CLINICAL CASE

Perianal presentation of Langerhans cell histiocytosis in children

Présentation périanale d’une histiocytose langerhansienne chez l’enfant

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Summary Langerhans cell histiocytosis, previously known as histiocytosis X, is a disease whose clinical presentation varies. Although it is uncommon, Langerhans cell histiocytosis may involve the perianal region. We report the case of a 2-year-old boy who presented with perianal ulcerated vegetative lesions and seborrhic dermatitis of the scalp. Biopsy of the lesions showed Langerhans cell histiocytosis. This patient did not have any other organ involvement, which is rare. The outcome was favourable with vinblastine and corticoids.

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Résumé L’histiocytose langerhansienne, anciennement dénommée histiocytose X est une maladie caractérisée par une expression clinique variable. Bien que rare, l’histiocytose langerhansienne peut toucher la région péri-anale. Nous rapportons l’observation d’un garçon de deux ans qui présentait une lésion péri-anale ulcérovégétante et des lésions de dermatite séborrhéique du cuir chevelu. La biopsie des lésions a confirmé le diagnostic d’histiocytose langerhansienne. Ce patient n’avait pas d’autres atteintes ; cette condition est rare. L’évolution a été favorable sous vinblastine et corticoïdes.

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Introduction Langerhans cell histiocytosis (LCH) is caused by a clonal proliferation of Langerhans cells. The condition can present as solitary bone lesions or as a systemic disorder with a wide clinical spectrum. We report the case of a 2-year-old boy who presented with perianal Langerhans cell histiocytosis.

Case report

A 2-year-old boy was admitted to our department to manage a 7-month history of perianal lesions. He did not have any other symptoms, in particular fever, cough, dysuria, abdominal pain, joint pain, or blurring of vision. On physical
examination, his weight and height were normal for his age and the pulse, temperature, blood pressure, and respiratory rate were normal. He had seborrheic dermatitis of the scalp, and bilateral auricular discharge with polyps. Anorectal examination showed a large ulcerated and vegetative lesion (Fig. 1). Results of the general physical examination were otherwise unremarkable. There were no oral lesions, lymphadenopathy, hepatosplenomegaly or dysmorphic features. Laboratory evaluation revealed mild anaemia with a haemoglobin of 10 g/dL, hematocrit of 32%, a mean corpuscular volume of 62 fL, a platelet count of 475,000/mL, an erythrocytic sedimentation rate of 12 mm/h, C-reactive protein < 6 mg/l, total serum protein concentration of 69 g/L, serum albumin concentration of 43 g/L, alanine aminotransferase was 20 U/L, aspartate aminotransferase was 5 U/L.

The perianal lesion swab was positive for *Escherichia coli* and group B. *streptococcus*. Culture of auricular discharge was positive for *E. coli* and *Proteus mirabilis*. Crohn’s disease, sexual abuse or an immune deficiency were suspected at this time. There was no sexual abuse. The patient received antibiotics without any improvement. HIV, syphilis and herpes serologies were negative. Abdominal ultrasound and small bowel barium were normal. Colonoscopy and colonic biopsies specimen were normal. Biopsy of the mass revealed granulation tissue with sheets of histiocytes, which had the characteristic cytological features of Langerhans’ cells and were strongly positive for CD1a antigen immunostain (Figs. 2 and 3). Biopsy of the scalp showed the same pattern. Based on these findings, a diagnosis of LCH was made. To rule out LCH involvement in other systems, a full skeletal survey was performed, and no bone lesions were found. Chest X-ray and computed tomography of the temporal bones were normal. A bone marrow biopsy revealed no features of histiocytosis. The patient was registered as a low-risk patient with multisystem disease (LCH group II) and was treated with prednisone and vinblastine with improvement of both the perianal lesion, as well as the seborrheic dermatitis and auricular discharge, 1 month after beginning treatment.

**Discussion**

LCH is a systemic disorder that largely affects children, with an incidence of 1/200,000 to 1/350,000 [1]. LCH, previously known as histiocytosis X, is a disease whose clinical presentation varies. Histologically, LCH is characterized by the accumulation of Langerhans cells along with lymphocytes, eosinophils, and macrophages. The histopathology of LCH has been extensively described [2,3]. On light microscopic analysis, LCH is suggested by the presence of large mononucleated cells with abundant eosinophilic cytoplasm and “coffee bean” or convoluted nucleus. The gold standard of the diagnosis of LCH has been the identification of the ultrastructural Birbeck or Langerhans cell granule. The Langerhans cells express CD1a and S100 protein, so the demonstration of CD1a by immunohistochemistry helps confirm the diagnosis. The etiology of LCH is obscure. Molecular studies have demonstrated clonal cell proliferation. However, LCH has not been considered a malignant disorder because of the frequent occurrence of spontaneous
remissions and the absence of aneuploidy or karyotypic abnormalities in lesional cells [4,5]. LCH is currently thought to represent a cytokine-mediated reactive cellular proliferation. So far, no studies have identified immunologic or viral etiologies.

LCH occurs in many clinical forms, affecting different systems and different sites in the same system with widely variable clinical outcomes [6,7].

The most characteristic dermatological presentation is scalp involvement. The scalp is erythematous with greasy scales, quite similar to seborrheic dermatitis. On the trunk, the lesions are discrete, yellow-brown, scaly papules, often with areas of purpura. Our patient had scalp involvement mimicking seborrheic dermatitis. Mucocutaneous involvement has been consistently reported to be more common in multisystem disease than in single-organ disease; 82% of patients with skin involvement had coexisting LCH lesions in other body systems [8]. Only six cases of pediatric perianal lesion related to LCH have been reported; three of them had multisystemic disease, one patient had perianal lesions with gastrointestinal involvement without any other systemic localisation and two patients had focal perianal lesions without systemic involvement [9—14]. To the best of our knowledge, this is a first case of LCH with cutaneous-mucosal involvement including the perianal region, scalp and external auditory meatus, with no other organ involvement. Perianal lesions related to LCH in adults have also been reported [15].

The common differential diagnosis of an anal lesion includes perianal Crohn’s disease with abscess or fistula, sentinel skin tags, Verrucous vulgaris, Condyloma acuminatum, Streptococcus proctitis cellulitis, and Hidradenitis suppurativa [11]. Granulation tissue may also be a result of a laceration related to perianal trauma, and child abuse should always be considered as well. In our case, B. staphylococcus and E. coli were isolated from the perianal lesion swab; however they were not the cause of the lesion. Involvement of the ears is common and presents as persistent aural discharge, polypoid involvement of the external auditory canal, or deafness [16]. Our patient had bilateral aural discharge and polypoid involvement of the external auditory canal that responded to treatment. The therapeutic regimen for LCH depends on the localization and extent of the disease. The optimal management of mucocutaneous LCH is not clearly defined. Observation, curettage, excision, intralesional corticosteroids, radiation therapy, nonsteroid anti-inflammatory agents and systemic chemotherapy have been used with different outcomes. In our case the outcome was favourable with corticosteroids and vinblastine.

Although the perianal area is an unusual site of skin involvement in histiocytosis, this disorder should be considered in any child with chronic unexplained perianal disease, and a biopsy of these lesions should be obtained.

Conflicts of interest

There are no conflicts of interest.

References