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A rare case of OEIS complex -newer approach to diagnosis of exstrophy bladder by color doppler and its differentiation from simple omphalocele

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Abstract

The objective of this article is to present a new approach to diagnose and differentiate similar ventral masses by color Doppler. Two cases of ventral masses, a rare case of OEIS complex (Omphalocele-exstrophy-imperforate anus-spinal defects) with unusual presentation of exstrophy bladder and another of simple omphalocele, were studied by color Doppler for diagnosis and differentiation between the nature of similar masses. Ventral mass with absent bladder, normal kidneys, and normal amniotic fluid index raised the suspicion of exstrophy bladder. Color Doppler depicting altered intrafetal course of umbilical arteries and umbilical arteries coursing along the sides of ventral mass substantiated the diagnosis. The spatial relation between umbilical artery and aorta (which has no mention in the current literature) in sagittal view has been identified as an acute angle in a normal fetus and coined as "K angle" arbitrarily by the author. Color Doppler reveals altered (widened) "K angle" in exstrophy bladder compared to normal fetuses. Other combined anomalies pointed to the diagnosis of OEIS complex. The second case of simple omphalocele depicts normal intrafetal course of umbilical arteries and normal acute umbilical artery—aorta angle (K angle) on color Doppler. Color Doppler aids the early diagnosis of ventral defects. New method by umbilical artery-aorta angle (K angle) assessment on color Doppler helps differentiate exstrophy bladder from omphalocele.

Key words: Color Doppler; exstrophy bladder; OEIS; omphalocele

Introduction

The OEIS complex is a rare disorder comprising a combination of defects – omphalocele, exstrophy bladder/ cloaca, imperforate anus and spinal defects. Incidence is 1 in 2–4 lakh live births with only a few reported cases of prenatal diagnosis. The diagnosis of its exstrophy

bladder component is challenging because of variable ultrasound appearance. The current case of OEIS complex presents a new approach to diagnose bladder exstrophy by color Doppler. Another case of simple omphalocele is reported to differentiate it from exstrophy bladder by this newer method of umbilical artery—aorta angle (K angle) assessment besides the course of perivesical umbilical arteries.

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Case Reports

Case 1

A 24-year-old primigravida came for her first anomaly scan at 17 weeks. Personal history was unremarkable. Ultrasound revealed normal fetal head, heart, stomach and kidneys. Liquor was adequate. Urinary bladder was not visualized [Figure 1]. A cystic outpouching was seen from lower anterior abdominal wall in sagittal and transverse view suggestive of exstrophy bladder [Figures 1 and 2A].

Color Doppler revealed perivesical umbilical arteries along the cystic mass in transverse and sagittal views^[1] [Figure 2A and B], altered course of intrafetal umbilical arteries [Figure 2A and B], widening of angle between umbilical artery and aorta "K angle" in sagittal view [Figure 2B].

Normal intrafetal course of umbilical arteries and normal (acute) umbilical artery-aorta angle (K angle) seen in Figure 2C and D.

Omphalocele, imperforate anus and meningocele were also seen on ultrasound [Figure 3]. Maternal serum alphafeto protein was raised and karyotyping was normal. Based on these findings a diagnosis of OEIS complex was made.^[2]

Patient refused amniocentesis. Patient was counselled and she opted for termination of pregnancy.

The fetus revealed omphalocele, exstrophy bladder, imperforate anus and meningocele which supported our prenatal ultrasound diagnosis of OEIS complex [Figure 4]. Associated findings such as club foot, low insertion of

st

Figure 1: OEIS complex – Omphalocele (red arrow), exstrophy-bladder (yellow arrow), St (Stomach), M (Meningocele)

umbilical cord and ambiguous genitalia were also seen at termination of pregnancy [Figure 4].

Case 2

A 26-year-old primigravida came for her anomaly scan at 12 weeks. This was her first conception in 9 years of marriage. There was no history of diabetes, thyroid disease or intake of drugs. There was no history of consanguinity. Fetal head, spine, heart, stomach, kidneys and all four limbs were normal. Ductus venosus flow was normal. No evidence of tricuspid regurgitation seen. Both renal arteries were seen and liquor was adequate.

A soft tissue ventral mass (with liver, partial stomach and bowel) was seen bulging in the region of umbilicus. Color Doppler revealed normal intrafetal course of umbilical arteries in sagittal and transverse view [Figure 5], umbilical arteries running along the lower margin of the mass in transverse view [Figure 5B], umbilical vein through the mass [Figure 5] and normal (acute) umbilical artery-aorta angle (K angle) in sagittal view [Figure 5C].

Dual marker test was normal with low risk of aneuploidies. Patient was counselled and she opted to continue the pregnancy.

