

DOI: <https://dx.doi.org/10.18203/2320-1770.ijrcog20252755>

Case Report

A case of rudimentary uterine horns with cervical agenesis with intramural fibroid and bilateral endometrioma

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Received: 04 July 2025

Accepted: 05 August 2025

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ABSTRACT

The objective of the study was to report a case of rudimentary horns of cervical agenesis with intramural fibroid and bilateral endometrioma. A 35-year-old nulligravida with a rare Mullerian anomaly of uterus, cervix and vagina with coexisting fibroid and endometrioma. A definite diagnosis and resection of Mullerian structure and endometrioma. This reports a rare case of occurrence of benign tumor of mesenchymal origin from a non-functioning uterine horn along with features of degeneration with cervical agenesis along with presence of bilateral ovarian endometrioma reinforcing the possibility of coelomic metaplasia theory of its origin.

Keywords: Mullerian anomaly, Rudimentary horn, Fibroid, Endometrioma

INTRODUCTION

Primary amenorrhea is defined as failure to reach menarche. Most of the underlying cause of primary amenorrhea can be classified into anatomic and sexual development abnormalities, ovarian insufficiency, hypothalamic or pituitary disorder or other endocrinal gland disorder, yet majority of cases are due to gonadal dysfunction (43%) followed by Mullerian agenesis (10-15%).¹

Mullerian anomalies are rare pathology of female genital tract development with prevalence of 5% of women.² The development of urogenital system starts in 7th week with development of mullerian duct followed by their canalization and their fusion from caudal to cephalad followed by resorption of investing septa. Failure at any step leads to abnormal development. Mayer-Rokitansky-Kuster-Hauser (MRKH) with aplasia of uterus and upper part of vagina in women with normal karyotype secondary sexual character with estimated incidence of 1 in 4500.³

90% of MRKH syndrome have uterine remnants with functional myometrium nodules which poses the histological features of myometrium, smooth muscle fibre and fibrous stroma and are therefore susceptible for same gynaecological condition as the former like fibroids.⁴ Ovaries are normal in cases of MRKH syndrome and thus estrogen dependent pathological conditions can develop. Distortion of anatomy accompanies Mullerian abnormality also predisposes the occurrence of endometriosis and chronic pelvic pain.⁵

CASE REPORT

A 35-years-old nulligravida came to gynaecology outpatient department with complaint of pain and feeling of heaviness in lower abdomen with gradually increasing lump for 5 years. Pain was dull aching in nature and relieved on medication. She had not attained menarche, nor had history of cyclical lower abdominal pain. She has been married for 20 years and has an active sexual life.

On general examination she was stable and well oriented, height 152 cm with average built. Her secondary sexual characters were well developed (breast-tanner stage V and pubic hair-stage V). Her systemic examination did not reveal any CNS, cardiac, respiratory or skeletal abnormality.

On per abdominal examination, a suprapubic smooth, firm mass of around 20-22 weeks size felt, mobile from side to side, non-tender with no other organomegaly. On local examination external genitalia appeared normal. On per speculum examination, a blind vagina of approximately 4 cm in length with cervix not visualized. On per vaginal examination, nodule felt at vaginal vault, a mobile firm mass approximately 20×15 cm felt, non-tender with uterus not felt separately. Based on history of primary amenorrhea and enlarging adnexal mass and clinical examination suspicion of ovarian or para ovarian cyst/mass or retroperitoneal mass was made. Transabdominal ultrasound followed by MRI was done which showed uterus like structure 3.1×3×4.6 cm, cervix-like structure 9mm, hypointense lesion 9.7×11.5×16.6 cm with hyperintense area suggestive of cystic/myxoid degeneration, f/c/w large intramural and subserosal fibroid with degeneration along with bilateral ovarian haemorrhagic cyst and upper abdominal imaging was normal. Hormonal profile containing follicle-stimulating hormone (FSH), luteinizing hormone (LH), and estradiol and anti-Müllerian hormone (AMH) were normal indicating toward functional HPO axis (FSH- 23.1 μ IU/ml; LH-7.90 mIU/ml; AMH- 1.9 ng/ml; and E₂-166.8 pg/ml) (Figure 1).

