Rare disease

Herlyn–Werner–Wunderlich syndrome: a rare cause of pelvic pain in adolescent girls

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Summary

The Herlyn–Werner–Wunderlich syndrome is a rare congenital anomaly characterised by uterus didelphys with blind hemivagina and ipsilateral renal agenesis. It usually presents after menarche with progressive pelvic pain during menses secondary to haematocolpos. Awareness is necessary in order to diagnose and treat this disorder properly before complications occur. MRI is the preferred modality for the delineation of uterine malformation. When renal anomalies are encountered, a screening should also be made for congenital abnormalities of the reproductive tract and vice versa. The authors report a case of a girl with this condition who had a prenatal diagnose of right renal agenesis and presented at 13 years old with pelvic pain caused by haematocolpos.

BACKGROUND

The Herlyn–Werner–Wunderlich (HWW) syndrome is a rare variant of müllerian duct anomalies characterised by the combination of uterus didelphys with obstructed hemivagina and ipsilateral renal agenesis. This syndrome was described for the first time in 1922 and was suspected in a young woman with regular menstruation and gradually increasing pelvic pain and a pelvic mass after menarche.¹ The true incidence of this anomaly is unknown, but it has been reported between 0.1% and 3.8%.^{2–3} The exact cause and pathogenesis are uncertain; however, the diagnosis and treatment at an early stage can relieve acute symptoms and preserve normal fertility.⁴

We describe the case of a 13-year-old girl with a known single kidney, recurrent abdominal symptoms during menstruation and presentation to the Emergency Department with acute abdomen.

CASE PRESENTATION

A 13-year-old girl, presented to the Emergency Department with a 4-day history of increasing right lower quadrant (RLQ) and hypogastric pain, nausea and sporadic vomiting without fever, diarrhoea or urinary symptoms. She was on the fourth day of menstrual bleeding. Her medical history revealed a right-sided renal agenesis diagnosed prenatally. The menarche had occurred 5 months before, her cycles were regular and the menstrual bleeding usually lasted about 4–5 days with severe dysmenorrhoea. She had not been sexually active yet. The main physical examination finding was tenderness on RLQ and hypogastric palpation, and doubtful Blumberg sign. The laboratory tests revealed a normal white blood cell count and a C reactive protein level of 2 mg/dl. An abdominal ultrasonography complemented by pelvic CT was performed on the day of admission, confirmed the absence of the right kidney and showed an image suggestive of haematocolpos on the right side and

two uterine bodies. The patient was then referred to the gynaecological department where she was medicated with a continually administered oral contraceptive to suppress menses and relieve symptoms until a MRI was obtained and a corrective surgery planned. The pelvic MRI showed two separate uteri with two separate cervixes (uterus didelphys bicollis) (figure 1A) and two proximal vaginas. The right vagina was distended and filled by a slightly hypointense material on T2-weighted images (figure 1B,C) and hyperintense material on T1-weighted images (figure 1D) that did not suppress on fat-suppressed T1-weighted sequences (figure 1E), suggesting a blood collection (haematocolpos). Only the distal portion of the left vagina was identified. Although a longitudinal vaginal septum was not directly visualised, its presence was admitted.

The patient underwent surgery with resection of the vaginal septum without perioperative or postoperative complications. She was released 2 days after surgery, showing an uneventful recovery.

INVESTIGATIONS

Referenced in the case presentation.

TREATMENT

The patient underwent surgery with resection of the vaginal septum.

OUTCOME AND FOLLOW-UP

The patient underwent surgery with resection of the vaginal septum without perioperative or postoperative complications. She was released 2 days after surgery, showing an uneventful recovery.

