CASE REPORT

Congenital abnormalities of the urogenital tract: the clue is in the cord?

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SUMMARY

Congenital abnormalities of the female urogenital tract are not uncommon, with an estimated incidence of 2–4% across the female population. Within this population, up to 40% will have associated renal tract abnormalities. A previously well 12-year-old girl presented to the emergency department with abdominal pain, vomiting and a palpable pelvic mass. Ultrasound and MR scans were performed. The imaging revealed a didelphys uterus, an obstructed hemivagina and ipsilateral renal agenesis, characteristic of Herlyn-Werner-Wunderlich syndrome. The patient was noted at birth to have a single umbilical artery, which is associated with an increased risk of congenital abnormalities and useful information for the early identification of abnormalities that have implications for renal function and future fertility.

BACKGROUND

Congenital abnormalities of the female urogenital tract are not uncommon with an incidence of 2–4%;¹ up to 40% of this population additionally have associated renal tract abnormalities.² These abnormalities have potentially serious medical and psychosocial sequelae later in life, particularly at the time of adolescence and menarche.

The following case report focuses on one such condition, Herlyn-Werner-Wunderlich syndrome, a very rare congenital abnormality of the female renal and urogenital tract. The article aims to highlight the investigation and management of such uncommon cases.

Herlyn-Werner-Wunderlich syndrome arises as a result of partial embryonic agenesis of the paramesonephric duct, although an exact mechanism remains to be characterised.

CASE PRESENTATION

A previously healthy 12-year-old girl presented to the emergency department with a 3-day history of vomiting and left sided severe pelvic pain. Her vomit was non-bilious and did not contain blood. On examination her abdomen was tender in the left hypochondrium and a pelvic mass could be palpated. The patient last opened her bowels 2 days earlier and had no other history of note. Although she had not lost any weight, the patient had been unable to eat or drink over the past 3 days due to her symptoms. She was noted to be tachycardic and tachypnoeic on admission to the children's ward.

Her menstrual history revealed she began menstruating at 11 years of age, with a regular 3/26 cycle, which was of normal flow with little pain.

Her last menstrual period was 5 days prior to admission. The patient did not have any atypical menstrual history and was not sexually active.

The patient was delivered at 37 weeks by caesarean section following placental abruption. At birth, medical staff noted the presence of a single umbilical artery (SUA) and a postnatal ultrasound scan was arranged. The ultrasonographer reported inability to visualise the left kidney.

INVESTIGATIONS

On admission to the emergency department, the patient received a midstream urine dip test (which showed proteinuria with the presence of blood), a full blood count, and a urea and electrolyte blood test. Blood tests revealed an elevated C reactive protein level of 103 mg/L. A urine pregnancy test was negative.

Following initial investigations and clinical examination, an urgent transabdominal and pelvic ultrasound was arranged, which identified a large left-sided adnexal mass. The left kidney could not be visualised, in keeping with the previous findings several years ago.

As a result of the ultrasound findings, a CT scan of the patient's pelvis and abdomen was then performed. The scan identified a large adnexal cystic abnormality, distention of the left fallopian tube and haematocolpos. The left kidney could not be visualised. These findings prompted an urgent gynaecological referral.

Following her initial ultrasound and CT scan, the patient was referred to the gynaecology team with a suspected ovarian cyst. The gynaecology team requested an abdominopelvic MR scan in order to localise the lesion and investigate possible renal agenesis, prior to going to the operating theatre. The MRI revealed the presence of a uterine didelphys with obstructed hemivagina due to a transverse septum; characteristic findings of a rare urogenital tract abnormality, Herlyn-Werner-Wunderlich syndrome.

DIFFERENTIAL DIAGNOSIS

The formulated differentials included constipation, bowel obstruction, urinary tract infection and ovarian cyst. Renal syndromes were also considered in view of proteinuria.

TREATMENT

Intravenous fluids, analgesia and empirical antibiotics were prescribed. A nasogastric tube was inserted in addition to a urinary catheter in order to measure urine output.

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Intraoperatively, a large haematocolpos was drained through the transverse vaginal septum and the septum resected. During the postoperative period the patient remained stable and pain-free. Following a 3-day observation period, with no post-operative complications, she was discharged home.

OUTCOME AND FOLLOW-UP

An ultrasound and dimercaptosuccinic acid radionuclide scan were arranged following discharge to establish the degree of left renal agenesis and remaining renal function. Once these scans had been compiled, outpatient follow-up at 6 weeks post-discharge was arranged at a tertiary centre. To date, the patient has normal renal function and has had no further manifestations of her condition. However, it remains to be seen as to the impact of this on her future fertility.

DISCUSSION

Although rare,³ Herlyn-Werner-Wunderlich syndrome should be suspected in patients who have an atypical combination of didelphys uterus, an obstructed hemivagina and ipsilateral renal agenesis. It is important to consider congenital abnormalities such as HWW among the differential diagnoses to identify these rare cases in early adolescence. The timeline of acute presentations may evolve with complications (including sepsis and pyosalpinx), which may threaten life and future fertility.

The embryological mechanisms surrounding the development of urogenital tract abnormalities remain poorly characterised. Three discrete embryological ducts form the renal and urogenital tracts and must undergo fusion for normal in-utero development to occur. The proximal two-thirds of the vagina, uterus, fallopian and cervix arise from the müllerian ducts. The müllerian ducts fuse with the urogenital sinus comprising the distal third of the vagina and the urethra. Initially, the müllerian ducts are separated by a septum, although it remains unclear how the process of fusion occurs; it is thought likely it occurs in a bidirectional craniocaudal fashion to form the completed structure in the midline. Incomplete fusion of these tracts leads to the characteristic septal defects found in urogenital tract abnormalities. The renal system arises from the mesonephric ducts, which migrate caudally in-utero to provide connection with the bladder.⁴

Two large studies^{5 6} demonstrated that the presence of an SUA is associated with an increased risk of congenital malformation, particularly renal tract abnormalities. Although the presence of an SUA is not diagnostically specific or sensitive, due to

this increased risk it is wise to ensure the renal and reproductive tracts are fully visualised on imaging to exclude malformation.

Finally, management of these inherently complex multisystem conditions should take place within specialist tertiary units.¹ These centres are likely to have access to a wide variety of clinical expertise and experience, which is of vital importance to the young patient, for whom this condition is very distressing.

Learning points

- ▶ Herlyn-Werner-Wunderlich syndrome is a very rare multisystem congenital abnormality.
- ▶ Patients with a single umbilical artery are at increased risk of congenital abnormalities, particularly those affecting the renal tract.
- ▶ Abnormalities of the female reproductive tract require careful management to avoid complications threatening life and fertility.

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