Case Report

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Langerhans cell histiocytosis presenting as a cecal mass in an adolescent: a rare surgical case report

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Introduction and importance: Langerhans cell histiocytosis (LCH) is a rare, idiopathic, multifocal disease that commonly occurs in the skin and skeletal system, although involvement of the gastrointestinal tract is exceptional. Only a limited number of cases have been reported in the literature, particularly at the intestinal level, often with incidental diagnosis after colonoscopic polypectomy, and presentation as a cecal mass is extremely uncommon.

Case presentation: A 15-year-old boy presented with abdominal pain and a non-tender right iliac fossa mass. Hematological and biochemical tests were within normal limits. Initial imaging studies, including ultrasound and contrast-enhanced computed tomography, revealed a cecal mass associated with mesenteric lymphadenopathy, with radiological features suggestive of lymphoma. Because of intolerance to bowel preparation, colonoscopy could not be performed. In addition to a history of treated intermittent distension with vomiting, which raised concern for recurrent sub-acute intestinal obstruction, a laparoscopy-assisted surgical approach was therefore chosen. Intraoperatively, a cecal mass with multiple focal swellings of the mesocolon was identified. A right hemicolectomy with ileo-transverse anastomosis was performed according to oncological principles. Post-operative PET imaging showed no evidence of residual disease, and the patient was subsequently enrolled in a follow-up program. This case highlights the rarity of intestinal LCH and the importance of considering it in the differential diagnosis of abdominal masses in pediatric patients. Radiological imaging studies have shown neither residual disease nor recurrence during 9 months of regular follow-up.

Clinical discussion: The World Health Organization classifies LCH within the hematopoietic and lymphoid tumors placing it on a spectrum between benign and malignant behavior depending on other factors, with unifocal or multifocal lesions and single- or multisystem involvement, which likely predict prognosis and guide post-surgical treatment.

Conclusion: The rarity of intestinal LCH presenting as a cecal mass highlights the importance of considering it in the differential diagnosis of abdominal masses in pediatric patients.

Keywords: cecal mass, gastrointestinal tract, LCH

Case presentation

A 15-year-old boy presented with abdominal pain, anorexia, nausea, and a non-tender right iliac fossa mass, with a past surgical history of recurrent attacks of abdominal distension with vomiting that were treated conservatively. Ultrasound of the abdomen revealed a right iliac fossa mass (5.5 × 5 × 5.7 cm) with multiple lymph nodes losing their central hili in the vicinity, raising suspicion of lymphoma, as shown in Figure 1a–c.

Dynamic computed tomography of the abdomen showed a cecal mass measuring 6 × 6 × 5.4 cm, occupying the whole cecal lumen, with an enlarged appendix and regional mesenteric lymph nodes that enhance with contrast, raising the same suspicion.

Due to intolerance to bowel preparation, colonoscopy could not be performed despite a history of treated intermittent distension with vomiting that could be alarming for recurrent sub-acute intestinal obstructions. A laparoscopic-assisted surgical approach was therefore chosen.

Intraoperatively, a cecal mass with multiple focal swellings of the mesocolon was identified. A right hemicolectomy with

HIGHLIGHTS

- Langerhans cell histiocytosis (LCH) is a rare disease. Cecal mass as a presenting complaint may be extremely rare.
- Surgical excision of the involved part with post-operative follow-up may be the cornerstone step of management.
- PET imaging is of great value in assessing multifocality and staging of disease, with tremendous importance in elaborating the need for other modalities of treatment.
- Prognosis depends on systemic involvement; the more systemic involvement, the worse the prognosis.
- The rarity of intestinal LCH presenting as a cecal mass highlights the importance of considering it in the differential diagnosis of abdominal masses in pediatric patients.

