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Prenatal hydrocolpos in a male☆

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ABSTRACT

Authors report on a case of prenatal diagnosis of hydrocolpos in a genetically male subject. Postnatal sonography and endoscopy confirmed the presence of a urogenital sinus and a vaginal cavity. Despite these findings, karyotype unexpectedly revealed a 46 XY, SRY positive, male subject. Even in the presence of an established diagnosis of fetal hydrocolpos, caution should be taken before conveying information to the parents about the fetal sex. Fetal karyotyping needs to be obtained whenever possible.

1. Case

A 25-year-old primigravida underwent prenatal sonography at 31 weeks of gestation. US evaluation by radiologist expert in antenatal diagnosis revealed ambiguous genitalia and a cystic lesion measuring 50 × 60 × 60 mm in the pre-sacral region with fluid–debris level inside. Mild bilateral pyelo-caliceal dilation was associated (Fig. 1A–C). Amniotic fluid was within normal range. The parents refused amniocentesis. Due to religious reasons a cesarean section was performed at 37 weeks of gestation because of sudden decrease of the amount of amniotic fluid. Physical examination of the newborn demonstrated clitoris and labia hypertrophy, with single external perineal opening. Gonads were neither visible nor palpable. The baby was thus referred to endocrinology and pediatric urology service for further assessment. At US examination a fluid filled thick walled structure was seen posterior to the bladder. The structure changed in size during the examination, along with bladder filling and voiding (Fig. 2A–C). Because of acute urinary retention, a supra-pubic tube was positioned. Cystoscopy revealed an 18 mm long urogenital sinus with narrow vaginal opening leading to a distended vaginal cavity. Bladder cavity was normal. Karyotype analysis showed a 46 XY, SRY positive pattern. No other genetic testing was performed. 17-OH hydroxyprogesterone level was within the normal ranges for the neonatal period. HCG stimulation test led to a rise in the level of plasmatic testosterone to 159.5 ng/dl.

At two months of age followup US examination disclosed the left testis in the abdomen, close to the internal inguinal ring, slightly movable toward the inguinal channel. It was 15 mm long and showed normal parenchymal echogenicity (Fig. 3A–D). After multidisciplinary conference, the baby underwent surgical exploration which disclosed both left and right testis in an abdominal position. Testes were normal looking, though reduced in size and with complete dydimo-epididymal dissociation. Biopsy was not performed. Left testis was pulled down and a Fowler-Stephens procedure was carried out on the right side.

2. Discussion

Congenital hydrocolpos is a condition in which the obstructed vagina is distended by fluid secretions and mucus. Antenatally it usually presents as a midline pelvic cystic mass. Despite increasing expertise in fetal sonography and technological improvement of the equipments, differential diagnosis with other conditions is still

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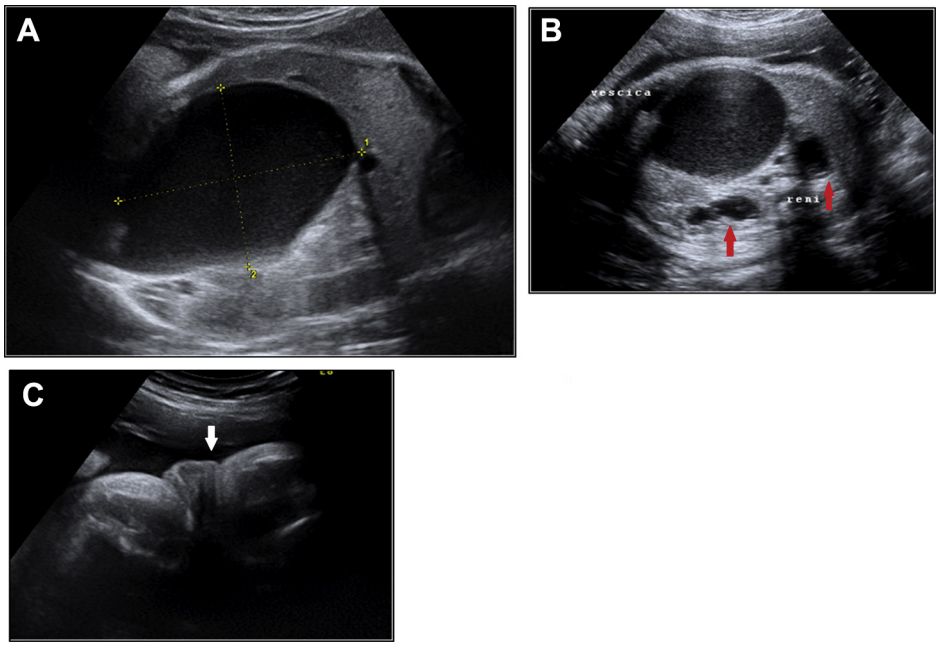

