CASO CLÍNICO

1. Hospital Nacional Docente Madre Niño HONADOMANI San Bartolomé, Lima, Perú
2. Facultad de Medicina San Fernando, Universidad Nacional Mayor de San Marcos, Lima, Perú
   a. Resident Physician of Gynecology-Obstetrics ORCID 0000-0001-8739-0491
   b. Medical Epidemiologist ORCID 0000-0003-0976-906
   c. Head of the Department of Gynecology-Obstetrics, Associate Professor ORCID 0000-0003-0709-8015

Funding Source: Study conducted with the authors' own resources.

Conflict of interest: The authors declare that they have no conflict of interest.

No technology related to artificial intelligence has been used in this article.

Received: 24 June 2024

Accepted: 3 September 2024

Online publication: 14 December 2024

Correspondencia:
Darwin Ayala Céspedes
Av. Alfonso Ugarte 825, Lima 1, Perú
976943858
Dayalac90@gmail.com

Cite as: Ayala Céspedes D, Cvetkovic-Vega A, Ramírez Cabrera J. Mayer-Rokitansky-Küster-Hauser syndrome: report of two cases. Rev peru ginecol obstet. 2024;70(4). Doi: https://doi.org/10.31403/rpgo.v70i2699

Mayer-Rokitansky-Küster-Hauser syndrome: report of two cases

Síndrome de Mayer-Rokitansky-Küster-Hauser: comunicación de dos casos

Darwin Ayala Céspedes[1,a], Aleksandar Cvetkovic-Vega[1,b], Juan Ramírez Cabrera[1,2,c]

DOI: https://doi.org/10.31403/rpgo.v70i2699

ABSTRACT

Mayer-Rokitansky-Küster-Hauser syndrome (SMRKH) is a congenital pathology characterized by aplasia or agenesis of structures that develop from the mullerian ducts in genotypically and phenotypically normal females. The prevalence of this syndrome is uncertain. There are only two population-based studies evaluating it. We present two clinical cases, in an adolescent and older adult woman. These patients require clinical, epidemiological, hormonal and imaging studies by a multidisciplinary team that includes a psychologist to support the patient's acceptance of the situation, as well as considerations related to subsequent surgeries and reproductive consequences.

Key words: Disorders of sexual development, Paramesonephric ducts

RESUMEN

El síndrome de Mayer-Rokitansky-Küster-Hauser (SMRKH) es una patología congénita caracterizada por aplasia o agenesia de las estructuras que se desarrollan en los conductos mullerianos en mujeres genotípica y fenotípicamente normales. La prevalencia de este síndrome es incierta. Existen solo dos estudios poblacionales que la evalúan. Presentamos dos casos clínicos, en una adolescente y en una adulta mayor. Estas pacientes requieren estudios clínicos, epidemiológicos, hormonales e imagenológicos por un equipo multidisciplinario que incluya un psicólogo para apoyar a la paciente aceptar la situación, así como los planteamientos relacionados a cirugías posteriores y las consecuencias en la reproducción.

Palabras clave. Trastornos del desarrollo sexual, Conductos paramesonéfricos

Introduction

Mayer-Rokitansky-Küster-Hauser syndrome (MRKHS) is a congenital pathology characterized by aplasia or agenesis of developing structures of the mullerian ducts in genotypically and phenotypically normal females[1]. Its etiology is unknown. Studies conducted in family groups suggest the presence of a first-degree genetic component[2]. Although this genetic component has not been extensively investigated[3], the most studied genes correspond to HOXA and WNT4, which are involved in dysregulation of the activation of the mullerian inhibitory substance[4].

The prevalence of this syndrome is uncertain. There are only two population-based studies evaluating it. The first, carried out between 1953 and 1957, reports one case in every 10,558 female births, and the second, from Finland, reports one case in 4,961 births[5]. In Peru there are no prevalence studies of the syndrome, but a series of 6 cases and a review of the literature was published in 2000[6].

Regarding the classification of MRKHS, there is type I which is mainly characterized by utero-vaginal aplasia, and type II is related to extragenital abnormalities which, in most cases, are renal (30%-40%), as well as skeletal, atrial or cardiac abnormalities in a lower percentage[3,7].

