Educational Material IQAP 1221

A 38-year-old man complained of malaise. The complete blood count showed Hb 13.9 g/dL, RBC 4.04 x 10^{12} /L, Hct 0.405 L/L, MCV 100.0 fL, WBC 4.0 x 10^{9} /L and platelet 70 x 10^{9} /L.

The peripheral blood smear shows a small number of abnormal lymphoid cells which possess appreciable amount of cytoplasm with circumferential cytoplasmic projections (Figures 1-2). The nuclei are oval to round with partially condensed and indistinct nucleoli. There are neutropenia, monocytopenia and thrombocytopenia. Red cells are morphologically unremarkable.

The lymphoid cells express B-lineage specific markers including CD19, CD20, and CD22 together with hairy cell markers, i.e. CD11c, CD25 and CD103 by multi-parametric flow cytometry. They are positive for acid phosphatase and are resistant to tartrate treatment (TRAP positive). The morphology, cytochemistry and immunophenotypic profile are classical for hairy cell leukaemia.

Hairy cell leukaemia (HCL) is an uncommon B-cell lymphoproliferative disease affecting adults. The most common laboratory finding is cytopenia. Monocytopenia is consistently present, except in HCL variant. Hairy cells are often seen in peripheral blood films. Diagnosis is based on morphology, immunophenotypic profile and cytochemistry (TRAP). Immunohistochemistry with tartrate-resistant acid phosphatase and annexin A1 are currently available to confirm bone marrow infiltration in trephine biopsy. Whole exome sequencing of HCL cells has recently revealed the presence of the *BRAF* V600E mutation (Tiacci *et al*, 2011).



Figure 1. An abnormal lymphoid cell with cytoplasmic projections (400x magnification).



Figure 2. An abnormal lymphoid cell with cytoplasmic projections (1,000x magnification).

Tiacci E, Trifonov V, Schiavoni G, Holmes A, Kern W, Martelli MP, Pucciarini A, Bigerna B, Pacini R, Wells VA, Sportoletti P, Pettirossi V, Mannucci R, Elliott O, Liso A, Ambrosetti A, Pulsoni A, Forconi F, Trentin L, Semenzato G, Inghirami G, Capponi M, Di Raimondo F, Patti C, Arcaini L, Musto P, Pileri S, Haferlach C, Schnittger S, Pizzolo G, Foà R, Farinelli L, Haferlach T, Pasqualucci L, Rabadan R, Falini B. BRAF mutations in hairy-cell leukemia. N Engl J Med 2011; 364: 2305-2315.