

International Journal of Clinical Obstetrics and Gynaecology

ISSN (P): 2522-6614
ISSN (E): 2522-6622
© Gynaecology Journal
www.gynaecologyjournal.com
2022; 6(6): 01-03
Received: 01-08-2022
Accepted: 05-09-2022

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Primary amenorrhea with mullerian duct anomalies with hypoplastic uterus-mayer rokitansky kustner hauser syndrome a case report

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DOI: <https://doi.org/10.33545/gynae.2022.v6.i6a.1219>

Abstract

Aim: To report a case of Mayer rokitansky kustner hauser syndrome in the Government Maternity Hospital (Tertiary Health Care Center).

Materials and Methods: A 17-year-old resident of Pileru, unmarried presented to the gynecology OPD at government maternity hospital, Tirupati with complaints of not attaining menarche. On examination her vitals are stable, height 150cm, weight 48kgs, BMI 21.3 kg/m². Bilateral breast tanner stage 4, pubic hair tanner stage 4. Per abdomen soft, external genitalia healthy, per rectal examination not able to palpate uterus.

Keywords: Mayer rokitansky hauser kustner syndrome, tanner staging

Introduction

Mayer Rokitansky Hauser Kustner syndrome is a form of Mullerian agenesis and is a cause of Primary Amenorrhea. It is characterized by absence of uterus, cervix, and upper vagina. 25% will have short vagina pouch. Secondary sexual characters will be Normal. Karyotype is 46XX. It occurs in 1 in 5000 to 1 in 20000. In this the ovaries are normal as they are developed from ovarian ridge.

MRKH syndrome may be isolated [type 1] but more frequently associated with renal, vertebral, and cardiac defects [type 2]. Vaginal Aplasia is treated by creating a neo vagina. Counselling should be done to the patients and their family members. There is a possibility of Conception by donor oocyte and surrogacy.

Case Report

A 17-year-old female, resident of Pileru, Tirupati, studying class 12, reported to the gynecology OPD at government maternity hospital Tirupati, with complaints of not attaining menarche.

On physical examination patient was conscious and coherent. Pulse rate: 96 bpm, blood pressure 110/70mm of Hg, Temperature: 98.6F Respiratory Rate: 18/min.

The patient had investigations at 17 years of age with Normal hormonal studies. On Ultrasound uterus not visualized. Bilateral ovaries normal. CT abdomen and pelvis uterus not adequately visualized, ovaries normal, MRI pelvis -uterus not visualized, rudimentary thin structure 1.8*0.3 cm with no proper endometrial -mullerian duct anomaly with severe hypoplasia of uterus. Karyotyping done-46XX.

