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Case Report

Uterine rudimentary horn and ovarian endometriosis with a special emphasis on VACTERL association: a report of two cases

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ABSTRACT

Mullerian duct anomalies are a wide and complex spectrum of congenital abnormalities that are often associated with renal as well as axial skeletal anomalies or can be a part of multiple malformation syndrome and may cause numerous other gynecological complications. We herein describe two young girls who presented with a common complaint of abdominal pain and had normal secondary sexual characteristics.The first case had a history of imperforate anus, and urinary incontinence due to renal anomalies including ectopic kidney, renal agenesis, duplex ureter along with sacral agenesis which were confirmed on imaging. VACTERL (V-vertebral anomalies, A-anorectal malformations, C-cardiovascular anomalies, T- tracheoesophageal fistula, E-esophageal atresia, R-renal anomalies, and L-limb defects) association was considered. To the best of our knowledge, this is the fourth such case with concomitant VACTERL association. Surgical resections were performed in both cases, after clinical-radiological correlation. Histopathological examination of specimens revealed a unicornuate uterus with a rudimentary horn and ovarian endometriosis. In conclusion, early identification and prompt intervention through a multidisciplinary approach are paramount for managing such anomalies as they can have many detrimental implications.

Keywords: Mullerian duct anomalies, Unicornuate uterus, Rudimentary uterine horn, Endometriosis, VACTERL

INTRODUCTION

The mullerian ducts, also called paramesonephric ducts, are of mesodermal origin. These ducts give rise to the upper third of the vagina, cervix, uterus, and fallopian tubes. Any deviation from normal development can result in mullerian anomalies. The reported incidence of mullerian duct anomalies (MDAs) in routine clinical practice has been documented as 0.1–0.5%.[1] These are usually asymptomatic in presentation and are classified into seven classes based on the type of fusion defect. The diagnosis is mainly based on radiology, and ultrasonography (USG) is the initial diagnostic modality. However, magnetic resonance imaging (MRI) is considered the gold standard. Some of these defects can be treated surgically. It is worth noting that the septate uterus is the most common type of uterine anomaly, accounting for nearly half of all cases. On the other hand, the unicornuate uterus is a congenital uterine malformation that makes up almost 2.5% to 13% of all anomalies.[2] Nevertheless, it is important to be aware that a unicornuate uterus with a rudimentary horn, ovarian endometriosis, and congenital renal agenesis are rare combinations that can be easily misdiagnosed due to the lack of typical clinical manifestations. This report highlights a VACTREL association with uterine rudimentary horn and ovarian endometriosis, providing valuable insights into this rare amalgamation.

CASE REPORTS

Case 1

A 12-year-old nulliparous adolescent girl presented with a chief complaint of dribbling urine from birth and abdominal pain associated with menstruation. Her physical examination was within normal limits. Local examination revealed separate urethral and vaginal openings. She had a history of anorectal malformation (imperforate anus) in the past since birth, for which posterior sagittal anorectoplasty was done 10 years ago. She attained her menarche at the age of 11 years. Radiological investigations were done to delineate the cause. Renal scintigraphy revealed perfusion with no cortical defect in bilateral kidneys. However, the right kidney showed decreased perfusion. Magnetic resonance urography showed an ectopic low-lying right kidney with a bilateral duplicated pelvicalyceal system with a bifid proximal right ureter having ectopic insertion into the right side of the vagina. This arose the possibility of a urethrovaginal fistula. MRI of the pelvis confirmed the presence of a unicornuate uterus with an obstructed non-communicating functional horn on the right side with right-sided hematometra, hemosalpinx, and right ovary showing endometrioma. MRI of the whole spine revealed lumbosacral transition vertebrae with lumbarisation of the S1 vertebra and partial sacral agenesis (Type 2) in the form of non-visualisation of S4-S5 (Figure 1).