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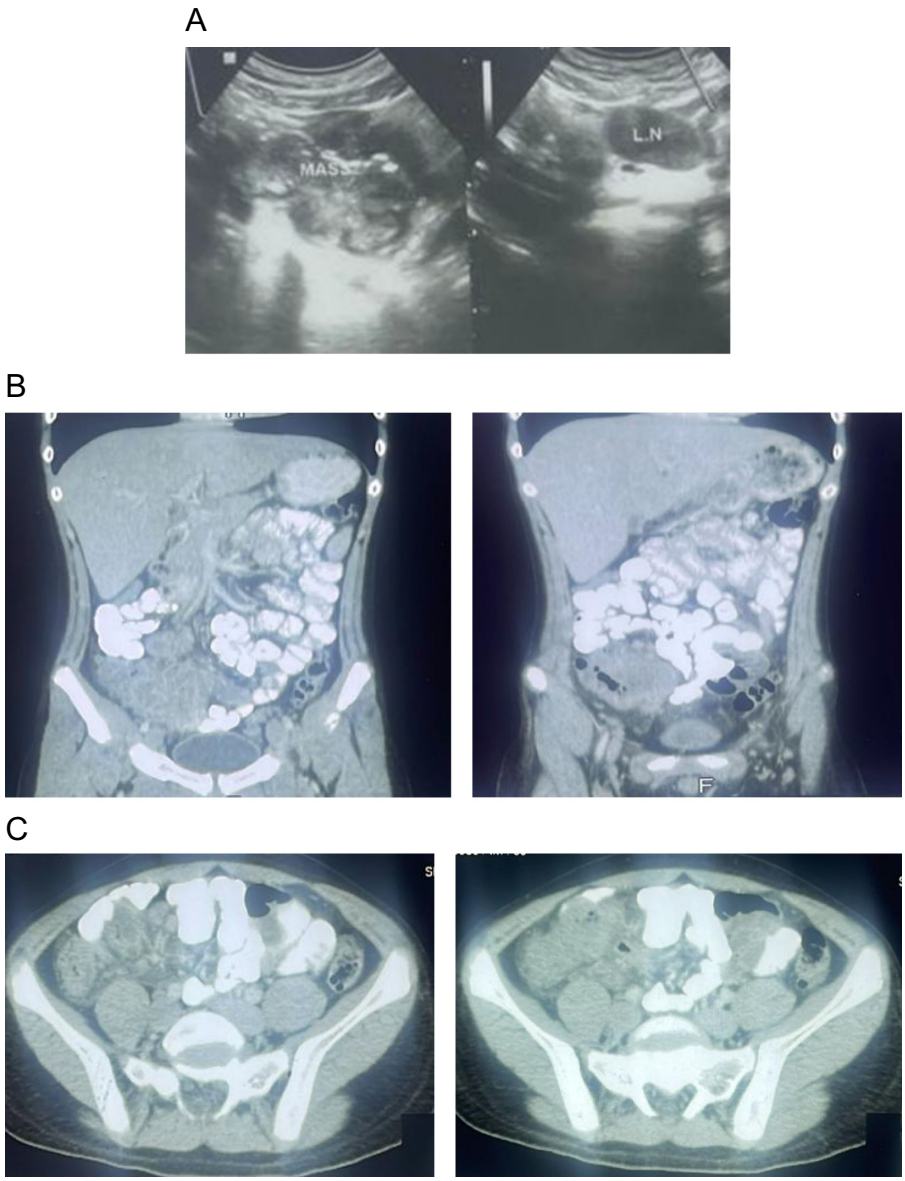


Figure 1. (a) Ultrasound showing cecal mass with mesenteric lymph node involvement. (b) Dynamic computed tomography scan of the abdomen, sagittal view. (c) Horizontal view showing cecal mass and mesenteric lymph nodes that enhanced with contrast.

ileo-transverse anastomosis was performed according to oncological principles. Post-operative PET imaging showed no evidence of residual disease, and the patient was subsequently enrolled in a follow-up program.

Radiological imaging studies have shown neither residual disease nor recurrence over 9 months of regular follow-up.

Pathology

Gross

Cecal mass (6.8 × 6.7 × 6.2 cm) completely obstructed the ileal lumen, with an ulcerated mucosa and involvement of neighboring regional lymph nodes; the appendix looked enlarged and congested; however, the terminal ileum was apparently not diseased, as shown in Figure 2.

Microscopy

Histopathology revealed characteristic Langerhans cell hyperplasia admixed with a large number of eosinophilic cells with rare mitotic figures. The tumor infiltrated the whole ileal and cecal wall, including the muscularis externa, with periserosal interruption toward the fatty tissue. All proximal, distal, and deep margins are tumor-free. Three out of 14 pericolic lymph nodes are involved by tumor, with preserved active follicles. Immunohistochemistry for CD68, S100, and CD1a, which are easy and rapid techniques, showed positive staining. Molecular testing was not done because of its unavailability in our country, as shown in Figure 3a–c.

