

Maculopapular Cutaneous Mastocytosis in a Diverse Population



Arnold S. Kirshenbaum, MD, Hanna Abuhay, BS, Hyejeong Bolan, RN, Dean D. Metcalfe, MD, and Melody C. Carter, MD
Bethesda, Md

Cutaneous mastocytosis (CM) is most frequently diagnosed in children, as well as in the majority of adults with systemic mastocytosis. Most commonly, both pediatric and adult patients present with the maculopapular cutaneous mastocytosis (MPCM) variant, also known as urticaria pigmentosa.¹ Large polymorphic lesions over trunk, head, and extremities tend to occur in childhood-onset CM, and small monomorphic lesions over thigh and trunk, which may also occur in children, predominate in adulthood-onset CM.² White, non-Hispanic patients are the predominate population at most centers including the National Institutes of Health. We thus asked whether the ability to identify MPCM in other groups might differ and confound the clinician from properly identifying and making the diagnosis of CM for patients undergoing evaluation.

In this retrospective examination of the appearance of MPCM in various populations and following informed consent, records of 10 patients (3 African American, 3 Hispanic, 2 white, non-Hispanic, and 2 Asian patients) reported herein were reviewed

for race/ethnicity, mastocytosis variant, age at time photographs of lesions were obtained, age of onset of disease, serum tryptase levels, and presence of *KIT* D816V in peripheral blood (Table I). Pediatric patients had onset of CM at birth or during the first year of life, and adult patients with CM had indolent systemic mastocytosis (CM/ISM) diagnosed during early to mid-adult years. Of the pediatric patients, 1 was African American, 2 were Asian, 2 were Hispanic, and 1 was white, non-Hispanic. Of the adult patients, 2 patients were African American, 1 patient was Hispanic, and 1 patient was white, non-Hispanic. Pediatric patients had tryptase levels ranging from 2.2 to 218.0 ng/mL. Adult patients had tryptase levels ranging from 15.8 to 218.0 ng/mL. One pediatric patient with CM and probable ISM (patient 7) was positive for *KIT* D816V as well as the pediatric patient diagnosed with ISM (patient 4); of the remaining 4 pediatric patients with CM, 1 was negative and 3 were not studied for the mutation. All adult patients with CM/ISM were positive for *KIT* D816V.

TABLE I. Demographic and clinical data of patient population

Patient	Race/ethnicity*	Variant†	Age in photo (y)	Age of onset	Pertinent medical history‡	Serum tryptase (ng/mL)	<i>KIT</i> D816V mutation§
1	AA	ISM	60	49 y	AP, D, DA, DZ, P, SB	90.0	+
2	AA	ISM	38	32 y	P	15.8	+
3	AA	CM	5	6 mo	AP, U, V	5.4	NA
4	W	ISM	9	Birth	AP, DA, F, FA, H, P, U	218.0	+
5	W	ISM	28	26 y	AP, D, DA, F, P	218.0	+
6	H	CM	6	Birth	AP, B, BP, DA, F, FA, N, P, SB, U	4.6	NA
7	H	CM/pISM	1	Birth	FA, P, U	19.4	+
8	H	ISM	69	59 y	H	30.4	+
9	A	CM	75	3 wk	B, F, P	63.9	NA
10	A	CM	2	3 mo	FT, Hy, P, U	2.2	–

*A, Asian; AA, African American; H, Hispanic; W, White, non-Hispanic (designated per National Institutes of Health Institutional Review Board guidelines).

†CM, Cutaneous mastocytosis; maculopapular cutaneous mastocytosis variant (MPCM) diagnosed by World Health Organization (WHO) definition of cutaneous mastocytosis; ISM, indolent systemic mastocytosis diagnosed by WHO criteria; pISM, probable ISM.

‡AP, Abdominal pain; B, blisters; BP, bone pain; D, diarrhea; DA, drug allergies; DZ, dizziness; F, flushing; FA, food allergies; FT, fatigue; H, hypotension; Hy, hyperpigmentation; N, nausea; P, pruritus; SB, shortness of breath; U, urticaria; V, vomiting.

§Peripheral blood *KIT* D816V. NA, Not available.

Mast Cell Biology Section, Laboratory of Allergic Diseases, National Institute of Allergy and Infectious Diseases, National Institutes of Health, Bethesda, Md
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Corresponding author: Arnold S. Kirshenbaum, MD, NIH/NIAID/Laboratory of Allergic Diseases, Bldg. 10, Room 11S239, 10 Center Drive, Bethesda, MD 20892-1960. E-mail: akirshenba@niaid.nih.gov.

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FIGURE 1. Cutaneous lesions in adult-onset mastocytosis. **A**, African American male. **B**, African American female. **C**, Hispanic male. **D**, White, non-Hispanic male. All lesions are monomorphic.