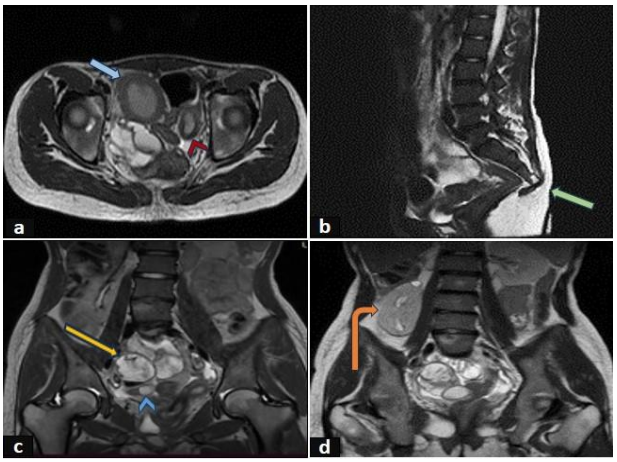

Figure 1: T2-weighted MRI scans (a) Axial plane: rudimentary horn (arrow), main uterine cavity (arrowhead); (b) Saggital plane: showing partial agenesis of the sacrum and agenesis of the coccyx (arrow); (c) Coronal plane: endometrioma (arrow) with dilated fallopian tube and hydrosalpinx (arrowhead); (d) Coronal plane: ectopic position of right kidney with anteriorly located renal pelvis i.e. malrotation (curved arrow).

The patient underwent laparotomy in pediatric surgery operation theatre for the right horn of the uterus excision with hematometra drainage. Intraoperative findings were consistent with the radiology. Bilateral ureteric reimplantation was also done. Excised specimens were sent for histopathological correlation. Gross examination revealed a grey-brown mass measuring 6×5×3 cm with an attached tubal segment measuring 4cm long (Figure 2). On the cut section, an endometrial cavity was identified which showed the presence of blood. The attached ovary appeared bulky and measured 2.5×1×1 cm in size. On cutting, chocolaty fluid oozed out. The ureteric end received in a separate container measured 0.4×0.2×0.1 cm. Microscopy exhibited a non-functional endometrium, unremarkable myometrium, and an ovary showing the presence of endometrial-type glands, and stroma along with areas of hemorrhage as well as hemosiderin-laden macrophages (Figure 3). Bilateral fallopian tubes were unremarkable and showed the presence of lumen. The patient was discharged on postoperative day 7 with stable vitals.

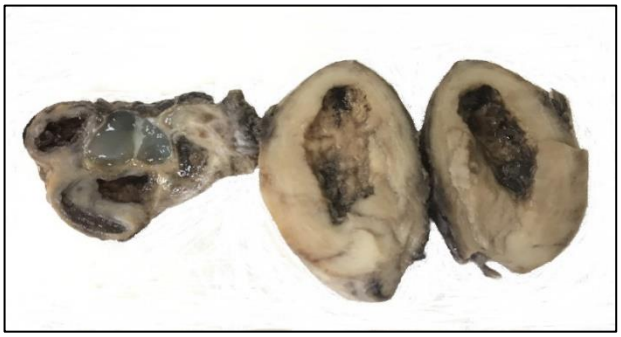

Figure 2: Rudimentary uterine horn with an ovarian cyst.

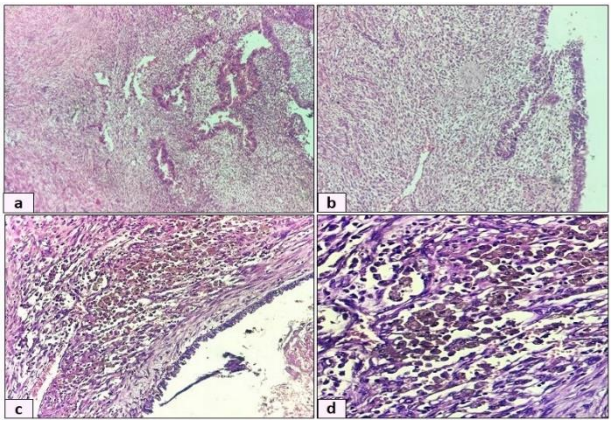

Figure 3: Photomicrographs showing (a) and (b) Non-functional endometrial glands and epithelial lining (H and E, x10); (c) and (d) Hemosiderin-laden macrophages along with endometrial epithelium, suggestive of Endometriosis (H and E, x20).

