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Case Report

Left ovarian torsion in type 2 Mayer-Rokitansky-Küster-Hauser syndrome: a rare case report

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ABSTRACT

Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome is a rare congenital anomaly of Müllerian duct development, occurring in approximately 1 in 4,500-5,000 female births, and is the second most common cause of primary amenorrhea after gonadal dysgenesis. Ovarian torsion complicating MRKH syndrome is exceptionally rare, with only a small number of cases described in the literature to date. We report the case of a 20-year-old unmarried woman with primary amenorrhea who presented with acute lower abdominal pain of three days' duration, nausea, vomiting, and a palpable abdominal mass. She was initially managed as suspected adnexal torsion or a degenerating uterine fibroid or pregnancy related complication. Ultrasonography and magnetic resonance imaging (MRI) demonstrated absence of the uterus and cervix, vaginal agenesis, right renal agenesis, and a large left ovarian cyst with a twisted vascular pedicle showing the "whirlpool sign", consistent with Type 2 MRKH syndrome complicated by left ovarian torsion; a preoperative chest radiograph was unremarkable. Emergency exploratory laparotomy confirmed an 8×7 cm left ovarian cyst with two complete turns of torsion of the pedicle, a fibrous band, and a small rudimentary uterine remnant. Detorsion, cystectomy, ovarian reconstruction with ovarian drilling were performed while preserving of viable ovarian tissue. Histopathology confirmed a benign (serous) simple ovarian cyst, and recovery was uneventful. Ovarian torsion is a rare but important complication of MRKH syndrome that can be easily overlooked because of the syndrome's atypical pelvic anatomy. A high index of suspicion, supported by ultrasonography and MRI, together with prompt fertility- and function-preserving surgery, remains key to a favourable outcome. This case adds to the limited published literature on this association.

Keywords: Mayer-Rokitansky-Küster-Hauser syndrome, Müllerian agenesis, Ovarian torsion, Primary amenorrhea, Renal agenesis, Ovarian cystectomy

INTRODUCTION

Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome, also known as Müllerian agenesis or congenital absence of the uterus and vagina, results from failure of Müllerian duct development in phenotypically normal 46,XX females with normal secondary sexual characteristics and normal ovarian function.¹ It is the second most common cause of primary amenorrhea after gonadal dysgenesis, with a reported incidence of approximately 1 in 4,500-5,000 female births.^{1,2} The condition is classified into an isolated form, Type 1, in which only the uterus and upper vagina are affected and which accounts for roughly 44% of cases, and Type 2, in which extragenital anomalies-

most commonly renal, followed by skeletal, cardiac and auditory defects-coexist (also termed the MURCS association), accounting for the remaining, and marginally larger, share of cases (approximately 56%).^{2,3} Type 2 disease is therefore, if anything, slightly more frequent among MRKH patients than the isolated form, even though MRKH as a whole remains a rare condition.

Because the ovaries develop independently of the Müllerian ducts, they are usually structurally and functionally normal in MRKH syndrome. Ovarian anomalies of any kind-including hypoplasia, malposition and cysts-have nonetheless been reported in a minority of cases, while frank ovarian torsion remains an

exceptionally rare complication, described only sporadically in the literature.^{4,5} Absent or abnormal uterine and ligamentous support, together with elongated or redundant adnexal pedicles, has been proposed as a predisposing mechanism.^{4,6} Diagnosis can be challenging, since the absence of a uterus alters the expected pelvic anatomy and may lead to misdiagnosis as a degenerating fibroid or a nonspecific adnexal mass.

We report a case of Type 2 MRKH syndrome-with associated vaginal agenesis and right renal agenesis-complicated by left ovarian torsion with a large ovarian cyst in a young woman presenting with acute abdomen and primary amenorrhea. Given how few such cases have been documented, we present this report to add to the existing literature on this rare association and to reinforce a diagnostic and management approach for similar future presentations.

CASE REPORT

Presentation

A 20-year-old unmarried woman presented with acute-onset lower abdominal pain of three days' duration, accompanied by nausea, vomiting, and progressive abdominal distension. She reported primary amenorrhea, with no history of cyclical abdominal pain. She had attained normal pubertal development, and her past medical and surgical history was unremarkable. She was initially admitted with a provisional diagnosis of adnexal torsion or degenerating uterine fibroid.

Examination

General examination revealed a normally built woman with well-developed secondary sexual characteristics; breast and pubic hair development corresponded to Tanner stage V. Abdominal examination demonstrated a tender, firm, mobile abdominopelvic mass extending up to the level of the umbilicus, with localized guarding but no signs of generalized peritonitis. External genitalia were normally developed.

