

# Cutaneous Mastocytosis Associated With Congenital Alopecia

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**Abstract:** Mastocytosis is a rare disorder that shows accumulation of mast cells in tissues. Atypical clinical features may mimic impetigo, Langerhans cell histiocytosis, and carcinoid syndrome; however, only 1 case of scarring alopecia associated with mastocytosis has been reported. We present the first case of cutaneous mastocytosis associated with congenital alopecia areata in a 3-year-old Korean girl. This case showed an atypical clinical presentation of congenital alopecia areata, but histopathological results confirmed the diagnosis of cutaneous mastocytosis.

**Key Words:** cutaneous mastocytosis, congenital alopecia

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## INTRODUCTION

Cutaneous mastocytosis (CM) represents a group of clinical disorders with proliferation and accumulation of mast



**FIGURE 1.** Well-demarcated alopecic patch on scalp (arrow).



**FIGURE 2.** Tan to brownish macules, yellowish vesicles, and a protruding mass suggestive of mastocytoma on the back.

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cells in the skin. According to the World Health Organization classification, there are 4 types of diseases in CM, including urticaria pigmentosa, maculopapular CM, diffuse CM, and mastocytoma of skin, but various other forms have also been reported, such as bullous mastocytosis.<sup>1,2</sup> Alopecia areata (AA) is thought to be caused by autoimmune response, which leads to nonscarring hair loss. Traditionally, AA has been categorized as an acquired autoimmune disease; however, few cases of congenital AA have been reported recently. It is quite difficult to diagnose congenital AA because tinea capitis, traction alopecia, telogen effluvium, and other disease conditions mimicking AA should be ruled out. Also, early-onset AA is associated with poor prognosis, and the prognosis and treatment option of congenital AA are yet uncertain.

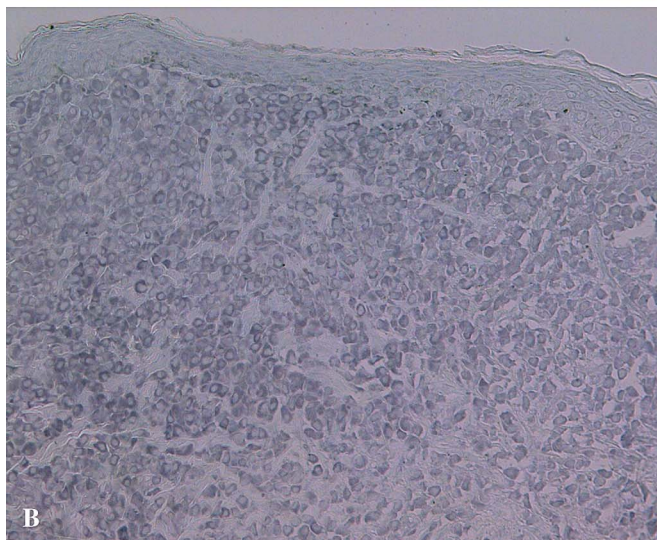
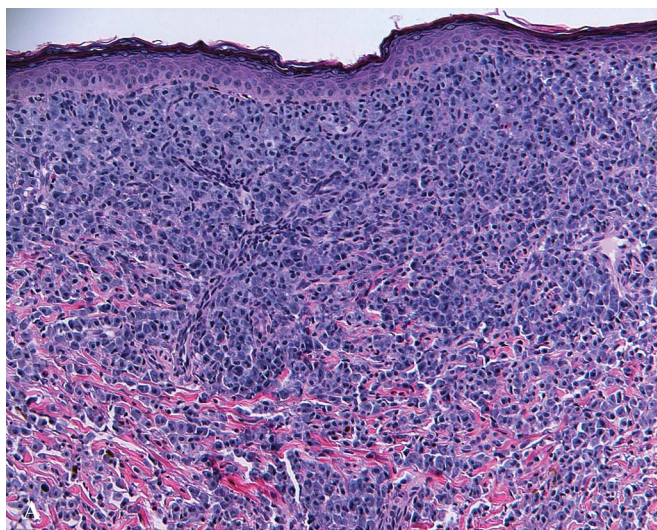


**FIGURE 3.** A, Dense dermal mast cell infiltration: cells show round nuclei surrounded by ample cytoplasm producing a “fried egg” appearance (hematoxylin and eosin,  $\times 100$ ). B, Special stain for toluidine blue demonstrated mast cell granule in the dermis (toluidine blue,  $\times 100$ ).

