Case 11

A 54-year-old man from Chiang Mai

Chief complaint: Asymptomatic reddish-brown spots on the upper arms, trunk and shoulder for 2 years

Present illness: The patient gradually developed asymptomatic reddish brown spots on the upper arms, trunk and shoulder for 2 years. These lesions tend to increase in intensity after having spicy food or exposure to hot weather. He had no systemic symptoms.

Past history: He denies of any underlying disease.

Personal history: He used to be a social alcohol drinker, though, denies alcohol intake for the past 10 years. No history of smoking.

Family history: Not remarkable

Physical examination:

HEENT: not pale, no jaundice, no parotid gland enlargement
LN: no lymphadenopathy
Heart and Lungs: WNL
Abdomen: no superficial vein dilatation, no distension, liver and spleen are not palpable
Extremities: no edema, no palmar erythema

Skin examination:
Multiple discrete well defined, slightly irregular-bordered, erythematosus to brownish macules with telangiectasia on the upper and outer aspect of the arms, shoulder, upper chest and upper back. Drier’s sign was negative.

Laboratory investigation:

CBC: Hb14.5 g/dl  Hct 43.2%  WBC 8300 ( N 62, L28,M7,E3)
BUN 20 mg/dl Cr 0.9 mg/dl
LFT :AST 19,ALT 33 ALB 41.7, GGT 32 TB 0.5 DB 0.2
Serum tryptase level: 7.34 ng/ml
U/S liver: mild fatty liver, otherwise normal

Diagnosis: Telangiectasia macularis eruptiva perstans

Presenter: Kumutnart Prabudhanitisarn
Consultant: Penpun Wattanakrai

Histopathology (S07-12308) (Fig 11.2, 11.3)
- Mild perivascular inflammatory infiltrate around telangiectatic blood vessels in the upper dermis
- Special stain (Giemsa) demonstrating mast cells predominate in some foci

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Discussion:

Mastocytosis is a rare condition characterized by abnormal growth and accumulation of mast cells that are functionally normal in many organs of the body. It usually presents in the skin, but may affect other tissues, particularly the bone marrow, lymph node, bone and gastrointestinal tract[1].

Clinical classification of mastocytosis consists of four types. The great majority of cases presented are indolent (type I) mastocytosis (either cutaneous or systemic mastocytosis) and that progression to an associated hematological disorder (type II) is uncommon. Both type III (aggressive mastocytosis with lymphadenopathy) and type IV (mast cell leukemia) are rare[1]. Cutaneous mastocytosis, classified according to a consensus nomenclature[2], composes of urticaria pigmentosa (UP) and maculopapular cutaneous mastocytosis, telangiectasia macularis eruptiva perstans (TMEP), diffuse cutaneous mastocytosis and mastocytoma of the skin.

Mutation in c-kit proto-oncogene (stem cell factor) has been implicated in the pathogenesis of mast cell disorder. Activated c-kit mutation leads to oncogenic transformation and enhance mast cell proliferation seen in mastocytosis[3]. Immunohistochemical examination of bone marrow biopsy sections has demonstrated that c-kit is strongly expressed on normal and abnormal mast cells in patients with systemic mast cell disease[4]. All sporadic adult-onset mastocytosis patients have c-kit mutation, however, mutation in c-kit seem to be lacking in most children and patients with familial disease[3]. The broad clinical classification and variation of disease suggest that different mechanism determine benign or malignant courses. Established surrogate markers of disease severity include extent and density of cutaneous lesion, typtase[5], histamine, IL6[6], CD25 and CD117 level[7].

TMEP is a rare form of cutaneous mastocytosis characterized by widespread, dark red or brown, 2-10 mm in diameter, telangiectatic macules on the trunk and extremities[1]. TMEP is regarded as pure cutaneous form, nevertheless, systemic involvement has been reported[8]. This type of mastocytosis is observed exclusively in adults. Due to the sparse number of mast cells, Darier's sign is usually absent. Lack of excessive mast cell on skin biopsy may warren special staining to demonstrate metachromasia of mast cell consisting of toluidine blue, giemsa, chloroacetate esterase, avidin and immunohistochemical staining of typtase.

The management of mastocytosis is based on underlying organ involvement. Avoiding physical stimuli (temperature extremes, massage, alcohol) and medications (eg NSAID, aspirin, opiates, polymyxin B sulfate, iodinated contrast media, dextrans and some muscle relaxants) is mandatory. Antihistamines are the mainstay of therapy. Mast cell stabilizer, UVA1, electron beam radiation, topical and systemic corticosteroid may be used in selected cases. IFN-α2a, 2b[9] and cladribine[10] have been report anecdotally to ameliorate some features of systemic mastocytosis. Flash lamp-pumped pulse dye laser can be used in TMEP with excellent outcome, although the response was temporary[11]. Proper antihistamine should be commenced prior to laser therapy to avoid potential complications of laser induced mediator release.
References: