



## Herlyn-Werner-Wunderlich syndrome: Two case report

Victoria Fontana<sup>a</sup>, Gabriela S. Aboud<sup>a</sup>, Liliana Sabbaj<sup>b</sup>

### ABSTRACT

Herlyn-Werner-Wunderlich syndrome, also known as obstructed hemivagina and ipsilateral renal anomaly (OHVIRA), is a rare, congenital Müllerian duct anomaly characterized by the association of septate uterus, obstructed hemivagina, and ipsilateral renal agenesis.

The most common clinical presentation is an abdominal mass secondary to hematocolpos, pain, and dysmenorrhea. It is associated with infertility, endometriosis, and menstrual and obstetric alterations.

The ultrasound is the technique of choice for the initial assessment, while the magnetic resonance imaging remains the most accurate method for diagnosis. The resection of the vaginal septum is the recommended treatment.

Here we describe 2 clinical cases to highlight the importance of an early diagnosis to prevent potential complications in the future.

**Keywords:** unilateral renal agenesis; urogenital anomalies; Müllerian duct; anomalies; septate uterus.

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<sup>a</sup> Clinical Pediatrics; <sup>b</sup> Outpatient Clinic; Hospital General de Niños Ricardo Gutiérrez, City of Buenos Aires, Argentina.

**Correspondence to** Victoria Fontana: [fontanav.4@gmail.com](mailto:fontanav.4@gmail.com)

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## INTRODUCTION

Müllerian duct anomalies are a broad spectrum of anomalies including uterine and vaginal agenesis or duplication. Their incidence ranges from 0.17% in the general population to 3.5% in infertile women.<sup>1</sup> Herlyn-Werner-Wunderlich syndrome, also known as obstructed hemivagina and ipsilateral renal anomaly (OHVIRA), is a rare, congenital Müllerian duct anomaly resulting from an alteration in the development of the Wolffian ducts with consequent failure of Müllerian duct fusion. It is characterized by the association of a septate uterus, obstructed hemivagina, and ipsilateral renal agenesis.<sup>2</sup> Its prevalence has not been determined; only case reports have been published in the bibliography.<sup>1</sup>

It is a multifactorial polygenic condition whose etiology and pathogenesis are unknown and does not present with chromosomal abnormalities. It may be associated with other genital, urological, rectal, cardiac, or skeletal dysplasias.<sup>3</sup> The most common clinical presentation is an abdominal mass secondary to hematocolpos, pain, and dysmenorrhea. It is associated with infertility, endometriosis, and menstrual and obstetric alterations.<sup>4</sup> Reports in pediatrics are scarce, and an early diagnosis prevents short- and long-term complications.

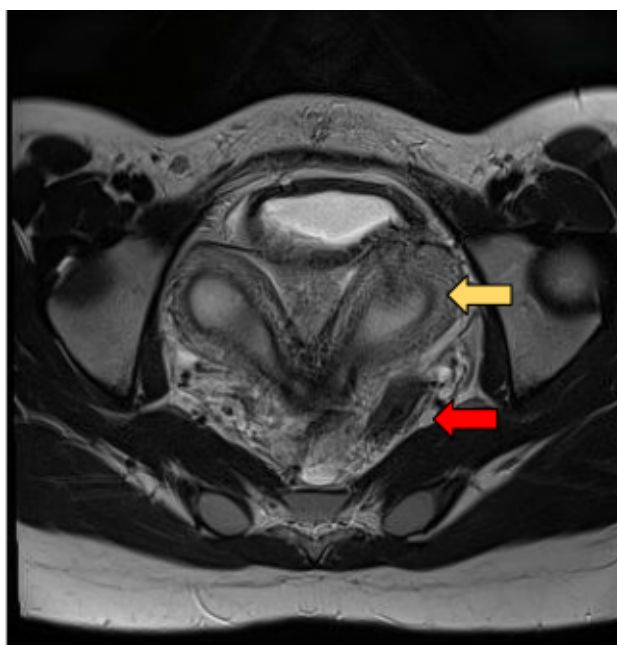
## CASE REPORT 1

A 14-year-old female patient with antenatal diagnosis of right renal agenesis, without pediatric follow-up, consulted due to secondary enuresis for the past 3 months, without urinary symptoms. She had the menarche at 12 years of age; at the time of consultation, she had regular cycles and dysmenorrhea.

