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Mullerian Remnant Leiomyoma Mimicking Ovarian Neoplasm in a Woman with MRKH Syndrome

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Abstract: ***Occurrence of leiomyoma arising from the rudimentary horn of uterus is an extremely rare condition, only few cases have been reported till date. Herein, we present a case of MRKHS TYPE II with leiomyoma, mimicking ovarian neoplasm on imaging studies, when the patient presented with complaints of vague lower abdominal pain. A differential diagnosis of ovarian neoplasm and leiomyoma must be considered in a case of mullerian agenesis presenting with mass per abdomen.***

Keywords: Mullerian Anomaly, MRKHS, Leiomyoma

1. Introduction

Mayer - Rokitansky - Kuster - Hauser (MRKH) syndrome, also referred as mullerian agenesis is a rare congenital mullerian duct anomaly. It is considered to affect 1 in 5000 female live births globally (1). Nearly 16% cases of primary amenorrhea are attributable to MRKH syndrome, making it the second most common causes of primary amenorrhea after gondal dysgenesis (2).

The first case of myoma in association with MRKHS was described by Beecham and Skiendzielewski in 1977 (3).

This article provides a narrative review of the current literature on such presentation of a case of MRKHS type II, its diagnostic dilemma and management.

2. Case Report

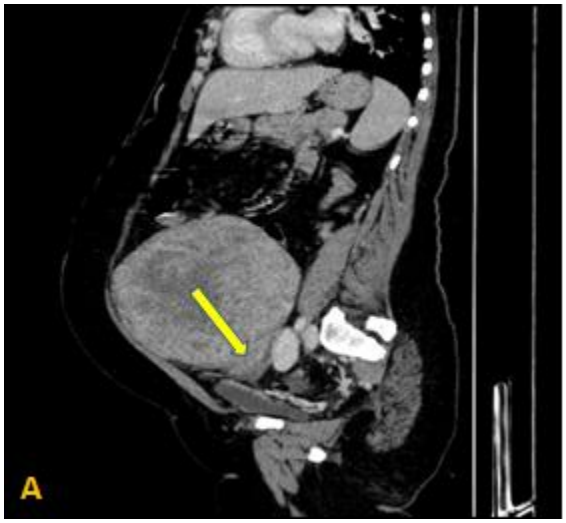
A 41 year old unmarried female, a known case of primary amenorrhea was referred to Father muller medical college and hospital, gynecology OPD with complaints of insidious onset of lower pain abdomen since 1 week, which was diffuse in nature and non - radiating, relieved on taking analgesics and not associated with vomiting or bowel or bladder disturbance.

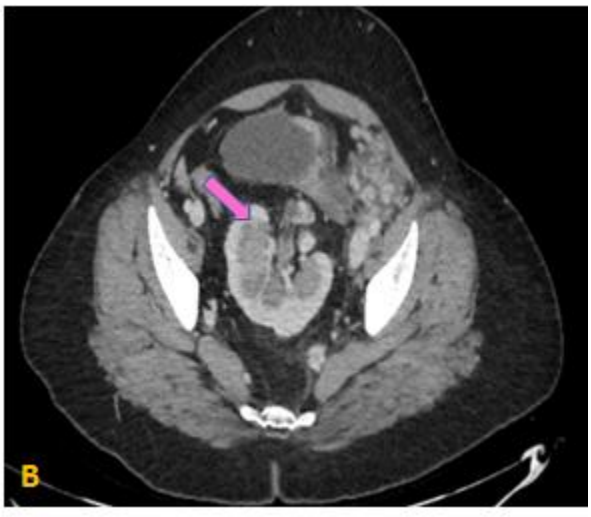
She had been evaluated for primary amenorrhea at the age of 16. However, no reports were available

On examination, a female body contour with Tanner stage 4 breast, axillary and pubic hair development were noted. On abdominal examination, a large non tender mass extending up to supra - umbilical region measuring 15 X 10 cm with regular border and firm consistency was palpable. There was no evidence of hepato - splenomegaly and ascites. External genitalia were normal with blind vaginal pouch of 1cm length

CECT abdomen and pelvis revealed a large heterogeneous enhancing lesion measuring 17.2 X 12.4 X14.3 cm arising from left hemi pelvis likely ovarian malignancy with a was noted in the pelvis. Uterus and bilateral ovaries could not be visualized. Laboratory investigations showed normal serum tumor marker levels.

