Langerhans cell histiocytosis in a patient with perianal lesion. A case report

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ABSTRACT

Langerhans cell histiocytosis is an expression of myeloid dendritic cells, associated with a significant inflammatory component and varied systemic involvement. The most common age at presentation is between 1 and 4 years, and it prevails among male subjects. Here we describe the case of a 5-year-old boy who presented with a granulomatous lesion with perianal fistula and lung and external ear involvement. An interdisciplinary approach helped to make a diagnosis, provide the necessary interventions, and start an adequate treatment.

Keywords: Langerhans cell histiocytosis; fistula; perineum; anal sphincter.

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INTRODUCTION

Langerhans cell histiocytosis (LCH) is a rare disease characterized by uncontrolled proliferation and accumulation of dendritic cells.1,2

Children are the most commonly affected age group. The peak of incidence of LCH occurs between 1 and 4 years old, with a mild predominance among male subjects.2,3

Possible risk factors include a family history of cancer, recurrent infection during childhood, and parental occupational exposure to metals, granites, or wood dust.4

Its clinical presentation may affect a single system, with bone as the most frequent site, or multiple systems, with involvement of the lung, liver, or hematopoietic system. In the localized form, signs and symptoms depend on the affected site; they usually manifest with pain, tenderness, and increased volume.5,6

Histopathology varies according to the organ affected and the stage of LCH. The proliferation of Langerhans cells forming granulomas together with lymphocytes, macrophages, and eosinophils is observed, although the presence of the latter is not necessary to make a diagnosis.7

Prognosis is related to age; patients younger than 2 years, despite being considered high-risk, have a good response to treatment if they do not have multisystem involvement. Patients with single organ involvement generally have a favorable prognosis with a high chance of spontaneous remission.8 Hypoalbuminemia, anemia, and other cytopenias are considered risk factors for a poor prognosis.1,9

Here we describe the case of a child diagnosed with Langerhans cell histiocytosis and perianal fistulas in association with an external auditory canal lesion and lung involvement.

CASE REPORT

This was a 5-year-old boy with normal growth and development parameters. At 18 months old, he developed a perianal fistula that resolved spontaneously.

The reason for consultation with the Department of Pediatric Surgery of our hospital was painful bowel movements and perianal irritation for the past 2 months. The patient’s physical examination was difficult due to his irritability, but his parents provided a photograph showing multiple small fistulas and a larger fistula at 3 o’clock. Antibiotic treatment was started with amoxicillin-clavulanic acid, sitz baths, and topical corticosteroid cream. Due to suspected inflammatory bowel disease, the patient was referred for a consultation with the Department of Pediatric Gastroenterology, where perinuclear anti-neutrophil cytoplasmic antibody (ANCAp), blood anti-Saccharomyces cerevisiae antibody (ASCA), and fecal calprotectin tests were requested; results were negative.

A week later, the patient showed little response to treatment and continued with painful bowel movements. The physical examination was difficult, even with the parents’ help. Treatment with metronidazole was started.

A month later, he was assessed again but no improvement was observed; he continued with painful bowel movements. A new physical examination was performed under anesthesia. A tortuous, granulomatous perianal lesion was observed at the perianal level, distorting the anatomy and involving the external anal sphincter. No lesions were observed in the rectal mucosa (Figures 1 and 2). A sample was collected for biopsy.

The consultation with the Department of Mental Health due to suspected abuse was not conclusive.

A computed tomography (CT) scan of the abdomen and pelvis with oral and intravenous contrast was unremarkable. The results of general lab tests, tumor markers, immunoglobulins, and serologies were normal.

The pathology report described a lesion compatible with a histiocytic granulomatous process (eosinophilic granuloma—Langerhans cell histiocytosis). A referral for immunohistochemistry was recommended.

Due to diagnostic suspicion, a new targeted case history was taken. The parents reported a history of recurrent suppurative otitis, currently with bloody discharge; the otoscopy was difficult. A skull x-ray for round osteolytic injuries was normal.

