

BRIEF REPORT

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CUTANEOUS MASTOCYTOSIS IN TWINS: MULTIPLE MASTOCYTOMAS AND URTICARIA PIGMENTOSA IN TWO PAIRS OF MONOZYGOTIC TWINS

Abstract: We report two sets of monozygotic twins with cutaneous mastocytosis: one set with urticaria pigmentosa and the other set with multiple mastocytomas. This is the first report of multiple mastocytomas in twins to our knowledge.

Cutaneous mastocytosis is mostly a sporadic disorder; however, multiple family cases and more than 10 pairs of twins and one set of triplets with urticaria pigmentosa have been described (1,2). We report two sets of monozygotic twins with cutaneous mastocytosis, one set with multiple mastocytomas, which is the first reported to our knowledge.

CASE REPORTS

Two pairs of Chilean male monozygotic twins were evaluated. They were born after uncomplicated pregnancies in nonconsanguineous marriages and had no family history of skin or hematological disorders.

Cases A and B

At the age of 5 years, tan papules developed on the neck, trunk, and extremities of twin A, and about 6 months later, an identical eruption developed on twin B. Physical examination revealed numerous reddish-brown, 2- to 10-mm macule-papules on the neck, trunk, and extremities (Fig. 1). No history of systemic symptoms existed, and their systemic examination was unremarkable. Skin biopsy of both children showed a diffuse infiltrate of mast cells in the upper dermis, and the overall picture was diagnostic of urticaria pigmentosa. Other laboratory tests were normal.

Cases C and D

A pair of 7-month-old twins presented with asymptomatic nodules on the face, trunk, and extremities since age of 2 months. Physical examination revealed well-developed infants, and each one had a few (<10) round-to-oval, reddish-brown, 5- to 15-mm nodules with peau d'orange surface appearance over the face, trunk, and extremities (Fig. 2) with positive Darier's. No macules or papules were observed. No history of systemic symptoms existed, and no further abnormalities were disclosed by thorough physical examination of both twins (lymphadenopathy or hepatosplenomegaly were not found). Skin biopsy of both children revealed a dense infiltrate of mast cells in the upper dermis, compatible with cutaneous mastocytosis, and considering that only nodules were present on the skin, the diagnosis of multiple



Figure 1. Multiple reddish-brown, 2- to 10-mm macules and papules on the trunk of twin A (A) and twin B (B).

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Figure 2. Twins C and D (**A**). One reddish-brown, 15-mm nodule on the left thigh of the twin C (**B**), and multiple round-to-oval, 5- to 15-mm papulo-nodules with peau d'orange on the trunk and extremities of twin D (**C**). No macules or papules are observed.

mastocytomas was made in both twins. Other laboratory tests were normal.

Evolution

No treatment was indicated, and both pairs of twins had normal physical and neurological development, and after 5 years of follow-up all lesions disappeared.

COMMENTS

Familial cutaneous mastocytosis is rare, and about 50 families and more than 10 pairs of identical twins affected have been reported (1,2).

The adult nonfamilial form of cutaneous mastocytosis is almost exclusively associated with mutation in codon

816 of c-kit, but in the familial and childhood variants, no single gene has been identified (3–6).

Sato-Matsumura et al (2) found no genomic mutations in exons 11 or 17 of c-kit in a set of monozygotic twins with urticaria pigmentosa. However, a recent study by Foster et al reported two pairs of twins with a heterozygous substitution in exon 10. This region encodes the transmembrane domain of the receptor, resulting in the substitution of leucine for methionine at codon 541. This data suggest that the single nucleotide polymorphism resulting in a Met-541-Leu c-kit mutation may predispose to pediatric mastocytosis (7).

Our twins with urticaria pigmentosa were not completely concordant on the disease onset since the second twin developed the lesions 6 months later than his co-twin, similar to other reports (8–10). We can hypothesize

that although both twins share the same mutations responsible for mastocytosis, different environmental factors (e.g., viral infections) could act differentially upon each child, precipitating the disorder in different ways (10). On the other hand, this discordance also suggests a multifactorial inheritance pattern (8–10).

Finally, although familial cases of urticaria pigmentosa and diffuse cutaneous mastocytosis have been reported (8–10), to our knowledge this is the first report of mastocytomas occurring in twins, which suggests this form of mastocytosis is also genetically determined.

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