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Leiomyoma in a Diagnosed Case of MRKH Syndrome: A Case Report

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Abstract: ***Introduction*: Primary amenorrhea is the failure to reach menarche. The second most frequent cause of primary amenorrhea, after gonadal dysgenesis, is the Mayer - Rokitansky - Küster - Hauser (MRKH) syndrome. Here, we present a rare instance of leiomyoma in a diagnosed case of MRKH syndrome. *Case Report*: A 35 - year - old woman presented to our Gynaecology OPD for abdominal pain and progressive abdominal distention for 8 months. Upon primary clinical assessment, she reported primary amenorrhea and primary infertility. There is no historical past illness. Her Physical Examination showed normal breast development and normal female body contour. Her external genitalia was normal. The patient was diagnosed with a case of MRKH. On USG bulky and heterogeneous uterus with a sub - endometrial cyst of myometrium suggestive of adenomyosis, endometrial hyperplasia, and posterior fundal wall fibroid mass 20 × 13 cm on right adnexa up to gastric region showing low vascularity, uterine or broad ligament fibroid was found. *Conclusion*: Women with MRKH syndrome who present with abdominal pain and mass. Ultrasonography is the first imaging to evaluate the pelvic mass and genitourinary system. Magnetic resonance imaging is a more accurate modality to confirm the pelvic mass, and evaluate the genitourinary system and may give signs of malignant tendencies. Complete removal by laparoscopic is recommended to manage this case.***

Keywords: MRKH syndrome, Leiomyoma, Primary amenorrhea

1. Introduction

Mullerian agenesis, a congenital condition affecting the female genital tract, is the second most common cause of primary amenorrhea, the first being gonadal dysgenesis [1, 2]. The most common presentation of Mullerian agenesis is the congenital absence of the vagina, uterus, or both, which also is referred to as Mullerian aplasia or vaginal agenesis or Mayer - Rokitansky - Kuster - Hauser syndrome (MRKH) [1]. It affects one in 4, 000–5, 000 female births. [2]. Usually undetectable till puberty, as secondary sexual characters are fully developed due to normally functioning ovaries. The diagnosis is often made either radiologically or laparoscopically in a woman with normal hormonal tests and female karyotype (46 XX) presenting with primary amenorrhoea. [2]

Three types of the syndrome are defined in the literature, Type 1 - Restricted to reproductive organs, Type - 2 having an atypical presentation with symmetric uterine remnants with abnormal uterine tubes commonly associated with ovarian disease, congenital renal, bone, and hearing defects. Type - 3 is known as the MURCS type involving uterovaginal hypoplasia or aplasia, renal, bone, and cardiac malformations. Environmental and genetic factors are thought to play a role though the definite etiological basis of the syndrome has not been understood to date. [3]

Benign lesions like myomas or fibroids commonly occur in the normal uterus. Occurrence from the rudimentary uterus in patients with MRKH syndrome is very rare. Only a few cases have been reported in the literature. Diagnosis and management are challenging to treat this condition. [4]

Here, we present a 38 - year - old woman, presenting with primary amenorrhea, diagnosed with MRKH syndrome with co - existing huge leiomyoma. Informed consent was obtained from the patient subject to report this case.

2. Case Report

A 35 - year - old woman presented to our Gynaecology OPD for abdominal pain and progressive abdominal distention for 8 months. Upon primary clinical assessment, she reported primary amenorrhea and primary infertility, which was never evaluated. Her previous medical and surgical history were unremarkable. Her family history was negative. For 8 months, she has been feeling abdominal pain, on a scale of 6 on the Visual Analog Scale, and was treated with analgesics. There is no history of any chronic illness. Her physical examination showed female body contour and a normal hair pattern with bilateral breast of Tanner Stage IV/V.

On pelvic examination, the external genitalia was normal. Normal vagina mucosa, vagina tract and cervix could not be visualised. Palpable, immobile firm pelvic mass extending from the suprapubic area and reaching up to the lower border of the liver of size 25 × 15 × 10 cm more toward the right abdomen and pelvis.

Transabdominal ultrasonography revealed a bulky and heterogeneous uterus with a sub - endometrial cyst of myometrium suggestive of adenomyosis, endometrial hyperplasia, and posterior fundal wall fibroid mass 20 × 13 cm on right adnexa upto gastric region showing low vascularity, uterine or broad ligament fibroid. (Fig.1). Both ovaries, kidneys and ureters were normal. She was diagnosed

with MRKH syndrome (uterus dysgenesis, proximal vaginal agenesis), and suggested for further MRI evaluation.

