A rare case of hydrometrocolpos from persistent urogenital sinus in patient affected by adrenogenital syndrome

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Abstract

Persistent urogenital sinus (PUGS) is a congenital pathological condition characterized by an abnormal communication between the urethra and vagina, which has an estimated incidence of 0.6/10,000 female births. It could be the only known malformation or part of a syndrome. PUGS is commonly shown by a pelvic mass, related to a distended bladder, hydrometrocolpos which is due to an obstruction leading to the dilation of the vagina and uterus (i.e., imperforate hymen, transverse vaginal septum or atresia, and PUGS) or both. We present a case of female patient with classical congenital adrenal hyperplasia, diagnosed on the 7th day of life, with ambiguous genitalia, untreated surgically only with hormone therapy by parental decision. The patient, at the age of 5 years and 5 months, came to our observation for a pelvic ultrasound, which revealed retrovesical neoformation with anechoic content and regular walls. We performed the ultrasound examination that showed the dilation of the cervix and the vaginal canal with anechoic finely corpuscolated content in the declining portion, compatible with hydrometrocolpos from probable persistence of the urogenital sinus. The voiding cystourethrography (VCUG) confirmed the ultrasound diagnosis, with evidence of urogenital sinus. In conclusion, ultrasound is the first diagnostic tool, but need to be completed by other technical procedures, which VCUG or magnetic resonance imaging to observe the site of fusion of the urinary and genital tract.

Keywords Ultrasound (US) · Persistent urogenital sinus (PUGS) · Voiding cystourethrography (VCUG) · Magnetic resonance imaging (MRI) · Hydrometrocolpos

Sommario

La persistenza del seno urogenitale (PUGS) è una rara condizione patologica congenita, caratterizzata da un'anomala comunicazione tra uretra e vagina, la cui incidenza è di 0.6/10.000 neonate. Clinicamente, può rappresentare l'unica malformazione rilevabile o far parte di un quadro sindromico. Si presenta comunemente come una massa pelvica, associata a distensione vescicale e/o idrometrocolpo, ossia dilatazione della vagina e dell'utero causata dall'accumulo di secrezioni cervicali conseguente ad un'ostruzione (imene imperforato, setto vaginale trasverso, atresia vaginale e PGUS). Viene presentato il caso di una paziente di sesso femminile, affetta da Sindrome Adreno-Genitale (SAG) forma classica, diagnosticata al 7° giorno di vita, con genitali ambigui (ipertrofia

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clitoridea, grandi labbra scrotalizzate) non trattati chirurgicamente per decisione parentale, in terapia ormonale sostitutiva. La paziente, all'età di 5 anni e 5 mesi, giunge alla nostra osservazione per una richiesta di ecografia della regione pelvica richiesta, come consulenza, per una neoformazione retrovescicale a contenuto anecogeno e pareti regolari. L'esame ecografico ha mostrato una dilatazione del collo e del canale vaginale a contenuto anecogeno finemente corpuscolato nella porzione declive, compatibile con idrometrocolpo da verosimile persistenza del seno urogenitale. La cistouretrografia ha confermato il reperto ecografico, con evidenza del seno uro-genitale. In conclusione, l'ecografia è la metodica di prima istanza utilizzata, tuttavia necessita di ulteriori metodiche, quali la Cistouretrografia e/o la Risonanza Magnetica per rilevare il punto di comunicazione tra il tratto urinario ed il tratto genitale.

Introduction

Persistent urogenital sinus (PUGS) is a congenital pathological condition characterized by an abnormal communication between the urethra and vagina, with an estimated incidence of 0.6/10,000 female births [1].

PUGS could be the only known malformation or part of a syndrome. It may be associated with abnormalities of sexual differentiation, rectal vaginal fistula, bladder agenesis, absence of vagina and hydrometrocolpos [2, 3].

