Cytology of systemic mastocytosis
Barbara J Bain

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Morphology Update

Cytology of systemic mastocytosis

Barbara J. Bain and A J Marks

Department of Haematology,
St Mary's Hospital Campus of Imperial College Faculty of Medicine,
St Mary's Hospital,
Praed Street,
London W2 1NY.

b.bain@ic.ac.uk

A 65-year-old woman with a 15-year history of urticaria pigmentosa on her trunk, arms and legs was referred for further investigation when she was found to have elevation of the serum tryptase to 190 µg/l (normal range 2–14). She had noticed worsening of her skin lesions but was otherwise in good health. A blood count, blood film and liver and renal function tests were normal. A bone marrow aspirate showed significant numbers of cytologically abnormal mast cells. Many of these had long and elegant tails, sometimes even three per cell (Image, top left). Some of the spindle shaped cells (Image bottom left) and other mast cells (Images right) were hypogranular. A morphological diagnosis of systemic mastocytosis was confirmed by demonstration of a \textit{KIT} D816V mutation in bone marrow cells.

Neoplastic mast cells may be undetectable in bone marrow aspirates from patients with systemic mastocytosis, even when trephine biopsy sections show significant infiltration. When present, they are usually cytologically abnormal so that the diagnosis can be made from marrow films. Hypogranular and spindle-shaped cells are characteristic. In addition, there may be aberrant expression of CD2 and CD25 and when mast cells are present in significant numbers in the aspirate a \textit{KIT} mutation, usually \textit{KIT} D816V, is likely to be detected.