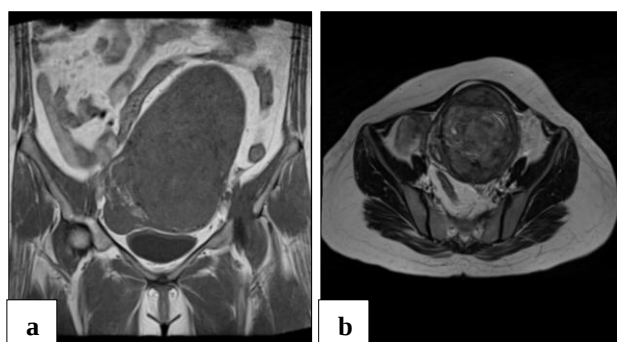


Figure 1: Preoperative lower abdomen MRI scan showing a large fibroid and a rudimentary horn seen separate from the mass (a) coronal plane, and (b) transverse plane.

Based on the radiological findings of rudimentary uterus with cervical agenesis a diagnosis of MRKH syndrome was made and informed to the patient and attendant. Patient was planned for exploratory laparotomy. On laparotomy, rudimentary bicornuate uterine horn were noted, both attached through fibrous cord like structures to a knob like structure probably hypoplastic cervix. Right horn was atrophic while intramural with subserosal extending fibroid of 20×10 cm (FIGO-6) seen arising from left horn. Both horns had separate tube and ovary. Bilateral

salpinx appeared normal while both ovaries were enlarged with endometrioma 10×8 cm and 10×12 cm in right and left ovary respectively adherent to pouch of Douglas and bowel loops. Adhesiolysis done, broad ligament dissected, ureter was mobilized laterally and since ovaries were unsalvageable, we proceeded for total hysterectomy along with removal of fibroid in toto with bilateral salpingo-oophorectomy, the third clamp was applied distal to nodular knob like swelling and uterus with non-canalized cervix removed, vaginal cuff remained intact (Figure 2).

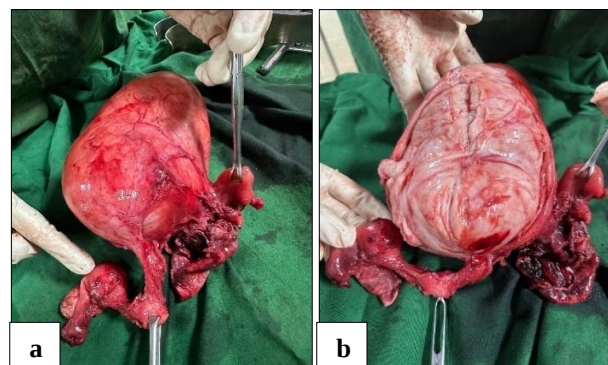


Figure 2 (a and b): Gross specimen showing rudimentary cervix with rudimentary horns with fibroid arising from left horn and bilateral endometrioma of the ovaries.

Endometriotic lesion over the peritoneum was fulgurated. Entire specimen was sent en bloc for histopathological examination. Postoperatively patient recovered well and is planned for combined hormonal replacement therapy.

Histopathology report was consistent with featured of cervical agenesis with presence of only fibrous tissue without epithelial lining or endocervical glandular component, both tubes had normal histology, ovaries showed presence of hemosiderin laden macrophages along with endometrial stroma suggesting presence of endometrioma. Mass in uterus showed smooth muscles arranged in fascicles with thick blood vessel suggesting fibroid, endometrium of rudimentary horn showed disordered proliferative phase and endometrial cavity did not have any hemosiderin (Figures 3-5).