It is commonly reported as a pelvic mass, associated with bladder relaxation and/or hydrometrocolpos, which is the dilatation of the vagina and uterus caused by accumulation of cervical secretions following an obstruction (unmapped imen, transverse vaginal septum, vaginal atresia and PGUS), which in turn can result in urinary retention, compression of inferior vena cava up to peritonitis [4, 5].

Case report

Female patient with classical congenital adrenal hyperplasia, diagnosed on the 7th day of life, with ambiguous genitalia (clitoral hypertrophy, labia majora fused) treated using

hormone replacement therapy with no surgical intervention by parental decision.

The patient, at the age of 5 years and 5 months, came to our observation for a pelvic ultrasound as previously advised, after an ultrasound carried-out at another facility for the occurrence of thelarche, which found an retrovesical neoformation with anechoic content and regular walls of 44×24 mm.

Our ultrasonographic assessment was done by ultrasound device Samsung RS80A with convex probe CA1-7A and linear probe L3-12A.

Results

The US examination showed a dilation of the cervix and of the vaginal canal (Dl: 69 mm, Dap: 32 mm, Dt: 45 mm), with anechoic finely corpuscolated content in the declining portion, compatible with hydrometrocolpos from probable persistence of the urogenital sinus (Figs. 1, 2).

The voiding cystourethrography (VCUG) confirmed the ultrasound diagnosis, with evidence of urogenital sinus of about 4 cm (Fig. 3).

Fig. 1 Longitudinal sonographic image of pelvis demonstrates a dilatation of the cervix and of the vaginal canal (Dl: 69 mm, Dap: 32 mm) with anechoic finely corpuscolated content in the declining portion

Fig. 2 Transverse sonographic image of pelvis demonstrates a dilatation of the cervix and of the vaginal canal (Dap: 32 mm, Dt: 45 mm) with anechoic finely corpuscolated content in the declining portion

Fig. 3 Voiding cystourethrography (VCUG), lateral view: after the opacification of the bladder and the vagina, the contrast medium shows an oblong structure which is the vagina overcome by the uterine cavity (urogenital sinus) of about 4 cm

Discussion

PUGS is commonly shown by a pelvic mass, related to distended bladder, hydrometrocolpos which is due to an obstruction leading to the dilation of the vagina and uterus (i.e., imperforate hymen, transverse vaginal septum or

atresia, and PUGS) or both [6]. If the main symptom is hydrometrocolpos, its early diagnosis in the neonatal period is very important since hydrometrocolpos may determine compressive symptoms (dilatation of upper urinary tract with urinary retention, compression of the inferior vena cava, or elevation of the diaphragm) [7]. Hydronephrosis

can also be associated leading to the development of renal dysplasia. Sometimes, fluid may pass from hydrometrocolpos to abdominal cavity through the tubes causing peritoneal irritation [8]. If the abdomen is too enlarged by the pelvic or abdominal mass, the resulting high intra-abdominal pressure can cause elevation of the diaphragm which may result in pulmonary hypoplasia associated with the reduction of the thoracic volume [9]. Most patients are identified for the presence of ambiguous genitalia, but in some cases the genitals are normal. Ambiguous genitalia requires a combined strategy of clinical procedures, specific laboratory examinations such as hormonal and karyotyping tests combined with imaging tools to guide the therapeutic approach. The early diagnosis of PUGS in the neonatal period is important to prevent early complications in the newborn patient [10].

In conclusion, the radiological assessment of PUGS is useful for clinicians since the exact anatomical evaluation of this abnormality is a crucial factor for surgical planning. Imaging studies are essentially based on ultrasonography, VCUG and magnetic resonance imaging (MRI).

Ultrasound is the first diagnostic tool. In the suspect of PUGS, the most important evaluation consists in the whole genitourinary system study, which should be performed to identify hydronephrosis and hydrometrocolpos [11, 12].

However, even if many important findings can be observed, the fusion site of the urinary and genital tract cannot be identified on ultrasound scans. Therefore, ultrasound examination needs to be associated with other technical procedures, such as voiding cystourethrography and/or magnetic resonance imaging [13, 14].

