PHOTO ESSAY

An Atlas of Lumps and Bumps, Part 49: Solitary Cutaneous Mastocytoma

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Solitary Cutaneous Mastocytoma

Mastocytosis is a rare heterogenous group of disorders characterized by a clonal proliferation and accumulation of mast cells in one or more organs, mostly skin and bone marrow and, less frequently, gastrointestinal tract, liver, spleen, and lymph nodes.[1-3] Depending on the sites of organ involvement, two main forms of mastocytosis are recognized, namely, cutaneous mastocytosis (when mast cell accumulation is limited to the skin) and systemic mastocytosis (when mast cell accumulation involves extracutaneous tissues particularly the bone marrow, often combined with skin involvement).[4] Cutaneous mastocytosis can be subclassified into one of the three subtypes, namely, solitary cutaneous mastocytoma, maculopapular cutaneous mastocytosis (also known as urticaria pigmentosa), and diffuse cutaneous mastocytosis.[5]

Somatic activating mutations in the *c-KIT* proto-oncogene (most commonly D816V) that encodes the mast cell growth receptor KIT (also called CD117), a transmembrane protein that binds to stem cell factor and promotes cell division, have been shown to result in abnormal proliferation of mast cells and melanocytes.[1,2,6-9] A proliferation of melanocytes accounts for the hyperpigmented lesions seen in cutaneous mastocytosis.[8,10,11] Disease manifestations such as pruritus, flushing, blistering, wheals, vomiting, abdominal pain, hypotension, and dyspnea are caused by spontaneous or induced activation of mast cells with resulting release of vasoactive mediators.[12,13]

The prevalence of mastocytosis has been estimated to be 1 in 10,000; 80% of these patients have cutaneous mastocytosis.[1,13] Solitary cutaneous mastocytoma accounts for 10 to 15% of children with cutaneous mastocytosis.[1,14,15] There is no gender or race predilection.[16]

Solitary cutaneous mastocytoma most often presents at birth or develops within the first few months of life.[4,13,14] Typically, solitary cutaneous mastocytoma presents with an indurated, nontender,

erythematous, yellow **(Figure 1)**, orange **(Figure 2)**, or brown **(Figure 3)**, oval or round macule, papule, plaque or nodule.[1,17,18] The lesion is usually 1 to 5 cm in diameter and often has a peau d'orange (pebbly, orange peel-like) appearance and a leathery or rubbery consistency.[1,6,17] The lesional margins can be sharp or indistinct.[5]

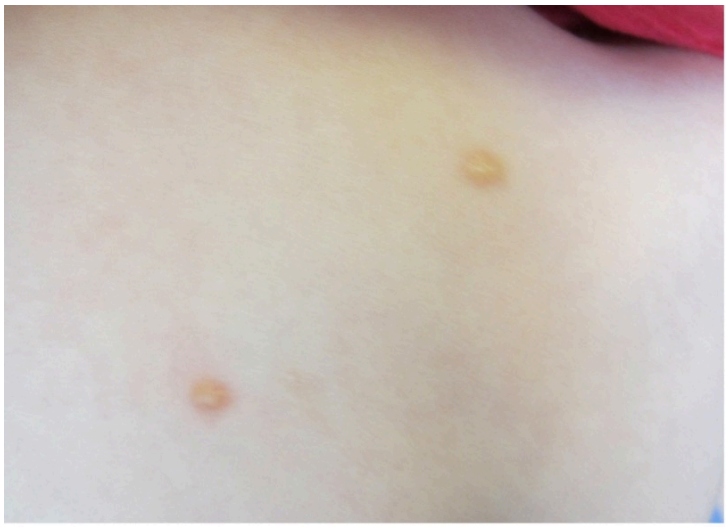

Fig. 1. *The prevalence of mastocytosis has been estimated to be 1 in 10,000. Occasionally, more tha one mastocytoma (up to a maximum of three) can be found.*