A magnetic resonance imaging of the pelvis showed a large soft tissue ovoid mass lesion with a well - defined capsule with central areas of necrosis located in the pelvis and abdomen, slightly of the midline, possibly arising from the right broad ligament suggestive of right - sided broad ligament fibroid. Non - visualization of the uterus, cervix, and upper 2/3rd of vagina suspecting aplastic?. Bilateral kidney was in a normal position other intraabdominal organs were normal. The patient was submitted to an open surgical intervention (Fig.2),

Intra Operatively, a huge myoma of 30× 20×15cm and of 7 kg weight arising probably from the rudimentary horn/ almost merging with the cervix and extending up to the liver. The uterus and cervix were aplastic with two rudimentary horns and two fundi, and bilateral tubes and ovaries were normal. We carried our intracapsular myomectomy with bilateral fimbriectomy. During the operation the bleeding was minimal. The histopathology showed uterus dysgenesis and broad ligament fibroid as shown in Figs.

3. Discussion

MRKH syndrome is a congenital disorder of the female reproductive system, which is usually diagnosed by clinical findings and supported by imaging. Diagnosis is usually made in adolescents when they do not have a menstrual period (primary amenorrhea) [3, 4]. The types of MRKH syndrome varied from restricted to reproductive organs only and involved genitourinary, bone, and cardiac malformation [2].

Most patients having MRKH syndrome require a functioning vagina to improve their quality of life regarding the sexual relationship. Unlike our case, the patient does not have any impact on the sexual relationship. We may ensure that this condition causes infertility, therefore surrogacy and adoption were their options for childbearing [4].

We should keep in mind that she has normal functioning ovaries and fallopian tubes, there is a chance of getting the child from the surrogate technique by retrieving the ovum [5]. Uterine remnants could present in varying sizes and consist of fibromuscular tissue, a small number of smooth muscles (myometrium), and stroma tissue arranged in glands (endometrium).

Therefore, it could resemble a tumour growing from that tissue, mostly leiomyomas, following the same pathogenic mechanisms as in the normal uterus and acting as targets for the ovarian hormones. Mostly these tumours were asymptomatic (diagnosed accidentally during check - up) or can give symptoms like chronic pelvic pain or discomfort [6, 7].
The basic pathogenesis of leiomyoma is estrogen - dependent growth of smooth muscles and fibroblasts and high sensitivity to it as compared to normal myometrium [8].

A rare incidence of leiomyoma in uterus remnant could be a decreased concentration or sensitivity of the estrogen receptors or genetic predisposition compared to the normal uterus with leiomyomas [9]

Therefore, removing all the masses and adjacent remnants is strictly required. The other advantage of the laparoscopic approach is clear visualization of adjacent pelvic organs, mostly the genitourinary system (bladder, ureters) [10, 11, 12].

4. Conclusion

For women with MRKH syndrome who present with abdominal pain and mass, ultrasonography is the first imaging to evaluate the pelvic mass and genitourinary system. Magnetic resonance imaging is a more accurate modality to confirm the pelvic mass and subsequently evaluate the genitourinary system and may give signs of malignant tendencies. Complete removal by laparoscopy is recommended to manage this case.

