ANDALAS OBSTETRICS AND GYNECOLOGY JOURNAL
Address for Correspondence:
Editorial Room Andalas Obstetrics and Gynecology Journal, 3rd floor of KSM of Obstetrics and Gynecology, RSUP DR. M. Djamil Padang, Jl. Perintis Kemerdekaan Padang, Sumatera Barat 25127

eISSN : 2579-8324 pISSN : 2579-8323

Website:
http://jurnalobgin.fk.unand.ac.id/index.php/JOE

CASE REPORT

Mayer-Rokitansky-Küster-Hauser syndrome

Arga Kafi Perdana Kusuma1, Bobby Indra Utama2

1. Obstetrics and Gynecology Department, Faculty of Medicine, Andalas University, Dr. M. Djamil Central General Hospital Padang, West Sumatera, Indonesia

2. Bobby Indra Utama, Division of Urogynecology and Reconstructure Aesthetic, Department of Obstetrics and Gynecology, RSUP Dr M. Djamil, Unversitas Andalas, Padang, Indonesia

Corespondence: *Arga Kafi Perdana Kusuma, email : argakafiperdana@gmail.com*

Abstract

Mayer–Rokitansky–Küster–Hauser syndrome (MRKH) is a syndrome characterized by uterine, cervix, and the two third of upper vagina aplasia which is the cause of incomplete development of the Müllerian duct. Reported a case of woman 28 years old, patient has not menstruated until now. The phenotype of the patient appears to be female, and with normal stature. Breast, axilla dan pubic hair distribution, fatty in buttocks and thigh developed normally. Fallopian tubes, uterine and 2/3 upper part of vagina were not formed. On gynecological clinical examination, found vaginal introitus with a vaginal sonde was 2 cm. On abdominal ultrasound examination, the uterus was seen as a line, right ovary measuring 2.93 x 2.59 cm and left ovary measuring 2.52 x 2.28 cm. The patient then underwent a diagnostic laparoscopic procedure, and found both right and left ovaries within normal limits, intact tubes, but no uterus was visible. Patient are planned for vaginoplasty.
Keywords: *MRKH syndrome, mullerian duct anomalies, vaginoplasty*

ANDALAS OBSTETRICS AND GYNECOLOGY JOURNAL
eISSN : 2579-8324 pISSN : 2579-8323
Address for Correspondence:
Editorial Room Andalas Obstetrics and Gynecology Journal, 3rd floor of KSM of Obstetrics and Gynecology, RSUP DR. M. Djamil Padang, Jl. Perintis Kemerdekaan Padang, Sumatera Barat 25127
Website:
http://jurnalobgin.fk.unand.ac.id/index.php/JOE

INTRODUCTION

MRKH (Mayer – Rokitansky – Küster -Hauser) syndrome is a syndrome characterized by aplasia of the uterus, cervix and upper vagina originating from the Mullerian ducts but the ovaries function normally. 1,2,3,4 The etiology of MRKH syndrome is unclear. However, embryologically there is a developmental disorder of the Mullerian duct fusion at the eighth week of pregnancy. The incidence is about 1:4000 and 1:5000.5

Clinical symptoms of MRKH syndrome are usually marked by no menstruation (primary amenorrhea) but with normal thelarche and adrenarche, sexual intercourse disorders and infertility. In general, amenorrhea is divided into 2, namely physiological amenorrhea (pre puberty, pregnancy, lactation, post menopause) and pathological amenorrhea consisting of primary amenorrhea and secondary amenorrhea. Primary amenorrhea is that until the age of 14 years you have not experienced menstruation accompanied by the development of secondary sex signs or until the age of 16 you have not experienced menstruation, but the secondary sex characteristics develop normally. 3 According to WHO, there are 3 groups that cause amenorrhea, namely group I, namely the hypothalamus-pituitary, group 2 is Polycystic Ovary Syndrome (PCOS), group 3 is gonadal failure

MRKH classification is divided into 3 types: Type I, where the reproductive organs have abnormalities (utero vaginal aplasia). Type II presents with asymmetric utero-vaginal aplasia or hypoplasia, with hypoplasia or absence of one or both fallopian tubes and the presence of malformations of the ovaries and/or the renal system. Type III or MURCS (Mullerian duct aplasia, Renal dysplasia, and Cervical Somite Anomalies), the most commonly associated anomalies are urological (15-40% of cases) and skeletal anomalies (20-40%) while hearing, cardiac, and digital defects (syndactyly or polydactyly) is rare

Physical examination found the following: secondary sexual characteristics after normal puberty and normal height. Speculum examination of the vagina can be difficult because of the degree of vaginal agenesis. Vulva, labia majora, labia minor and clitoris within normal limits. Laboratory studies include: chromosomal analysis to rule out X chromosome karyotype abnormalities and Androgen Insensitivity Syndrome (AIS). 8 Diagnosis can be made using ultrasound, MRI, Laparoscopy, and Pyelography. MRI examination is the gold standard in this case because it produces excellent images of the superficial and deep tissue and can clarify inconclusive ultrasound results related to uterine cavitation.