We present two clinical cases of MRKHS, one in a 16-year-old adolescent girl and the other in a 44-year-old female patient, with similar clinical findings and who were referred to the Hospital Nacional Docente Madre Niño San Bartolomé.

Case 1

A 16-year-old nulliparous patient from Huancavelica, in the central-western highlands of Peru, with no menarche, reported chronic pelvic pain in 28-30-day cycles since the onset of puberty 4 years earlier. The pain was colicky, of 8/10 intensity, which decreased with analgesics. On physical examination she presented moderate intensity pain on deep palpation in the hypogastrium with no signs of peritoneal irritation. Breast development was Tanner 4, and the distribution of pubic hair was Tanner 3. External genitalia revealed absence of continuity of the vaginal introitus (Figure 1), hymen of normal characteristics, permeable urethra and vulva with unaltered labia majora and labia minora.

The patient was hospitalized for studies. The dosage of hormones (TSH, T3, T4, LH, FSH, estradiol, prolactin and testosterone) was within normal parameters. Imaging tests were requested and displayed no renal or abdominal abnormalities. Pelvic ultrasound showed a predominantly solid mixed tumor measuring 57 x 33 x 35 mm in the right adnexa (Figure 2) and the right ovary measuring 30 x 19 x 20 mm displaced to the left, with no visualization of the left ovary. Magnetic resonance imaging (MRI) presented the uterus of normal shape and size in intermediate position, the cavity of 10.4 cm distended by fluid, and small Nabothian cysts on both sides of the midline smaller than 8 mm. The conclusions were uterus in intermediate position with serometra and alteration of cervical morphology. The karyotype study showed no chromosomal alterations (46,XX).

Examination under anesthesia was performed, in which the labia majora and minora, urethra and hymen were of normal characteristics but without hymenal orifice. The cervix could not be palpated by rectal examination, the uterine body of 4 x 4 cm was lateralized to the right. Diagnostic laparoscopy showed the presence of a uterine sketch of 4 x 4 x 4 cm, absence of the left horn (Figure 3), tube and right ovary of 3 x 2 x 1 cm with a paratubal Gardner cyst of 2 x 2 cm, left tube elongated and hypotrophic in its proximal third, adhered in its middle third to the pelvic wall, and presence of a Gardner's cyst adhered to the left tube and ovary, 3 x 2 x 1 cm, of normal appearance. After a favorable postoperative period, she underwent laparotomy 5 days later to remove the rudimentary right hemiuterus distended by hematometra, with bilateral salpingectomy. Agenesis of the left hemiuterus was seen and the ovaries had a normal appearance. The patient evolved favorably, pending reconstructive surgery of the neovagina.

The patient was hospitalized for 2 days, analgesics were administered, and her return to Huancavelica was arranged. In the follow-up by telephone during the following 2 months, she reported no discomfort or incidence.

Case 2

A 44-year-old nulliparous woman from the city of Jauja in the central highlands of Peru, without menarche, reported chronic pelvic pain in cycles for 18 years, in addition to primary infertility. She had undergone a right cystectomy 17 years earlier. The gynecological examination revealed the absence of the vaginal canal (Figure 4), normal hymen, permeable urethra and vulva with unaltered labia majora and labia minora.

Imaging tests showed no renal or abdominal abnormalities. Pelvic ultrasound revealed hematometra with a volume of 38.87 mL, absence of uterine cervix and hypotrophic ovaries (Figure 5).

The MRI showed an enlarged uterus with right lateralization and alteration of its morphology due to the presence of an extensive liquid collection that distended the endometrial cavity and collapsed the cervical canal, with a volume of 35 mL.

Examination under anesthesia recognized labia majora and minora, urethra and hymen of normal characteristics, vagina 3 cm deep, absence of cervix uteri. The opening of the cavity revealed multiple parieto-epiploic and right parieto-uterine adhesions, rudimentary right uterine horn of 8 x 6 cm with hematometra of approximately 40 mL (Figure 6), agenesis of the uterine cervix, absence of the left uterine horn, ovary and left tube of normal appearance, ovary and right tube absent due to surgical history.

