CASE REPORT

Appendiceal agenesis discovered incidentally at laparotomy for suspected acute appendicitis: a case report and review of the literature

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Abstract

The vermiform appendix is a vestigial organ in humans. The congenital absence of the appendix was first described by Morgagni in 1719. Since then, more than a hundred such cases have been reported. The estimated incidence of this condition, based on autopsy studies and laparotomies, is 1 in 100,000, which is exceptionally rare. Nevertheless, given that acute appendicitis is one of the most common causes of abdominal pain, and is the most common indication for emergency abdominal surgery, the practising surgeon needs to be aware of this clinical entity as it can potentially lead to diagnostic confusion and even iatrogenic injuries. Hereby, we report the case of an 18-year-old girl, who presented with symptoms suggestive of acute appendicitis and was found to have congenital absence of the appendix intra-operatively.

Keywords: Appendicitis, Vermiform Appendix, Congenital Abnormalities, Anatomic Variation.

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Introduction

The vermiform appendix is an evolutionarily vestigial organ present in humans. The first medical report of congenital absence of the vermiform appendix was published by Morgagni in 1719.[1] Since then, more than a hundred such cases have been documented in the medical literature.[2] The estimated incidence of this condition, based on autopsy studies and laparotomies, is 1 in 100,000, which is exceptionally rare.[3] Nevertheless, given that acute appendicitis is one of the most common causes of abdominal pain, and is the most common indication for emergency abdominal surgery, the practising surgeon needs to be aware of this clinical entity as it can potentially lead to diagnostic confusion and even iatrogenic injuries.[4] Hereby, we report the case of an 18-year-old girl, who presented with clinical features suggestive of acute appendicitis and was found to have congenital absence of the appendix intra-operatively. Written informed consent was obtained from the patient's father for publication of this case report.

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Case Report

An 18-year-old girl presented to the casualty department of Hayatabad Medical Complex (Peshawar, Pakistan) on 13th September 2024, with a one-day history of abdominal pain. She had been in her usual state of health until two days prior to presentation. She developed abdominal pain of insidious onset, initially mild and ill-localized, which later intensified and migrated to the right lower quadrant of abdomen. The pain was described as sharp and was aggravated by food intake, coughing or movement. She denied any urinary complaints or alteration in bowel habits. She did endorse having some nausea and one episode of vomiting. She also confirmed having poor appetite after the onset of abdominal pain. There was no history of documented fever spikes or known contacts with sick individuals. Her past medical history was only significant for iron deficiency anaemia. There was no history of previous surgeries. There was no family history of malignancy in her parents or siblings. She had menarche at the age of 14 and her last menstrual period was about 15 days prior to current presentation. Her menstrual cycles were regular every 4 weeks, with period lasting 2 to 3 days. She was unmarried and denied any history of tobacco use.

On initial presentation, her vital signs were remarkable for tachycardia (pulse rate 104 beats per minute) and low-grade fever (oral temperature of 37.6°C). Physical examination revealed a young girl of average built, lying in bed in mild distress due to pain. Oral examination was only remarkable for hyperaemic pharynx and dry mucous membranes. Tonsils were normal in size without any exudates. Lungs auscultation was clear, and cardiac examination revealed no murmurs or gallops. On abdominal examination, she had a scaphoid abdomen with no visible surgical scars. Deep tenderness to palpation in the right lower quadrant as well as at McBurney's point was elicited. Rebound tenderness and Rovsing sign (right lower quadrant abdominal pain upon

Table-1: Results of laboratory investigations for our patient.

Investigation	Result	Reference range
Haemoglobin	13.9 g/dL	12.0 – 16.5 g/dL
Haematocrit	41.3%	36.0% – 50.0%
Mean corpuscular volume	90 fL	80 – 96 fL
Red blood cell count	4.58×10^{12}/L	$4.1 – 5.3 \times 10^{12}$/L
Total leucocyte count	14×10^{9}/L	$3.6 – 11.2 \times 10^{9}$/L
Platelet count	236×10^{9}/L	$150 – 400 \times 10^{9}$/L
Sodium	131 mmol/L	136 – 144 mmol/L
Potassium	3.6 mmol/L	3.6 – 5.1 mmol/L
Chloride	106 mmol/L	101 – 111 mmol/L
Bicarbonate	28 mmol/L	22 – 32 mmol/L
Blood urea nitrogen	19 mg/dL	8 – 20 mg/dL
Creatinine	0.91 mg/dL	0.44 – 1.03 mg/dL
Glucose	109 mg/dL	70 – 140 mg/dL
Calcium	8.9 mg/dL	8.9 – 10.3 mg/dL
Alanine aminotransferase	27 IU/L	15 – 41 IU/L
Aspartate aminotransferase	27 IU/L	14 – 54 IU/L
Total bilirubin	0.8 mg/dl	0.3 – 1.2 mg/dL

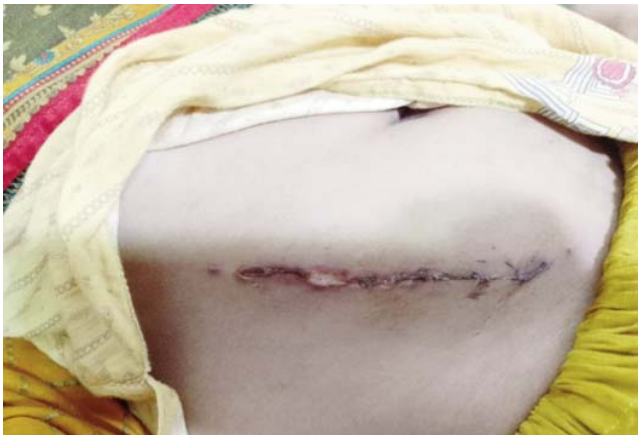

Figure-1: Post-operative picture of the patient's surgical wound at follow-up visit.

applying and releasing pressure to the left lower quadrant) were also present. Shifting of tenderness with change in posture was also noted. Bowel sounds were slightly hypoactive.

