 781–784, 2006.
4. HARRIS R. D., NYBERG D. A., MACK L. A., WEINBERGER E.: Anorectal atresia: prenatal sonographic diagnosis. *Am. J. Roentgenol.* 149(2): 395–400, 1987.
5. POHL-SCHICKINGER A., HENRICH W., DEGENHARDT P., BASSIR C.: Echogenic foci in the dilated fetal colon may be associated with the presence of a rectourinary fistula. *Ultrasound Obstet. Gynecol.* 28(3): 341–344, 2006.
6. TAIPALE P., ROVAMO L., HIILESMAA V.: First trimester diagnosis of imperforate anus. *Ultrasound Obstet. Gynecol.* 25(2): 187–188, 2005.
7. LAM Y. H., SHEK T., TANG M. H.: Sonographic features of anal atresia at 12 weeks. *Ultrasound Obstet. Gynecol.* 19: 523–524, 2002.
8. KAPONIS A., PASCHOPOULOS M., PARASKEVAIDIS E., MAKRYDIMAS G.: Fetal anal atresia presenting as a bowel dilatation at 16 weeks of gestation. *Fetal Diagn. Ther.* 21(4): 383–385, 2006.
9. MANDELL J., LILLEHEI C. W., GREENE M., BENACERRAF B. R.: The prenatal diagnosis of imperforate anus with rectourinary fistula: dilated fetal colon with enterolithiasis. *J. Pediatr. Surg.* 27(1): 82–84, 1992.
10. SHIMOTAKE T., HIGUCHI K., TSUDA T., AOI S., IWAI N.: Infrared spectophotometry of intraluminal meconium calculi in a neonate with imperforate anus and rectourethral fistula. *J. Pediatr. Surg.* 41(6): 1173–1176, 2006.
11. STOLL C., ALEMBIK Y., ROTH M. P., DOTT B.: Risk factors in congenital atresias. *Ann. Genet.* 40(4): 197–204, 1997.
12. PEŇA A., LEVITT M. A.: Imperforate anus and cloacal malformations. In: Pediatric surgery. Ashcraft K. W., Holder T. M., Holcomb W. (eds), WB Saunders, Philadelphia, 2005, 496–517.
13. ŠKÁBA R.: Anorektální malformace. In: Dětská chirurgie (in Czech). Šnajdauf J., Škába R. (eds), Galén, Prague, 2005.
14. CHEN C. J.: The treatment of imperforate anus: experience with 108 patients. *J. Pediatr. Surg.* 34(11): 1728–1732, 1999.
15. RINTALA R. J., PAKARINEN M. P.: Imperforate anus: long and short term outcome. *Semin. Pediatr. Surg.* 17(2): 79–89, 2008.