Treatment of vaginal agenesis can be done with non-surgical and surgical techniques. The Frank technique is a non-surgical technique that is performed by pressing on the dilatation of the vaginal cavity for a certain period of time, requiring patience and discipline for the patient to do it. Meanwhile, surgical techniques can be performed using the McIndoe technique, Williams Vaginoplasty, Flap Rotation procedure, Intestinal Neovagina technique, and the Vecchietti technique. Based on the literature, the cause in this case is thought to be the result of aplasia or failure of the fusion of the mullerian and wolfii ducts.

ANDALAS OBSTETRICS AND GYNECOLOGY JOURNAL
eISSN : 2579-8324 pISSN : 2579-8323
Address for Correspondence:
Editorial Room Andalas Obstetrics and Gynecology Journal, 3rd floor of KSM of Obstetrics and Gynecology, RSUP DR. M. Djamil Padang, Jl. Perintis Kemerdekaan Padang, Sumatera Barat 25127
Website:
http://jurnalobgin.fk.unand.ac.id/index.php/JOE

CASE REPORT

1. History and Physical Examination

A 28 year old woman came to the Obstetrics and Gynecology Polyclinic at RSUP Dr. M. Djamil Padang with complaints of not menstruating even though the patient is 28 years old. The patient never went to the doctor because the complaint was considered normal. The patient began to see a doctor when she was 20 years old, the results of an ultrasound examination showed uterine hypoplasia. Then the patient is given progesterone medication but the complaints are still felt, the patient does not experience menstruation. The patient returned to visit another obstetrician and was told that her uterus and vagina were not fully formed. The patient was recommended for surgery but the patient refused due to fear so he did not continue treatment anymore. Eight years later, the patient began to venture back into treatment. The patient never felt monthly abdominal pain.

No complaints about a lump in the stomach, decreased appetite and weight, urinary or defecation disorders. The patient also did not complain of any disturbance of daily activities. Phenotypically, the patient appeared to be a woman, had normal height, and had a body like women in general. Examination results of vital signs within normal limits. Head and neck examination did not reveal a webbed neck or facial abnormalities. The results of physical examination of the lungs, heart and abdomen were within normal limits. The four extremities also did not show any abnormalities such as polydactyly, syndactyly, or the absence of fingers. The patient's breasts were well developed (Tanner stage 4) and had normal external genital organs, clitoris, labia majora and labia minor within normal limits. Distribution of axillary and pubic hair growth was also within normal limits (Tanner stage 4). In addition, there is also fat growth on the buttocks and thighs.

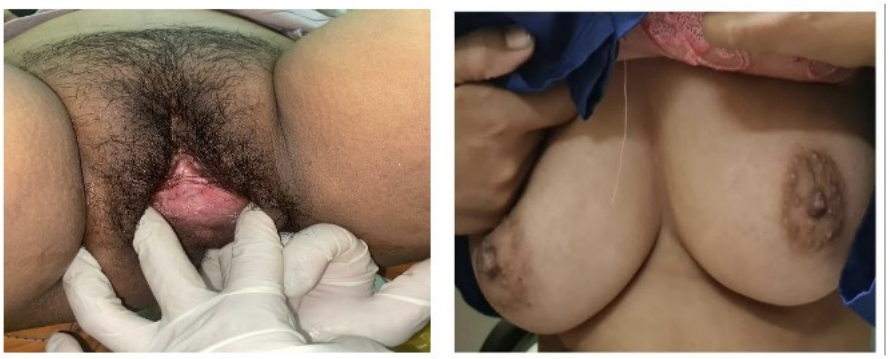

Figure 1. Physical appearance of the patient

Gynecological examination, on clinical gynecological examination, inspection of the vulva and urethra was within normal limits, the vaginal introitus was visible with a 2 cm long vaginal probe. On rectal toucher examination, the anal sphincter was normal, the ampulla was not collapsed, adnexal masses were not palpable, and the uterus was not palpable. The patient also feels that he is a woman completely.