## CASE REPORT

A 3-year-old Korean girl presented with a 7-month history of well-demarcated tan- to reddish brown-colored macules and papules on the face and trunk, which spreaded gradually; also, she had about 1.5-cm sized brown-colored protruding nodule on her back. The patient was born at term with absence of scalp hair. One year before presentation, her grandparents noted patches of pigmented hair growth on her scalp, but she still had well-demarcated, round to oval, completely bald patches over her scalp. Physical examination revealed well-demarcated, distinct alopecic patches on scalp (Fig. 1), 1.5-cm sized round well-demarcated raised lesion on the back, erythematous to tan-colored maculopapules, and few excoriation marks over face and trunk, with a yellowish vesicle that suggests the diagnosis of urticarial pigmentosa (Fig. 2). The pull test on the surrounding areas of alopecic patches did not detach any hair. Clinically, yellowish to brownish nodules with erythematous patches were noticed on the alopecic patches of the patient; however, normal hair-bearing scalp showed no skin changes at all (Fig. 3A,B). Neither the parents nor the other members of her family reported any episode of AA and CM. The parents denied taking any kinds of drugs during the pregnancy. The patient's grandparents had cosmetic concerns, and the patient had a sense of itching, provoked by stroking the lesions, changes in body temperature, and strenuous activities. Other physical examination results were unremarkable. Laboratory data showed a normal blood cell count and serum chemistry; peripheral blood



**FIGURE 4.** Yellowish nodules with erythematous patches on the alopecic patches of the patient.

smear revealed normocytic and normochromic anemia with poikilocytosis, and elevated plasma levels for egg allergy were noted.

Histological examination of a biopsy taken from the alopecic patch of scalp, normal-appearing scalp, and mastocytoma on the back showed significant hyperplasia of mast cells in the papillary dermis around the blood vessels (Fig. 4). Special stain with toluidine blue showed abundant mast cell granules, confirming the diagnosis of urticaria pigmentosa. Based on clinical and histological findings, this case was diagnosed as CM associated with diffuse congenital AA. The patient's mastocytosis was treated with topical hydrocortisone 1% and oral antihistamines, and after 9 months of treatment, not only urticaria pigmentosa but also alopecia showed improvement (Fig. 5). Close patient follow-up is planned because there have been reports of transition of CM to systemic mastocytosis.

## DISCUSSION

CM has 4 major types, including urticaria pigmentosa, solitary and multiple mastocytomas, diffuse and erythrodermic mastocytosis, and telangiectasia macularis eruptiva perstans.<sup>3</sup> The first case report of alopecia associated with mastocytosis linear was described in 1971 by Bureau et al.<sup>4</sup> Thereafter, a handful cases of alopecia associated with mastocytosis have been reported<sup>5</sup>, and to our knowledge, mastocytosis presented with congenital alopecia has never been reported.

Xu et al<sup>6</sup> have suggested that alopecia and mast cell disease might be associated. Mast cell granules have many cytokines, growth factors such as fibroblast growth factors, and vascular endothelial-derived growth factors. The receptors

of these growth factors have a role in regulating development and cycling of hair follicle; therefore, it is possible that alopecia may be consequence of mast cell disease.

According to the previous reports, CM patients may have mutations of c-kit a receptor of growth factor. The stem cell factor on mast cells,<sup>1,6,7</sup> causes unregulated growth factor activation which results in increased proliferation and decreased apoptosis of mast cells. And recently it was also reported that c-kit has a role in hair growth; therefore, upregulated c-kit in the hair follicle cells can trigger AA and androgenetic alopecia.<sup>8</sup> C-kit is also expressed in melanocytic lineage cells,<sup>8</sup> so hyperactivation of c-kit can cause hyperpigmentation of affected skin, and this finding can help to explain tan- to reddish brown-colored maculopapules, which is a typical finding in CM, especially in urticaria pigmentosa.

In this case, the severity of the urticaria pigmentosa was closely associated with that of alopecia; alopecia and urticaria pigmentosa responded to the treatment. This suggests that severe mastocytosis can cause dense inflammation of hair-bearing skin, including scalp; therefore, mastocytosis can be a rare cause of alopecia, especially in young children.

