



## Cicatricial Alopecia in a Pediatric Patient with Anetodermic Mastocytosis

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### Abstract

Mastocytosis is a rare condition characterized by a progressive accumulation of mast cells in cutaneous tissue and extracutaneous organs. The skin is involved in the majority of cases, hence, presented with heterogenic clinical expression. No more than 50 patients with anetodermic mastocytosis have been reported worldwide, most of which were adults. Herein, an anecdotal case of a 7-year-old patient with disseminated anetodermic lesions and cicatricial alopecia, histological verified as mastocytosis, is presented. Remarkably, Darier's sign, usually pathognomic for mastocytosis, was negative. PUVA treatment was considered the right choice of treatment. A comprehensive review of literature demonstrates the very rare nature of the disease and the exceptional involvement of the scalp in this age group.

**Keywords:** Anetoderma; Mastocytosis; Cicatricial alopecia

### Introduction

Mastocytosis represents a spectrum of clinical disorders characterized with the accumulation of mast cells in different tissues and organs [1]. Systemic mastocytosis usually involves bone marrow, spleen, liver, lymph nodes and gastrointestinal tract and may present with or without skin involvement. The most common manifestation of skin mastocytosis is Urticaria Pigmentosa (UP). It is characterized by small or larger brown-red maculopapules on the trunk and extremities [2]. Mild trauma results in erythema and urticarial lesions around the mastocytic maculae within a few minutes. This phenomenon is known as Darier's sign and is considered pathognomic for cutaneous mastocytosis [3]. The prognosis of cutaneous mastocytosis is usually benign and the lesions disappear spontaneously without residual features. Rare and atypical presentations of skin mastocytosis include bullous, pseudoxanthomatous, congenital dermatographism and anetodermic lesions [3]. The possible causative mechanism of anetoderma is the elastic and collagen fibers fragmentation upon mast cell degranulation and release of many destructive inflammatory mediators, resulting in skin laxity [3].

We report a patient with multiple anetodermic lesions on the scalp, neck, trunk and upper limbs with a diagnosis of mastocytosis and a negative Darier's sign.

### Case Presentation

A 7-year-old boy was admitted in our department with a 3-year history of an asymptomatic rash on the scalp, neck, trunk and upper limbs. Past medical, drug and family history was negative. The patient's mother reported development of reddish pruritic nodules that changed to anetodermic lesions. No systemic symptoms such as bone or abdominal pain, flushing, diarrhea were present. The clinical findings revealed multiple well-defined atrophic yellowish macules and papules, ranging in the size between 0.2 cm to 2 cm in diameter (Figure 1). Multiple atrophic patches of cicatricial alopecia were seen on the scalp (Figure 2). Histopathologic findings of a scalp lesion revealed dense infiltrate of monomorphic mast cells (Figure 3), verified by specific staining Toluidine blue (Figure 4). The pathological infiltrate demonstrated sheet-like distribution with accentuated folliculotropism. Degeneration and reduction of elastic fibers was found throughout the dermis (Figure 5), corresponding to the anetodermic clinical features of the skin lesions. The diagnosis of cutaneous mastocytosis was coined. Routine hematological tests were within normal ranges with the exception of C-reactive protein (9.5<25 IU/ml). Serum tryptase was also referential. Abdominal ultrasonography did not reveal spleno- and hepatomegaly. A radiological bone examination showed

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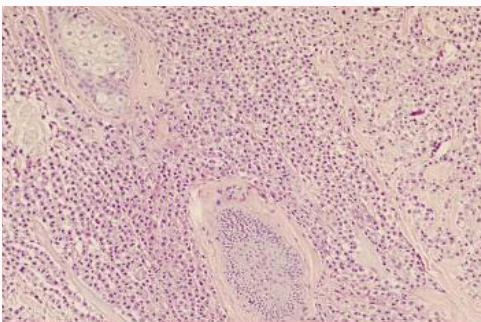
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**Figure 1:** Multiple well-defined atrophic hyperpigmented macules and papules on patient's neck and upper trunk.



