Meconium is not enough: look for the hole!

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SUMMARY

Anorectal malformations are usually diagnosed at birth, and the diagnosis is usually readily apparent in boys and girls by direct inspection of the perineal region. However, some of the less complex lesions, often referred to as 'low lesions', such as rectovestibular fistula in girls, may not be diagnosed at birth. These children are able to pass meconium, and it is only when the narrow fistula fails to pass formed stool later in infancy or childhood does the child become symptomatic. It is important to emphasise that the diagnosis of these defects should be made during neonatal predischARGE examination. Such a delayed diagnosis may change their management, complicate surgical repair and may contribute to both functional and psychological problems for the patient and family.

BACKGROUND

Anorectal malformations (ARM) constitute the most varied group of congenital defects. The incidence of ARM is 1 of every 5000 newborns.¹ Vestibular fistula is the most common form of ARM in girls and it is associated with the best prognosis. It is estimated that 93% of patients with vestibular fistula will develop voluntary bowel movements by the age of 3 years.² We present a late detected case of vestibular fistula, which was missed twice on separate occasions by healthcare professionals.

CASE PRESENTATION

A 2-year-old female child was referred to our unit, from her General Practitioner, with diagnosis of an anorectal malformation. She was under medical treatment for chronic constipation, which started in

the second year of life, and showed no improvement despite the use of laxatives. The diagnosis was missed during neonatal baby checks and at the 6-week baby check conducted at the local community hospital. There were no social concerns.

During the consultation in our unit, examination of the perineal region revealed a flat perineum with absence of normal anal opening, and a fistula orifice was seen anterior to the posterior fourchette of the vagina (figure 1A,B). Abdominal examination was otherwise unremarkable and the diagnosis of anorectal malformation with vestibular fistula was made.

INVESTIGATIONS

The child underwent a renal USS and echocardiography (ECHO), as part of her normal work up, and this did not reveal any associated anomalies. A barium enema was performed to confirm the diagnosis and delineate the length of the fistula (figure 2A). Distended distal colonic bowel was visualised and the fistula was 2.3 cm long; there was no common channel and therefore a cloacal anomaly was excluded.

TREATMENT

Single-stage operative repair was completed with a view to perform a colostomy if bowel preparation was not adequate, or at the time of surgery, if major dissection was necessary. The child was admitted 2 days before the procedure for bowel preparation with sodium picosulphate 2.5 mg Ter Die Sumendum (three times a day, TDS) for 2 days, and Phosphate Fleet enema 50 ml in the morning for 2 days prior to surgery. The patient was positioned prone and posterior sagittal ano-rectoplasty approach was performed.

Figure 1 (A) Perineal examination with retraction of the labia majora reveals the presence of the vestibular fistula. Vagina orifice and urethra (urinary catheter already positioned) are clearly distinct from the fistula. (B) The perineum is flat and the absence of the normal anal orifice is clearly noticed.

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Figure 2 (A) The barium enema shows the length of the fistula; no connection was seen with urinary tract or the vagina and therefore a cloacal anomaly is excluded. (B) Posterior sagittal ano-rectoplasty and the pulled-through anus.

Upon stimulation, good muscle complex was observed and, during dissection, a long common wall with the vaginal posterior wall was freed, and the neo-anus was pulled through the sphincter muscles, without the need of a covering colostomy (figure 2B).

OUTCOME AND FOLLOW-UP

The patient was kept nil by mouth for 48 h following the procedure, after which she was commenced on clear fluids. She opened her bowel on the third postoperative day. Normal diet was restarted on day 4 postoperatively and she was discharged home on day 5 with a healthy appearing neoanus.

DISCUSSION

The diagnosis of anorectal malformation in female infants is relatively easy to make by direct inspection of the perineal area. Delayed diagnosis has been reported especially in vestibular fistulae³ and even during adulthood.⁴ Kim *et al*³ reported late diagnosis in 13% of all new cases of anorectal malformation; of these patients, 9 of 15 cases (60%) were females with vestibular fistula seen in 8 of these patients. The main symptom was worsening constipation, usually associated with the introduction of solid food. The age range of presentation was 3–11 months in eight cases, and one girl was diagnosed at 14 years of age.

We believe the diagnosis should not be missed as all neonates are fully evaluated by healthcare professionals during the neonatal period prior to discharge. All neonates should be carefully evaluated and clinical examination should include evaluation of the perineum. Attention should be taken to note the location and appearance of the anus. Delayed diagnosis of the anorectal malformation has the potential to contribute to unnecessary patient suffering and, to some extent, to diminished parental

confidence in their physicians. Vestibular fistula in newborns can be corrected mostly without the need for a stoma, which is difficult to obtain later in life.⁵

Learning points

- ▶ The newborn perineum should be thoroughly examined, even if they have passed meconium.
- ▶ Late diagnosis of anorectal malformations (ARM) can complicate definitive surgery.
- ▶ Missing ARM at birth can potentially raise medico-legal issues.

Competing interests None.

Patient consent Obtained.

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