DISCUSSION

Müllerian (paramesonephric) duct anomalies are congenital anomalies of the female genital tract which result from

Figure 1 Selected MRI images of the pelvis and abdomen. Coronal T2-weighted images: (A) Uterus didelphys bicollis and (B) a distended and blood-filled right hemivagina (arrows). Transverse images: The haematocolpos (arrows) is slightly (C) hypointense on T2-weighted images, (D) hyperintense on T1-weighted images and (E) does not suppress on fat-suppressed T1-weighted sequences.

non-development or non-fusion of the müllerian ducts or failed resorption of the uterine septum during the sixth to ninth weeks of fetal life causing a wide-ranging series of reproductive ducts malformations.^{5 6}

The paramesonephric ducts of the genetically female embryo fuse together in the midline and form the uterus, cervix and the upper four-fifths of the vagina. The lower 20% of the vagina are formed from sinovaginal bulbs which are protrusions of the urogenital sinus.⁷ The urinary and genital systems arise from a common ridge of mesoderm arising along the dorsal body wall and rely on normal development of the mesonephric system. Hence, abnormal differentiation of the mesonephric and paramesonephric ducts may also be associated with anomalies of the kidneys.⁷ Renal agenesis is the most common anomaly although horseshoe or pelvic kidney, cystic renal dysplasia, duplication of the collecting system and ectopic ureters have all been described.^{7 8} Renal agenesis is predictive of an ipsilateral obstructive müllerian anomaly greater than 50% of the time.⁸ These anomalies have a right-sided dominance, twice as often as on the left side.^{9 10} Such relationship between female genital and urogenital anomalies should lead us to examine the urogenital system when a genital anomaly is identified and vice-versa.⁴

Typically, HWW syndrome presents with acute or chronic pelvic pain shortly following menarche.² These patients menstruate normally and may have no specific symptoms, except dysmenorrhea.⁴ A progressively enlarging pelvic mass has also been described.⁵ If treatment is delayed, complications may develop, such as endometriosis caused by retrograde menstruation, infections and pelvic adhesions, which in turn might cause obstruction of the genital organs. Clinical suspicion and awareness of the syndrome are therefore imperative to making a timely diagnosis and preventing these complications.¹¹ In one study, it was demonstrated that the delay in diagnosis was due to several factors. As the patient menstruates regularly from the non-obstructed horn, the diagnosis of outflow obstruction is more difficult. Second, it is an uncommon condition and therefore not often thought of as a diagnostic possibility. Third, when these adolescents present to their paediatrician or family physician with symptoms of cyclic dysmenorrhoea, they are usually given anti-inflammatory drugs and oral contraceptives. These treatments inadvertently cause a delay in diagnosis as they diminish or eliminate menses.²

CT and ultrasound are the most widely used diagnostic tools.¹¹ However, MRI is considered to be more sensitive for imaging soft-tissue anatomy and delineating subtle

findings seen in congenital anomalies. Hence, it should be obtained before any surgical intervention.² Laparoscopy is not mandatory but could be helpful in confirming the diagnosis when radiologic imaging is inconclusive, especially in those cases with endometriomas warranting resection.³ As obstructive genital lesions may be associated with other anomalies such as coarctation of the aorta, atrial septal defects and abnormalities of the lumbar spine, a complete physical examination and abdominal tests may be indicated.²

Resection of the vaginal septum is the treatment of choice of obstructed hemivagina.⁵

Women with uterus didelphys have a reasonable chance of getting pregnant, but the abortion rate is high (74%) and premature delivery is common (22%).¹² A caesarean section is required in 82%.¹⁰

Evaluation of the genital tract by means of MRI scanning is recommended in all girls with known renal abnormalities detected antenatally or thereafter, before the onset of menstruation. This enable us to diagnose some patients before menarche and carry out a surgical correction of the obstruction before any damage has occurred because of haematocolpos, haematometra and retrograde menstruation.¹⁰

Learning points

- ▶ The HWW syndrome is a rare congenital anomaly characterised by uterus didelphys with blind hemivagina and ipsilateral renal agenesis.
- ▶ Awareness of the HWW syndrome is necessary in order to diagnose and treat this disorder properly before complications occur.
- ▶ When renal anomalies are encountered in female infants, a screening should be made for congenital abnormalities of the reproductive tract and vice versa.
- ▶ MRI is the preferred modality for the delineation of uterine malformation.

Competing interests None.

Patient consent Obtained.

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