Benign cephalic histiocytosis in a 2-year-old boy with an inconspicuous clinical presentation at onset

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Key Clinical Message:

We report a case of a natural course of benign cephalic histiocytosis (BCH) with favorable outcome under watch and wait strategy requiring no extensive diagnostic work-up given the benign and self-limited course of the disease and highlighting the clinical characteristics of BCH and its distinction from other non-Langerhans cell histiocytosis.

Introduction

In 1971 Gianotti et al. first described benign cephalic histiocytosis (BCH) and up to the present approximately 70 cases have been reported in the literature. (1, 2) BCH belongs to the group of non-Langerhans cell histiocytoses (NLCH) and clinically and histologically resembles juvenile xanthogranuloma (JXG) and generalized eruptive histiocytosis (GEH). Skin lesions are characterized by erythematous, yellowish to brownish macules and papules, which are mostly located on head and neck. The etiology of BCH is as yet unresolved.

Case Report

A two-year-old boy with a fair complexion presented with multiple, light brown macules, evenly distributed on the cheeks and the forehead (Fig. 1a-b). The family history was negative for skin diseases. According to the mother, small macular lesions had appeared at the age of 9 months on the left temple without any preceding inflammation. Subsequently, new lesions developed, particularly after the summer. Differential diagnoses included juvenile warts, freckles and urticaria pigmentosa. The Darier sign was negative. Since the lesions were restricted to the face and increased after environmental sun exposure also a diagnosis of xeroderma pigmentosum (XP) was considered. However, genetic testing for a mutation in the *ERCC* 3 gene was negative. Five months later the number of lesions had increased and the macules had evolved into monomorphic flat reddish brown papules. (Fig. 2a-b) The clinical presentation now indicated a diagnosis of benign cephalic histiocytosis (BCH). Confirmation of the clinical diagnosis by histopathology was not possible, as the mother did not consent to a biopsy from the face, where at that time the lesions were exclusively located. A complete blood count, comprehensive blood chemistry and an abdominal ultrasound were all unrevealing. In light of the benign nature of BCH and its propensity for spontaneous resolution a watchful waiting approach was chosen.

At follow-up examinations 9 and 16 months later a few new lesions had developed in the pelvic area and on the dorsal forearms and none of the pre-existing lesions had regressed. Two and a half year later part of the extracephalic lesions had completely disappeared and the lesions on the head were fading (Fig. 3a-b).

Discussion

The diagnosis of BCH is made on clinical grounds. Diagnostic criteria are the manifestation in early childhood (average 15 months), the typical clinical morphology and the predominant distribution in the head and neck region. However, contrary to its original designation as benign*cephalic* histiocytosis, several recent case reports have shown that extracephalic skin involvement, particularly on the upper trunk, is also commonly found. (2-4)

BCH is a benign disease without systemic involvement that runs a self-limited course. The lesions usually resolve within 50 months with occasional mild atrophy and hyperpigmentation. (3, 5) Recently, improvement of skin lesions has been reported in a 5-year-old boy with extensive, progressive BCH involving the face, trunk and extremities after twice-daily treatment with topical 1% rapamycin. (6)

Clinical differential diagnoses include Langerhans cell histiocytosis (LCH) and the two prevailing types of NLCH, juvenile xanthogranuloma (JXG) and generalized eruptive histiocytosis (GEH). LCH is a rare cutaneous disease that usually presents with treatment-resistant eczematous lesions on the scalp and/or intertriginous sites resembling seborrheic or diaper dermatitis. LCH may take a malignant course and affect other organs such as the lungs, liver or hematopoietic system. It can be differentiated from NLCH by immunohistochemistry, which reveals positive staining for the Langerhans cell makers S100, CD1a and langerin. (3)

JXG is characterized by many small pink to red–brown, dome shaped papules scattered on the upper part of the body that rapidly become yellow (micronodular JXG) or by a few nodular lesions (large nodular JXG). JXG mostly occurs in infants or young adults and may be associated with extracutaneous involvement of the eyes, liver and lung. An association with neurofibromatosis I, juvenile chronic myelomonocytic leukemia and LCH has also been described.

GEH typically affects adults and presents as recurrent crops of generalized numerous red to brown papules with a widespread distribution. GEH can be associated with hematological neoplasia. Clinical and histopathological distinction between an early phase of small nodular JXG or GEH and BCH may occasionally be difficult or even impossible. Thus it has been proposed that these diseases might be variants of a single clinical entity. (7-9)

In the present case the diagnosis of BCH was based only on clinical criteria that included a disease onset in early childhood (on average 15 months), the typical clinical morphology of the lesions and their predominant distribution in the head and neck region. If feasible, however, a skin biopsy should always be obtained to allow for differentiation from other types of cutaneous histiocytosis.

In summary, our case brings to attention the clinical characteristics of BCH and points to the fact that BCH may not be confined to the cephalic region. We also discuss the distinction of BCH from other types of NLCH and underline the benign course of this disease, which does not require an extensive diagnostic work-up but only a regular clinical monitoring.

Author Contributions:

All authors have made substantial contributions to conception and design, or acquisition of data, or analysis and interpretation of data; and have been involved in drafting the manuscript or revising it critically for important intellectual content; and have given final approval of the version to be published. Each author has participated sufficiently in the work to take public responsibility for appropriate portions of the content; and agreed to be accountable for all aspects of the work in ensuring that questions related to the accuracy or integrity of any part of the work are appropriately investigated and resolved.

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Figures

Figure 1a-b: Light brown macules and flat papules at first presentation.

Figure 2a-b: Reddish brown papules 5 months after the initial presentation.

Figure 3a-b: Regression to light brown macules and flat papules after 2 years.