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Case Report

Case Report

Unicornuate uterus with a rudimentary non-communicating cavitary horn in association with VACTERL association: a rare case report

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Abstract

Mullerian anomalies are malformations that affect the embryological development of paramesonephric ducts and are associated with multiple urogenital defects due to shared embryology, including VACTERL association, which coexists in about one-third of these patients. We report a rare case of a unicornuate noncommunicating horn uterus with a rudimentary second horn in a 16-year-old girl.

Keywords: unicornuate uterus; Müllerian duct anomalies; VACTERL; case report; embryological development

Introduction

The true incidence of Müllerian duct anomalies (MDAs) is believed to be between 0.1% and 3.8% [1]. Although its incidence may be as high as 25% in women with recurrent miscarriages and subfertility [2]. MDAs are congenital anatomic abnormalities of the female genital tract that arise from nondevelopment or nonfusion of the Müllerian ducts or failed resorption of the uterine septum [3, 4]. MDAs may lead to symptoms such as pelvic pain, dysmenorrhea, abnormal bleeding at the time of menarche, recurrent pregnancy loss, and/or premature delivery [4]. Patients with MDAs are at increased risk of having renal, skeletal, or abdominal wall abnormalities [5]; these abnormalities should also be identified and documented.

The European Society of Human Reproduction and Embryology and the European Society for Gynecological Endoscopy classification system for female genital malformations organize uterine anomalies into six main classes: Class U0 involves all cases with a normal uterus. Class U1 or dysmorphic uterus incorporates all cases with a normal uterine outline but with an abnormal shape endometrial cavity excluding septa. Septate uterus is the class U2 anomaly. Class U3 or bicorporeal uterus comprises all cases of fusion defects. Class U4 or hemi-uterus includes all cases of the unilateral formed uterus. Class U5 or aplastic uterus incorporates all cases of uterine aplasia. Class U6 is kept for still unclassified cases. Cervical and vaginal anomalies are classified in independent supplementary subclasses [6].

We report a rare case of a noncommunicating unicornuate left uterus with right rudimentary noncommunicating (functional) cavitary horn (class 2b) in a 16-year-old girl who was diagnosed with VACTERL association.

Case report

We report a rare case of a noncommunicating unicornuate left uterus with right rudimentary noncommunicating (functional) cavitary horn (class U4a) in a 16-year-old girl who was diagnosed with VACTERL association. She was presented to our center with 2 years history of cyclic severe abdominal pain with no menstrual bleeding, and she had well-developed secondary sexual characteristics. Abdominal CT scan with IV contrast imaging revealed an enlarged high-density fluid-filled left horn of left uterus in keeping with left hematometra at lateral side, there is convoluted cystic structure with high-fluid density measuring that 6.6 × 6 × 7.5 cm suggests hydrosalpinx, the left ovary cannot be seen separately from the structure, also, a 2.5 × 3 × 3 cm fluid-filled rounded structure in the right hemipelvis mostly represented noncommunicate Rt rudimentary horn, and right ovary appears normal. Vagina seen hypoplastic and duplication of inferior vena cava in which left inferior vena cava (IVC) appears smaller than right IVC. She had single left kidney normal in shape and its ureter coursing posteriorly in relation to the left IVC.

Congenital vertebral anomalies are seen in multilevel: butterfly vertebra at T9 and T11, fusion of anterior elements of L3-L4 vertebrae with loss of intervening disc space, lumbarization of the S1 with loss of lumbar lordosis, and hemivertebra of S2 vertebral body.

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She underwent vaginal exploration that showed blunt vaginal pouch without any communication with uterus; then, laparotomy for exploration founded right noncommunicating uterine horn, normal right ovary, left small isolated horn with hematometria that evacuated with needle (about 100 cc) in try to relieve symptoms, left ovarian chocolate cyst was excised, and both horns not connected with vagina.

Ten months after first presentation, when family accepted surgical management, total abdominal hysterectomy for both right rudimentary horn and small left unicornuate horn, with left salpengioopherectomy, right ovary preserved.

Discussion

In embryological development, the Müllerian (or paramesonephric) ducts are first identifiable at 5–6 weeks gestation when they begin to grow medio-caudally toward the urogenital sinus. The cranial part and the mid horizontal part of each duct develop into fimbria and fallopian tube, while the caudal part of the duct conjoins in the midlines with its contralateral side to create uterovaginal canal that will develop to the uterus, cervix, and upper third of vagina. Initially, the two Müllerian ducts are composed of solid tissue, and then internal canalization of each duct produces two canals separated by a septum that is usually regressed at around 20 weeks. The uterine endometrium is derived from the lining of the fused Müllerian ducts, whereas the endometrial stroma and myometrium are derived from adjacent mesenchyme [7–9].