Targeted imaging for fetal anomalies scan at 19 weeks revealed the ventral mass with the same color Doppler findings. No other abnormality was detected. Quadruple test was normal. This is an ongoing pregnancy of 25 weeks now.

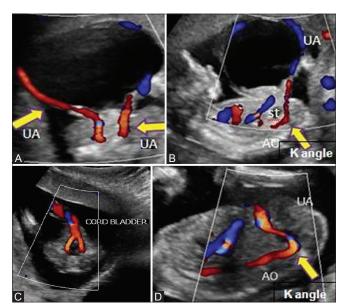


Figure 2 (A-D): (A and B) Color Doppler in exstrophy bladder showing altered course of intrafetal umbilical arteries and widening of umbilical artery (UA) – aorta (AO) angle "K angle" in transverse and sagittal views (C and D) Normal course of umbilical artery and normal umbilical artery – aorta "K angle" (arrow) in normal fetus

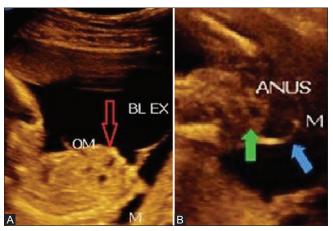


Figure 3 (A and B): OEIS complex (A) Omphalocele (red arrow) seen as an irregular mass bulging in the region of umbilicus with bowel loops extending into it;(B) Imperforate anus (green arrow) seen as oval hypoechoic area instead of signature appearance of anal sphincter (echogenic center), Spinal defect-meningocele (blue arrow)

Discussion

The OEIS complex has a sporadic occurrence with a possible role for environmental and genetic causes. The estimated incidence is 1–200,000 to 400,000 live births. [2]

The OEIS complex refers to the combined occurrence of:[1,2]

- Omphalocele
- Exstrophy bladder/cloacal exstrophy
- Imperforate anus
- Spinal anomalies.

Renal anomalies are often associated. Etiology is unclear. Teratogenic effect of diazepam and diphenylhydantoin have been proposed. Single defect in blastogenesis and mutations in homeobox genes, such as *HLXB9*, have been suggested. Higher incidence of OEIS in monozygotic twins suggests a possible genetic contribution to this defect.^[2]

Maternal serum alpha fetoprotein levels are raised and karyotying is normal.

Management

If diagnosed early termination of pregnancy is suggested. However, if diagnosed late, the neonates require multidisciplinary approach. Multiple reconstructive surgeries involve closure of spinal defect, closure of omphalocele, creation of a new bladder and construction of anal orifice with adequate sphincter mechanism.

Differential diagnosis of OEIS complex include:

- Limb body wall complex
- Pentology of Cantrell
- Amniotic band syndrome

A ventral defect accompanied with meningocele should raise the suspicion of OEIS complex and warrant a hunt for



Figure 4 (A-C): OEIS complex (A) Omphalocele (red arrow), Exstrophy-bladder (yellow arrow) (B) Imperforate anus (green arrow), Spinal defect-meningocele (blue arrow) (C) Low insertion of umbilical cord, ambiguous genitalia, club foot

more combined anomalies. Current case of OEIS complex depicts all four cardinal findings.

The case also documents associated findings which compliment the diagnosis –low insertion of umbilical cord, ambiguous genitalia and club foot.

Isolated exstrophy bladder has an incidence of 1 in 40,000 births. It is twice as common in males. Anterior abdominal wall and anterior wall of bladder are deficient and posterior wall of bladder protrudes through the defect. The bladder is turned inside out and mucosa of bladder is exposed with urine constantly trickling into amniotic fluid.

Bladder exstrophy seen as fluid filled cyst in the current case possibly takes into account the evolution of this anomaly.^[3] It is possible to formulate the hypothesis that the natural history of this congenital anomaly begins with a failure of the closure of the lower abdominal wall (due to the persistent cloacal membrane which may remain intact till 22 week of gestation) through which emerges a megacystis (bladder ectopia), which subsequently explodes and everts exposing its internal lining in the lower part of the abdominal wall [Figure 6].

Exstrophy bladder may have a variable ultrasound appearance–cystic/soft tissue ventral mass. There may not be any ventral mass at nuchal scan as the intra-abdominal pressure may not be enough to cause the posterior wall to bulge in early pregnancy. [4] Nonvisualization of urinary bladder with normal kidneys and amniotic fluid index raises the suspicion of extrophy bladder. Color Doppler

for umbilical arteries and low insertion of umbilical cord substantiates the diagnosis.

Color Doppler findings in exstrophy bladder as seen in current case:

- Altered intrafetal course of umbilical arteries
- Umbilical arteries along the sides of bulging mass
- Widening of umbilical artery–aorta angle (K angle)

Normally, umbilical artery forms an acute angle with aorta.

