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LETTER TO THE EDITOR

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WILEY

A case of Japanese pediatric cutaneous mastocytosis with disparate clinical classifications of maculopapular cutaneous mastocytosis and mastocytoma

Dear Editor,

Cutaneous mastocytosis (CM) is a disease in which mast cells localized in the cutaneous and extracutaneous tissue hyperproliferate[1]. Herein, we described a case of a Japanese pediatric patient with two disparate clinical features of CM and a D816V mutation. This study was approved by Tokyo Medical University Ethics Committee

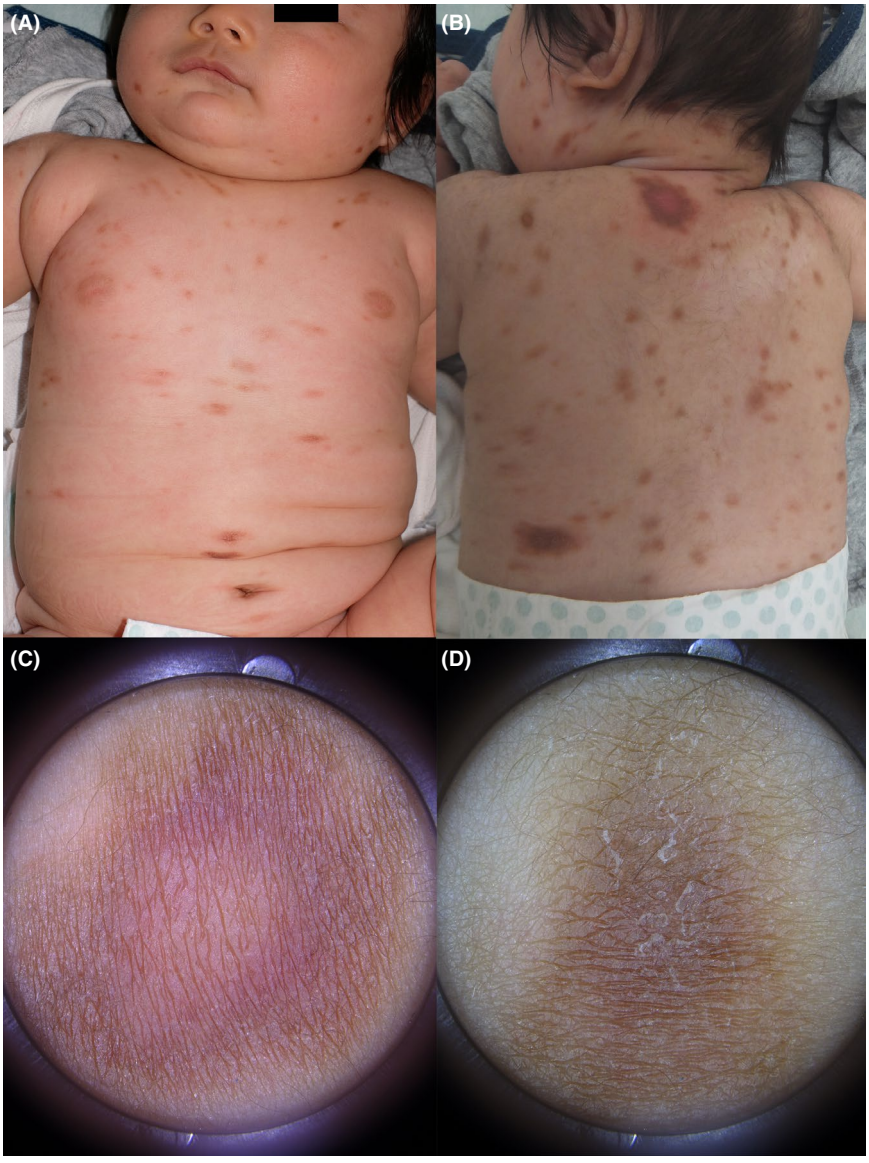

FIGURE 1 Clinical features and dermoscopic findings of cutaneous mastocytosis. A, Face and trunk; B, back; C, dermoscopic finding of the mastocytoma of the back; and D, dermoscopic findings of the maculopapular cutaneous mastocytosis of the trunk

