CASE REPORT

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Diagnosis of Mayer-Rokitansky-Küster-Hauser syndrome

Diagnóstico de síndrome de Mayer-Rokitansky-Küster-Hauser

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ABSTRACT

Mayer-Rokitansky-Küster-Hauser (MRKH) syndrome is a rare congenital malformation characterized by agenesis or hypoplasia of the uterus and the upper third of the vagina, in patients with a forty-six, XX karyotype and normal secondary sexual development. We present the case of a 12-year-old female patient with primary amenorrhea and hypogastric pain. Imaging studies revealed hematometra secondary to distal vaginal obstruction. Magnetic resonance imaging and laparoscopy confirmed the presence of a functioning hypoplastic uterus, absence of the cervix, and a short vaginal canal, findings consistent with type I MRKH. Emergency surgical drainage was performed. Comprehensive management of this syndrome requires a multidisciplinary approach for diagnosis, surgical intervention, and psychological support, due to its reproductive, emotional, and social impact.

Keywords: Amenorrhea, abnormalities, Mullerian Ducts, Peru. (Medical Subject Headings NLM)

RESUMEN

Los Trastornos del Desarrollo Sexual (DSD) son condiciones congénitas que afectan el desarrollo sexual cromosómico, gonadal o fenotípico. Uno de los DSD más comunes es el Síndrome de Insensibilidad a los Andrógenos (SIA), con una prevalencia de 1:60,000 nacimientos vivos. Se caracteriza por genitales externos de apariencia femenina, ausencia de útero y ovarios, y presencia de testículos intraabdominales en individuos con cariotipo 46,XY. Presentamos el caso de una mujer de 29 años con un diagnóstico inicial de síndrome de Rokitansky, que posteriormente fue confirmado como SIA mediante secuenciación genética, se realizó gonadectomía profiláctica para prevenir la malignidad. Este caso resalta la importancia de un diagnóstico oportuno para prevenir complicaciones como tumores gonadales y problemas psicológicos relacionados con la identidad de género. La intervención temprana y el manejo adecuado son fundamentales para el bienestar físico, sexual y emocional de las pacientes con SIA.

Palabras clave: Síndrome de Insensibilidad a los Andrógenos, Trastornos del Desarrollo Sexual, Diagnóstico Genético, Disgenesia Gonadal, Enfermedad Rara

Introduction

Mayer–Rokitansky–Küster–Hauser (MRKH) syndrome is a congenital anomaly characterized by complete or partial agenesis of the uterus and the upper two-thirds of the vagina in individuals with a normal female karyotype (46,XX) and normal development of secondary sexual characteristics. Clinically, the condition typically presents as primary amenorrhea and may be associated with renal, skeletal, and other systemic malformations[1,2].

The estimated prevalence of MRKH syndrome is approximately 1 in 4,500 to 5,000 live female births, making it the second most common cause of primary amenorrhea[3]. In the Peruvian context, although large-scale epidemiological data are lacking, reported clinical cases highlight the importance of considering this diagnosis in patients presenting with primary amenorrhea despite normal pubertal development[4].

This syndrome is classified into two types. Type I, or typical form, presents as isolated Müllerian duct aplasia, with congenital absence of the uterus, cervix, and upper two-thirds of the vagina, without associated anomalies. In contrast, type II, or atypical form, is associated with renal, vertebral, and occasionally auditory or cardiac malformations, in addition to Müllerian aplasia[5,6].

The diagnosis of MRKH syndrome requires a comprehensive evaluation that incorporates imaging modalities such as pelvic ultrasonography and magnetic resonance imaging, exploratory laparoscopy, anatomopathological evaluation of the ovaries, and karyotype analysis (7). At present, no curative treatment is available; therefore, management should be multidisciplinary, integrating medical, surgical, and psychological interventions tailored to the individual needs of each patient (8). Written informed consent for publication of this case report was obtained from the patient's mother, with assurance of confidentiality and use of the information exclusively for academic purposes.