Case 2

A 14-year-old young nulliparous female presented with a history of abdominal pain for 5 years. She attained menarche at the age of 12 years. Her physical examination revealed a soft, well-defined mass of 14 weeks in size in

the hypogastric region arising from the pelvis. It was immobile, well-defined, and tender. Outside USG showed a right ovarian cystic lesion. However, on getting the radiological investigations done in our hospital, it was revealed that there was also a presence of a rudimentary horn with hematometra which was missed initially. The initial USG pelvis revealed a normal-sized uterus. Bilateral ovaries were normal in size and echotexture. The right ovary showed a cystic lesion of size 8.7×8.4 cm. The left ovary measured 3.8×2.3 cm with no free fluid. These features were suggestive of the right ovarian chocolate cyst. However, her follow-up USG showed Didelphys/bicornuate uterus. Endometrial thickness measured 4 mm. 2nd cavity measured 5.8×3.8 cm and no continuity were seen with the rest of the uterus; retained fluid was present. MRI pelvis showed a left unicornuate uterus with a non-communicating right uterine horn having functioning endometrium and the presence of hematometra. Right, endometrioma with right-sided hematosalpinx was also noted. It was classified under class U4a of The European Society of Human Reproduction and Embryology (ESHRE) and the European Society for Gynaecological Endoscopy (ESGE) classification system for female genital malformations. MRI findings validated the USG findings. Right rudimentary horn excision with right salpingectomy and right ovarian endometriotic cystectomy was done. The specimen was sent for histopathological examination. Gross examination revealed a specimen of the unicornuate non-communicating horn of the uterus with an ovarian cyst and fallopian tube altogether measuring 10×8×4 cm. The uterine horn alone measured 5×3×2.5 cm. The uterus on the cut section showed the presence of a cavity filled with blood. Endometrial and myometrial thickness measured 0.1 cm and 1.5 cm respectively. Another cyst was identified at the junction of this cavity and the ovarian cyst contained chocolaty fluid. The ovarian cyst flap measured 10×1 cm. Cyst wall showed congestion and focal thickened areas measuring 5×2 cm. However, no papillary excrescences were identified on gross (Figure 4).

Figure 4: Unicornuate non-communicating horn of the uterus with an endometriotic cyst.

The fallopian tube measured 2 cm in length. It was grossly unremarkable and showed the presence of a lumen on cut. Two paratubal cysts measuring 0.1 to 0.5 cm in diameter were also identified. On cutting, serous fluid oozed out of them. On microscopy, the endometrium was non-functional with unremarkable myometrium and the ovary showed features of endometriosis. Bilateral fallopian tubes were unremarkable with a lumen and showed the presence of paratubal cysts. The patient was discharged on postoperative day 10 with an uneventful postoperative period.

DISCUSSION

Unicornuate non-communicating, cavitary horn is a type 2 mullerian anomaly. It occurs due to partial or complete development arrest in one of the paramesonephric ducts. It is usually accompanied by dysmenorrhoea, hematometra, and endometriosis. Since it may obstruct and present with abdominal pain, it requires surgical intervention. It has been well-established in the literature that MDAs often occur in close conjunction with renal anomalies in approximately 29% of the cases, typically in cases of a unicornuate uterus.[3] The association is attributed to the embryology of the two systems. The urinary and reproductive systems both originate from the intermediate mesoderm along the posterior wall abdominal cavity. Initially, the excretory ducts of both systems enter the cloaca, a common canal. The most common renal defect is agenesis, while ectopic kidney, horseshoe kidney, and duplex system are other associated defects.