Clinical discussion

The Langerhans cell histiocytosis (LCH) tumor is a rare disease characterized by infiltration of histiocytes that resemble

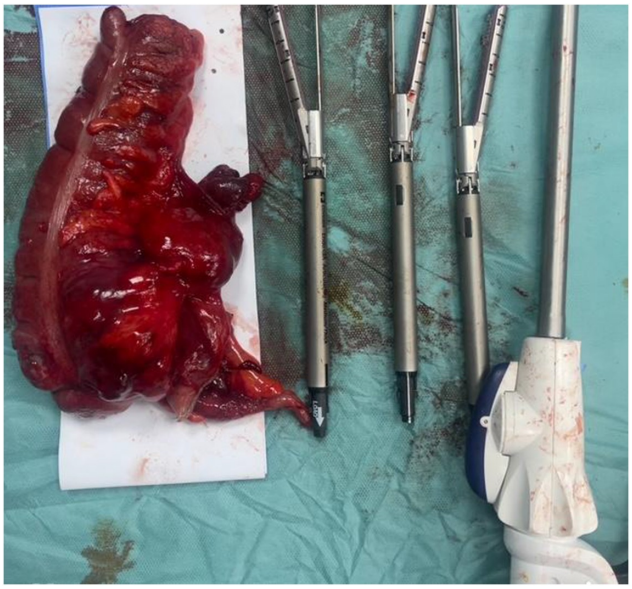


Figure 2. Right hemicolectomy showing a gross cecal mass with mesenteric swellings, and the appendix looks enlarged and congested.

Langerhans cells of the skin and dendritic tissue, which explains the tumor's name.

The World Health Organization classification of hematopoietic and lymphoid tumors categorizes disorders of histiocytes into three specific groups – dendritic cell disorders, macrophage-related disorders, and malignant histiocytic disorders[1] – with a debated pathogenesis, either reactive or neoplastic. The latter is associated with the BRAF V600E mutation in 57% of LCH cases[2].

The incidence is three to five cases per million in children and one to two cases per million in adults[3,4]. Bones, skin, and lungs are the most commonly affected organs, with extremely rare gastrointestinal involvement from the mouth to the anus, as well as the surrounding skin along the gastrointestinal tract[5].

The gastrointestinal presentation lies on a spectrum ranging from nausea, bloating, bleeding, and ulcerative colitis-like features, and the condition was incidentally diagnosed by histopathology after colonoscopic polyp snaring[6].

Singhi and Montgomery[7] and his colleagues at John Hopkins reported a case series of 12 gastrointestinal tract LCH cases in 2011, of whom 10 were adults. The characteristics of these patients are reported in Table 1. Endoscopically, one patient had stomach involvement, and the rest had colorectal involvement either as polyps (seven patients) or as ulcers (two patients). Interestingly, the two patients who presented with ulcers later developed skin lesions within 2 years of follow-up.

A right iliac fossa mass, with its differential diagnoses – especially cecal lymphoma – is an extremely rare presentation. Imaging studies, including a dynamic contrast-enhanced computed tomography scan of the abdomen, revealed a cecal mass with regional lymph nodes that showed contrast enhancement.

Due to intolerance to bowel preparation, colonoscopy could not be performed. In addition to a history of treated intermittent distension with vomiting that could be alarming for recurrent