On the basis of history and CECT abdomen, a provisional diagnosis of left ovarian neoplasam with MRKH type II was established.





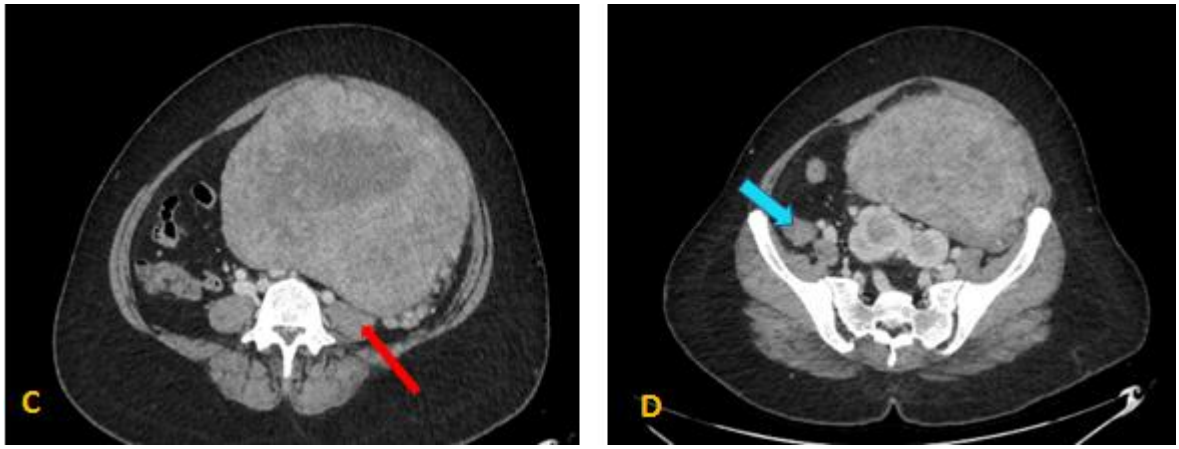


Image 1: A (yellow arrow) - Small triangular structure at the base of the mass - likely uterine/Mullerian remnant, also absent body of uterus, cervix, vagina; B (pink arrow) - pelvic ectopic crossed fussed kidney; C (red arrow) Rounded structure posterior to the mass adjacent to vessels that are branches of aorta - likely left ovary; (blue arrow) - right ovary

Patient was posted for laparotomy with frozen section. Intraoperatively, a solid mass of 16X 13 cm noted in the midline, firm consistency with smooth surface and irregular borders arising from the left rudimentary horn of the uterus. Left ovary with polycystic appearance seen in close proximity to the mass. Right rudimentary horn of uterus and right polycystic ovary were seen separately from the mass. Bilateral fallopian tubes and broad ligaments found to be fused with the lateral portion of round ligament which was stretched over the mass. Specimen was subjected for frozen section was reported with a suspicion of borderline ovarian malignancy of left ovary. Hence, we proceeded with bilateral salphingo - ophorectomy in addition to rudimentary uterus excision. Postoperative recovery of the patient was satisfactory and uneventful.

Later, Histopathological examination confirmed the smooth muscle nature consistent with leiomyoma of rudimentary horn of uterus. Bilateral ovaries were found to be unremarkable.

