

Anorectal Malformation with Associated Duodenal Obstruction and Uterus Didelphys

MV Vincent¹, D Madhuri²

ABSTRACT

Anorectal malformations have long been associated with multiple other anomalies, commonly referred to as the VACTERL complex. We present a case of a newborn baby girl with an unusually high number of associated anomalies, including the rarely encountered condition of uterus didelphys.

Keywords: anorectal malformation, duodenal obstruction, pelvic kidney, uterus didelphys

Malformación anorrectal asociada con obstrucción duodenal y útero didelfo

MV Vincent¹, D Madhuri²

RESUMEN

Las malformaciones anorrectales se han asociado por mucho tiempo con otras anomalías, conocidas comúnmente como asociación VACTERL. Presentamos el caso de una niña recién nacida con un número inusualmente alto de anomalías asociadas, incluyendo una condición raramente encontrada conocida como útero didelfo.

Palabras clave: Malformación anorrectal, obstrucción duodenal, riñón pélvico, útero didelfo

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INTRODUCTION

Anorectal malformations (ARMs) present as a disease spectrum which can occur in isolation or in conjunction with other well-described congenital anomalies. We present the case of a newborn baby girl with an ARM and multiple associated anomalies. The associated anomalies included malrotation without volvulus, duodenal atresia (Type 1), absent gallbladder, meckel's diverticulum, a right pelvic kidney, and uterus didelphys.

CASE REPORT

A newborn baby girl was referred to the surgical unit of The Queen Elizabeth Hospital in Barbados, after her

newborn examination revealed an absent anal opening. The baby was born by emergency Caesarean section after her mother presented with meconium-stained liquor with associated fetal decelerations at 40 weeks' gestation. The mother was a 26-year-old primigravida with a history of polycystic ovarian disease. She first presented antenatally at 32 weeks' gestation when her initial antenatal scan revealed the presence of a fetal double bubble, a finding which persisted throughout her pregnancy.

At birth, the baby weighed 2.88 kg with Apgar scores of 8 at one minute and 9 at five minutes. She was immediately transferred to the neonatal intensive care unit where she required no cardiorespiratory support.

From: ¹Department of Surgery, The Queen Elizabeth Hospital, Barbados, West Indies and ²Department of Radiology, The Queen Elizabeth Hospital, Barbados, West Indies.

Correspondence: Dr MV Vincent, Department of Surgery, The Queen Elizabeth Hospital, Martindale's Road, Bridgetown, St Michael, Barbados, West Indies. Email: michvincent@yahoo.com

On examination, three unusual perineal openings were noted. She possessed an anal dimple with the anal orifice located anteriorly (a vestibular fistula), through which copious amounts of meconium was seen to be evacuating. Her vaginal orifice revealed the presence of a midline septum, and a normal urethral opening was noted (Fig. 1). She had no limb deformities or dysmorphic facial features.

A nasogastric tube was passed and placed on free drainage, after which a portable chest X-ray was performed. This revealed the presence of a persistent double bubble as well as the presence of 13 ribs bilaterally (Fig. 2). Further investigations were subsequently ordered, and the following anomalies were revealed.

An echocardiogram (on day 1 of life) showed significant persistent pulmonary hypertension. The right heart chamber was also dilated and a patent foramen ovale present with a small secondary atrial septal defect with left-to-right shunting.

Abdominal and pelvic ultrasound scans revealed a right pelvic kidney and uterus didelphys (duplication of the vagina, cervix and uterus).

On day 3 of life, the baby underwent exploratory laparotomy and the formation of an umbilically-sited distal descending colostomy, under triple-antibiotic cover (ampicillin, gentamycin and flagyl). The protective colostomy was placed in preparation for the second stage of management of her ARM – posterior sagittal anorectoplasty.

Intraoperatively, she was noted to have an absent gallbladder. The proximal duodenum was moderately dilated and bulbous with the presence of an intraluminal membrane. The caecum and appendix were located in the right upper quadrant with the majority of the small bowel located in the right side of the abdomen, and the presence of a meckel's diverticulum measuring only 3 mm in length. The sigmoid colon and rectum were both distended with copious amounts of meconium.



