
Students' Corner

Letter to the Editor

Smouldering Systemic Mastocytosis: A rare disorder with a difficult diagnosis

Madam, until recently, the Mastocytosis (MC) was viewed primarily as an effector cell in allergic diseases. However, it has been shown that MC's play a crucial role in many processes such as wound healing, tumour control, and transplant tolerance.¹

The term mastocytosis denotes a heterogeneous group of disorders characterised by abnormal accumulation and growth of MC's.² Systemic mastocytosis (SM), however, can develop at any age and is characterised by multifocal infiltrates of MC's in visceral organs with or without skin involvement.²

A particular category named 'smouldering mastocytosis' was introduced into the mastocytosis consensus classification at the 2001 meeting.³

Indolent SM, is a rare disease that may go unnoticed or may be misdiagnosed. The disease is a clonal disorder of the MC characterized by the proliferation and accumulation MC's within various organs of the body, most frequently the skin. Interaction between the cytokine stem cell factor (SCF) and its cognate receptor, c-kit (KIT), plays a vital role in regulating MC growth and differentiation.⁴ Activating mutations of the c-kit proto-oncogene play a casual role in the pathogenesis. The clinical symptoms and signs are owing to the buildup of these clonally derived MC's in diverse tissues, as well as the discharge of MC mediators, including histamine, heparin, prostaglandins, leucotrienes and cytokines. Manifestations range from exclusively cutaneous or indolent systemic disease, to systemic disease with an associated clonal haematological non-MC lineage disorder, to aggressive systemic mastocytosis, to MC leukaemia or sarcoma at the other end of the spectrum (WHO classification). The typical skin lesions are urticaria pigmentosa, which are monomorphic, pigmented, maculopapular or nodular lesions in a prevalent symmetrical distribution, frequently indicating Darier's sign (wheal and adjacent erythema developing in a lesion as a result of

rubbing).

There is currently no effective curative therapy for systemic mastocytosis. In general, for cutaneous and indolent systemic mastocytosis, treatment is aimed at symptomatic relief; this is possible with the aid of agents like antihistamines and MC stabilizers.⁴ The novel agents' dasatinib, nilotinib and imatinib (tyrosine kinase inhibitors) may be effective for patients with severe symptoms or with aggressive mastocytosis⁵ although these are not of proven benefit. IM, in particular, is ineffective in the presence of a D816V mutation. Trials testing novel agents such as PKC-412 and stem cell transplant as a form of treatment are also underway for patients with the aggressive form of the disease.

Because of the non-specificity of some of the symptoms, SM can be difficult to diagnose. It is important for pathologists and clinicians to be aware that SM can be mistaken for an acute illness being infectious in nature.

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