Clinical case

A 12-year-old female patient with no relevant personal or family medical history was admitted to the Gynecology Emergency Department of the Ramiro Prialé Prialé National Hospital in the city of Huancayo, presenting with abdominal pain of insidious onset, severe intensity, continuous in nature, located in the left iliac fossa and radiating to the midline of the hypogastrium, without relief from conventional analgesics. She reports no previous menstruation (primary amenorrhea).

The general physical examination revealed a face with mild dysmorphic features, showing hypertelorism and low-set ears. Secondary sexual characteristics corresponded to Tanner stage II, with incipient breast development and sparse pubic hair.

Abdominal examination revealed a palpable mass in the hypogastric region with well-defined edges, firm consistency, and tenderness on deep palpation, findings suggestive of retained fluid or blood. External gynecological examination demonstrated a circular, scalloped hymen with regular edges and a permeable hymenal orifice measuring approximately 8 mm in diameter. Gentle insertion of a sterile swab identified a vaginal canal of approximately 3 cm in depth with distal resistance, raising suspicion of a transverse vaginal septum.

Additional tests are requested, with the following results:

- Karyotype: 46, XX (normal female).
- Luteinizing hormone (LH): 0.02 mIU/mL.
- Follicle-stimulating hormone (FSH): 1.55 mIU/mL.
- Blood type: A Rh positive.
- Complete blood count: within normal parameters, no signs of anemia or infection.

Diagnostic studies were expanded using medical imaging.

Pelvic ultrasound revealed a cystic, elongated formation with posterior enhancement and a "ground glass" appearance, with walls measuring 4.6 mm and no flow on Doppler imaging. The lesion was located in the left adnexa and extended toward the midline, suggesting a uterus with retained blood content.

Subsequently, a computed tomography (CT) scan of the abdomen and pelvis was performed, with and without intravenous contrast, which showed a structure consistent with an enlarged uterus, slightly lateralized to the left, with hyperdense endometrial content (272 HU) causing thinning of the myometrium, findings consistent with hematometra. In order to rule out associated structural malformations, an abdominopelvic X-ray was performed, which showed no musculoskeletal or renal abnormalities.

For better anatomical characterization, this was supplemented with a pelvic magnetic resonance imaging (MRI), which showed distension of the uterine cavity due to a fluid collection compatible with hematometra or serometra, probably secondary to distal vaginal obstruction. No renal or skeletal abnormalities were identified[9,10]. The findings were consistent with severe hypoplasia or partial agenesis of the vagina, suggestive of Mayer-Rokitansky-Küster-Hauser syndrome type I[9, 11].

Image 1. Own creation. Abdominal ultrasound showing the uterus with fluid content that appears "frosted glass."

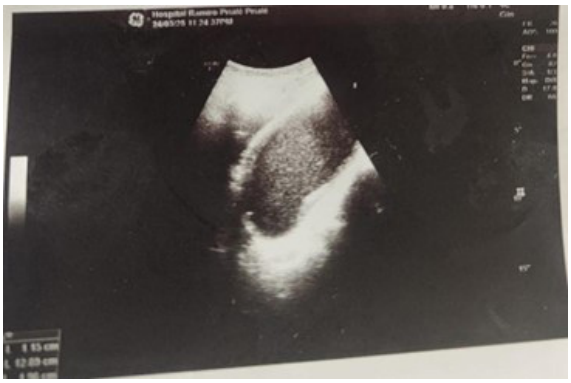
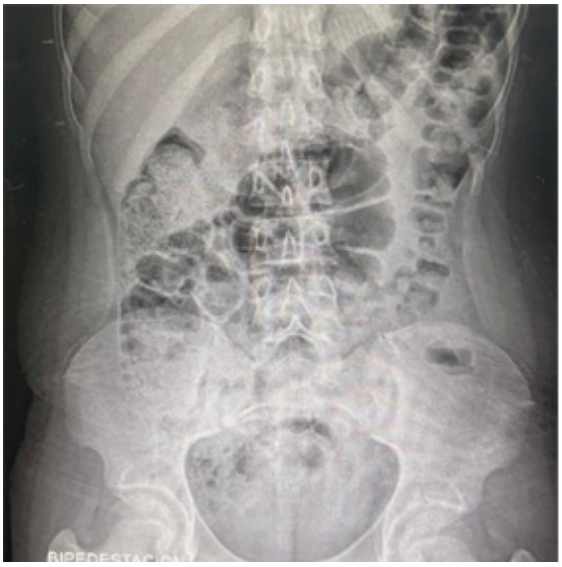


