Manipal Journal of Nursing and Health Sciences

Volume 7 | Issue 1 Article 10

August 2021

Mayer Rokitansky Kuster Hauser syndrome: A case of Mullerian Agenesis

Anu Baby
Manchester University NHS Foundation Trust ,United Kingdom., babyanu1988@gmail.com

Simi Kurian
Department of Obstetrics and Gynecology Government Medical College Kannur, Kerala, drsimikurian@gmail.com

Rani Jose
Deparment of Medical and Surgical Nursing, Government Nursing College, Kannur, Kerala, India, ranijbinu@gmail.com

Case report

Mayer Rokitansky Kuster Hauser syndrome: A case of Mullerian Agenesis

Anu Baby*, Simi Kurian, Rani Jose

Email: babyanu1988@gmail.com

Abstract

Developmental anomalies of the Mullerian duct are one of the fascinating congenital disorders encountered in which Mayer Rokitansky Kuster Hauser syndrome (MRKH) is one of the wide variety of malformations. The most common presentation in MRKH syndrome is primary amenorrhea with normal development of secondary sexual characteristics and normal female karyotype (46, XX). The ovaries and fallopian tubes are usually functional, but the uterus and upper two-third vagina are either underdeveloped or absent. MRKH syndrome can either be an isolated utero-vaginal aplasia (Type I) or associated with extragenital anomalies (Type II). A case of Type I MRKH syndrome is reported here.

Keywords: Amenorrhea, Mullerian agenesis, Mullerian duct, MRKH

Introduction

Puberty is a time when a girl grows both physically and emotionally from a child into a teenager and eventually into an adult. The major landmark of puberty for females is menarche and the conclusion of puberty is reproductive maturity. Mullerian agenesis or MRKH is a congenital malformation characterized by failure of the Mullerian ducts to develop during embryogenesis. The estimated prevalence is one in 4000-5000 live births (Naem, Shamandi, & AL-Kurdy, 2020). Primary amenorrhea due to aplasia of the uterus and upper two-thirds of the vagina forms the most typical presenting feature. Most adolescents with MRKH have normal female karyotype (46, XX) (Londra, Chuong, & Kolp, 2015). Two types of MRKH have been identified- MRKH Type I (typical) and Type II (atypical). The atypical type is also associated with renal, vertebral, auditory, or cardiac defects. Treatment options for vaginal agenesis include surgical creation of neovagina and vaginal dilation. Surrogacy, adoption, and uterus transplantation provide motherhood options for women with MRKH (Herlin, Petersen, & Brännström, 2020). This condition is named MRKH after August Franz Joseph Karl Mayer, Carl Freiherr von Rokitansky, Hermann Kuster, and G A Hauser.

Anu Baby[1], Simi Kurian[2], Rani Jose[3]

[1] *Registered Nurse, Manchester University NHS Foundation Trust ,United Kingdom.*

[2] *Associate Professor, Dept of Obstetrics and Gynaecology, Government Medical College, Kannur, Kerala, India*

[3] *Professor, Dept of Medical and Surgical Nursing, Government Nursing College, Kannur, Kerala, India*

Manuscript received: 22 June 2020

Revision accepted: 25 January 2021

*Corresponding Author

Case report

A 19-year-old female presented with primary amenorrhea and normal secondary sexual characteristics. She had no complaints of any cyclic abdominal pain. General physical examination findings were normal. Her genitalia examination revealed normal major and minor labia, normal pubic hair development, and external urethral meatus. The vaginal examination was not performed on her initially as she was a young female. An endocrine panel was run and her Luteinising Hormone (LH), Follicle Stimulating Hormone (FSH), prolactin, estradiol, progesterone, and testosterone levels were found to be within normal limits. The thyroid function test showed no abnormalities.

A sonographic study of the pelvis reported a hypoplastic uterus with normal-sized ovaries and fallopian tubes.

How to cite this article: Baby, A., Kurian, S & Jose, R. (2021). Mayer Rokitansky Kuster Hauser syndrome: A case of Mullerian Agenesis. *Manipal Journal of Nursing and Health Sciences*, 7 (1). 59-61.

She was put on Primolut-N (norethisterone) 5 mg, once daily, for a week, and no positive response was obtained. Therefore, she was sent for a repeat ultrasound scanning and was advised to have a filled bladder before undergoing the test. Special focus was made on bladder filling as she verbalized doubts of not having done so during her initial scan. The second imaging study showed normal ovaries and adnexa but rudimentary uterus. A streak-like structure measuring 33.1 x 9.4 x 14.7 mm with poor myoendometrial differentiation was identified in the normal position of the uterus. Her vaginal examination revealed a 2.5 cm long blind pouch. Diagnostic laparoscopy also confirmed the diagnosis of Mullerian agenesis. The left ovary could not be visualized due to obliteration of the left half of the pelvis by the sigmoid colon. The right ovary was found to be normal in size and had cysts.

A detailed review of the history stated no known cases of MRKH or any other gynaecological malformations in her family. There was no history of delayed menarche in the family. Her mother confirmed no known exposure to any medication or maternal illness during pregnancy. Chromosomal analysis revealed a female karyotype with a pericentric inversion of chromosome 9- 46, XX, inv(9) (p11q13). This pericentric inversion, however, is so common that cytogeneticists consider them as a normal variant. The incidence is said to be about 1% to 3% in the general population and is often familial (Muthuvel, Ravindran, Chander, & Subbian, 2016). She was given therapeutic counselling and vaginoplasty was recommended.

