Diagnostic clues to avoid pitfalls in the management of rare association of anorectal malformation with ileal atresia

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Clinical image

A full-term, 2700 g, one-day-old baby presented with imperforate anus, bilious vomiting and abdominal distension. Physical examination, ultrasonography abdomen and echocardiography revealed no associated congenital anomalies. Local examination of the perineum revealed white epithelial pearls in the penoscrotal raphe with well-developed gluteal folds (Fig. A), consistent with anocutaneous fistula, a low anorectal malformation (ARM). Since no meconium was seen in the perineum even after 20 hours of birth, a prone cross-table X-ray was done which suggested high ARM (Fig. B), as distal gas shadows were above the pubococcygeal line. Laparotomy was performed because of discrepancy in the clinical and radiological findings, which revealed distal ileal atresia and microcolon, with absent cecum and appendix. After resection of atretic bowel, double-barrel ileo-colostomy was performed. Postoperative course was uneventful. Distal cologram showed a low ARM with no genitourinary fistula (Fig. C). Subsequently, patient underwent ileo-colic anastomosis with anoplasty. At last follow-up, the baby was eighteen months old and was thriving well.

Associated gastrointestinal anomalies are seen in 10%-25% of ARM, with tracheoesophageal fistula and duodenal atresia being the commonest.[1] Association of ARM with intestinal atresia beyond duodenum, though described in literature, are rare with less than 10-reported cases in the English literature.[1,2] Besides, such associations are more likely to occur with high ARM, unlike low ARM in this case.[2] Correct preoperative diagnosis in such patients is difficult and requires a high index of clinical suspicion. Diagnostic clues that shall alert the clinician to suspect proximal intestinal atresia in ARM are: a) presence of mucus with no meconium at anoplasty or colostomy, b) microcolon at colostomy c) inconsistency in clinical and radiological findings, and d) associated polyhydraminos on antenatal ultrasonography.[1-3] Missed diagnosis carries a high morbidity and often needs reexplorations as reported in five of the six previously reported cases.[2-4] In a newborn with ARM, a prone cross-table lateral X-ray is usually obtained after 24 hours of birth if no meconium is visible in the perineum.[5] Although in the majority of cases with low ARM, it may be non-contributory; but in few patients like those with proximal bowel atresia and who have delayed presentation with perforation,[6] it may provide subtle diagnostic clues, which aid in optimal surgical management with no additional risk or delay in treatment.

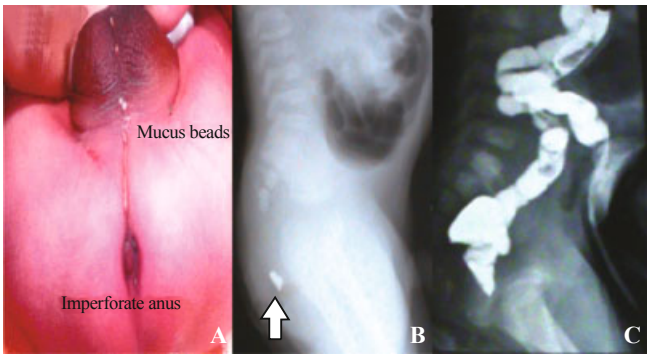

Fig. A: Clinical photograph showing absent anal opening with mucus beads in the midline raphe, suggestive of an anocutaneous fistula (low anorectal malformation); **B**: Lateral view of a prone cross-table X-ray showing distal most gas shadow above the pubococcygeal line, suggestive of high anorectal malformation (open black arrow indicates the marker at the site of anus); **C**: Distal cologram shows low termination of the bowel.

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References

1 Asabe K, Handa N. Anorectal malformation with ileal atresia. Pediatr Surg Int 1997;12:302-304.
2 Goodwin S, Schlatter M, Connors R. Imperforate anus and colon atresia in a newborn. J Pediatr Surg 2006;41:583-585.
3 Lin Steve CL, Khong PL, Tam PKH. Double intestinal atresia in imperforate anus: A diagnostic conundrum for Paediatric Surgeons. Asian J Surg 2004;27:52-53.
4 Asabe K, Nagasaki A. Double atresia of the hindgut with ileal stenosis: A case report. Asian J Surg 2004;27:49-51
5 Levitt MA, Pena A. Anorectal malformations. In: Coran AG, eds. Pediatric surgery, 7th ed. Philadelphia: Elseiver Saunders, 2012: 1289-1309.
6 Raveenthiran V. Spontaneous perforation of the colon and rectum complicating anorectal malformations in neonates. J Pediatr Surg 2012;47:720-726.

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