Embryologically, the urinary system and the genital system are closely linked. Both develop from the intermediate mesoderm along the posterior wall of the abdominal cavity, and initially, the excretory ducts of both systems enter a common cavity, the cloaca. Three slightly overlapping kidney systems are formed in a cranial-to-caudal sequence during intrauterine life in humans: the pronephros and mesonephros which are transitory structures but critical to the development of the metanephros. The paired mesonephric ducts drain the mesonephros into the cloaca and are necessary for lower vaginal formation. The permanent kidneys develop from two sources: the ureteric bud and the metanephrogenic blastema. The ureteric bud is a diverticulum from the mesonephric duct near its entrance into the cloaca and it is the primordium of the ureter, renal pelvis, calices, and collecting tubules [7, 9].

During the fourth to seventh weeks of development, the cloaca divides into the urogenital sinus anteriorly and the anal canal posteriorly. The urogenital sinus gives rise to lower vagina, bladder (except trigon), and urethra. The trigone is derived from the caudal ends of the mesonephric ducts. The ovaries develop from primitive germ cells, mesothelium of the posterior abdominal wall, and adjacent mesenchyme, and are independent of Müllerian duct differentiation. Therefore, the ovaries are usually normal in a patient with MDAs [7–9].

The unicornuate uterus (Class U4) is a result of abnormal or failed development of one Müllerian duct. It accounts for ~2.4%–13% of all Müllerian anomalies [10] and is divided into two subclasses depending on the presence or not of a functional rudimentary cavity. Class U4a or hemi-uterus with a rudimentary (functional) cavity characterized by the presence of a communicating or noncommunicating functional contralateral horn. Class U4b or hemi-uterus without rudimentary (functional) cavity characterized either by the presence of nonfunctional contralateral uterine horn or by aplasia of the contralateral part [6].

Clinically, noncommunicating rudimentary horns with functional endometrium are the most significant subtype. The correct diagnosis of this entity has important clinical implications as they are likely to be associated with dysmenorrhea and pelvic pain from haematometra or from endometriosis due to retrograde menstruation [11]. Furthermore, pregnancies in these rudimentary horns can occur following transperitoneal migration of sperm or zygote and generally will result in a life-threatening uterine rupture [12]. Whenever diagnosed, surgical excision (preferably laparoscopic) of the functional rudimentary horn is recommended even if the horn is communicating.

The acronym *VATER* association was first described by Quan which followed by the acronym VACTERL described by Baumann [13, 14]. This association comprises a group of birth defects which tend to co-occur. These defects are vertebral anomalies, anorectal malformations, cardiovascular anomalies, tracheoesophageal fistula, esophageal atresia, renal anomalies, and limb defects [13, 14]. The presence of at least three of the aforementioned anomalies is diagnostic for VACTERL association [14]. In the presenting case, the patient has Duplication of inferior vena cava in which left IVC appears smaller than right IVC. She had single left kidney normal in shape, and its ureter coursing posteriorly in relation to the left IVC.

Congenital vertebral anomalies are seen in multilevel.

To our knowledge, this is one of very rare documented cases that describe a VACTERL association with the coexistence of unicornuate uterus with a noncommunicating rudimentary functional horn. Nunes *et al.* [5] reported the first case in a 28-year-old nullipara female known to have a missing vertebra, imperforate anus, anovaginal fistula, right renal agenesis, and an extra digit on one hand. She had a regular cycle since menarche at age 15. She presented with a 6-month history of progressive constipation and sciatica followed by acute urinary retention which was revealed to be due to hematometra/hematosalpinx of a noncommunicating right uterine horn and fallopian tube [5]. Jessel *et al.* [15] described a case of distal vaginal agenesis and right unicornuate uterus with left nonobstructed rudimentary horn in a 14-year-old girl with a known history of VACTERL association and repaired imperforate anus.

Heinonen reported no VACTERL association in his series with 42 cases of unicornuate uteri [16]. Heinonen reported the presence of Kidney abnormalities, bony anomalies, auditory defect, Hirschsprung's disease, an absence of the gallbladder, and annular pancreas [16].

To finish, the development of the urinary system is closely related to the genital tract and anomalies of these organs are often coexisting. Less commonly, MDAs may coexist with developmental anomalies of the distal gastrointestinal tract particularly cloaca anomaly [17]. A retrospective study of female patients treated for imperforate anus revealed that primary vaginal anomalies occurred in 22 of 72 (32%) patients assessed, and uterine anomalies occurred in 18 of 51 (35%) patients assessed. A bicornuate uterus and uterus didelphys were the most common abnormalities [18]. In our case, and the case described by Nunes *et al.*, the patients had a unicornuate uterus with rudimentary horn and the diagnosis of MDAs was not recognized until they develop symptoms later. Hence, it is important that an experienced gynecologist is involved at an early stage in the care of female infants with anorectal malformations.

