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INTERACTIVE CLINICAL CASE

Renal agenesis: early diagnosis of a rare and asymptomatic syndrome

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Abstract

Based on the renal agenesis, a suspicion of Herlyn-Werner-Wunderlich syndrome (HWWS) arose. Asymptomatic and with nonspecific findings, it was decided to take an expectant approach.

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CASE REPORT

A 2-month-old female patient was taken to the pediatric nephrology ward at Conceição Children's Hospital (HCC) after experiencing a febrile urinary tract infection, pre- and postnatal renal alterations, and an atrophied right kidney. Imaging revealed a cyst in the cul-de-sac, possible dilation of the vaginal cavity, and a double uterus. On physical examination, she presented with a normal vulva and anterior anus. A digital rectal examination indicated a palpable mass corresponding to the vagina.

1) Which age group sees the most frequent diagnosis of Herlyn-Werner-Wunderlich syndrome?

A) Neonates
B) Preschoolers
C) Adolescents
D) Adults

Magnetic Resonance Imaging (MRI) subsequently ordered showed no significant alterations, only a small presacral cyst-like structure indicating fluid in the vaginal cavity

Question comment 1:

Although the most serious chronic complications are related to adulthood, such as infertility, pelvic inflammatory disease, among others, some signs can already be seen in the prenatal period, such as renal agenesis. The most common age range for symptoms to appear is in adolescence, with prepubertal symptoms being very rare.[1,2]

Diagnosis before menarche is generally due to renal complications, which identify other alterations in the triad that tend to be more asymptomatic.[2,3,4]

Therefore, it is important to suspect the syndrome from the prenatal period when renal agenesis is identified. In addition, diagnosis can be delayed by the use of oral contraceptives and nonsteroidal anti-inflammatory drugs, since they mask the main symptoms that lead the patient to investigation and increase the chances of secondary complications of the syndrome.[2,5,6]

Magnetic Resonance Imaging (MRI) subsequently ordered showed no significant alterations, only a small presacral cyst-like structure indicating fluid in the vaginal cavity.

2) What is the gold standard for diagnosing Herlyn-Werner-Wunderlich syndrome?

A) Hysteroscopy
B) Ultrasound
C) Magnetic Resonance Imaging
D) Computed Tomography

Question comment 2:

During physical examination, a suprapubic mass that is painful to the touch may be identified, often accompanied by urinary incontinence during mobilization.[7] Bimanual examination reveals a hemivaginal septum bulging due to hematocolpos. In cases where diagnosis is challenging in young women with no history of sexual activity, anal examination may be an effective alternative, highlighting vaginal bulging.[2,5]

Despite the complexity of the diagnosis, it is important to emphasize that techniques such as hysteroscopy and tomography are rarely used in this context. Hysteroscopy, which offers direct visualization of the uterine cavity, and tomography, which provides detailed images of internal structures, are often not the preferred choices due to limitations in detecting certain specific abnormalities related to the syndrome.[8]

For further investigation, additional tests are performed, including pelvic ultrasound, ultrasound of the kidneys and urinary tract, with pelvic magnetic resonance imaging being considered the gold standard in this context. The images obtained clearly show the presence of uterus didelphys, distension of the uterine horn due to the presence of hematometra and hematocolpos, as well as evidence of hydronephrosis and ipsilateral renal agenesis.[2,5,6]

These findings provide crucial information for the accurate diagnosis of the condition. The combination of detailed physical examination and advanced imaging techniques is essential to fully understand the extent of the abnormalities and guide the most appropriate therapeutic approach. The use of ultrasound and magnetic resonance imaging, in particular, allows a comprehensive view of the structures involved, contributing to more effective clinical management.[2,6,8]

Renal agenesis heightened the suspicions of Herlyn-Werner-Wunderlich syndrome. Since the patient did not exhibit symptoms and had nonspecific findings, the team opted for an expectant approach.

3) Since the signs and symptoms of Herlyn-Werner-Wunderlich syndrome are gynecological, which alternative covers the most common findings?

A) Amenorrhea
B) Menstruation with normal or decreased flow, dysmenorrhea, and abdominal pain.
C) Increased menstrual flow
D) Dyspareunia

Question comment 3:

In prepubertal individuals, the predominant finding is a palpable pelvic mass, indicative of hydrocolpos, often associated with the identification of ipsilateral renal agenesis. Given that most diagnoses occur during adolescence, the typical presentation includes dysmenorrhea in 63.9% of cases, pelvic pain in 35.4% of cases, commonly of progressive evolution,

and irregular menstrual cycles, although they may be regular in cases of incomplete obstruction.[7]

Unlike other genital tract anomalies that often result in amenorrhea, HWWS is characterized by a normal menstrual pattern or by light bleeding, as documented in previous studies. Additionally, in the first months, the symptoms may be uncharacteristic.[2,8] It is important to note that the use of contraceptives in patients with dysmenorrhea may result in the suppression of the menstrual cycle, as well as the symptoms of cramping pain and increased abdominal volume. This interaction between the syndrome and contraceptive use underscores the need for a thorough assessment of medical history and drug therapy when managing patients affected by HWS.[2,8]

At the age of 12, during menarche, an ultrasound revealed two uterine cavities filled with fluid, suggesting hematocolpos. The patient was admitted for further investigation and surgery. An MRI confirmed the presence of a bicornuate uterus, a vaginal septum, and fluid in the right vagina. The team attempted drainage under general anesthesia; however, the absence of vaginal bulging made the procedure impossible. The patient continued to receive multidisciplinary outpatient follow-up.

4) What potential complications arise from the syndrome?

A) Infertility
B) Endometriosis
C) Pelvic inflammatory disease
D) All of the above

Question comment 4:

Because it is a rare pathology, it is important that both pediatricians and gynecologists consider it for early detection in girls and adolescents with renal agenesis, since a delay in diagnosis and definitive surgical treatment of the condition can lead to a poor prognosis.[7] The most common chronic complications are precisely linked to Mullerian malformation, such as: infertility, inability to maintain pregnancy until delivery, female pelvic inflammatory disease, and endometriosis.[1,7] The most common acute complications are hematocolpos, pyohematocolpos, and rupture of uterine abscess.[1]

Due to obstruction, most patients with HWWS require surgical treatment.[7]

Considering Herlyn-Werner-Wunderlich syndrome in the differential diagnosis of newborns with renal agenesis allows for early diagnosis, facilitating the introduction of appropriate interventions and improving the management and quality of life for patients.

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