**Figure 2:** Atrophic patches of scar alopecia.

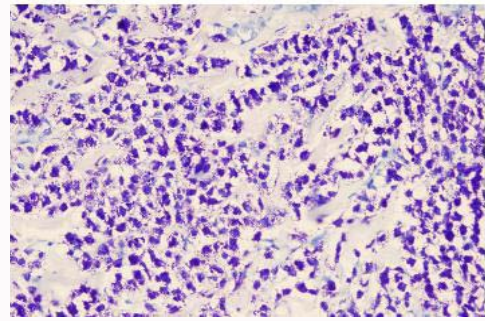


**Figure 3:** Dense dermal infiltrate in sheet-like distribution with accentuated folliculotropism (HE, x100).

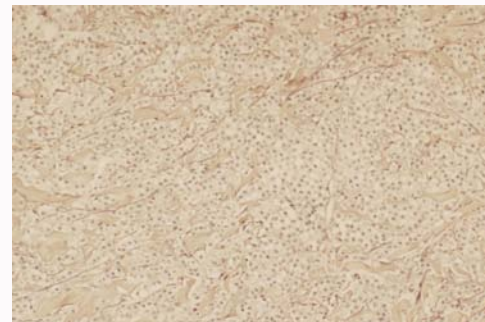
no alterations. Genome testing of c-kit mutations (including codons 816, 820 and 839) was negative. PUVA therapy was considered most appropriate to alleviate patient's symptoms and prevent formation of new lesions. Thirty sessions were performed (cumulated dosage 400 J/cm<sup>2</sup>) and no appearance of new lesions was observed. The dermatological status was quite improved.

## Discussion

Anetoderma is a rare benign dermatological condition, clinically characterized by skin atrophy as a result of dermal elastic tissue destruction [4]. Anetoderma is either primary (Jadassohn-Pellizzari type) evolving on previously unaffected skin, or secondary (Schweninger-Buzzi type), which is a complication of other inflammatory dermatoses such as lupus erythematosus, syphilis, amyloidosis, tuberculosis, perifolliculitis, acne, varicella, systemic administration of drugs (penicillamine) or mastocytosis [5,6]. Anetodermic mastocytosis is considered exceptionally rare, with



**Figure 4:** Monomorphic infiltrate of mast cells (Toluidin, x200).



**Figure 5:** Lack of elastic fibers throughout the whole dermis (Elastica, x100).

no more than 50 cases described worldwide [7]. First, Hallopeau [8] published a case in 1892. Thereafter, two comprehensive clinical reviews and anecdotal case reports demonstrated adult onset, female sex predisposition, chronic recalcitrant disease course, and predilection to trunk and extremities [9]. Pseudopelade has been observed in only 6 patients, all of them adults [7]. Herein, we dare speculate to describe the first pediatric case of anetodermic mastocytosis with cicatricial alopecia.

The pathogenesis of the disorder is still obscured. Presumably, the release of mast cell mediators with their local and systemic actions cause associated systemic and dermatological inflammatory symptoms, including the possible elastic fibers degeneration with subsequent skin laxity. However, residual elastolysis is extremely rarely described worldwide [7] and all cases have demonstrated positive Darier's phenomenon as an indirect sign of enhanced release of histamine, leukotrienes and prostaglandins on site of mast cell accumulation [3]. Those mediators provoke skin inflammation and should act as promoters to more aggressive dermal destruction. Unexpectedly, our patient did not have a positive Darier's sign. There was no evidence of systemic involvement either. These peculiar features do not correspond to the greater dermal damage, characterized by definitive elastic fibers degeneration and raise the hypothesis that anetodermic mastocytic changes depend on inflammatory mediators' destructive quality profile and not on their total amount.

Tissue culture experiments showed mast cell mediators, which inhibit enzymes that play a significant part in collagen synthesis impeding the hydroxylation and glycosylation of the polypeptide chain [10]. Heparin is reported to increase the fragmentation of collagen [11]. The proteolytic enzymes - tryptase and chymase-weaken the tissue matrix [12]. The inflammatory response is obviously dependable on the effector cell subtypes. Accumulation of eosinophils, neutrophils and macrophages upon mast cell degranulation possibly

increase the elastase activity and promote elastic fibers fragmentation.

Treatment choice is extremely difficult in children mastocytosis. It has to minimize the side effects risk providing proper alleviation of irritative skin symptoms. A recent consensus of therapeutic approach in pediatric mastocytosis favors its benign nature and evokes stepwise symptomatic modalities [13]. The objective is focused on stabilizing the mast cell degranulation in order to control disease symptoms. Recommendations for either single use of H1 anti-histamines or a combination of H1 and H2 antihistamines are usually given. Oral cromolyn sodium is effective in children with systemic symptoms such as diarrhea, abdominal pain, nausea, and vomiting. Oral methoxypsoralen therapy with long-wave Psoralen plus Ultraviolet A radiation (PUVA) is considered appropriate in bullous cutaneous mastocytosis and in non-hyperpigmented skin lesions. PUVA therapy leads to apoptosis in the immature mast cells and decreases the symptom of pruritus [14]. Serum histamine levels and low histamine excretion is demonstrated after PUVA sessions. Based on the good responses observed in other patients with anetodermic mastocytosis [6], we consider PUVA as the most favorable therapeutic approach for our patient.

## Conclusion

Mastocytosis in children is a rare inflammatory skin condition with unknown pathogenesis, natural history and prognosis. Accumulation of patients and proper analysis of their epidemiological, clinical, histological, genetic and immunological characteristics is a must to encompass the various patterns of this condition. Here in, we present a very rare case of pediatric anetodermic mastocytosis with exceptional scalp involvement to highlight another aspect of disease peculiarity.

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