The patient was hospitalized for 2 days, with favorable evolution and was discharged with indications.

She went for a check-up one week after the operation, to remove the stitches, with no discomfort or incidences.

Figure 1a. Case 1: Unaltered external genitalia. Absence of continuity in the vaginal introitus.

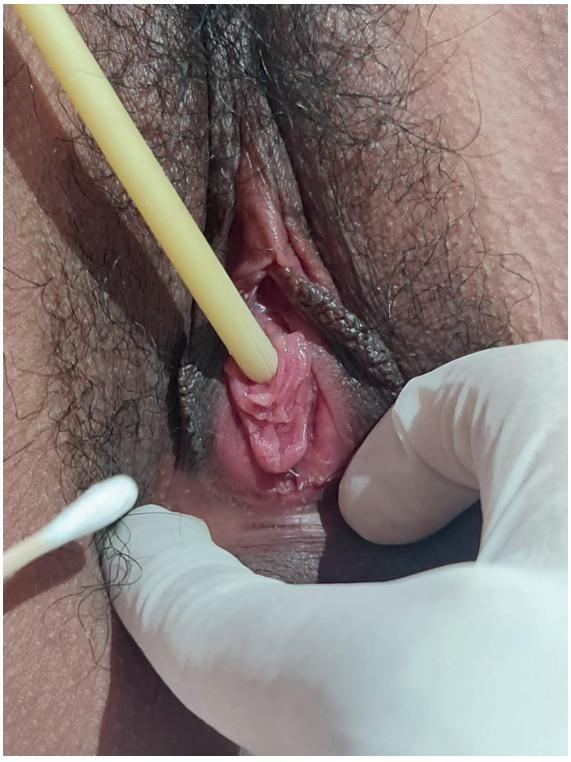


Figure 1b. Case 2: Unaltered external genitalia. Absence of continuity in the vaginal introitus.

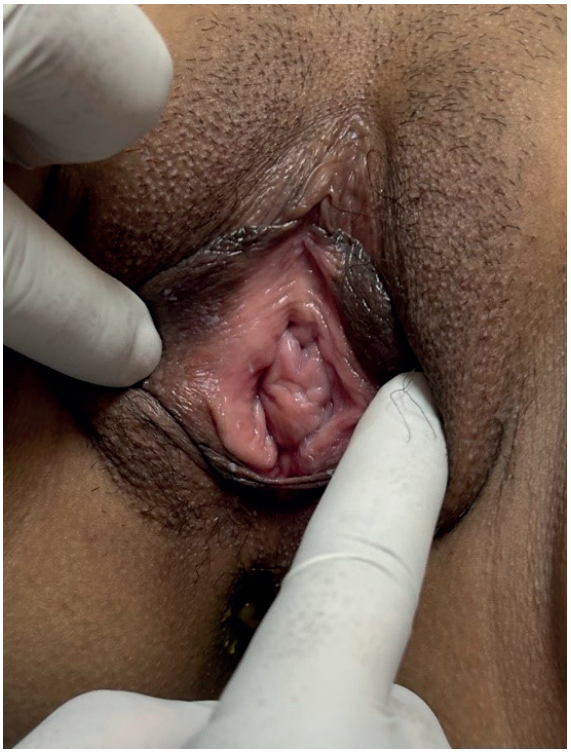


Figure 2a. Case 1: Right adnexal mixed tumor with anechoic content reported as hematometra.

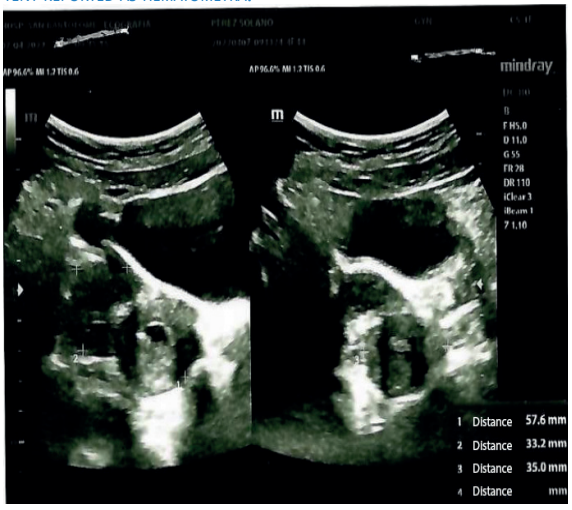


Figure 2b. Case 2: Rudimentary right uterine horn of 8 x 6 cm with hematometra.