Fig. 1. Prenatal sonography at 31 weeks gestation. A. longitudinal scan of fetal abdomen: voluminous median fluid containing cavity with low-level echoes; B. transverse scan of fetal abdomen: median cavity and bilateral dilated pyelocalyceal system; C. transverse scan of fetal pelvis: ambiguous genitalia.

required. In particular, sacro-coccygeal teratomas with pelvic extension, anterior meningocele or megacystis-microcolon-intestinal hypoperistalsis syndrome need to be ruled out [1]. Pathogenetic mechanisms of hydrocolpos include vaginal or hemivaginal atresia, imperforate hymen, and more complex cases of urogenital sinus and cloaca, due to failure of urethro-vaginal separation with vaginal orifice behaving as a valve system and leading to obstruction [2].

In complex cases, ascites, due to urine reflux through the Fallopian tubes into the fetal peritoneum, may result. Polyhydramnios is usually associated. In our case the diagnosis of hydrocolpos was made antenatally on the basis of the finding of a midline cystic mass with internal echoes and mild bilateral pyelo-caliceal dilation, suggesting urinary retrostasis from ureteral compression. Ambiguous genitalia were also detected. Neither further malformations nor ascites were evident. Regardless of the underlying anomaly, the presence of hydrocolpos mostly invariably recalls female anatomy. Not surprisingly, the majority of case descriptions do not report fetal or neonatal karyotyping [3–5]. The birth of a female baby was all that was expected. In our case, postnatal findings of hypertrophy of labia and clitoris were consistent with prenatal report of ambiguous genitalia and diagnostic endoscopy revealed a short,

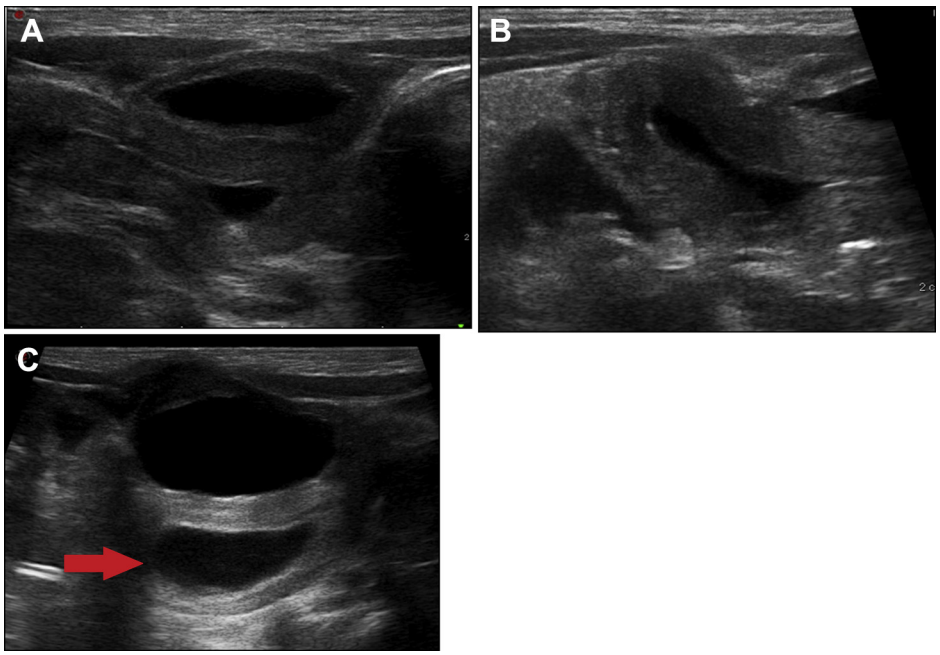

Fig. 2. Sonography at birth. A and B. longitudinal scan; C. transverse scan of the pelvis: thick walled variably distended median structure posterior to the bladder.