As shown in [Figure 1, A and B](#), adult African American patients and the adult Hispanic patient ([Figure 1, C](#)) had monomorphic lesions over the trunk similar to the white, non-Hispanic patient ([Figure 1, D](#)), which were easily identifiable. Pediatric African American and Hispanic patients ([Figure 2, A and B](#)) also exhibited easily identifiable monomorphic lesions over the trunk. Two Asian patients ([Figure 2, C and D](#)) both demonstrate a consistent pattern of skin lesions, although subtler in one and nodular lesions in the other. One Hispanic patient ([Figure 2, E](#)) exhibited polymorphic small and larger lesions, which were confluent in some areas. All lesions were

characteristically similar to the white, non-Hispanic pediatric patient ([Figure 2, F](#)).

These images thus demonstrate that the macroscopic features of CM lesions in various populations should not, in general, prevent proper identification and diagnosis of MPCM.

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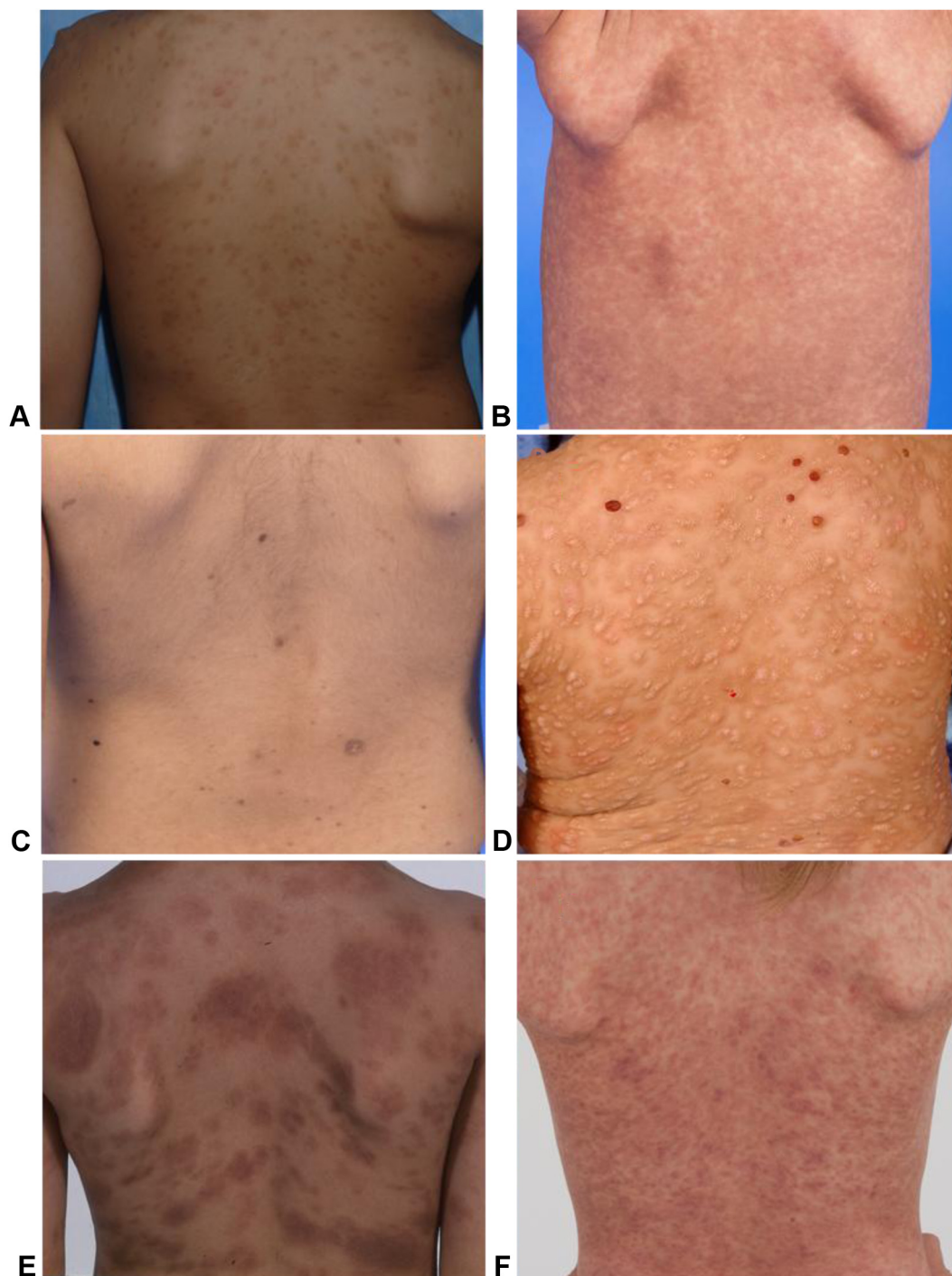


FIGURE 2. Cutaneous lesions in pediatric-onset mastocytosis. **A**, African American male. **B**, Hispanic male. **C**, Asian male. **D**, Asian female. **E**, Hispanic female. **F**, White, non-Hispanic female. **A-D** and **F** are monomorphic, and **E** is polymorphic.

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