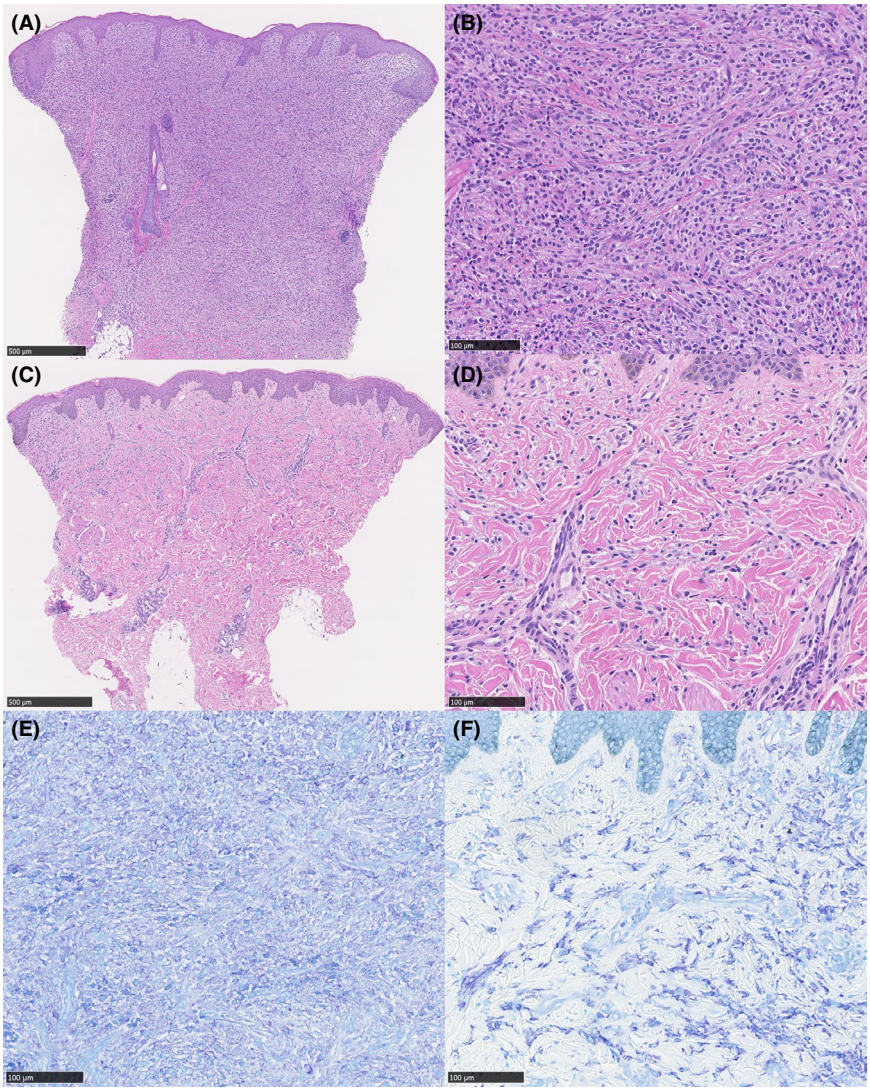

FIGURE 2 Histological observations of the mastocytoma and maculopapular cutaneous mastocytosis. A, Mastocytoma at lower magnification (hematoxylin and eosin staining, scale bar: 500 mm). B, Mastocytoma at higher magnification (hematoxylin and eosin staining, scale bar: 100 mm). C, Maculopapular cutaneous mastocytosis at lower magnification (hematoxylin and eosin staining, scale bar: 500 mm). D, Maculopapular cutaneous mastocytosis at higher magnification (hematoxylin and eosin staining, scale bar: 100 mm). E, Mastocytoma at higher magnification (Toluidin blue staining, scale bar: 100 mm). F, Maculopapular cutaneous mastocytosis at higher magnification (Toluidin blue staining, scale bar: 500 mm)

(No. T2019-0160). A 4-month-old Japanese male patient was born with two nodules on the back and multiple flat macules distributed over the entire body. The clinical examination revealed two lesions that 1.5-cm reddish-brown nodules on the upper and left back and multiple, flat, brownish macules scattered over the entire body (Figure 1A,B). Both types of lesion were positive for Darier's sign. Dermoscopy revealed a pattern of yellowish-orange dots in the two nodules and fine brown reticular lines in the multiple flat macules (Figure 1C,D). A skin biopsy of a nodule and macule was performed. Histology of the nodule demonstrated the presence of ballooned cells in the dermis (Figure 2A,B) and that of the macule likewise demonstrated the presence of ballooned cells in the upper dermis around the vessels (Figure 2C,D). Toluidine blue showed metachromatic staining (Figure 2E,F) in the ballooned cells. Based on these findings, the preliminary diagnosis of mastocytoma and maculopapular cutaneous mastocytosis (MCM) was made. Direct sequencing of mRNA isolated from the mastocytoma and MCM was analyzed mutations of the KIT gene in exons 9, 11, and 17, including five hot spot areas (codons 509, 522, 560, 816, and 820). This case was diagnosed both the mastocytoma and MCM contained *KIT* D816V mutations. The patient experienced no developmental abnormalities, and anaphylaxis due to UV and temperature were denied. A blood test was normal, and splenomegaly was denied. At a follow-up examination six months after receiving H_1receptor blockers, the patient denied organomegaly and severe symptoms, and subsequent follow-up visits to the hospital at intervals of six months were scheduled.

The prognosis of CM is better in pediatric patients than in adults. In approximately 70% to 80% of affected children, the disease resolves by adulthood. About 20%–30% of pediatric cases of CM involve a *KIT* D816V mutation[2]. In pediatric CM, the risk of anaphylaxis and aggressive disease is considered to be low. Previous studies have reported three similar cases of the co-occurrence of MCM and mastocytoma[3], all of which harbored a *KIT* gene mutation in exon 9 although in the present case the mutation was detected in exon 17. The present study is the first to report a case of pediatric CM with a *KIT* D816V mutation with the co-occurrence of the two clinical features of nodules and macular plaques. Recent research has revealed that the *KIT* D816V mutation in mastocytosis patients may be a predictor for persistent factors[4]. The relations two disparate

clinical features of CM and D816V mutation cannot explain now. And two disparate of CM is very rare. Thus, long-term follow-up of affected children is necessary, and prospective studies evaluating the prognostic value of sequencing *KIT*[5] are warranted.

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The patients in this manuscript have given written informed consent to publication of their case details.

CONFLICTS OF INTEREST

The authors declare no conflicts of interest.

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