A rare case of multiple leiomyomas on rudimentary uterus in a woman with Mayer Rokitansky Kuster Hauser (MRKH) syndrome: A challenging diagnosis and laparoscopic approach

Achmad Kemal Harzif\textsuperscript{a,∗,1}, Sonia Ambalagen \textsuperscript{b,∗}, Fisytanisa Elya Charilda \textsuperscript{c}, Heidi Dewi Mutia\textsuperscript{c}

\textsuperscript{a} Division of Reproductive Immuno-Endocrinology, Department of Obstetrics and Gynecology, Faculty of Medicine Universitas Indonesia, Dr. Cipto Mangunkusumo Hospital Jakarta, Indonesia
\textsuperscript{b} Department of Obstetrics and Gynecology, Dr. Cipto Mangunkusumo Hospital Jakarta, Faculty of Medicine Universitas Indonesia, Pangeran Diponegoro Street No. 71, Kenari, Jakarta, Indonesia
\textsuperscript{c} Indonesian Reproductive Medicine Research and Training Center (INA- REPROMED), Faculty of Medicine Universitas Indonesia, Dr. Cipto Mangunkusumo Hospital Jakarta, Pangeran Diponegoro Street No. 71, Kenari, Central of Jakarta, 10430, Indonesia

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**A B S T R A C T**

Mayer Rokitansky Kuster Hauser (MRKH) syndrome is a congenital disorder involving reproductive, genitourinary, bone, and cardiac malformation. The incidence is 1 in 4000–5000 females livebirths. The phenotype is female 46 XX karyotype, normal secondary sexual characteristics, and normal functional ovaries. The occurrence of leiomyoma in uterine remnant in MRKH syndrome is a very rare case, even though several cases have been reported. The diagnosis and management approach, in this case, is quite challenging. Here, we report a 38 years old female who represents multiple leiomyomas on the rudimentary uterus, then we did laparoscopic removal of the fibroids and adjacent rudimentary uterus. © 2021 Published by Elsevier Ltd on behalf of IJS Publishing Group Ltd. This is an open access article under the CC BY-NC-ND license (http://creativecommons.org/licenses/by-nc-nd/4.0/).

1. Introduction

Mayer-Rokitansky-Kuster-Hauser (MRKH) syndrome is a congenital disorder that occurs in females and affects the reproductive organs. This condition appears as underdevelopment or absence of the upper vagina, cervix, and uterus, but the external genitalia is normal [1]. Mostly this condition undetectable till the girl has puberty because this condition primarily causes primary amenorrhea with normal developing secondary sexual characteristics, as the ovaries are present and functional [1,2]. This syndrome is quite uncommon, with an incidence of one in 4000–5000 female births, and is the second most frequent cause of primary amenorrhea. The patients always present karyotype 46, XX [2].

This syndrome is classified into three types according to the involvement of structures other than the ones related to reproductive organs. The most common one is type I, represented by abnormalities restricted to reproductive organs. The second one is Type II is atypical, with the presence of symmetric uterine remnants and normal uterine tubes, mostly associated with ovarian disease, congenital renal, bone abnormalities, and hearing defects. The third one is called as MURCS type, involving urogenital hypoplasia or aplasia, renal, bone, and cardiac malformations. The etiology of this syndrome is not fully understood, and environmental and genetic factors are thought to play a role [1–3].

Myomas or fibroids are rather common benign lesions in the normal uterus that can arise from the remnant uterus. The occurrence of myoma arising from the rudimentary uterus is a very rare finding and only a few cases have been reported. Diagnosis and further management are challenging conditions to treat this condition [4].

Here, we represent a 38 years old woman, for the first time diagnosed MRKH syndrome concurrent with multiple leiomyomas. Informed consent was obtained from patient’s subject to report this case. This case report was reported in line SCARE 2020 criteria [5].

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2. Case presentation

A 38 years old woman presented to our Endocrinology Clinic for abdominal pain. Upon primary clinical assessment, she reported primary amenorrhea and primary infertility, which have never been evaluated. Otherwise, her previous medical and surgical were unremarkable. Her family history was negative. For 3 years, she has been felt abdominal pain, a scale of 6, and was treated with analgesics. There is no historical past illness. Then she was referred for further evaluation, found 6 cm mass on left ovary, suggested surgery, but she refused. The abdominal pain and discomfort consistently occurred. No complaint regarding sexual intercourse. She did not have symptoms related to the bladder or the bowel. Her physical examination showed female body contour and normal hair pattern. On pelvic examination, the external genitalia was normal. Normal vagina mucosa and tract, unseem cervix. Palpable firm boundary mass on suprapubic area size 5 cm, and on left adnexa size 6 cm, mobile.