We report yet a rare case of VACTERL association and a unicornuate uterus with noncommunicating functional rudimentary horn, in hope of expanding the knowledge of a rare occurrence. This case also highlights the importance of considering the diagnosis of MDAs in patients with a history of other anomalies and/or a history of early-age secondary dysmenorrhea.

Conclusion

This case also highlights the importance of considering the diagnosis of MDAs in patients with a history of other anomalies.

Conflict of interest statement

There is no conflict of interest to declare.

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Consent

Written informed consent was obtained from the patient family for reporting this case and its associated images. The consent is available for review on request.

References

1. Gidwani G, Falcone T. *Congenital Malformations of the Female Genital Tract: Diagnosis and Management*. Philadelphia: Lippincott Williams and Wilkins, 1999.
2. Saravelos SH, Cocksedge KA, Li T-C. Prevalence and diagnosis of congenital uterine anomalies in women with reproductive failure: a critical appraisal. *Hum Reprod Update* 2008;**14**:415–29. https://doi.org/10.1093/humupd/dmn018.
3. Nahum GG. Uterine anomalies. How common are they, and what is their distribution among subtypes? *J Reprod Med* 1998;**43**:877–87.
4. Stampe SS. Estimated prevalence of müllerian anomalies. *Acta Obstet Gynecol Scand* 1988;**67**:441–5. https://doi.org/10.3109/00016348809004257.
5. Nunes N, Karandikar S, Cooper S, *et al.* VATER/VACTERL syndrome (vertebra/anus/cardiac/trachea/esophogus/radius/renal/limb anomalies) with a noncommunicating functioning uterine horn and a unicornuate uterus: a case report. *Fertil Steril* 2009;**91**:1957. e1911–2. https://doi.org/10.1016/j.fertnstert.2008.12.140.
6. Grimbizis GF, Gordts S, Di Spiezio SA, *et al.* The ESHRE/ESGE consensus on the classification of female genital tract congenital anomalies. *Hum Reprod* 2013;**28**:2032–44. https://doi.org/10.1093/humrep/det098.
7. Sadler TW. *Langman's Medical Embryology*. Philadelphia: Lippincott Williams & Wilkins, 2011.
8. Robboy SJ, Taguchi O, Cunha GR. Normal development of the human female reproductive tract and alterations resulting from experimental exposure to diethylstilbestrol. *Hum Pathol* 1982;**13**:190–8. https://doi.org/10.1016/S0046-8177(82)80177-9.
9. Moore KL, Persaud TVN, Torchia MG. *Before We Are Born: Essentials of Embryology and Birth Defects*. Philadelphia: Elsevier Health Sciences, 2015.
10. Grimbizis GF, Camus M, Tarlatzis BC, *et al.* Clinical implications of uterine malformations and hysteroscopic treatment results. *Hum Reprod Update* 2001;**7**:161–74. https://doi.org/10.1093/humupd/7.2.161.
11. Markham SM, Waterhouse TB. Structural anomalies of the reproductive tract. *Curr Opin Obstet Gynecol* 1992;**4**:867–73. https://doi.org/10.1097/00001703-199212000-00015.
12. Kadir R, Hart J, Nagele F, *et al.* Laparoscopic excision of a noncommunicating rudimentary uterine horn. *BJOG* 1996;**103**:371–2. https://doi.org/10.1111/j.1471-0528.1996.tb09744.x.
13. Quan L, Smith DW. The VATER association:-vertebral defects,-anal atresia,-T-Efistula with esophageal atresia,-radial and-renal dysplasia: a spectrum of associated defects. *J Pediatr* 1973;**82**:104–7. https://doi.org/10.1016/S0022-3476(73)80024-1.
14. Baumann W, Greinacher I, Emmrich P, Spranger J. Vater or vacterl syndrome (author's transl). *Klinische Padiatrie* 1976;**188**:328–37.
15. Jessel RH, Laufer MR. Management of lower vaginal agenesis in a patient with unicornuate uterus. *J Pediatr Adolesc Gynecol* 2013;**26**:e21–3. https://doi.org/10.1016/j.jpag.2012.10.007.
16. Heinonen PK. Unicornuate uterus and rudimentary horn. *Fertil Steril* 1997;**68**:224–30. https://doi.org/10.1016/S0015-0282(97)81506-3.
17. Pradhan S, Vilanova-Sanchez A, McCracken KA, *et al.* The Mullerian black box: predicting and defining Mullerian anatomy in patients with cloacal abnormalities and the need for longitudinal assessment. *J Pediatr Surg* 2018;**53**:2164–9. https://doi.org/10.1016/j.jpedsurg.2018.05.009.
18. Hall R, Fleming S, Gysler M, McLorie G. The genital tract in female children with imperforate anus. *Am J Obstet Gynecol* 1985;**151**:169–71. https://doi.org/10.1016/0002-9378(85)90004-3.