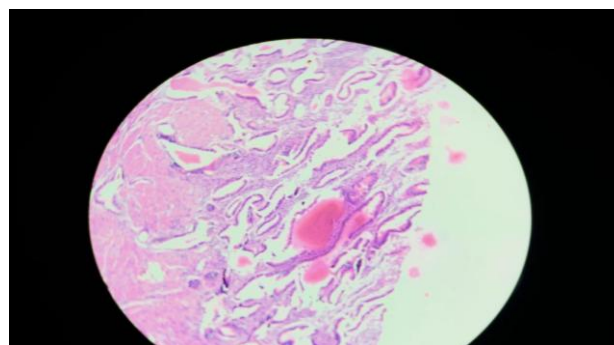


Figure 3: Uterine horn showing disorder proliferative phase endometrium.

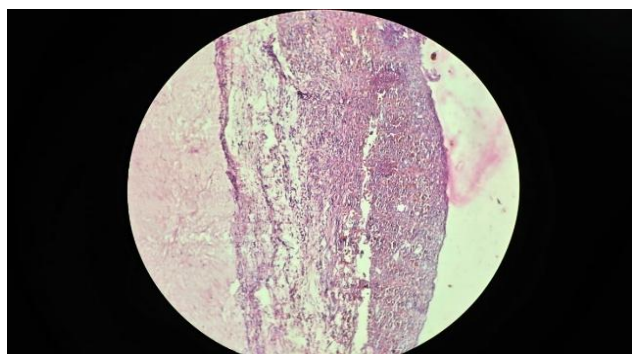


Figure 4: Ovarian cut section showing endometrioma.

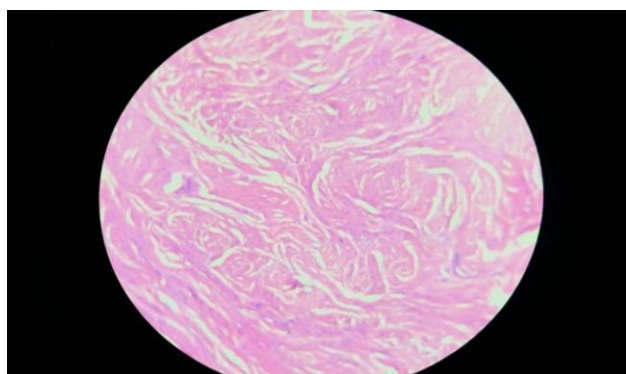


Figure 5: Cervix with fibrous tissue.

DISCUSSION

Agenesis of Mullerian duct (i.e. paramesonephric duct) result in complete or partial absence of Mullerian structures. MRKH syndrome account 10-15% of primary amenorrhea with normal karyotype, normal secondary sexual character and normal serum FSH and estrogen. MRKH syndrome is characterised by absence of uterus with blind vagina (absence of upper 2/3rd of vagina). It can occur in isolation i.e. MRKH syndrome type I or in association with other malformation like renal (unilateral agenesis, ectopia of kidney or horseshoe kidney), skeletal-vertebral (Klippel Feil anomaly, fused vertebral, scoliosis), hearing defect, cardiac and digital anomalies termed as MRKH syndrome type II.³

MRKH syndrome usually have nonfunctional myometrial nodule in its uterine remnant. This poses a risk of development of gynaecological problems such as leiomyoma and adenomyosis. The ovaries in cases of MRKH syndrome are also functional hence predisposed to polycystic ovary, endometriomas or even ovarian tumors.⁶

Fibroids are benign mesenchymal tumour arising from smooth muscle fibres and fibrous stroma of myometrium. Fibroid arising in MRKH syndrome is a rare association consist of transformed proliferating myometrial cells and disordered extracellular matrix. These smooth muscle cells response under hormonal influence from normally functioning ovaries which contain high concentration or sensitivity of estrogen receptors, progesterone receptors

and aromatase in remnant of myometrial tissue. Somatic genetic mutation or clonal chromosomal abnormalities in patient with MRKH syndrome contribute to different levels of estrogen signal regulation.⁷ Epigenomic changes in uterine fibroid like aberrant DNA methylation and histone tail modification can dysregulate gene expression, estrogen stimulate growth of uterine fibroid through its receptor ER α while progesterone through its apoptosis inhibitor BCL-2 protein increases fibroid bulk by inhibiting their programmed cell death.⁸ Usual presentation of fibroid in patient with MRKH syndrome are pelvic mass and pressure symptoms. Hence a pelvic mass in MRKH syndrome has to be differentiated from ovarian fibroma, gastrointestinal stromal tumor (GIST), extravesical leiomyoma of urinary bladder and adnexal lesions. However, the possibility of fibroid should not be ruled out.