Investigations

Laboratory investigations showed a total leukocyte count of 8,740 cells/ μ L with 62.4% neutrophils, and a C-reactive protein of 5.1 mg/L. Serum FSH was 4.22 mIU/mL and LH was mildly elevated at 22.26 mIU/mL; screening for HIV, hepatitis C and hepatitis B was non-reactive. Tumor markers, thyroid profile and other sex hormones were within normal limits

Transabdominal ultrasonography performed prior to referral showed that the uterus could not be visualized, with a clear pouch of Douglas; a right adnexal structure measuring approximately 5.1 $\times$ 1.7 cm was noted, along with a well-defined unilocular left ovarian cyst measuring 4.8 $\times$ 4.8 cm (queried as a Para ovarian cyst), and the right

kidney could not be visualized. A chest radiograph performed as part of the preoperative work-up was unremarkable (Figure 1).

MRI of the abdomen and pelvis corroborated these sonographic findings and additionally demonstrated absence of the uterus and cervix with vaginal agenesis, consistent with Müllerian agenesis, along with right renal agenesis (Figures 2 and 3). A large midline pelvic cyst measuring approximately 9.7 $\times$ 8.7 cm was seen compressing the dome of the urinary bladder. The left ovary was enlarged with a twisted vascular pedicle showing the characteristic "whirlpool sign", confirming left ovarian torsion, while the right ovary appeared normal.

Taken together-a 46,XX karyotype, normal secondary sexual characteristics, absent uterus and cervix with vaginal agenesis, and an associated right renal agenesis-these findings established a diagnosis of Type 2 MRKH syndrome complicated by left ovarian torsion with a large ovarian cyst.

Management

Emergency exploratory laparotomy revealed an enlarged left ovary containing an approximately 8 $\times$ 7 cm simple cyst, with two complete turns of torsion of the ovarian pedicle (Figures 4 and 5). Mild ascitic fluid was present in the peritoneal cavity. A fibrous band with attached fimbriae was noted at the ovarian pedicle, along with a small nodular structure measuring approximately 2 $\times$ 1 cm, suggestive of a rudimentary uterine remnant (Figure 6). Vaginal agenesis was also confirmed intraoperatively (Figure 7).

Detorsion of the left ovary was performed, followed by cystectomy. Ovarian reconstruction was undertaken and ovarian drilling was performed; the fibrous band was excised, and viable ovarian tissue was preserved (Figure 8). The postoperative course was uneventful, and the patient was discharged in stable condition. Histopathological examination of the cyst wall confirmed a benign (serous) simple ovarian cyst.

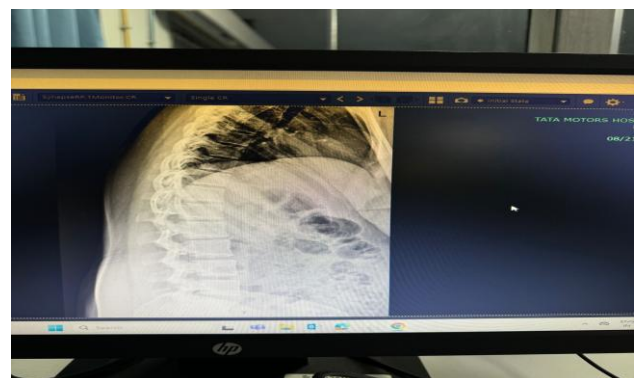


Figure 1: Preoperative chest radiograph-no significant cardiopulmonary cervical somite abnormality.

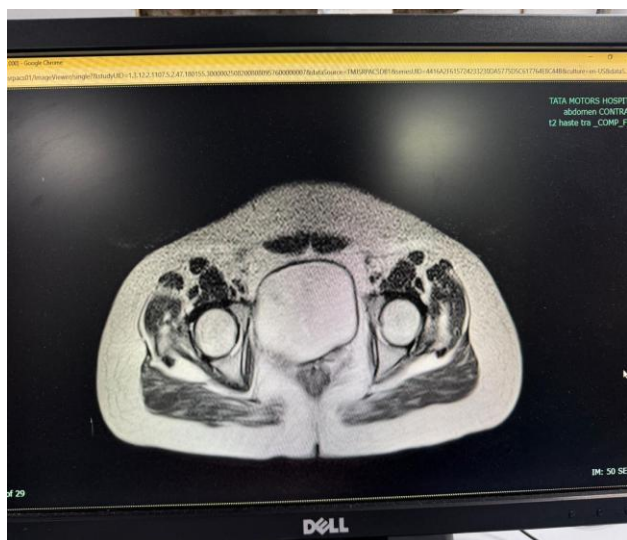


Figure 2: MRI pelvis-large midline pelvic cyst with absence of the uterus.

