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Case Report

Challenging diagnosis of the Langerhans cell histiocytosis in pediatrics: A case report

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ABSTRACT

LCH is a rare, sporadic, and challenging disease manifested by single or multiorgan involvement. Thyroid enlargement, as the initial manifestation, is mostly presented in the pediatric population, which is resistant to conventional treatments for goiter. The timely diagnosis of LCH is important to prevent undesirable complications. This paper presents an 18-month-old girl with an enlarged thyroid in whom papillary cutaneous lesions were found on the scalp, neck, and diaper area, and solitary punched out lytic lesion of the skull. The definite diagnosis of LCH was made after 1.5 months of the initiation of the first symptom. The patient underwent chemotherapy with acceptable results.

Keywords: Langerhans cell histiocytosis, pediatric, thyroid, goiter, skin, skull lesions

Introduction

Langerhans cell histiocytosis (LCH) is the inflammatory neoplasia of the myeloid precursor cells caused by mutations of the mitogen-activated protein kinase pathway, which ultimately results in hard and soft tissue destruction. Children are not usually affected by this condition, with an incidence of 2.6-8.9 per million children aged below 15 years [1], the prevalence is probably higher because localized disease regresses spontaneously. The reported peak incidence of LCH is between 1 to 5 years of age. Boys are more affected than girls, with the male/female ratio of 3.7/1 [2]. LCH could be presented as a single system or multisystem involvement, with various affected organs. The definite diagnosis is challenging, as different organs could be affected and various symptoms could be presented. This paper is going to report a rare case of LCH and the challenges we encountered to reach the final diagnosis.

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Methodology

Case presentation

An 18-month-old girl was referred to us due to progressive neck mass enlargement since one month before. The patient was diagnosed with Hashimoto's thyroiditis because of increased thyroid-stimulating hormone (TSH) level (10 µg/dl) and positive thyroid peroxidase antibody (anti-TPO). The neck mass enlargement continued despite levothyroxine administration and she was referred for further evaluations. Upon arrival at the hospital, she was admitted to the intensive care unit due to cyanosis and seizure, which lasted for 5 minutes. The episodes of cyanosis and seizure also occurred repeatedly during the admission. The physical examinations revealed a large non-tender thyroid, sizing approximately 4*5cm with non-firm texture. There was no organomegaly. There was no history of growth impairment, weight loss, or fever, and lethargy were detected. Papillary cutaneous lesions were found on the scalp, neck, and diapered area **(Figure 1)**. Based on the dermatology consultant, the patient was highly suspicious of histiocytosis. The laboratory tests showed

increased TSH level (48 µg/dl), decreased T4 (11 µg/dl), and negative anti-TPO. All the liver function tests, serum electrolytes, creatinine, and osmolality levels were within the normal range. Right lobe thyroid biopsy was taken and pathological evaluations were consistent with LCH. As a result, skull x-ray **(Figure 2)**, thoracic computed tomography (CT) scans, whole-body bone scan, and abdominal sonography were performed. All these investigations except skull x-ray, which indicated solitary punched out lytic lesion, were normal.

a)

b)

c)

Figure 1. Papillary cutaneous lesions in A: scalp, B: Neck, C: diapered area.

Figure 2. lateral skull Xray (Adenoid view)

Results

The patient underwent chemotherapy based on LCH Iranian pediatric oncology protocol, with the administration of daily Prednisolone and weekly Vinblastine for 6 weeks, followed by daily 6-mercaptopurine and weekly Methotrexate for one year. The patient was visited 6-months after the initiation of the treatment, the thyroid size was acceptable with normal thyroid hormones. The skin lesions were also resolved.

Discussion

Up to 30% of patients with LCH have a multi-system disease, and bones (70%) and skin (33%) are the most commonly affected organs [3]. The most common endocrinologic presentation of classic LCH is Diabetes Insipidus (DI) due to hypothalamus-pituitary involvement. The thyroid is rarely involved in LCH, and it is often associated with multiorgan involvement of LCH. The prevalence of thyroid LCH is one in 238 LCH cases [4]. The typical age of most children with thyroid LCH is less than 10 years; however, the thyroid LCH is much more common in the adult population. The most common manifestation of thyroid LCH is thyroid enlargement, goiter [4]. It usually presents as a palpable nodule (25.8%) or diffuse enlargement (59%) in one or two lobes [5]. This nonspecific presentation could be misdiagnosed with other diseases, including Hashimoto's thyroiditis, endemic goiter, thyroid lymphoma, thyroid carcinoma, and lymphoid tuberculosis [6]. Eight percent of patients are presented with lymphocytic (Hashimoto's) thyroiditis, while others are euthyroid (41%), hypothyroid (20%), subclinical hypothyroid (11%), or subclinical hyperthyroid (1.5%) [7]. Furthermore, anti-thyroid autoantibodies are also positive in some cases of thyroid LCH [7]. All the clinical manifestations, laboratory, and imaging work-ups resemble thyroid disorders, which make the diagnosis more difficult. A definite diagnosis could be made

through accurate history taking and detection of the other symptoms, such as bone or skin involvement, and most importantly the thyroid biopsy. Similar to our patient's symptoms, in 2019, He et al. [8] reported a 3.5-year-old boy with an enlarged neck mass, respiratory distress, seborrheic-like dermatitis, and hepatosplenomegaly. The initial manifestation of their patient was enlarged thyroid, which caused respiratory distress and other organs' involvement due to delayed diagnosis. Bones, especially the skull, are most commonly affected by LCH, which can be presented by either no symptoms or pain or localized soft tissue mass [4]. Asymptomatic solitary punched out lesion was also detected in our patient; this shows the importance of comprehensive physical examination for detecting the involved organ. Besides, about 53% of children with the multiorgan disease have skin lesions [9], the most common cutaneous lesion during childhood is seborrheic dermatitis-like lesions [10], similar to the described case, which is presented as erythematous or brown discrete papules on the skull, neck, axilla, and trunk. The skin lesions could also resemble persistent Diaper dermatitis, which was also present in our patient [10].

Conclusion

This experience showed that in case the enlarged thyroid gland with all laboratory work-ups consistent with thyroid disorders persist in spite of adequate treatment, LCH should be considered as the possible diagnosis and the patient should be investigated carefully for other organ involvement. The immediate biopsy of the tissues precipitates the process of definite diagnosis and also the initiation of the proper treatment.

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