Functional ovaries in patient of MRKH syndrome predisposes them to ovarian pathology commonly being ovarian endometrioma which is a form of pelvic endometriosis wherein chocolate fluid containing cyst is present within ovary associated with decreased ovarian reserve. Prevalence of endometriosis in MRKH without functioning endometrial tissue is very rare. Various theories have been suggested for development of endometriosis with most accepted is theory of retrograde menstruation proposed by Sampson but in patient with non-functioning rudimentary uterine horn this theory fails to explain its occurrence. Another theory proposed by Meyer that coelomic membrane undergoes metaplasia forming typical endometrial like glands and stroma with evidence of histological finding of gradual transition from normal appearing ovarian surface epithelium and ovary epithelia inclusion to minimal formation of endometroid glandular epithelia and transition from normal ovarian stroma to endometriotic stroma supporting coelomic metaplasia in genesis of ovarian endometriosis⁵. This metaplastic process may involve both ovarian epithelial and stromal component. Role of steroid hormones, mainly estrogen is responsible for propagation and expansion of endometrium. Another theory proposed by Morris led to an alternative competing theory that metaplastic ovarian surface epithelium could be source of epithelial lining of chocolate cyst as the surface epithelium poses high intrinsic potential for Mullerian metaplasia and is more pronounced within the hormonal and chemical milieu of ovarian cyst.⁹

Endometriosis in pseudocyst formed by accumulation of menstrual debris from endometrial implants to peritoneal layers generates adhesion formation but local immunological system of peritoneal cavity plays a protective role with tremendous capability of resorption thus pelvic peritoneum shows slight adhesion formation.¹⁰

Treatment option for MRKH syndrome remain challenging the option for MRKH Syndrome related sexual dysfunction include non-surgical i.e. vaginal dilatation therapy for vaginal agenesis or surgical creation

of neovagina i.e. uterovaginoplasty. Reproductive options are limited like adoption. As ovarian development and function are usually unaffected, current option can be conception of biological children through gestational surrogacy or an experimental alternative of uterine transplantation to avoid ethical issue.

MRKH syndrome associated with gynaecological complication requiring total hysterectomy with bilateral salpingo-oophorectomy leading to iatrogenic premature or early menopause may need hormone replacement therapy. The reactivation or malignant transformation of residual endometriosis foci is main concern with use of HRT after pelvic clearance of endometriosis hence ESHRE recommends use of continuous combined HRT and avoidance of estrogen only HRT until the age of natural menopause.^{10,11}

CONCLUSION

Patient with Mullerian agenesis with pelvic pain or mass pose a diagnostic dilemma hence should be carefully evaluated. As congenital absence/hypoplastic uterus does not rule out gynaecological conditions. Diagnostic modalities like sonography with or without Doppler and MRI helps in confirming the diagnosis. Surgical management is the best treatment option along with reconstructive procedure depending on functional status of endometrium. Surrogacy is an option for patient desiring for biological parenthood.

ACKNOWLEDGEMENTS

The authors would like to thank the patient for forgiving consent to report her case. They would also like to acknowledge their colleagues who helped them make the diagnosis and the OT staff for capturing the images.

Funding: No funding sources

Conflict of interest: None declared

Ethical approval: Not required

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Cite this article as: Kumari A, Sah S, Mukherjee M, Anu D. A case of rudimentary uterine horns with cervical agenesis with intramural fibroid and bilateral endometrioma. Int J Reprod Contracept Obstet Gynecol 2025;14:3139-42.