Her laboratory investigations are detailed in Table 1 and were remarkable for neutrophilic leucocytosis (total leucocyte count 12×10^9 cells/L with 92% neutrophils). Her urinalysis was negative for leucocyte esterase or nitrite. Her urine pregnancy test was also negative. Intravenous access was established and she was resuscitated with 2 L of intravenous lactated Ringer's solution. She was also given IV diphenhydramine 25 mg for nausea, IV ketorolac 15 mg for pain and IV famotidine 20 mg prophylactically to prevent ketorolac-induced gastritis. Given an Alvarado score of 9, the pretest probability of acute appendicitis was considered very high and pre-operative imaging was not pursued. Therefore, the patient was taken directly to the operating room for open appendicectomy. Intra-operatively, despite tracing the taeniae coli to their junction, mobilization of the right colon and careful inspection of the ileocaecal junction, the appendix could not be visualized. In view of this, the incision was extended superiorly and onto the right upper quadrant, and the entire length of the small bowel as well as large bowel was inspected. No appendix could be visualized, even though the caecum appeared normal and lacked a mesentery. Epiploic appendages appeared normal. A few subcentimeter mesenteric lymph nodes were observed, which were deemed to be too small for biopsy and were not considered pathologic. No Meckel's diverticulum or intestinal malrotation could be identified. Fallopian tubes and ovaries were examined, and were only notable for two small (~1.5 cm) ovarian cysts. The abdominal wall, subcutaneous tissues and skin were closed in layers in usual manner.

The patient recovered from anaesthesia uneventfully. Her abdominal pain gradually improved and she was discharged home on oral analgesics. Her pelvic ultrasonography was performed, which did not reveal any tubo-ovarian pathology. The final diagnosis remained indeterminate, with possibilities including Mittelschmerz or mesenteric lymphadenitis. At a follow-up visit, the patient was doing well and her surgical wound was healing well (Figure 1).

Discussion

Congenital absence of the appendix is an extremely rare but well-described clinical entity.[1,3] Embryologically, the human appendix develops from the mid-gut, which forms a caecal diverticulum—progenitor of both adult caecum and appendix. By 8 weeks of gestation, the appendix becomes microscopically visible in the foetus and continues to elongate and rotate with the caecum during development, ultimately residing in the right lower quadrant.[5] Agenesis of the appendix results from atresia or arrested development of progenitor cells that give rise to the appendix, although intrauterine vascular accidents due to fibrous bands may also be implicated.[5] In order to make a diagnosis of appendiceal agenesis intra-operatively, all other anatomic variants of the appendix need to be excluded, especially intramural appendix (embedded within the wall of the caecum) or in unusual locations inside the abdominal cavity.[5,6] The Collin's classification categorizes appendiceal agenesis into five types depending on the presence or absence of caecal malformations—given the common origin of appendix and caecum from the caecal diverticulum.[7] In this case,

appendiceal agenesis was identified in the presence of a normal caecum—classified as Collin's type III.

In previously reported cases of appendiceal agenesis, the diagnosis was most commonly established in patients who presented with symptoms of suspected acute appendicitis and were found to have congenital absence of the appendix during laparotomy or laparoscopy.[2,4,6] Occasionally, the diagnosis of congenital absence of the appendix has been made on elective abdominal or pelvic surgeries performed for other indications.[10] In all such cases, obtaining a detailed history of prior surgeries is essential. It is also important to confirm the surgical history with the patient's parents to ensure that no prior abdominal surgeries were performed during childhood. In the present case, the surgical history was reconfirmed with both the patient and her parents, and it was established that the patient had never undergone any abdominal surgery in the past.

Acute appendicitis is a common cause of abdominal pain and, despite advances in imaging modalities, the negative appendicectomy rate remains as high as 20%.8 The Alvarado score is the most commonly used diagnostic tool for making a presumptive diagnosis of acute appendicitis, with surgery typically indicated for patients with a score of 7 or higher. However, a systematic review has shown that the Alvarado score tends to overestimate the likelihood of acute appendicitis in women.[9] In our patient, the Alvarado score was 9 and patient was taken directly to the operating room where a diagnosis of appendiceal agenesis was confirmed after meticulous inspection of the entire bowel. It is important for the operating surgeon to consider the possibility of anatomic malformations of the appendix, as unrecognized variants pose a high risk of iatrogenic injury.[4]

Conclusion

Appendiceal agenesis is an extremely rare congenital anomaly of the appendix. The practising surgeon needs to be aware of this rare anomaly as it may lead to diagnostic confusion and inadvertent iatrogenic injury when discovered at surgery.

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AUTHOR'S CONTRIBUTION:

MNK, MK, MBK, MT & AR: Concept, design, collected patient's data, drafting, reviewing, final approval and agreement to be accountable for all aspects of the work.