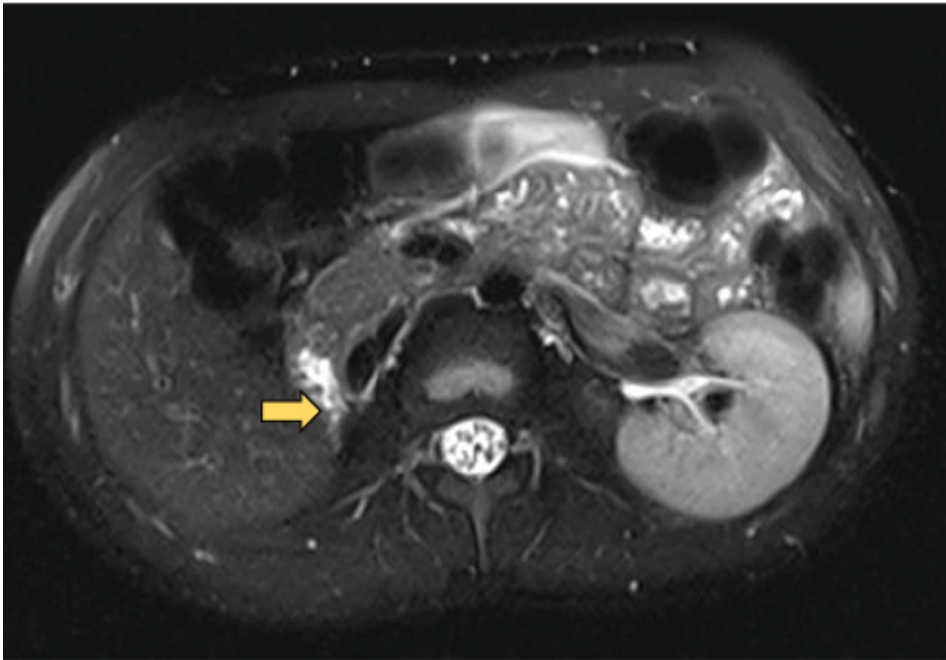
Due to her solitary kidney condition and for the study of urinary incontinence, urinalysis, urine culture, and lab tests with kidney function tests were done; results were within normal limits. An abdominal, renal, and bladder ultrasound showed the absence of right kidney and uterus with a septate appearance. The diagnosis was Herlyn-Werner-Wunderlich syndrome.

An interdisciplinary follow-up was started by the Departments of Pediatrics, Urology, Nephrology and Gynecology; a magnetic resonance imaging of the abdomen and pelvis was requested for assessment and treatment consideration (*Figures 1 and 2*). The patient received treatment with desmopressin for her urinary incontinence, as indicated by the Department of Urology; she showed a rapid clinical improvement and therefore an association with the syndrome was dismissed.

**FIGURE 1. Magnetic resonance imaging of the pelvis, case 1**



*Magnetic resonance imaging of the pelvis without contrast in T2. Axial section. The yellow arrow points to the septate uterus. The red arrow points to a localized fluid collection in the left, posterior side of the pelvis adjacent to the rectum-sigmoid colon, compatible with endometriosis.*

**FIGURE 2. Magnetic resonance imaging of the abdomen, case 1**

*Magnetic resonance imaging of the abdomen without contrast in T2. Axial section. The yellow arrow points to the absent right kidney.*

The magnetic resonance imaging showed a pararectal loculated fluid collection compatible with endometriosis; the patient is currently planned for surgery.

### CASE REPORT 2

A 12-year-old female patient with right renal agenesis, under follow-up by the Department of Nephrology, who had had the menarche 4 months prior to consultation. She attended the outpatient emergency service due to constant abdominal pain in the hypogastric region for the past 36 hours. Her abdomen was tender with deep palpation, without peritoneal reaction. The abdominal ultrasound showed bicornate uterus and right renal agenesis with an image suggestive of ipsilateral hematocolpos. The magnetic resonance imaging confirmed the diagnosis of hematometrocolpos, which was drained in the operating room with subsequent resection of the vaginal septum (Figure 3) and the absence of the right kidney (Figure 4). The patient had a favorable course, with no complications.

### DISCUSSION

Herlyn-Werner-Wunderlich syndrome is a rare, often asymptomatic congenital anomaly with late diagnosis. The typical triad consists of septate uterus, obstructed hemivagina, and ipsilateral

renal agenesis, but other renal anomalies may occur, such as ureteral anomalies, pelvic kidney, ectopic ureter implantation, even in the vagina.<sup>5</sup> These alterations are significantly more frequent on the right side, although there is no clear explanation for this finding.<sup>1,3,5,6</sup>

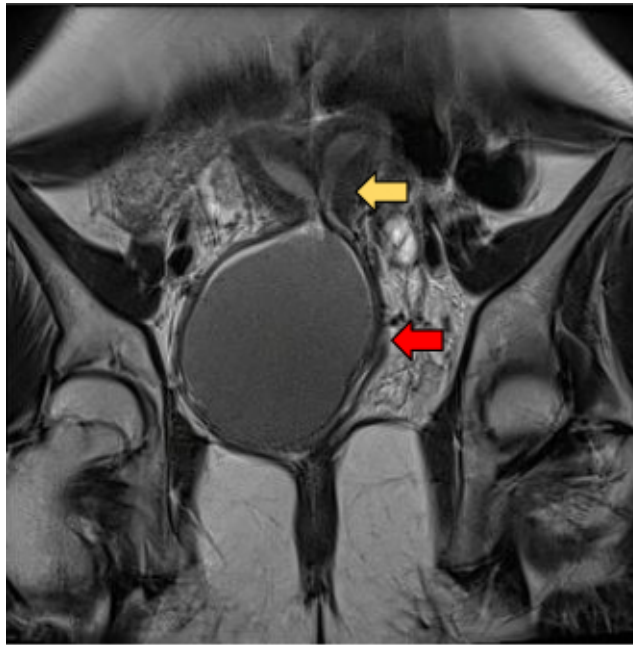
The main symptom is cyclic pain secondary to vaginal obstruction that appears after the menarche. The presence of regular menstrual cycles, as the vaginal obstruction is incomplete, may delay and confuse diagnosis. Other possible presentations are the onset of an abdominal, pelvic, or paravaginal mass secondary to a hematosalpinx, hematocolpos, or hematometra resulting from retained and partially clotted blood in the obstructed hemivagina. Some patients only have infertility or repeated miscarriages.<sup>6</sup>