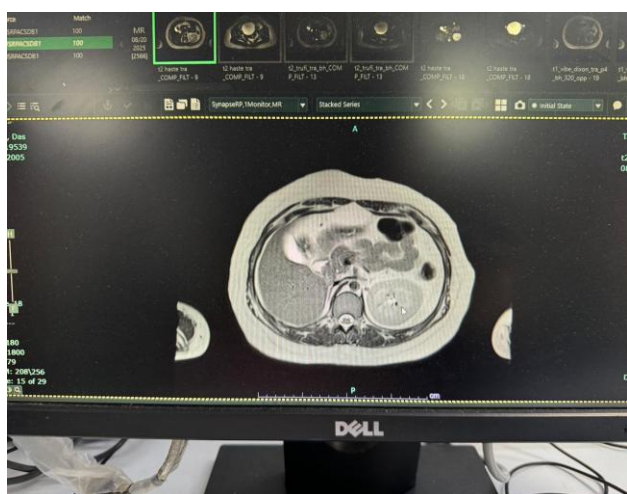


Figure 3: MRI abdomen-right renal agenesis.



Figure 4: Left ovarian cyst with rudimentary uterine remnant.



Figure 5: Delivered ovarian cyst with fibrous band and rudimentary uterine remnant with torsion.



Figure 6: Intraoperative examination confirming vaginal agenesis.



Figure 7: Left ovary after detorsion, cystectomy and reconstruction, with viable tissue preserved.

DISCUSSION

Ovarian involvement in MRKH syndrome is uncommon, since the ovaries originate from the genital ridge rather than the Müllerian ducts and are therefore usually spared. Ovarian anomalies of any kind-hypoplasia, streak change, malposition or cysts-have been reported in an estimated 5-10% of cases, but ovarian torsion itself remains an exceptional finding, documented in only a handful of published reports over the past decade.^{4,5,7} Absent normal uterine and broad-ligament support, combined with increased adnexal mobility, elongated or redundant infundibulopelvic and utero-ovarian pedicles, and-as in the present case-fibrous bands or rudimentary Müllerian remnants tethering the adnexa abnormally, is thought to predispose to torsion.⁴⁻⁶

It is worth clarifying a point of frequent confusion in the literature: MRKH syndrome as a whole is rare, but among patients with MRKH, the Type 2 (syndromic) form-with associated renal, skeletal, cardiac or auditory anomalies-is, if anything, marginally more common than isolated Type 1 disease, accounting for just over half of reported cases.^{2,3} Renal anomalies, including unilateral agenesis as seen in our patient, are the most frequent extragenital finding in Type 2 disease.^{2,3} An associated renal anomaly should therefore not itself be regarded as an unusual finding in MRKH, even though the syndrome overall-and ovarian torsion as its complication in particular-remains rare.

Clinically, ovarian torsion in a patient with MRKH is easily missed because the absent uterus alters the expected pelvic anatomy on examination and imaging, and can be mistaken for a degenerating fibroid or a nonspecific adnexal mass, as occurred at initial presentation in our patient. Ultrasonography remains a useful and accessible first-line tool. MRI, however, proved decisive, both confirming Müllerian agenesis and the associated renal anomaly and demonstrating features specific to torsion, including ovarian enlargement, a twisted pedicle and the “whirlpool sign”.⁶ A preoperative chest radiograph, while not diagnostic for MRKH itself, forms a routine part of anaesthetic fitness assessment and, together with MRI-based screening of the urinary tract, helps exclude other systemic or thoracic anomalies that may coexist in Type 2 disease. Combined and early use of these imaging modalities is therefore central to timely diagnosis.

Surgical management aims at prompt detorsion with ovarian preservation wherever feasible, particularly in young patients, to safeguard both endocrine function and future fertility potential through assisted reproductive technologies. Current evidence supports conservative management with detorsion even when the ovary appears congested or cyanotic at surgery, since ovarian viability often recovers once blood flow is restored, and simple untwisting carries a lower risk than adnexectomy.^{5,7} In our patient, detorsion with cystectomy, ovarian reconstruction and ovarian drilling achieved preservation of viable

ovarian tissue with an uneventful recovery, consistent with outcomes described in comparable published cases.^{4,5,7}

Given how few cases of ovarian torsion complicating MRKH syndrome have been reported to date, we present this case to add to the existing pool of published literature on this association, and to reinforce awareness of it among clinicians managing young women with primary amenorrhea and acute pelvic pain.

CONCLUSION

Ovarian torsion is a rare but clinically important complication of MRKH syndrome that can be easily overlooked because of the syndrome's atypical pelvic anatomy. A high index of suspicion in young patients presenting with primary amenorrhea and acute pelvic pain, supported by ultrasonography and MRI, allows timely diagnosis, while prompt, ovary-conserving surgery remains the cornerstone of management. This case adds to the limited literature on this association and underscores the value of a fertility-preserving surgical approach whenever feasible.

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