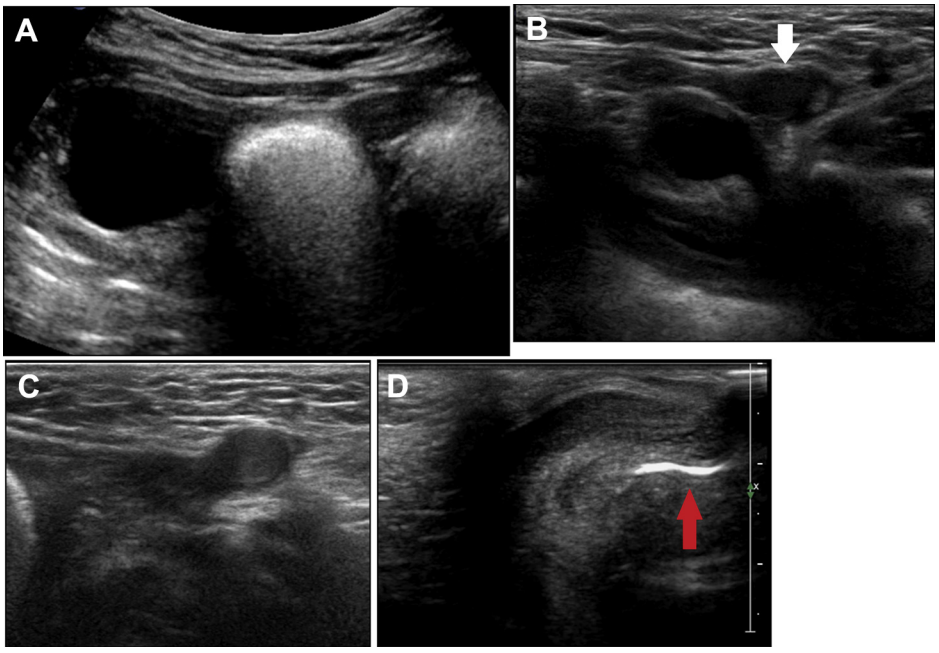

Fig. 3. Sonography at 2 months. A. transverse scan; B longitudinal scan of the pelvic region: left testis lateral to the bladder at the internal inguinal ring level; C. transverse scan of the left testis; D. longitudinal scan of the micropenis.

18 mm long, urogenital sinus with a dilated vaginal cavity with thick walls. In cases in which cytogenetic analysis was reported, normal 46, XX female karyotype was shown. Of interest, in one case severe polyhydramnios and increase of maternal ovarian steroidogenic enzymes were present [5] and in another one ascites was present [6]. It is unclear if it was the hydrocolpos per se or associated features which prompted genetic testing. At any rate, these features were absent in our case at the time of observation. Hydrometrocolpos has also been described as associated with complex syndromic conditions, characterized by different chromosome deletion; nevertheless, affected subjects always showed a 46 XX, female karyotype [7]. One case of hydrocolpos associated with ambiguous genitalia in 45, X0/46, XY mosaicism has been reported by Moore [8], and however, it is unclear from the description whether karyotype was obtained in the fetus or in the newborn. As previously mentioned, fetal karyotype was unavailable in our case, because the parents refused amniocentesis due to religious reasons. Karyotype of the newborn revealed 46 XY, SRY positive male pattern.

3. Conclusions

To our knowledge, there are no previous reports of congenital hydrocolpos in a pure 46, XY male. Actually, pseudovaginal cavities have been reported in literature in 46, XY DSD subjects with ambiguous external genitalia due to defect of 5α-reductase [9]. At present, we do not have an explanation about how the vaginal cavity, in the presence of a urogenital sinus, can become so enlarged during prenatal life. Presumably, an abnormal residue of the Mullerian ducts was distended by retained fluid (urine) collection, so resulting in fetal hydrocolpos. What message can be taken from this report? So far, prenatal diagnosis of hydrocolpos has been classically associated with female sex and this kind of information may have been consequently conveyed to the parents. On the basis of our findings, we recommend some caution during consultation in cases of antenatally diagnosed hydrocolpos. Fetal karyotyping analysis should be obtained whenever possible to better direct the diagnostic process as well as to convey more precise information to the prospective parents.

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