Telangiectasia macularis eruptiva perstans with congenital onset in a 5-year-old girl

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Summary

Telangiectasia Macularis Eruptiva Perstans (TMEP) is a clinical variant of cutaneous mastocytosis which is rarely observed in childhood. A 5-year-old girl was referred to our Department for further work-up of asymptomatic disseminated reddish-brown macules with congenital onset and persistent course. Histopathological examination revealed hyperpigmentation of basal keratinocytes, vascular ectasia and enhanced perivascular mast cell counts in the upper dermis, altogether leading to the diagnosis of TMEP. Congenital-onset TMEP has only sporadically been reported in the pediatric population, with two of three hitherto published cases arising within a familial background. Mucosal involvement, as present in our patient, has not been described before. In contrast to the common pediatric variants of cutaneous mastocytosis, the clinical course of infantile TMEP tends to be chronic and refractory to therapeutic interventions but remains benign without systemic involvement.

KEY WORDS: mastocytosis; telangiectasia macularis eruptiva perstans; mast cells.

Introduction

TMEP is an uncommon clinical variant of cutaneous mastocytosis which mostly presents in adults. It has only exceptionally been observed in the pediatric population (1) with less than a dozen pediatric cases hitherto published world-wide (1-6). Of all other subtypes of cutaneous mastocytosis – namely maculopapular mastocytosis, solitary or few mastocytomas, i.e. the most common variants of childhood, and diffuse cutaneous mastocytosis – TMEP has the most subtle clinical and histologic findings presenting only a sparse perivascular and interstitial mast cell infiltrate. Owing to the facts that a positive Darier sign is only rarely observed and that dermal mast cells are often subtle and more spindle-shaped in TMEP than in the other variants of mastocytosis, correct diagnosis of TMEP represents a challenging task for both the clinician and the dermatopathologist, and is often delayed or even missed.

Case report

A 5-year-old, otherwise healthy girl was referred to our Department for evaluation of a disorder of hyperpigmentation. Since birth, disseminated light-brown macules were present having raised the suspicion of being café-au-lait spots within the context of neurofibromatosis. Her mother’s pregnancy, labor and delivery had been normal, and there was no history of previous trauma, dermatitis or infection. During infancy and early childhood, the lesions had remained unchanged with respect to both clinical appearance and extent. Pruritus, occasional swelling of the lesions and flushing were denied. No family members suffered from similar lesions.

On clinical examination, multiple scattered non-scaling reddish-brown macules and patches measuring 5 to 25 mm were evident on the trunk, the face and the extremities (Figure 1 A, B). Most of them were sharply delineated, and some were surrounded by a white halo. Neurofibromas, axillary freckling and intraocular Lisch nodules were not detected on thorough whole-body examination. The lesions did not urticate on rubbing (negative Darier sign). Of note, within most of the lesions conspicuous small arborizing capillary vessels could be even better visualized on dermoscopy were noted (Figure 1 C). In addition, a 5-mm solitary telangiectatic macule was present at the right tip of the tongue (Figure 1 D) without further mucosal involvement. The remainder of the physical examination was without any pathological findings. Histopathological examination of a skin biopsy taken from a representative reddish-brown macule of the right arm was rather unremarkable at first glance. However, taking a closer look, basal hyperpigmenta-
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Without melanocytic hyperplasia was evident, explaining the brown hue on clinical grounds. Moreover, numerous ectatic vessels were present in the upper dermis being surrounded by a sparse lymphocytic infiltrate (Figure 2 A). Both admixed within this perivascular round-cell infiltrate as well as diffusely scattered between the collagenous fibers conspicuous round and spindle cells with broad basophilic cytoplasm were seen. These stained positive with Giemsa (Figure 2 B), chloracetate esterase and c-kit (CD117) (Figure 2 C), corresponding to mast cells with prominent metachromatic intracytoplasmic granules. Absolute dermal mast cell count was elevated with 10-15 mast cells/high power field (HPF) (normal reference count <5 mast cells/HPF), and mast cells were found in small clusters with predominance around the superficial vascular plexus.

Taking these histopathological findings together with the clinical presentation, a diagnosis of congenital-onset TMEP was made. Routine laboratory examination including complete blood cell count, blood biochemistry as well as serum tryptase were within normal limits.

Discussion

In our case, an uncommon disease manifestation as early as in the neonatal period distracted prior diagnosis. To our knowledge, only three additional cases of congenital-onset TMEP with two of them occurring in a context of familial mastocytosis have been documented before (2-4). The family history of the herein presented 5-year-old girl was negative for similar skin lesions. Our case gained additional peculiarity by showing a unique mucosal involvement which has not been described before.

In contrast to adult cases, pediatric cutaneous mastocytosis is believed to represent a transient dysregulation of local growth factors including aberrant expression of soluble stem cell factor (SCF, equivalent to mast cell growth factor, c-kit ligand) rather than a genetic mutation involving the tyrosine kinase c-kit proto-oncogene in the adult variants (7-9). The majority of pediatric cases of cutaneous mastocytosis/mastocytoma show a good prognosis with spontaneous gradual resolution by adolescence with less than 15% of cases exhibiting persistent signs and symptoms. Alike
our 5-year-old girl, neither of the previously published pediatric cases with TMEP showed any systemic symptoms. Albeit only few cases have been reported and long-term follow-up is lacking, the clinical course of pediatric TMEP appears to be rather chronic and refractory in nature. However, it obviously remains benign without systemic involvement rendering the need for therapeutic intervention unnecessary.

Disclosure
The authors declare they have no conflict of interest.

References