Pseudo-Gaucher cells in myelodysplasia

A J Stewart, R D G Jones

Abstract

A case of myelodysplastic syndrome is reported, in which the bone marrow contained many cells with the typical light microscopic morphology of Gaucher cells. In the absence of any evidence of inherited Gaucher’s disease, these cells are considered to be pseudo-Gaucher cells, which have been described previously in association with other haematological diseases. This is the first report of their occurrence in myelodysplastic syndrome.

Keywords: myelodysplasia; pseudo-Gaucher cells

Case report

A 72 year old woman presented with a four day history of general malaise and flu-like symptoms. Examination revealed no abnormality apart from mild hypertension and an area of cellulitis on her right shoulder. She had no significant past medical history and was taking no drugs. A full blood count showed a haemoglobin concentration of 94 g/l, white cell count $7.2 \times 10^9/l$, neutrophils $6.2 \times 10^9/l$, platelets $78 \times 10^9/l$, and MCV 84 fl. Examination of the blood film revealed mild red cell anisocytosis, but very abnormal neutrophils with abnormal nuclear segmentation, Döhle bodies, and hypogranularity (fig 1), suggesting myelodysplasia. There were no blasts in the peripheral blood.

Examination of a bone marrow aspirate taken during the admission confirmed the diagnosis of myelodysplasia. The marrow was markedly hypercellular. Micromegakaryocytes were present, and many of the larger megakaryocytes showed abnormal nuclear configurations. Myelopoiesis was left shifted with marked hypogranularity. Morphologically identifiable myeloblasts accounted for 4% of nucleated cells. (Of note, however, was the presence of a significant (23%) population of CD34 positive cells on immunophenotyping.) No pathological sideroblasts were identified. On the basis of these appearances, the myelodysplasia was subclassified as refractory anaemia according to the French-American-British (FAB) classification. Sea blue histiocytes were prominent, but particularly striking was the large number of cells with the typical morphological appearance of Gaucher cells (fig 2). Electron microscopic examination was, unfortunately, not performed. Cytogenetic analysis revealed a normal karyotype.

A late presentation of inherited Gaucher’s disease was excluded by a leucocyte β-glucosidase activity of 24.94 nmol/l/mg (reference range 4.9–14 nmol/l/mg).

She was treated with intravenous antibiotics. An abscess formed under the cellulitic area and required surgical drainage. Culture of pus grew Staphylococcus aureus. She recovered slowly after surgery and was allowed home. Thereafter, her myelodysplasia progressed rapidly: she became red cell transfusion dependent within three months of presentation, and blasts began to appear in her blood in increasing numbers. She died of sepsis 10 months after presenting. The appearance of her peripheral blood at death was that of acute leukaemia, although a bone marrow aspirate was not repeated.

Comment

Until 1966, when they were described in chronic myeloid leukaemia, the presence in marrow of Gaucher cells was felt to be pathognomonic of Gaucher’s disease. Since that original report, Gaucher-like or pseudo-Gaucher cells have...
been noted in a variety of conditions including acute lymphoblastic leukaemia,\(^1\) Hodgkin’s disease,\(^2\) thalassaemia,\(^3\) and multiple myeloma.\(^4\) These cells display the “wrinkled tissue paper” appearance of the cytoplasm and the eccentric, lobulated nucleus typical of true Gaucher cells,\(^5\) but they do not contain the tubular inclusions which are found in authentic Gaucher cells.\(^6\) The presence of pseudo-Gaucher cells probably reflects the increased load of leucocyte membrane derived glucosylceramide presented to macrophages under conditions of high cell turnover when the normal pathways for its removal may be saturated.

In myelodysplasia, increased leucocyte destruction occurs because of ineffective haemopoiesis. If this postulate is correct, the appearance of pseudo-Gaucher cells in myelodysplastic bone marrow is not unexpected. It has not, however, been reported before as far as we are aware.

Our patient’s myelodysplasia progressed more rapidly than expected from the morphological appearance of her marrow at presenta-

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