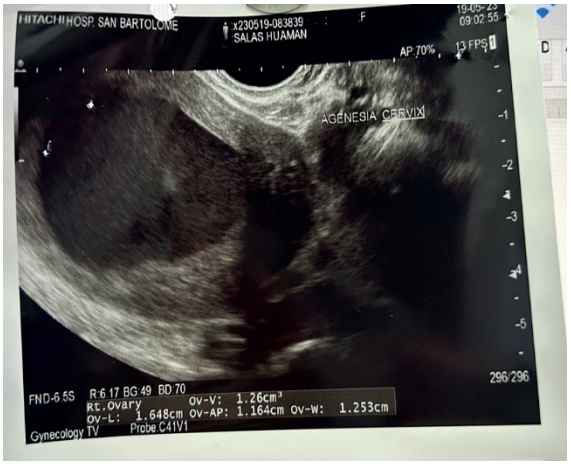


Figure 3a. Case 1: Presence of the right uterine horn of 4 x 4 x 4 cm with hematometra. Absence of left hemi-uterus. Ovaries of normal appearance.

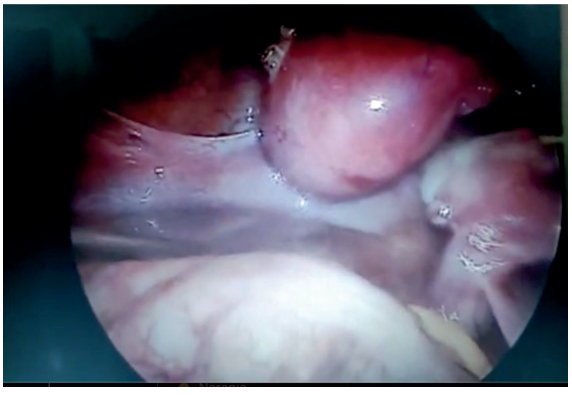
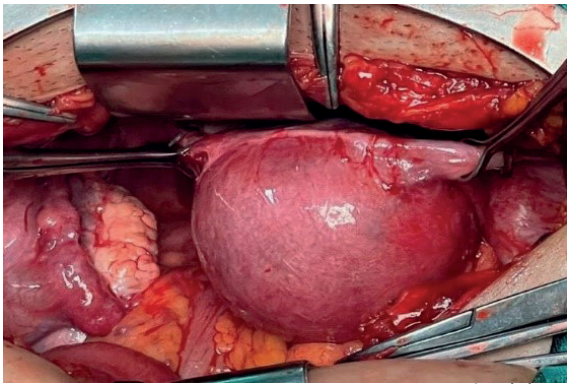


Figure 3b. Case 2: Right uterine horn distended 6 x 5 x 6 cm with hematometra. Absence of left hemiuterus. Ovaries and normal left tube. Adhesions due to a history of left adnexectomy.



Discussion

The pain characteristics presented by both patients are frequent correlates in these cases and are explained by the fact that the patient does not identify her problem until she notices the absence of menstruation and the associated pain, comparable to menstrual periodicity. This period of apparent normality is explained by the presence of a normal 46,XX karyotype with no identifiable phenotypic alterations and normal ovarian function[1]. In our second case, the patient had undergone surgery 18 years ago in the rural area, for a complicated adnexal cyst probably related to an endometriotic cyst in the right adnexa as a consequence of cervical agenesis that prevented the outflow of menstrual flow. There was no study of the patient's underlying pathology, the menstrual collection reappeared and a hematometra was formed. This patient was married, wanted to procreate but did not have access to a medical specialist to study her infertility. Cyclic pain with a duration similar to that of a menstrual period may be correlated with intrauterine hematometra due to the absence of the vaginal canal.