Fig. 1: Perineal openings showing catheters in the urethra and both orifices of the double vagina. Meconium is also noted emanating from the vestibular fistula.

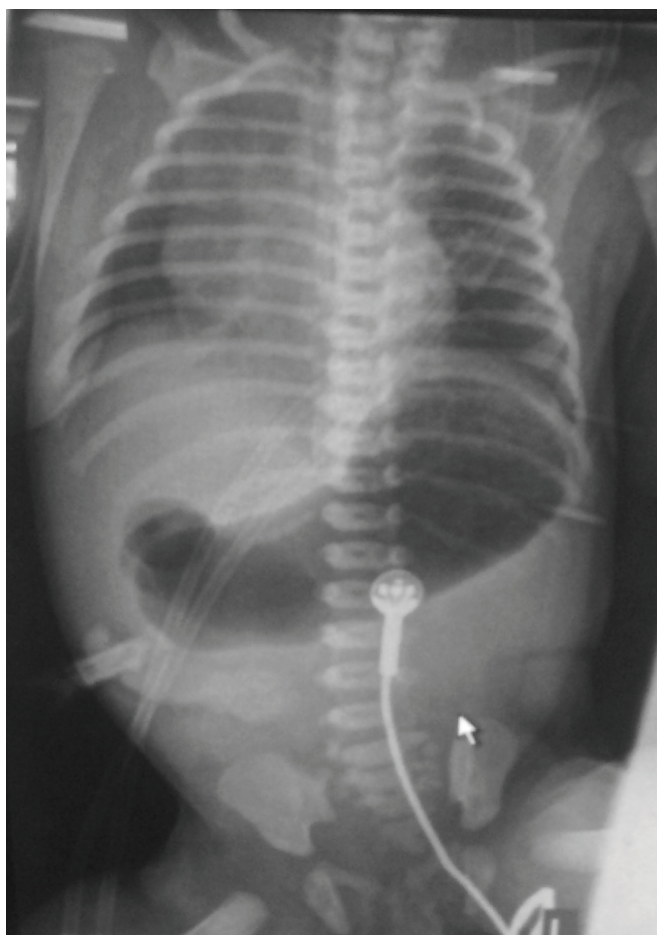


Fig. 2: Radiograph showing 13 ribs bilaterally. The classical 'double bubble' of duodenal obstruction is also apparent.

A diamond-shaped duodenojejunostomy was performed, together with appendicectomy, and an umbilically-sited distal descending colostomy with the distal end narrowed to a mucus fistula. Prior to the formation of the mucus fistula, the meconium within the rectum and sigmoid colon was emptied and both areas irrigated with saline.

Postoperatively, the baby was stable with the passage of meconium per stoma and moderate quantities of nasogastric aspirates (ranging from 90 to 130 ml per day), which was noted to be mildly to moderately bilious in nature. On day 3 postoperatively, the baby began having bradycardic episodes with associated desaturations, and was initially placed on ventilator continuous positive airway pressure (CPAP) *via* high flow nasal cannulation and then subsequently intubated and ventilated. Her blood cultures revealed the growth of a gram-negative bacillus (*Acinobacter*) and so antibiotics were changed initially to vancomycin, gentamycin and flagyl and finally to meropenem, gentamycin and flagyl based on sensitivities. However, the baby remained critically ill despite supportive care and developed generalized oedema, worsening neutropenia and worsening thrombocytopenia. She was also noted to have persistent desaturations while ventilated with evidence of persistent pulmonary hypertension of the newborn. Her urine output tailored, and she developed bloody secretions from the mouth and eventually succumbed to overwhelming sepsis from *Acinobacter* on day 6 of life.

DISCUSSION

Anorectal malformations have an estimated incidence of 1 in 5000 live births (1–3). The condition presents in a spectrum ranging from mild anal anomalies to complex cloacal malformations. The aetiology remains unclear, though a genetic component has been proposed, particularly for vestibular fistulas (1, 4, 5). In fact, patients with perineal or vestibular ARMs are two to three times more likely to have a family member with an ARM, and this is important with respect to genetic counselling (1, 4).