eISSN : 2579-8324 pISSN : 2579-8323

ANDALAS OBSTETRICS AND GYNECOLOGY JOURNAL

Address for Correspondence:
Editorial Room Andalas Obstetrics and Gynecology Journal, 3rd floor of KSM of Obstetrics and Gynecology, RSUP DR. M. Djamil Padang, Jl. Perintis Kemerdekaan Padang, Sumatera Barat 25127

Website:
http://jurnalobgin.fk.unand.ac.id/index.php/JOE

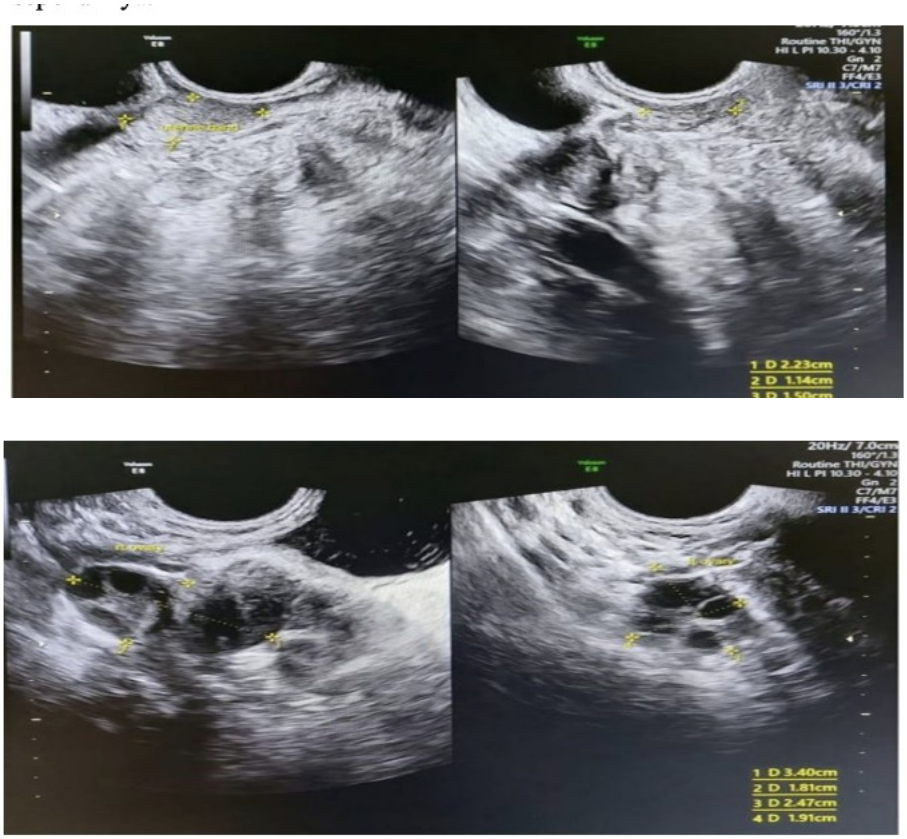

Figure 2. Ultrasound Examination Results

Diagnostic Evaluation

Transabdominal ultrasound examination showed results showing the uterus was only a line, the right ovary measuring 2.93 x 2.59 cm and the left ovary measuring 2.52 x 2.28 cm.

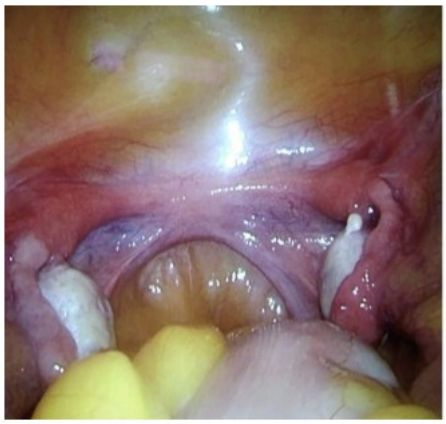

Figure 3. Laparoscopic Examination Results

Laparoscopic examination showed both ovaries and tubes within normal limits, but no uterus was seen. The patient was diagnosed with primary amenorrhea et causa Mayer–Rokitansky–Küster–Hauser (MRKH) syndrome. The patient was then planned for vaginoplasty.