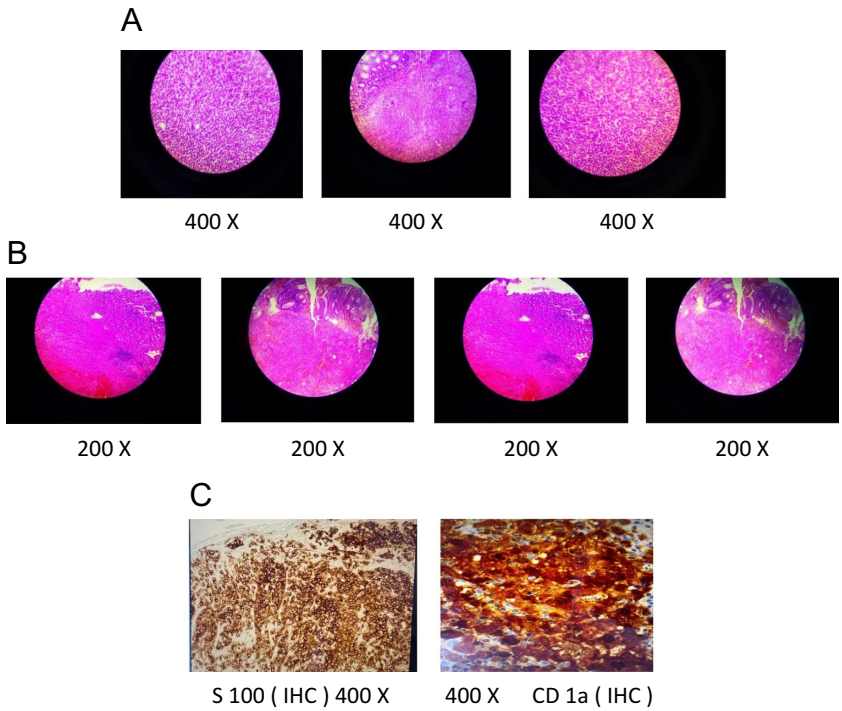


Figure 3. Section(200×) showing expansion of the lamina propria by diffuse lymphohistiocytic cells, with distortion and destruction of the mucosal glandular pattern. Section (400×) showing heterogeneous infiltrate composed of large numbers of eosinophils, mature lymphocytes, and Langerhans cells (12–15 μm) in diameter with abundant pale eosinophilic cytoplasm, irregular and elongated nuclei with prominent nuclear grooves and folds, pink chromatin and distinct nucleoli, rare scattered multinucleated cells, variable rare mitotic activity, lacking any associated interstitial fresh hemorrhage and/or tumor necrosis. Immunohistochemistry showed positivity for S100, CD68, and CD1a, with membranous and cytoplasmic reactions.

Table 1

Singhi and Montgomery[7] case series of GI tract LCH in adult population.

Age & gender	Clinical presentation	Endoscopic findings	Location	No. of lesions	Follow-up
60 M	Routine	Polyp	Cecum	1	Negative
60 F	Routine	Polyp	Cecum	1	Negative
40 F	Anemia	Polyp	AC	1	Adenoma
51 F	Routine	Polyp	TC	1	Negative
55 F	Routine	Polyp	Sigmoid	1	Negative
77 F	Constipation	Polyp	Sigmoid	1	Negative
56 F	Routine	Ulcerative	AC, TC, DC	Multiple	Cutaneous disease
54 F	Constipation	Ulcerative	Anus	1	Cutaneous disease
63 F	Cecal volvulus	N/A	AC and Cecum	Multiple	Negative
68 M	Dysphagia	Polyp	Stomach antrum	1	Negative

sub-acute intestinal obstruction, a laparoscopic-assisted surgical approach was therefore chosen.

Intraoperatively, a cecal mass with multiple focal swellings of the mesocolon was identified. A right hemicolectomy with ileo-transverse anastomosis was performed according to oncological principles.

A cecal mass diagnosed as LCH by histopathology is an extremely rare presentation compared with the above few gastrointestinal cases that establish this disease diagnosis by polypectomy during routine colonoscopy[8–11].

The pathogenesis is not fully understood because of its rare occurrence. LCH is clinically classified into three groups: unifocal involvement in a single organ system, multifocal involvement in a single organ system, or multisystem involvement. This classification is of paramount importance because each category is treated with its own regimen to minimize the toxicity and morbidity of the disease[12].

PET-imaging scan is recommended to evaluate the extent, severity, and follow-up of the disease, with a very low false-positive rate, and may ultimately be of benefit in mapping the nature of the treatment regimen[13].

The outcome of management depends on multiple factors, principally the fact that patients with multisystem involvement have a poor prognosis.

The case report, "Langerhans Cell Histiocytosis Presenting as a Cecal Mass in an Adolescent: A Rare Surgical Case Report," was written in accordance with the SCARE guidelines[14].

Conclusion

This case highlights the rarity of intestinal LCH and the importance of considering it in the differential diagnosis of abdominal masses in pediatric patients.

Ethical approval

Ethical approval was obtained from the ethics review board.

Consent

Informed consent was obtained from the participating patient for the publication of anonymized information in this article.

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Author contributions

H.S.H. and Y.J.K. contributed to the conception, preparation, critical revision, and final approval of manuscript.

Conflicts of interest disclosure

The authors declare that they have no related conflicts of interest in this work.

Research registration unique identifying number (UIN)

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All data relavent to this case report are included in the article.

Declaration of Generative AI During the preparation of this work

The authors did not utilize any generative AI or AI-assisted technologies.

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