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Images

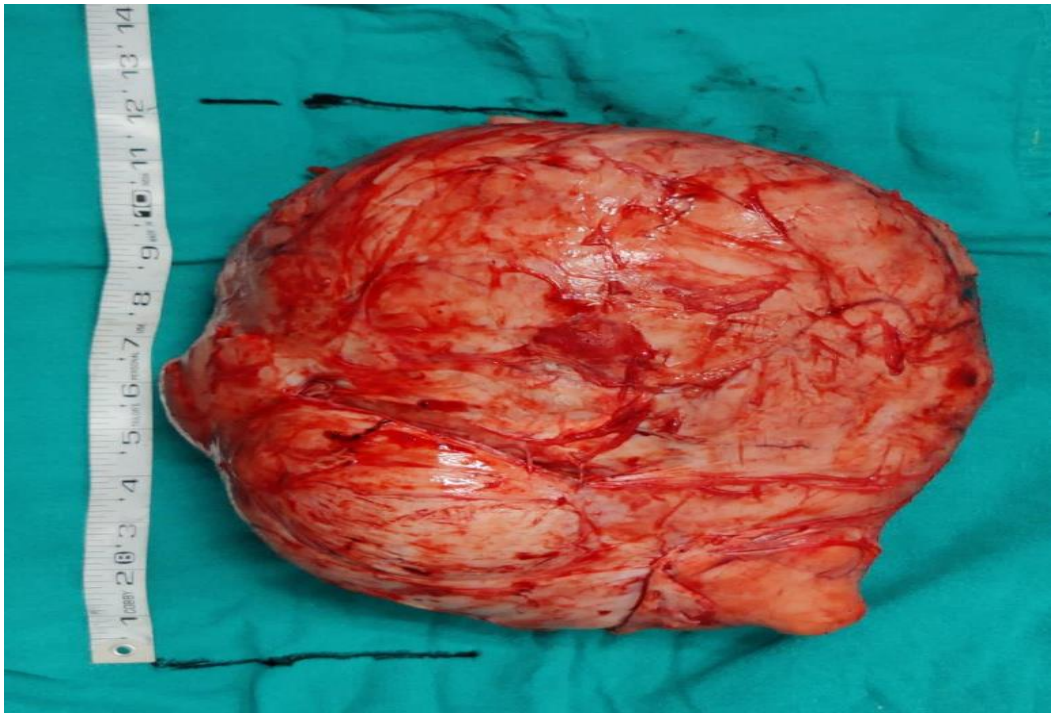

Figure 1

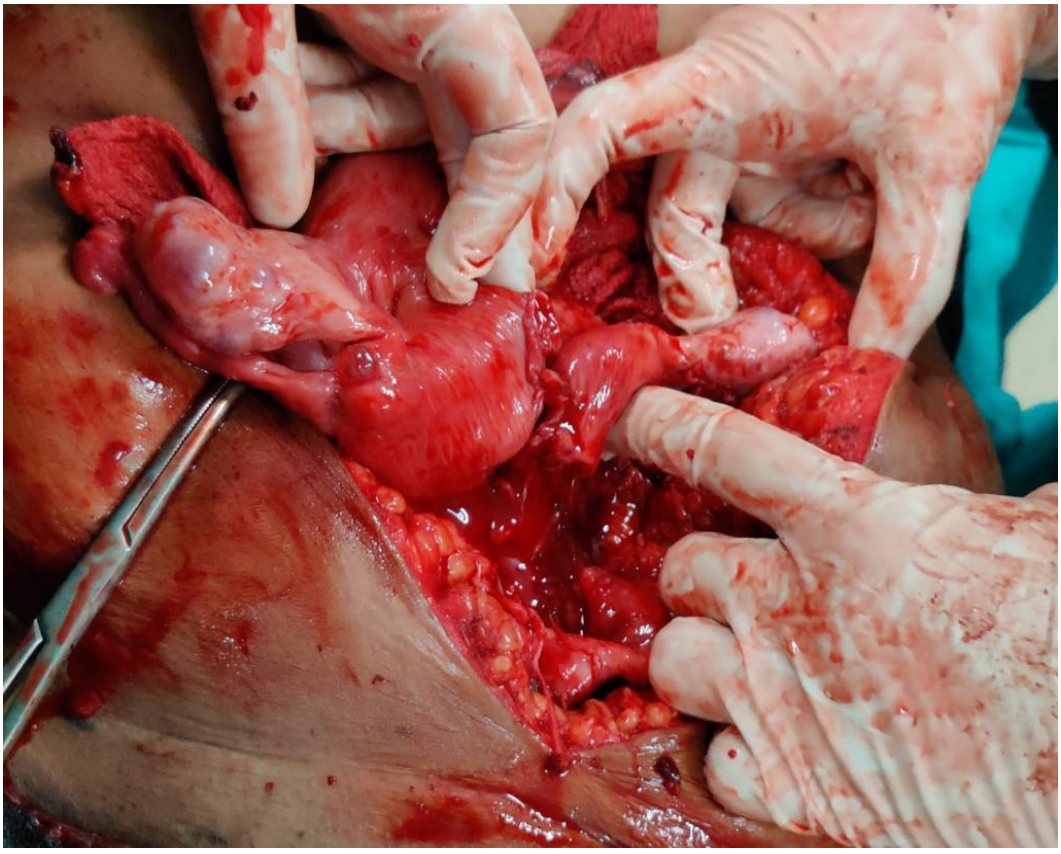

Figure 2