Systemic symptoms associated with increased mast cell counts and their inflammatory mediators can be combined with CM; therefore, systemic workup, including complete blood cell count with peripheral blood smear and a blood chemistry study, may be necessary to rule out systemic involvement of mastocytosis.

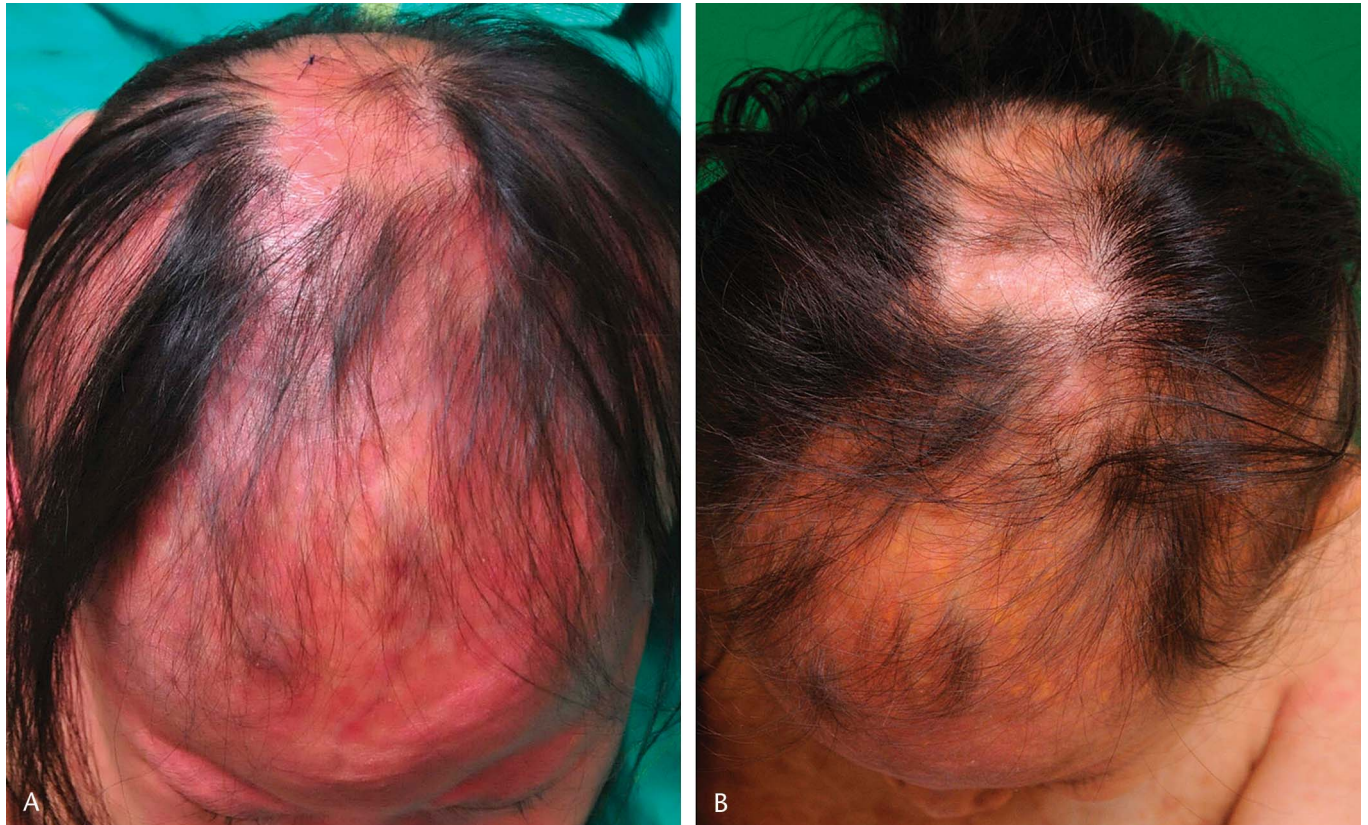


**FIGURE 5.** A, Congenital alopecic patches. B, Alopecic patches showed improvement after treatment.

Congenital AA associated with urticaria pigmentosa is rare but should be considered in the differential diagnosis of alopecia, especially in young children. We present our experience with the use of oral antihistamine in the management of urticaria pigmentosa associated with congenital alopecia. Further studies regarding the mechanism of alopecia associated with systemic mastocytosis may be required.

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