Discussion

MRKH results due to malformation of the Mullerian ducts which starts developing around 5th-6th gestational weeks. The uterus, fallopian tube, cervix, and upper two-thirds of the vagina originate from the Mullerian duct or the paramesonephric duct, and the lower part of the uterus origins from the urogenital sinus. The upper part of the Mullerian duct forms the two fallopian tubes whereas the caudal part of the two ducts fuse to form the uterus, cervix, and upper two-thirds of the vagina (Herlin et al., 2020).

Most cases of MRKH have an absent uterus and agenesis of the upper two-thirds of the vagina. Despite the absence of a uterus, there might be the presence of rudimentary uterine anlagen as we have seen in this case report. MRKH is the second most common cause of primary amenorrhea following gonadal dysgenesis (Al Dandan, Hassan, Alsaihati, Aljawad, & Almejhim, 2019). Females with this condition usually show normal development of secondary sexual characteristics, normal endocrine function, and have a normal chromosomal constellation (46 XX). Other reproductive structures (fallopian tubes and ovaries) and external genitalia are often normal. Typical MRKH (Type I) is characterized by an absent or rudimentary uterus and vagina with normal tubes and ovaries. Atypical (Type II) ones may also have renal (40%) and spinal anomalies (30-40%) more often, and hearing (10-25%) and cardiac problems less frequently (Morcel, Camborieux, & Guerrier, 2007). The atypical variant may also show variance in ovaries and fallopian tubes.

The etiology of MRKH is still unclear. Most cases of MRKH are seen with no family history and affected cases usually have 46 XX karyotype. Therefore, a genetic cause for this condition cannot be established clearly. However, few cases have demonstrated an autosomal dominant pattern of inheritance of this condition (Jain & Kamra, 2018). Decreased activity of Galactose-1- phosphate uridyl transferase enzyme (GALT) is also a proposed biological cause for vaginal agenesis (Gupta & Ansari, 2002). Sonography, MRI, and laparoscopy are the recommended tests for detecting these abnormalities. Although ultrasound is often the first diagnostic test, the results can be inconclusive. MRI is the recommended modality of the test as it can confirm the diagnosis and detect any other associated anomalies (Al Dandan et al., 2019).

Management includes intensive psychological counselling to both patients and their families. Vaginoplasty and vaginal dilation can be performed to allow for satisfactory sexual intercourse. A neovagina can be created using non-surgical methods (dilators) if at least a vaginal dimple is present and in case of its absence, several surgical measures are available (Jain & Kamra, 2018). Adoption and surrogacy are recommended for those wishing to have children. As ovulation usually occurs among women with Mullerian

agenesis, in vitro fertilization and surrogacy can be opted by those who desire their genetic offspring. The uterine transplant has now emerged as a fertility treatment modality for women with MRKH.

Conclusion

Patients with MRKH experience womanhood differently. The overwhelming amount of stress produced by MRKH often trounces one's coping ability creating damage to the psyche of the adolescents and younger adults already trying to meddle through the turbulence and perturbation lashing in their trail to the calm shore of adulthood. Preparing and strengthening patients and their families right from the period of diagnosis would help them in managing emotional and behavioural turmoil.

Sources of support: None
Conflict of interest: None declared
Source of support in the form of grants: None

References

Al Dandan, O., Hassan, A., Alsaihati, A., Aljawad, L., & Almejhim, F. (2019). A rare form of Mayer-Rokitansky-Küster-Hauser syndrome: Case report and review of literature. Case reports in women's health, 24, e00137. https://doi.org/10.1016/j.crwh.2019.e00137

Gupta, N. P., & Ansari, M. S. (2002). Mayer-Rokitansky-Kuster-Hauser (MRKH) syndrome - A review. Indian Journal of Urology. 18(2); 111-6. Available from: https://www.indianjurol.com/text.asp?2002/18/2/111/37398

Herlin, M. K., Petersen, M. B., & Brännström, M. (2020). Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome: A comprehensive update. Orphanet Journal of Rare Diseases,15; 214. https://doi.org/10.1186/s13023-020-01491-9

Jain, N., & Kamra, J. H. (2018). MRKH syndrome: a review of literature. International Journal of Reproduction, Contraception, Obstetrics and Gynecology, 7(12), 5219. https://doi.org/10.18203/2320-1770.ijrcog20184999

Londra, L., Chuong, F. S., & Kolp, L. (2015). Mayer-Rokitansky-Kuster-Hauser syndrome: a review. International journal of women's health, 7, 865–870. https://doi.org/10.2147/IJWH.S75637

Morcel, K., Camborieux, L., Programme de Recherches sur les Aplasies Müllériennes, & Guerrier, D. (2007). Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome. Orphanet journal of rare diseases, 2, 13. https://doi.org/10.1186/1750-1172-2-13

Muthuvel, A., Ravindran, M., Chander, A., & Subbian, C. (2016). Pericentric inversion of chromosome 9 causing infertility and subsequent successful in vitro fertilization. Nigerian medical journal: Journal of the Nigeria Medical Association, 57(2), 142–144. https://doi.org/10.4103/0300-1652.182080

Naem, A., Shamandi, A., & AL-Kurdy, B. (2020). A rare form of Mayer–Rokitansky–Küster–Hauser syndrome associated with ovarian endometrioma: a case report. Journal of Surgical Case Reports; 9, 393 https://doi.org/10.1093/jscr/rjaa393