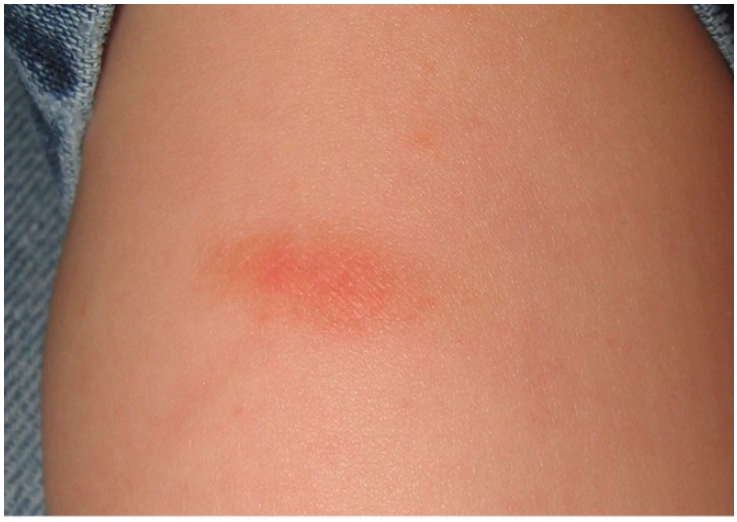

Fig. 2. *Solitary cutaneous mastocytoma accounts for 10 to 15% of children with cutaneous mastocytosis.*

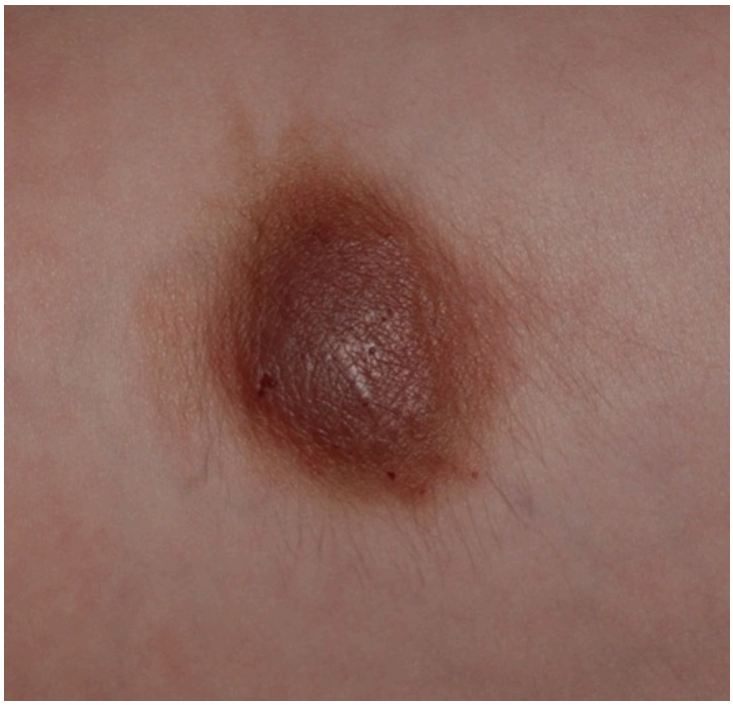

Fig. 3. *Typically, solitary cutaneous mastocytoma presents with an indurated, nontender, erythematous, yellow, orange, or brown, oval or round macule, papule, plaque or nodule.*

Occasionally, more than one mastocytoma (up to a maximum of three) can be found.[4,2,13] Pruritus is variable; the lesion can be asymptomatic or moderately pruritic.[1,12,17] The lesion usually increases in size (but not in number) for several months and then grows in proportion to the size of the child for a variable period of time before regressing over time.[1,19] Sites of predilection include the trunk, extremities, axillae, groins, followed by the head and neck.[1,19] The palms and soles are usually spared.[6] The lesion may spontaneously urticate and/or, more commonly, urticate when rubbed or stroked.[1,19,20]

Upon rubbing or stroking, the lesion becomes pruritic, erythematous, or edematous; this reaction is referred to as Darier sign **(Figure 4).**[1,19,21] The Darier sign is caused by degranulation of mast cells with subsequent release of mast cell mediators induced by physical stimulation. Although the Darier sign is pathognomonic,[16,21] it is only seen in 50% of patients with solitary cutaneous mastocytoma **(Figu 5**).[1,22,23] As such, the absence of a Darier sign does not exclude the diagnosis of a solitary cutaneou mastocytoma.[2] Some of the lesions may blister with irritation.[1,24] Although systemic symptoms such as flushing, dyspnea, hypotension, nausea, vomiting, abdominal pain, diarrhea, and headache are much more commonly seen in patients with systemic mastocytosis, they can also occur in patients with solitary cutaneous mastocytoma.[1,24] Therefore, large or extensive lesion of solitary cutaneous mastocytoma should not be vigorously rubbed and the Darier sign should not be elicited in case of a large mastocytoma as such a maneuver can precipitate severe symptoms.[1,25] Organomegaly and lymphadenopathy are characteristically absent.[1,25]