Medina and Rechkemmer[6] in Peru present cases with horseshoe pelvic kidney or renal agenesis. In our two cases, ultrasound and MRI did not show similar findings. This situation correlates with the report of Nguyen[8] in which it is mentioned that magnetic resonance imaging (MRI) showed an aplastic vaginal canal without identification of the cervix or uterus; the ovaries and neighboring organs did not show anomalies. This series did not report skeletal anomalies present, which are frequent according to the literature, but were not evidenced in our cases. Another frequent aspect is the presence of ovarian cysts[6,8,9]. In our first patient paratubal Gardner's cysts were observed.

In these cases, ultrasound has a fundamental guiding role in the gynecological, abdominal and renal evaluation to identify and describe the structure of the female sexual organs and look for malformations in other organs, most frequently renal followed by bone and cardiovascular[9]. This will allow establishing the probable classification of the syndrome. Finally, the MRI defines the organic structures in question in order to prepare for surgery.

In case 1, having confirmed the imaging diagnosis with MRI, the surgical alternative was considered as well as the construction of a neovagina when the patient begins her sexual life. Although hormonal therapy can prevent menstruation and attenuate cyclic pain, the presence of a rudimentary right hemiuterus and agenesis of the left hemiuterus tipped the risk/benefit balance towards removal of the aforementioned structures. The construction of neovagina with procedures such as those of McIndoe and intestinal vaginoplasty[10] with satisfactory results, still requires long term evaluation. The procedure was postponed in our patient until she decides to initiate her sexual function. In case 2, the patient had sexual activity without discomfort, despite having only the distal end of the vaginal canal.

Research on the prevalence of SMRKH in Peru, as well as the role of the genes involved, should be encouraged.

Referencias bibliográficas

1. Liszewska-Kapłon M, Strózik M, Kotarski Ł, Bagłaj M, Hirnle L. Mayer–Rokitansky–Küster–Hauser syndrome as an interdisciplinary problem. Adv Clin Exp Med. 2020;29(4):505-11. doi:10.17219/acem/118850
2. Jacquinet A, Millar D, Lehman A. Etiologies of uterine malformations. Am J Med Genet. 2016;170(8):2141-72. doi:10.1002/ajmg.a.37775
3. Triantafyllidi VE, Mavrogianni D, Kalampalikis A, Litos M, Roidi S, Michala L. Identification of Genetic Causes in Mayer-Rokitansky-Küster-Hauser (MRKH) Syndrome: A Systematic Review of the Literature. Children (Basel). 2022;9(7):961. doi:10.3390/children9070961
4. Londra L, Chuong FS, Kolp L. Mayer-Rokitansky-Kuster-Hauser syndrome: a review. Int J Womens Health. 2015;7:865-70. doi:10.2147/IJWH.S75637
5. Herlin MK, Petersen MB, Brännström M. Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome: a comprehensive update. Orphanet J Rare Dis. 2020;15:214. doi:10.1186/s13023-020-01491-9
6. Medina R, Rechkemmer A. Síndrome de Mayer-Rokitansky: Presentación de seis casos y revisión de la literatura. Rev Peru Ginecol Obstet. 2000;46(4):341-3. doi:10.31403/rpgo.v46i919
7. Fontana L, Gentilin B, Fedele L, Gervasini C, Miozzo M. Genetics of Mayer–Rokitansky–Küster–Hauser (MRKH) syndrome. Clin Genetics. 2017;91(2):233-46. doi:10.1111/cge.12883
8. Nguyen BT, Dengler KL, Saunders RD. Mayer–Rokitansky–Kuster–Hauser Syndrome: A Unique Case Presentation. Military Med. 2018;183(5-6):e266-e269. doi:10.1093/milmed/usx066
9. Herlin M, Bjørn AMB, Rasmussen M, Trolle B, Petersen MB. Prevalence and patient characteristics of Mayer–Rokitansky–Küster–Hauser syndrome: a nationwide registry-based study. Hum Reprod. 2016;31(10):2384-90. doi:10.1093/humrep/dew220
10. Altez C, Molina E, Ortega F, Angulo D. Neovagina con revestimiento peritoneal por laparascopia. Rev peru ginecol obstet. 2013;59(1):55-7. doi:10.31403/rpgo.v59i21