Transabdominal and transrectal ultrasonography revealed multiple uterine fibroids on the rudimentary uterus, ranging from 3 cm to 5 cm (Fig. 1). Both ovaries and kidneys were normal. She was diagnosed with MRKH syndrome (uterus dysgenesis, proximal vaginal agenesis), suggested for MRI evaluation. A magnetic resonance imaging of the pelvis showed hypoplastic uterus, with multiple subserosal fibroids, one with degenerated fibroids on suprapubic, anteriorly to the bladder, size 3.1 × 4.8 cm. Other intraabdominal organs were normal.

The patient was submitted to a laparoscopic intervention (Fig. 2), during which we found a band-like uterine horn structure, multiple fibroids were stretching over the rudimentary uterus, size ranging from 6 cm to 5 mm (smallest). Both ovaries and tubes were normal. We removed the whole rudimentary uterus following all the fibroids. During the operation the bleeding was minimal. The histopathology showed uterus dysgenesis and multiple leiomyomas as shown in Figs. 3 and 4.

3. Clinical discussion

MRKH syndrome is a congenital disorder of a female reproductive system, which is usually diagnosed by clinical findings and supported by imaging. Diagnosis is usually made in adolescents when they did 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 for getting the child from the surrogate technique by retrieving the ovum [6]. Uterine remnants could present in varying sizes, consist of fibromuscular tissue, a small number of smooth muscles (myometrium), and stroma tissue arranging in glands (endometrium). Therefore, it could resemble a tumor 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 tumors were asymptomatic (diagnosed accidentally during check-up) or can give symptoms like chronic pelvic pain or discomfort [7,8].

Finding a pelvic mass in MRKH cases during regular clinical and ultrasound examination could indicate in need of a diagnostic laparoscopy [7]. Aising leiomyomas on uterine remnants in MRKH cases is a theoretical possibility. However, till nowadays these cases may have been reported rarely earlier. The basic pathogenesis of leiomyoma is estrogen-dependent growth of smooth muscles and fibroblasts and high sensitivity to it as compared to normal myometrium [9]. As the presents of endogenous estrogen or exogenous estrogen could be the main problem to develop leiomyoma
in MRKH. In this case, there was no evidence of exogenous estrogen exposure to this patient, then only endogenous estrogen may be the factor. 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 [11] Unfortunately, we could not perform receptor studies to establish the exact pathogenesis. Laparoscopic removal as a first-line approach was indicated. Removing the multiple fibroids and adjacent uterus remnant were the best choice for today [12,13]. Several studies reported these cases, most of them using the laparoscopic approach for diagnosing and treating. Otherwise, laparotomy can be done in low resources areas, voluminous uterus, and masses which might be a limitation to do a laparoscopy. Other than that, a suspect for malignancy in leiomyoma could happen in this case as in a normal uterus, even though the possibility is quite low [14]. Therefore, removing all the masses and adjacent remnants is strictly required. The other advantages of the laparoscopic approach are clear visualization of adjacent pelvic organs, mostly genitourinary system (bladder, ureters) [12,14,15].

Differential diagnosis of leiomyoma of the rudimentary uterus in MRKH syndrome is ovarian fibroma, GIST (gastrointestinal stromal tumor), extravesical leiomyoma of the urinary bladder [15]. Other cases have been reported that uterine adenomyosis can be represented in a normal uterus which may be due to direct invasion of uterine mucosa into uterine musculature as compared to the rudimentary uterus, this theory could not be applied. Metaplasia of the stromal cells under the influence of autocrine factors or paracrine factors which are intermediaries of genetic, immunologic, and endocrine influences can lead to forming adenomyosis in MRKH syndrome [10].

Fig. 2. (A) Showed multiple fibroids on uterus remnants, both ovaries were normal; (B) Fallopian tubes were normal, largest uterine fibroid; (C) Uterine fibroids on right horn; (D) Multiple uterine fibroids.

Fig. 3. Histopathology result: (A) myometrium tissue: smooth muscles in uterine fibroids (40X); (B) Proliferation of smooth muscles, irregular pattern (400X); (C) No myototic activity.
4. Conclusion

Women with MRKH syndrome who present with abdominal pain and mass, leiomyoma of Mullerian remnant should be considered for diagnosis. 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 pristinely evaluating the genitourinary system (kidney, ureters, bladder), and maybe give signs for malignancy tendencies. Complete removal by laparoscopic is recommended to manage this case.

Declaration of Competing Interest

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Ethical approval

This study is exempt from ethical approval.

Consent

The patient was already informed with no data such as names, initials, and hospital numbers would be published.

Author contribution

Achmad Kemal Harzif: study concept and design, editing manuscript.
Sonia Priyadashini: data collection, analysis and interpretation.
Fistyanisa Elya Charilda: writing and editing manuscript.
Heidi Mutia Dewi: study concept and design, editing manuscript.

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