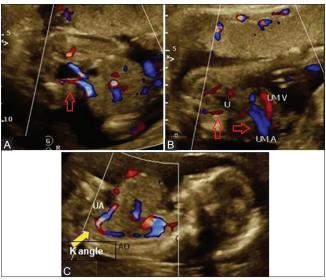


Figure 5 (A-C): Color Doppler in a case of omphalocele (A and B) showing normal intrafetal course of perivesical umbilical arteries (red arrow) in transverse view (C) Normal intrafetal course of umbilical artery with normal umbilical artery (UA) – aorta (AO) angle "K angle" (yellow arrow) in sagittal view

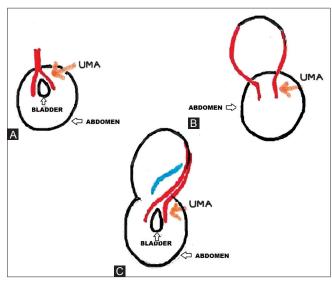


Figure 7 (A-C): Line diagram (transverse view)(A) Normal intrafetal course of umbilical arteries (arrow) in a normal fetus; (B) Altered intrafetal course of umbilical arteries (arrow) in exstrophy bladder; (C) Normal intrafetal course of umbilical arteries (arrow) in omphalocele

Low insertion of umbilical cord (umbilical cord insertion-to-genital tubercle length short for gestational age)^[4] helps in the early diagnosis of exstrophy bladder between 12 and 18 weeks when there is no bulge from lower anterior abdominal wall. In this case, low insertion of umbilical cord, was a postnatal diagnosis.

In challenging situations such as incomplete expression of OEIS complex a misdiagnosis of simple omphalocele may be made. Hence, a case of simple omphalocele is also taken up here to differentiate it from exstrophy bladder when both present as a ventral mass. Color Doppler in the current case of omphalocele^[5] revealed:

- Normal intrafetal course of umbilical arteries
- Normal acute umbilical artery–aorta angle (K angle)

In reference to the current literature, there is no mention of the spatial relation between the umbilical artery and aorta

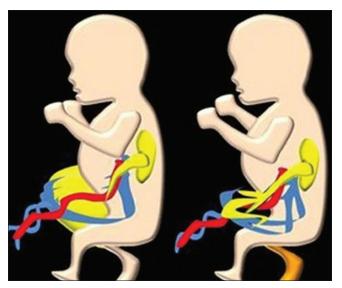


Figure 6: Schematic representation of the natural history of exstrophy bladder adapted from-http://www.sovuog.com.ve/trab_inves/case_blade_exstrophy.pdf [Last accessed on 2017 Sep 14]

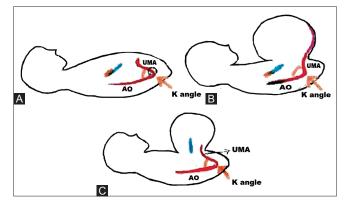


Figure 8 (A-C): Line diagram (sagittal view)(A) Normal intrafetal course of umbilical arteries and normal "K angle" in a normal fetus; (B) Altered intrafetal course of umbilical arteries and widened "K angle" in exstrophy bladder; (C) Normal intrafetal course of umbilical arteries and normal "K angle" in omphalocel

on color Doppler in sagittal view of a normal fetus or a case of ventral mass. This space has neither been identified nor been described. I identify this spatial relation as an acute angle in a normal fetus and have named it "K angle" arbitrarily. Further in description of the current case reports of ventral masses, the "K angle" is widened in exstrophy bladder due to the altered intrafetal course of umbilical arteries; of whereas it is normal (acute) in omphalocele where the intrafetal course of umbilical arteries remain normal.

Line diagrams depict course of intrafetal umbilical arteries and "K angle" in normal fetus [Figures 7A and 8A], exstrophy bladder [Figures 7B and 8B], and omphalocele [Figures 7C and 8C].

The role of umbilical artery–aorta angle (K angle) in the diagnosis of lower abdominal wall defects has not been reported in literature, which makes this article unique.

To conclude, in cases where imaging criteria for diagnosis of lower abdominal wall defects are difficult to discern, color Doppler helps confirm the diagnosis of exstrophy bladder and its differentiation from omphalocele. The observation was validated here by two case reports with similar ventral mass. This newer approach to diagnosis by color Doppler applied to fetus with non-visualization of urinary bladder allows early diagnosis of exstrophy bladder at nuchal scan (12–13 weeks) and hence ensures better planning for prenatal and postnatal treatment.

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Conflicts of interest

There are no conflicts of interest.

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