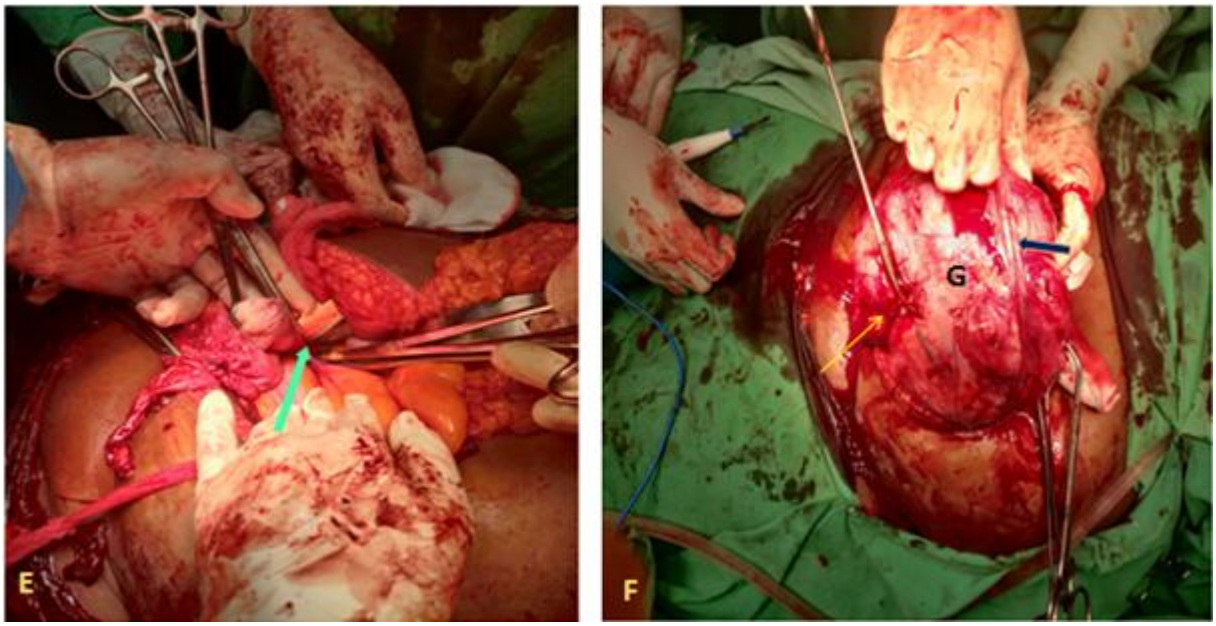


Image 2: E - Right ovary, fallopian tube and broad ligament - Left ovary; G - Mass arising from left rudimentary horn

3. Discussion

The first description of congenital absence of uterus and vagina dates back to 1562, where the Italian Anatomist Realdo Colombo in his main work 'De Re Anatomica' describes an entity 'vulva rara' in a woman with no womb and vagina, who complaints of pain upon coitus. (4)

MRKH syndrome is a rare a congenital female reproductive system anomaly associated with an absence or incomplete development of mullerian duct. Three different types of MRKH syndrome has been identified, type I (typical form) characterized by the congenital absence of the uterus and upper vagina with normal ovaries and fallopian tubes; type II (atypical form) associated with malformations of the ovaries or renal dysfunction and type III (MURCS syndrome) suggested as uterovaginal aplasia/ hypoplasia, renal dysfunction with other congenital anomalies such as renal, skeletal, hearing, cardiac, and ocular anomalies (5, 6). The diagnosis of MRKH syndrome is often delayed till puberty by the virtue of presence of normal secondary

sexual characters, 46 XX karyotype and normal ovarian function when the patient presents with primary amenorrhea

A review of the medical literature illustrated leiomyoma in 35 MRKH patients and the uterine remnant noted to be the origin of leiomyoma among 65 % of the cases (7). Other origins include multiple uterine remnants, fibrous myometrial bands, and the broad ligament, round ligament, vascular leiomyoma, and parametrial tissue (7, 8). In our case myoma arose from the left rudimentary horn of uterus.

Patients with MRKH syndrome with pelvic mass pose a diagnostic dilemma as highlighted in our case, where computed tomography appearance of fibroid of remnant uterus was mistaken for ovarian tumor, probably because of low sensitivity of the available imaging modalities to exclude the remnant uterine tissue with certainty. Furthermore, a large fibroid or a pedunculated subserosal fibroid may occasionally be misconceived for an ovarian tumor (9). Hence it is prudent for the interpreting radiologist to be aware of this diagnostic pitfall.

Exploratory laparotomy was performed in most of the reported cases so was in our case. It is recommended that the minimum surgical intervention is the excision of myoma and adjacent uterine remnant with or without bilateral salphingo - ophorectomy (10). Leiomyomas in MRKH patients are best explained by the fact that these patients maintain normal ovarian function with normal levels of estrogen and progesterone owing to the evolution of fibroids in the similar pattern as those in normal patients. It is noteworthy that, the presence of smooth muscle cells in the proximal end of mullerian duct despite it being endodermal in origin supports the possibility of fibroids in MRKHS patients with absent uterus. Additionally, a somatic genetic mutation has been found to regulate the levels of estrogen signaling which contributes to development of fibroids in MRKH patients. A genome sequencing approach may prove to be a valuable tool for investigating MRKH syndrome associated with fibroids

Although, the development of Leiomyomas in MRKH patients is a rare scenario, every gynecologist should be aware of this presentation and consider the possibility of ovarian neoplasm as well as fibroids in the differential diagnosis.

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