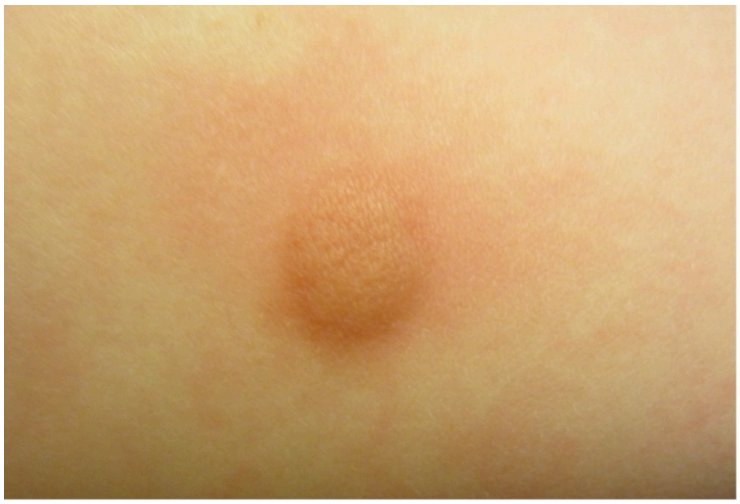

Fig. 4. *Upon rubbing or stroking, the lesion becomes pruritic, erythematous, or edematous, which is known as the Darier sign.*

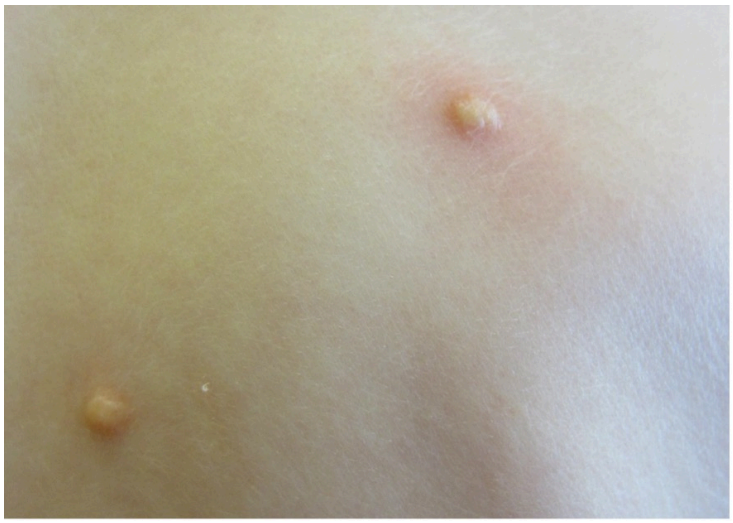

Fig. 5. *The Darier sign is only seen in 50% of patients with* *solitary cutaneous mastocytoma*.

The diagnosis is mainly clinical, based on the morphology of the lesion, the presence of a positive Darier sign, and absence of systemic involvement.[1,6] Dermoscopic features of a solitary cutaneous mastocytoma include light-brown blots, reticular vascular pattern, yellow-orange blots (attributable to dense papillary and reticular dermal mast cell infiltrate), and a pigment network (attributable to derma mast cell infiltrate and accumulation of melanin in basal keratinocytes).[20,23] When Darier sign is present, dermoscopy typically shows a decrease in yellowish hue and pigment network intensity alor with the appearance of peripheral erythema.[23] A skin biopsy is not essential but can be helpful if the diagnosis is in doubt.

Generally, the prognosis is good. Most children whose onset of disease is before two years of age tend to have spontaneous regression of skin lesions by 6 years of age and resolution of the disease l 10 years of age.[1,13,15,16]

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EDITOR'S NOTE:
This article is part of a series describing and differentiating dermatologic lumps and bumps. To acces previously published articles in the series, visit: https://www.consultant360.com/resource-center/atlas-lumps-and-bumps.

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