Image 2. Own elaboration: Abdominal pelvic X-ray, no associated malformations (musculoskeletal or renal) were evident.



The patient has not undergone surgery. Medical management with continuous menstrual suppression was chosen, as indicated by gynecology, with the aim of avoiding uterine bleeding and preventing cyclical pelvic pain. Since the start of treatment, the patient reports a progressive decrease in pain and is currently asymptomatic, afebrile, and performing daily activities appropriate for her age. At 1- and 4-week follow-ups, good adherence and tolerance to the drug were documented, with no clinically relevant adverse effects (no severe headache, no intercurrent metrorrhagia, no significant vasomotor symptoms). The follow-up ultrasound showed no new collections or cavity distension. Multidisciplinary follow-up (gynecology and psychology) is maintained, with education on warning signs (progressive pelvic pain, fever, abnormal bleeding) and periodic reevaluation to determine the appropriate time for anatomical correction based on pubertal maturity, residual anatomy, and the preferences of the patient and her mother.

Discussion

Our patient's clinical presentation—primary amenorrhea with normal pubertal development and cyclic pelvic pain—is consistent with that described for Mayer–Rokitansky–Küster–Hauser (MRKH) syndrome, where the diagnosis is usually established in adolescence and pain occurs when there is functioning endometrial tissue or distal obstruction[12]. This phenotype fits within the spectrum of MRKH type I, although in most series uterine aplasia is more extensive; when a functioning hypoplastic cavity is preserved, cyclic pain due to blood retention (hematometra) is an expected finding and explains the need for early decompression[13]. In accordance with what has been reported, general analysis and gonadotropic profile maintain values typical of puberty, given that the ovaries are functioning, which helps to differentiate MRKH from other causes of primary amenorrhea with gonadal insufficiency[14]. In terms of imaging, magnetic resonance imaging remains the method of choice for accurately delineating residual Müllerian anatomy, characterizing the uterine cavity and vaginal canal, and ruling out associated malformations; ultrasound and tomography may suggest the diagnosis, but MRI offers greater performance for therapeutic planning[10,11].

In the present case, the sequential use of ultrasonography and computed tomography, with subsequent confirmation by MRI, demonstrated distension of the uterine cavity secondary to distal obstruction in the context of a shortened vaginal canal, findings that are consistent with reports from cohorts evaluated at specialized centers[14]. The absence of associated renal or skeletal anomalies—features commonly observed in type I— also suggests a more favorable functional prognosis compared with more complex forms[15].

Regarding management, current evidence supports prioritizing a multidisciplinary approach that includes effective pain control, early surgical decompression to prevent secondary complications such as endometriosis and infection, and the use of hormonal suppression as a temporary measure prior to definitive corrective intervention. The immediate postoperative course

observed in this patient aligns with these established recommendations[16].

Overall, this case illustrates that, even within MRKH type I, the presence of a functioning hypoplastic uterus with distal obstruction may lead to hematometra and cyclic pelvic pain. Careful clinicoradiological correlation and timely intervention are essential to optimize clinical outcomes and to facilitate individualized planning for reconstructive procedures and reproductive counseling[9–16].

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