It has now been over 40 years since ARMs have been known to be associated with a variety of anomalies, commonly known as the VACTERL association, which comprises vertebral anomalies, anal atresia, cardiac malformations, trachea-oesophageal fistula, renal anomalies and limb abnormalities (1, 5, 6).

Genitourinary complications can occur in up to 50% of patients with an ARM (2, 4). In girls with a vestibular ARM, gynaecological anomalies occur in about 17% (4, 7), while those with more complex ARMs such as

cloacal anomalies have an even higher incidence of 53–67% (2, 3, 7–11). These gynaecological anomalies include hemivagina, vaginal septum, vaginal atresia/absent vagina, multiple vaginas, absent cervix, absent uterus, unicornuate uterus, bicornuate uterus, uterus didelphys, and duplication of the genitalia. Many of these gynaecological anomalies may be missed until adolescence or early adulthood, especially if no attempt is made to exclude them during the neonatal period. These include:

- vaginal septum (symptomatic in almost half of adult patients) which can be associated with the prevention of effective use of a tampon for menstrual flow, painful vaginal intercourse, cyclic pelvic pain, abnormal vaginal bleeding, bleeding during intercourse and an intermittent mucopurulent discharge (7, 8, 12);
- hemivagina which can lead to hydrocolpos with associated hydroureteronephrosis, haematocolpos, haematometra with the risk of endometriosis, and pelvic inflammation (8, 9, 11, 12);
- uterine anomalies (unicornuate uterus, bicornuate uterus, uterus didelphys) leading to an increased risk of preterm labour, preterm delivery and abnormal fetal presentations; and
- absent cervix leading to abnormal menstrual flow and infertility (7, 13).

As noted in this case, uterus didelphys consists of the presence of two uterine horns each with a cervix (14–16). This rare congenital anomaly is thought to arise from the failure of lateral fusion of the müllerian ducts and hence is often associated with ipsilateral renal anomalies, most commonly renal agenesis (12, 15–19).

If the anomaly is associated with separate vaginas, as in this case, the girl is usually asymptomatic as there is an outlet for physiological and menstrual secretions (9). However, an asymmetric bicornuate uterus with a rudimentary horn does not communicate with the vagina and will usually become symptomatic with the onset of menstruation. Common presentations include lower abdominal pain, a tender suprapubic mass, urinary retention and constipation (9, 11, 14–16). In such cases, there is cyclic bleeding from the hemi-uterus and vagina of the unobstructed side with obstructive symptoms and development of haematometra from the contralateral obstructed uterus (9, 16).

Thus, it has been proposed that all girls should undergo vaginoscopy at the time of posterior sagittal anorectoplasty (PSARP) or at the time of the formation

of a protective colostomy, with vaginoscopy and cystoscopy \pm laparoscopy being performed in those with more complex ARMs (2, 7, 8, 11). This is to ensure timely diagnosis of associated gynaecological anomalies, assist with surgical planning, and prevent the above-mentioned complications at puberty, with its associated long-term sequelae (2, 7, 9, 11, 12, 16). It is also generally thought that performing these procedures during the neonatal period will prevent the associated psychological trauma of undergoing 'vaginal surgery' during later childhood or adolescence (7, 8).

Girls with vestibular ARMs and an isolated vaginal septum can be effectively managed at the time of PSARP. A window or defect can be made within the septum to allow drainage of both hemivaginas, or the septum can be completely resected (7, 8, 16). Resection of the vaginal septum is most commonly performed using a scalpel or scissors. Other less invasive techniques include hysteroscopic resection, with the preservation of the hymen (18), or resection using combined laparoscopy and vaginoscopy (16).

In summary, ARMs in girls can be associated with a variety of genitourinary anomalies. Knowledge of these associations is important so that early surgical interventions can be provided and serious complications such as chronic pelvic inflammation and infertility can be minimized. Long-term follow-up into adulthood must also be provided as inevitably the growing patient and parents will have concerns with respect to future sexual intimacy and reproductive potential (8, 11).

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