ANDALAS OBSTETRICS AND GYNECOLOGY JOURNAL
Address for Correspondence:
Editorial Room Andalas Obstetrics and Gynecology Journal, 3rd floor of KSM of Obstetrics and Gynecology, RSUP DR. M. Djamil Padang, Jl. Perintis Kemerdekaan Padang, Sumatera Barat 25127
eISSN : 2579-8324 pISSN : 2579-8323
Website:
http://jurnalobgin.fk.unand.ac.id/index.php/JOE

DISCUSSION

The clinical symptoms complained of in this patient include not having menstruation until the age of 20 which shows a sign of amenorrhea, namely the absence of menstruation in a woman which includes one of the following 3 signs: No menstruation until the age of 14 years, accompanied by no growth or development of secondary sex characteristics; (2) Menstruation does not occur until the age of 16 years, accompanied by normal growth and development of secondary sex characteristics; (3) Menstruation has not occurred for at least 3 consecutive months in a woman who has had menstruation before. This patient is classified as suffering from primary amenorrhea, namely amenorrhea that occurs before menarche which is the first menstruation experienced by women. Menarche occurs at an average age of 13 years. Primary amenorrhea that occurs in these patients can be caused by various things, evaluation of the causes of amenorrhea is carried out based on the division of four compartments, namely disorders of the uterus and patency (outflow tact), disorders of the ovaries, disorders of the pituitary, and disorders of the hypothalamus / central nervous system.

The results of the overall physical examination in this patient showed secondary sexual physical growth which included breast growth (thelarche) and changes in axillary and pubic hair (adrenarche or pubarche). This physical growth occurs as a result of increased adrenal androgen production and occurs on average at the age of 7-8 years. These patients are also accompanied by changes in body shape, with accumulation of fat in the thighs, hips, and buttocks. In this case estrogen increases total body fat which is distributed in the thighs, buttocks, and abdomen. The physical changes associated with puberty occur sequentially, any deviation from the sequence or timing of events can be considered as an abnormality.

Meanwhile, from the results of the gynecological physical examination of the genital organs, the vulva and urethra were within normal limits, the vaginal introitus with a vaginal sonde was 2 cm long, and the uterus was not palpable. This suggests an anomaly in the uterus, cervix, and vagina. Anomalies of the female genitalia can be caused by several mechanisms, namely canalization, agenesis, fusion, and embryonic rests. Anomalies in the female genital organs are caused by defects in the lateral and vertical fusion processes of the urogenital sinus and the Mullerian duct. The process of fusion (merging) of the right and left Mullerian ducts will be completed at 12 weeks of gestation. Meanwhile, the canalization process will be completed at 5 months of gestation. Failure of vertical fusion between the mullerian ducts and the urogenital sinus will cause abnormalities in the canalization of the genital organs. Furthermore, failure to perform lateral fusion will result in organ duplication. Impaired resorption will result in the formation of a septum. In this case, 2 cm of uterus, cervix, and vagina were not found which is classified as Muelleri duct hypoplasia or agenesis. Failure to form will result in the genital organs not being formed at all. If both Mullerian ducts are involved, there will be no uterus, the two Fallopian tubes, and the upper third of the vagina. 10 This is consistent with the theory that patients with MRKH syndrome complain of never having menstruation (primary amenorrhea) due to abnormal vaginal aplasia or absence of a uterus. 11,12 In accordance with the theory that the ovaries continue to function normally so that there is secondary sexual growth like women in general. the diagnosis of this disease is enforced at the age of 10-18 years

ANDALAS OBSTETRICS AND GYNECOLOGY JOURNAL
Address for Correspondence:
Editorial Room Andalas Obstetrics and Gynecology Journal, 3rd floor of KSM of Obstetrics and Gynecology, RSUP DR. M. Djamil Padang, Jl. Perintis Kemerdekaan Padang, Sumatera Barat 25127
eISSN : 2579-8324 pISSN : 2579-8323
Website:
http://jurnalobgin.fk.unand.ac.id/index.php/JOE

Supporting examinations in the form of ultrasound imaging and diagnostic laparoscopy have been carried out in this patient, the results obtained were that the uterus was not found, both ovaries were within normal limits, and other organs were within normal limits. Such supporting examinations are necessary to enable determination of the anatomical characteristics of this syndrome. Ultrasound is the initial method of choice. This method can show the absence of the uterus between the bladder. Ultrasound, CT-scan and MRI examinations that have been carried out in this patient support the diagnosis of MRKH syndrome. 14 The overall physical examination and other supporting examinations can determine the type of MRKH syndrome. Based on the examination that has been carried out, no abnormalities such as urological, skeletal, hearing defects, cardiac, and digitalis (syndactyly or polydactyly) are found, thus indicating MRKH syndrome type 1.13

