Pseudo-Gaucher cells

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Abstract
A case of acute lymphoblastic leukaemia, associated with cells resembling Gaucher cells in the bone marrow, is reported. The patient had no evidence of inherited Gaucher’s disease and the ultrastructural appearance of the cells was consistent with pseudo-Gaucher cells described in other haematological diseases. This is the first report of these cells in association with acute lymphoblastic leukaemia.

Case report
A 12 year old boy presented with a short history of pallor and dyspnoea on exertion. Examination showed that he had a few palpable axillary lymph nodes and an enlarged liver and spleen. A full blood count showed that his haemoglobin concentration was 68 g/l, platelet count 66 × 10^9/l, with a white cell count of 295 × 10^9/l and 95% blasts. The bone marrow had been replaced with L1 lymphoblasts, and immunophenotyping was consistent with null cell acute lymphoblastic leukaemia. The marrow also contained large numbers of cells with the morphological appearance of Gaucher cells (fig 1). Electron microscopic examination of these cells showed that there were elongated lysosomes filled with fibrillar inclusions; occasional dense rounded structures were also present, but the tubular inclusions described in true Gaucher cells were not seen.

He entered remission following treatment according to the UKALL XI protocol, schedule C. Subsequent marrow aspirates have shown a reduction in the number of Gaucher cells but they were still present in the marrow 20 weeks after diagnosis (fig 2).

Leucocyte $\beta$-glucosidase activity was 8.0 μmol/g/h (normal range 5–15 μmol/g/h), excluding the diagnosis of inherited Gaucher’s disease. The $\beta$-glucosidase concentrations in the parents were 9.3 and 10.4 μmol/g/h.

Comment
Cells resembling Gaucher cells occur in a number of conditions in which cell turnover is increased, including chronic myeloid leukaemia,1 Hodgkin’s disease,2 and thalassaemia.3 These pseudo-Gaucher cells have distinct appearances on electron microscopy which distinguish them from true Gaucher cells. The ultrastructural appearances of the cells in this case were consistent with the pseudo-Gaucher cells described previously.1 The bone marrow is the result of overloading of $\beta$-glucosidase by excessive cell turnover; hence there is a relative rather than an absolute deficiency of the enzyme.

To our knowledge, these cells have not been reported previously in association with acute lymphoblastic leukaemia, nor have they been noted in the morphological review of about 3000 cases in the Medical Research Council UKALL trials (J Lilleyman, personal communication). The presence of these cells in our patient may be evidence that he is a carrier of Gaucher’s disease which is usually inherited as an autosomal recessive characteristic.


Figure 1 Acute lymphoblastic leukaemia with pseudo-Gaucher cells in bone marrow.

Figure 2 Bone marrow: same case as figure 1 five months from diagnosis and in remission. Pseudo-Gaucher cells still present.