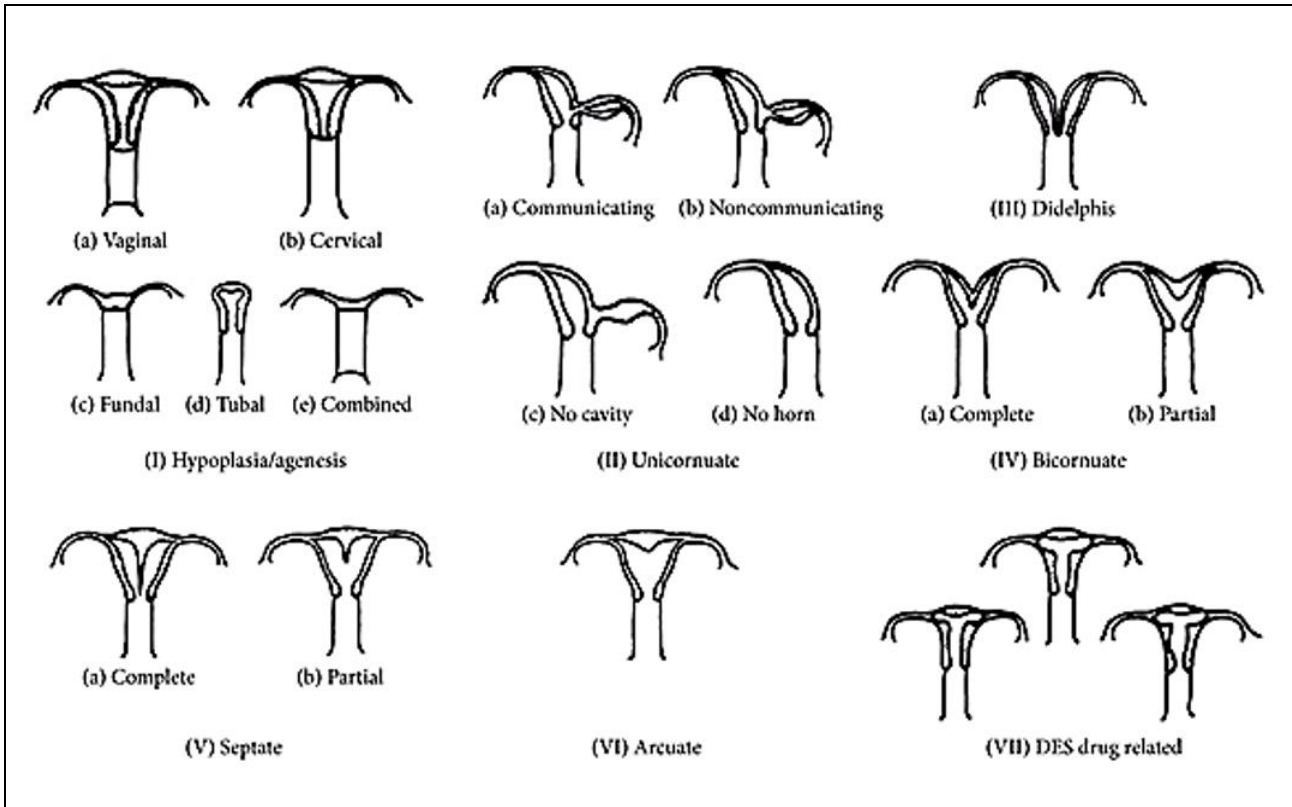
Case discussion

Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome is characterized by a physiological development of the secondary sexual characters and by a normal female karyotype 46 XX, but with a congenital aplasia of the uterus and of two-third superior parts of upper vagina [1]. Schematically, we may distinguish between a simple syndrome, of first type (I), and complex syndrome, of second type (II). In the second type, other associated malformations are found out Müllerian duct aplasia Renal Dysplasia and Cervical Somite anomalies (MURCS) with renal unilateral agenesis, renal ectopia, or horseshoed kidney [2]; skeletal alterations with a particular reference to vertebral anomalies with Klippel-Feil syndrome, melted vertebrae, and scoliosis [3]; anomalies of auditory system; only in some cases heart defects and syndactyly or polydactyly are associated with it Some studies [4,5] assume two different syndromes that are an isolated

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form of congenital agenesis of uterus and vagina and a more generalised condition, in which the agenesis of uterus and vagina is an important and specific feature within a more

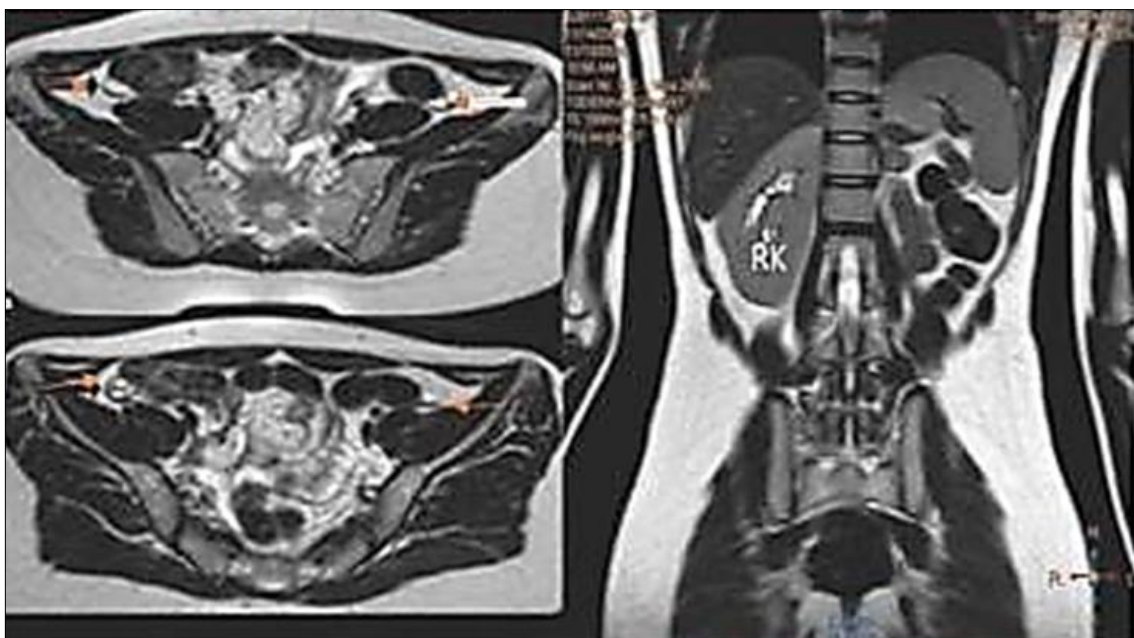
complex syndrome. Besides, atypical groups and other acronyms to indicate other associations of malformations, as genital-renal-ear-syndrome (GRES) may be taken into account.



Classification of anomalies of Mullerian duct developed by American Fertility Society (1988) and reproduced by Troiano and McCarthy (6)

To explain this condition, it has been suggested that in patients with MRKH syndrome, there is a very strong hyperincretion of Müllerian-inhibiting factor (MIF), which would provoke the lack of development of the Müllerian ducts from primitive structures (as what normally occurs in male phenotype). These

alterations are commonly associated with renal agenesis or ectopia. Specific mutations of several genes such as WT1, PAX2, HOXA7-HOXA13, PBX1, and WNT4 involved in the earliest stages of embryonic development could play a key role in the etiopathogenesis of this syndrome. Besides, it seems that the other two genes, TCF2 (HNF1B) and LHX1, are involved in the determinism of this pathology.



Absents uterus with ovaries

Right kidney with renal agenesis



Empty Pelvic fossa

Conclusion

MRKH syndrome is one of the most common causes of primary amenorrhea and ultrasound is useful for diagnosing any associated renal anomalies. MRI is more precise than USG and less invasive and expensive than laparoscopy. This condition has psychologically devastating consequences but now anatomical defects can be corrected surgically by assisted techniques. So correct evaluation of the patient and proper management is mandatory.

Conflict of Interest

Not available

Financial Support

Not available

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How to Cite This Article

Vasudha S and Sreelatha. Primary amenorrhea with müllerian duct anomalies with hypoplastic uterus-mayer rokitansky kustner hauser syndrome a case report. *International Journal of Clinical Obstetrics and Gynaecology*. 2022;6(6):01-03.

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