VCUG is a low-cost examination, which allows anatomic evaluation and provides important information for treatment of PGUS. This procedure consists in the injection of contrast medium through a bladder catheter and does not require anesthesia. However, the result of this examination may depend on the experience of the radiologist.

MRI is a fail-safe when ultrasound and VCUG results are doubtful or inadequate. This method directly demonstrates the presence of malformation and the precise site of the abnormal communication between the urinary and genital tract.

However, the association of ultrasound and VCUG can correctly assess most cases of PUGS.

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Compliance with ethical standards

Conflict of interest The authors declare that they have no conflict of interest.

Ethical approval All procedures performed in studies involving human participants were in accordance with the ethical standards of the institutional and/or national research committee and with the 1964 Helsinki declaration and its later amendments or comparable ethical standards.

Human and animal rights This article does not contain any studies with animals performed by any of the authors.

Informed consent Informed consent was obtained from all individual participants included in the study.

References

1. Valentini AL, Giuliani M, Gui B, Laino M, Zecchi V, Rodolfino E, Ninivaggi V, Manzoni C, Bonomo L (2016) Persistent urogenital sinus: diagnostic imaging for clinical management. What does the radiologist need to know? *Am J Perinatol* 33:425–432
2. Kitta T, Kakizaki H, Iwami D, Tanda K (2004) Successful bladder management for a pure urogenital sinus anomaly. *Int J Urol* 11:340–342
3. Taori K, Krishnan V, Sharbidre KG, Andhare A, Kulkarni BR, Bopche S, Patil V (2010) Prenatal sonographic diagnosis of fetal persistent urogenital sinus with congenital hydrocolpos. *Ultrasound Obstet Gynecol* 36:641–643
4. Vitale V, Cigliano B, Vallone G (2013) Imperforate hymen causing congenital hydrometrocolpos. *J Ultrasound* 16:37–39
5. Sidatt M, Wedih AOSM, Boubaccar AO, Litime AOE, Feil A, Moussa AO (2013) Hydrocolpos and hydrometrocolpos in newborns. *Arch pédiatrie organe Off la Société Fr pédiatrie* 20:176–180
6. Witters I, Meylaerts L, Peeters H, Coumans A, Wirjosoekarto S, Fryns JP (2012) Fetal hydrometrocolpos, uterus didelphys with low vaginal and anal atresia: difficulties in differentiation from a complex cloacal malformation: a case report. *Genet Couns* 23:513–517
7. Subramanian S, Sharma R, Gamanagatti S, Agarwala S, Gupta P, Kumar S (2006) Antenatal MR diagnosis of urinary hydrometrocolpos due to urogenital sinus. *Pediatr Radiol* 36:1086–1089
8. Brien JC, Shariat SF, Herman MP et al (2010) Preoperative hydronephrosis, ureteroscopic biopsy grade and urinary cytology can improve prediction of advanced upper tract urothelial carcinoma. *J Urol* 184:69–73
9. Mohmand H, Goldfarb S (2011) Renal dysfunction associated with intra-abdominal hypertension and the abdominal compartment syndrome. *J Am Soc Nephrol* 22:615–621
10. Chavhan GB, Parra D, Oudjhane K, Miller SF, Babyn PS, Pippi Salle FL (2008) Imaging of ambiguous genitalia: classification and diagnostic approach. *Radiographics* 28:1891–1904
11. Celayir YC, Kurt G, Ceyhan S, Cici I (2013) Spectrum of etiologies causing hydrometrocolpos. *J Neonatal Surg* 2:2–5
12. Bhargava P, Dighe M (2009) Prenatal US diagnosis of congenital imperforate hymen. *Pediatr Radiol* 39:1014
13. Berrocal T, López-Pereira P, Arjonilla A, Gutiérrez J (2002) Anomalies of the distal ureter, bladder, and urethra in children: embryologic, radiologic, and pathologic features. *RadioGraphics* 22:1139–1164
14. Ryu J, Kim B (2001) MR imaging of the male and female urethra. *Radiographics* 21:1169–1185