Persistent obstruction may lead to retrograde bleeding into the abdominal cavity causing endometriosis, pelvic adhesions, or infections.<sup>6</sup> These complications may affect the patient's quality of life due to the resulting dysmenorrhea.

This condition may be resolved surgically, so an early suspicion and diagnosis make it possible to prevent alterations in daily life that may result from dysmenorrhea, visits to the emergency department, surgery for hematocolpos, and any concern about infertility in adulthood.

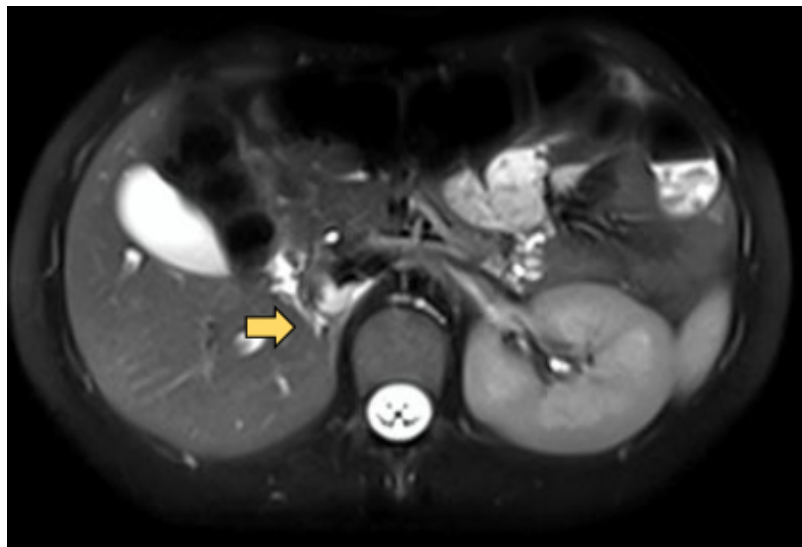
The ultrasound is the technique of

**FIGURE 3. Magnetic resonance imaging of the pelvis, case 2**



*Magnetic resonance imaging of the pelvis with contrast in T2. Coronal section. The yellow arrow points to the septate uterus. The red arrow points to hematocolpos in the right hemivagina.*

**FIGURE 4. Magnetic resonance imaging of the abdomen, case 2**



*Magnetic resonance imaging of the abdomen with contrast in T2. Axial section. The yellow arrow points to the absent right kidney.*

choice for the initial assessment with a 90-92% accuracy in uterine malformations; moreover, it is an inexpensive, fast, and non-invasive method.<sup>6</sup> In the ultrasound, a hematocolpos is seen as a heterogeneous fluid collection, with marked posterior acoustic

enhancement, better characterized with a magnetic resonance imaging, where the appearance will depend on the bleeding time.<sup>4</sup>

The magnetic resonance imaging is still the most accurate method for the diagnosis of Müllerian duct malformations, with an accuracy

and sensitivity close to 100%, although, due to its cost and lower accessibility, the initial assessment is usually an ultrasound.<sup>6</sup>

If a uterine abnormality is asymptomatic and detected during the prepubertal period, the patient will be instructed to return after the onset of thelarche, as the presence of estrogens may help to make a definite diagnosis. Estrogen stimulation increases the size of the uterus and allows a better examination of the uterus. In addition, prior to puberty, imaging studies may be controversial.<sup>7</sup>

Another important period for screening is at birth in patients with renal and urinary tract anomalies. In the neonatal period, the influence of maternal hormones facilitates the visualization by imaging studies of the internal genitalia and any potential anomaly, being an ideal moment for their assessment.<sup>8</sup>

The resection of the vaginal septum is the recommended treatment. In adolescents, a measure to postpone surgery would be the use of GnRH analogues to maintain amenorrhea, especially in cases of cervical atresia. Surgery is usually conservative with elective resection of the vaginal septum for the obstructed hemivagina, marsupialization of the blind hemivagina, and drainage of the resulting hematocolpos, in order to relieve symptoms and ensure successful reproductive outcomes.<sup>3</sup>

To conclude, the suspicion and diagnosis of renal malformations in female patients makes it necessary to rule out associated gynecological

conditions because their development is embryologically related.

OHVIRA is a rare congenital anomaly with great clinical significance and simple surgical resolution, so it is important to be aware of this condition for an early detection in female children and adolescents, since a delay in diagnosis and treatment may lead to future complications. ■

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