The treatment for mullerian duct anomalies is chosen based on the class of mullerian duct anomalies. According to the American Fertility Society (AFS), MRKH syndrome is a class Ic mullerian duct anomaly based on the presence of a mullerian aplasia. Ren

CONCLUSION

MRKH (Mayer-Rokitansky-Küster-Hauser) syndrome is a syndrome characterized by aplasia of the uterus, cervix and upper vagina originating from the mullerian ducts but the ovaries function normally. The incidence of MRKH syndrome is very rare, varying between 1:4000 and 1:5000. The diagnosis of MRKH syndrome was established from the results of anamnesis with primary amenorrhea, physical examination did not find the uterus, cervix and upper 2/3 of the vagina but found signs of secondary sexual growth as in women in general and from the results of imaging with ultrasound and diagnostic laparoscopy no uterus was found, but with both ovaries and fallopian tubes and other organs within normal limits. This patient was then decided to undergo vaginoplasty action

REFERENCES

1. Sultan C, Lauber AB, Philibert P. Mayer- Rokitansky-Kuster-Hauser Syndrome: Recent Clinical and Genetic Findings. Journal Gynecological Endocrinology. 2009;25(1):8-11.

2. Morcel K, Camborieux L, Guerrier D. Mayer- Rokitansky-Kuster-Hauser (MRKH) Syndrome. Orphanet Journal of Rare Diseases. 2007;2(13):1- 9.

3. Wiyasa A, Mukminah A. Penatalaksanaan Sindroma Mayer Rokitansky Kuster Hauser (Sindroma MRKH) Dengan Sindroma Klippel Feil. Jurnal Kedokteran Brawijaya. 2004;20(2):92-95.

4. Ekici A, Strissel P, Oppelt P. HOXA10 and HOXA13 Sequence Variations in Human Female Genital Malformations Including Congenital Absence of The Uterus and Vagina. Gene 2013. 2013;518(2):267-272.

eISSN : 2579-8324 pISSN : 2579-8323

ANDALAS OBSTETRICS AND GYNECOLOGY JOURNAL

Address for Correspondence:
Editorial Room Andalas Obstetrics and Gynecology Journal, 3rd floor of KSM of Obstetrics and Gynecology, RSUP DR. M. Djamil Padang, Jl. Perintis Kemerdekaan Padang, Sumatera Barat 25127

Website:
http://jurnalobgin.fk.unand.ac.id/index.php/JOE

5. Guerrier D, Mouchel T, Pasquier L, Pellerin I. The Mayer-Rokitansky-Küster-Hauser syndrome (congenital absence of uterus and vagina) – phenotypic manifestations and genetic approaches. Journal of Negative Results in Biomedicine. 2006;5:1-1.

6. Oppelt P, Renner S, Kellermann A. Clinical Aspects of Mayer–Rokitansky–Kuester–Hauser Syndrome: Recommendations for Clinical Diagnosis and Staging. Human Reproduction. 2006;21(3):792-797.

7. Hendarto H.Gangguan Haid/ Perdarahan Uterus Abnormal.In:Anwar M,Baziad A,Prabowo RP, editors. Ilmu Kandungan. 3th ed. Jakarta: Bina Pustaka Sarwono Prawirohardjo;2011.p.161-183.

8. Paraton H. Embriologi Sistem Alat-alat Urogenital. In: Anwar M, Baziad A, Prabowo RP, editors. Ilmu Kandungan. 3th ed. Jakarta: Bina Pustaka Sarwono Prawirohardjo; 2011.p.33-46.

9. Dhaheri SA, Kaabi JA, Aswad S, Souid AK, Langer RD. Mullerian Agenesis: Presenting As An Isolated Rudimentary Uterus. Oman Medical Journal. 2014;29(5):1-2.

10. Gari A. McIndoe Neovagina in Patients With Mullerian Agenesis: A Sigle Center Experience. Pak Journal Med Science. 2017;33(1):236-240.

11. Yakasai I, Daneji S, Natasha A, Yunus E. Late Presentation of Mayer-Rokitansky-Kuster-Hauser Syndrome in The Tropics-Northern Nigeria. British Journal of Medicine and Medical Research. 2015;5(8):1077-1082.

12. Medical and Science. 2014;2(1):106-108.

13. Pizzo A, Laganà AS, Sturlese E, Retto G, Retto A, Dominici RD, et al. Mayer-Rokitansky-Kuster

14. Hauser Syndrome: Embryology, Genetics and Clinical and Surgical Treatment. Obstetrics and Gynecology. ;2013:1-10