A loop colostomy was performed to defunctionalize the affected region. During the same surgical procedure, both ears were examined. A granulomatous or angiomatous lesion was observed that prevented visualization of the left tympanic membrane and involved the external ear. The lesion was resected and a compression tamponade was placed. The biopsy found its histology was similar to that of the perianal lesions.

The patient was referred to a tertiary care facility that offered Gastroenterology and Pediatric Oncology services to continue with his ancillary treatment.
tests and establish a diagnosis and eventual treatment. There, the diagnosis was confirmed by morphology and immunohistochemistry tests and treatment with meprednisone and vinblastine at 6 weekly doses was started. The study of lymphocyte populations showed T-cell lymphopenia, so prophylactic treatment with trimethoprim-sulfamethoxazole was added.

A lung CT showed an anarchically arranged lung parenchyma, multiple cysts, and abnormal lung parenchymal spaces. Function tests were requested, including a gait test, a spirometry, and diffusion of carbon monoxide; all were unremarkable.

Since the initiation of treatment with corticosteroids and chemotherapy, the patient's course was favorable. The initial lesion continues improving towards healing (Figure 3); after 5 months of treatment, the lesion reduced in size, there is erythema in the perianal region, and there are no fistulas or associated lesions.

The Department of Oncology assesses the patient in the City of Buenos Aires every 21 days. His primary pediatrician conducts a longitudinal follow-up at his place of residence. An interdisciplinary approach helped to make a diagnosis, provide the necessary interventions, and start an adequate treatment.

DISCUSSION

LCH is a rare group of disorders of unknown etiology, with various clinical presentations.
The age and sex of the clinical case described here are consistent with the most frequent descriptions in the bibliography. The most commonly affected organs are the skeleton (80%), the skin (33%), and the pituitary gland (25%). Other organs involved include the liver, spleen, bone marrow, lungs (15%), lymph nodes (5–10%), and the central nervous system excluding the pituitary gland (2–4%). In our patient, these organs were not affected, except for the lungs.

The main clinical manifestations in LCH are related to the affected site. In our patient, the lesions and fistulas located in the perianal region account for his painful bowel movements. Perianal involvement is a rare form of presentation. The patient’s recurrent suppurative otitis episodes from an early age were interpreted as associated with this condition once a pathological diagnosis was made. Perianal involvement is a rare form of presentation. The patient’s recurrent suppurative otitis episodes from an early age were interpreted as associated with this condition once a pathological diagnosis was made. The clinical presentation and prognosis of LCH vary widely from a self-limited unifocal bone lesion to a fatal multisystem disease with involvement of the lungs, liver, spleen, and hematopoietic system. In older children, LCH is usually localized, whereas a more aggressive form of disease with organ dysfunction is typical in children under 2 years of age.

The lung involvement in our patient gives rise to a guarded prognosis in relation to cancer. Other prognostic factors to consider in this child were his age at onset of perianal lesions before 2 years of age and his sex.

Other similar cases of LCH with perianal involvement have been reported. In one case, along with a skull lesion; in another, associated with gingival lesions and systemic disease; and a case of a child with a perianal mass presented with multiple lytic lesions of the skull, a right scapular lesion, and a right paratracheal lymph node. All cases responded to systemic chemotherapy.

Clinical and radiological findings are non-specific. A biopsy is necessary to establish a diagnosis. According to some authors, surgery is considered necessary only when there is neurological and/or soft tissue involvement, as in the case of our patient, which allows collecting an adequate specimen for a pathological study.

The pathology report helps to confirm diagnosis by microscopy and immunohistochemistry, which shows a positive characteristic marker, CD1a, as in our patient.

Treatment is determined on an individual basis and depends on the severity of the disease. While patients with localized involvement at a single site usually receive local therapy, those with multisystem LCH require systemic chemotherapy.

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Figure 3. Perianal lesion after 5 months of treatment
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REFERENCES