Hydrocolpos is vaginal distension caused by fluid accumulation due to vaginal obstruction, often by imperforate hymen or transverse vaginal septum. This condition can also be linked with anomalies such as imperforate anus or persistent urogenital sinus and can be associated with genetic syndromes such as Bardet–Biedl, McKusick–Kaufman, and Pallister–Hall.[4] Our case was also associated with a history of imperforate anus that had been corrected with surgery in the past. Imperforate hymen is a rare congenital defect of the female reproductive system, characterized by the absence of an opening in the hymen that covers the vaginal opening. It affects about 1 in 2,000 females. In females, the hymen is perforated during embryonic development to create a connection between the vestibule and the vaginal canal. If this process does not occur, it results in an imperforate hymen.[5] The presence of vaginal distension during birth, caused by thick mucus, can be a helpful indicator for diagnosing this condition at an early stage. However, even though it is easily diagnosed, it often remains unrecognized until puberty, when females begin to experience symptoms such as abdominal pain and menstrual detritus. Imperforate hymen can cause hematocolpos, which results in amenorrhea and cyclic pelvic pain. It may be associated with MDAs.[6] Heinonen et al reported multiple congenital abnormalities including Hirschsprung's disease, urinary system abnormalities, auditory defects, bony anomalies, an absent gallbladder, and an annular pancreas.[7] However,

some studies propose a non-association between the two conditions as well as renal anomalies.[8] According to a study conducted by Rawan AO et al imperforate hymen can be significantly associated with MDA as a component of VACTERL syndrome. VACTERL association is a non-random grouping of birth defects affecting multiple areas of the body, including vertebral anomalies (V), anorectal malformations (A), cardiovascular anomalies (C), tracheoesophageal fistula (T), esophageal atresia (E), renal anomalies (R), and limb defects (L). The presence of at least two or three component defects should be used to define VACTERL syndrome.[9] Our first case had a VACTERL association. After reviewing the pertinent literature, we have identified this as the fourth reported case of the VACTERL association worldwide. The three previously documented cases were reported by Nunes et al, Obeidat R et al, and Jose D. Roman.[9-11]

Endometriosis is characterized by the presence of endometrial tissue outside the uterus. Endometriotic cysts (endometriomas) most commonly involve the ovaries, where they can partially or almost completely replace the normal tissue. Repeated hemorrhage can cause the cyst to turn into a chocolate cyst. The cyst's contents usually contain semi-fluid or inspissated, chocolate-colored material.[12] The exact cause of endometriosis is still unclear, but the most widely accepted theory suggests that it is due to retrograde menstruation. During this process, menstrual blood containing endometrial cells flows back into the pelvic cavity through the fallopian tubes instead of leaving the body. These cells can then stick to the surfaces and walls of the pelvic organs, where they grow and thicken during each menstrual cycle, ultimately leading to bleeding. Other proposed mechanisms include metaplastic transformation of peritoneal cells, induction theory (combination of retrograde and coelomic metaplasia theories), and embryonic cell changes (under the influence of estrogen).[13-15] The presentation of a unicornuate uterus with a rudimentary uterine horn and ovarian endometriosis is highly variable. Thus, it is difficult to catch up with the defect till the consequences manifest.[16]

In the first case, though the patient had a history of dribbling/urinary incontinence from birth, due to low socio-economic status, the family could not avail of timely treatment. They got more concerned when she started having complaints of severe dysmenorrhea from the time she attained menarche. The second case caused discomfort to the patient in the form of dysmenorrhea and thus was managed with an early intervention. Treatment of MDAs is divided into- medical and surgical. Medical treatment includes pain relieving agents for dysmenorrhoea. Non-surgical vaginal lengthening procedures can be done for agenesis/atresia. Surgery is curative. Thus, a multidisciplinary approach including clinical history and examination along with radiological and pathological correlation helps in quenching the exact diagnosis and appropriate as well as timely management of such patients.

CONCLUSION

The two cases of adolescent girls with uterine rudimentary horn and ovarian endometriosis, emphasizing a rare VACTREL association highlight the importance of identifying these MDAs well in time, for early diagnosis, histopathological confirmation, and patient management. The presence of MDAs with other anomalies in young patients can be challenging. Thus, a multidisciplinary approach should be used to avoid complications like infertility, abortions, rupture causing hemoperitoneum, and hemorrhagic shock.

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