IMAGENS EM HEMATOLOGIA/IMAGES IN HEMATOLOGY

Translocation t(1;16) in Chronic Eosinophilic Leukaemia

Translocação t(1;16) em Leucemia Eosinofílica Crônica

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Chronic eosinophilic leukaemia (CEL) is a rare myeloproliferative disease in which clonal proliferation of eosinophil (Eo) precursors results in persistent increase of mature Eo in the bone marrow and peripheral blood.[1] Various chromosomal abnormalities have been reported in CEL,[2] especially aberrations of the long arm of the chromosome 5, involving breakpoints in the q31-35 region, where genes encoding for interleukin-3, interleukin-5, granulocyte macrophage-colony stimulating factor and platelet-derived growth factor β receptor are located.[3]

We present herein a case of CEL that presented a der(16)t(1;16). The patient was a 60-year-old female first referred to our unit in November 2000 due to skin lesions and leukocytosis with eosinophilia. At this time the peripheral blood examination showed: Hb: 15.2g/dl, WBC: 23.7x10^3/μl (1% myelocytes, 3% metamyelocytes, 38% neutrophils, 45% eosinophils, 5% basophils, 6% lymphocytes and 2% of monocytes) and Platelets: 121.0x10^3/μL.

Infiltration by mature Eo was seen by histological examination of a skin lesion. Bone marrow aspirate showed hypercellularity of the granulocytic lineage with 45% of mature Eo. The diagnosis of CEL was based on a blood sample and bone marrow aspirate and biopsy analysis using conventional criteria.[4] The *BCR-ABL* transcript was negative by the conventional method of molecular analysis.[5]

The bone marrow cells were cultured for obtaining karyotype according to a conventional method.[6] The karyotype 47,XX,der(16)(16q22+),+21 was identified in all analysed metaphases (Figure 1).

The t(1;16) was seen in all metaphases analysed by fluorescence *in situ* hybridisation (FISH), using a painting probe for chromosome 1 (WPC DNA Probe 1, Spectrum Orange) obtained from Vysis (Downers Grove IL, USA) (Figure 2).

The patient has been seen in our service for four years, under hydroxyurea (500 mg daily) therapy, without symptoms or transformation to a blastic crisis.

The t(1;16) is a frequent recurrent rearrangement in solid tumours such as breast carcinoma and Ewing's sarcoma,[7] but it has very occasionally been described in haematological malignancies.[8,9] To the best of our knowledge, this chromosomal abnormality has not yet been described in CEL.

The gene *CBFB* is located in the 16q22 region and is implicated in the abnormal eosinophilopoiesis observed in acute myelomonocytic leukaemia with eosinophilia (M4Eo).[10] Thus, we hypothesize that the same gene could have been involved in the origin of the patient's CEL disease.

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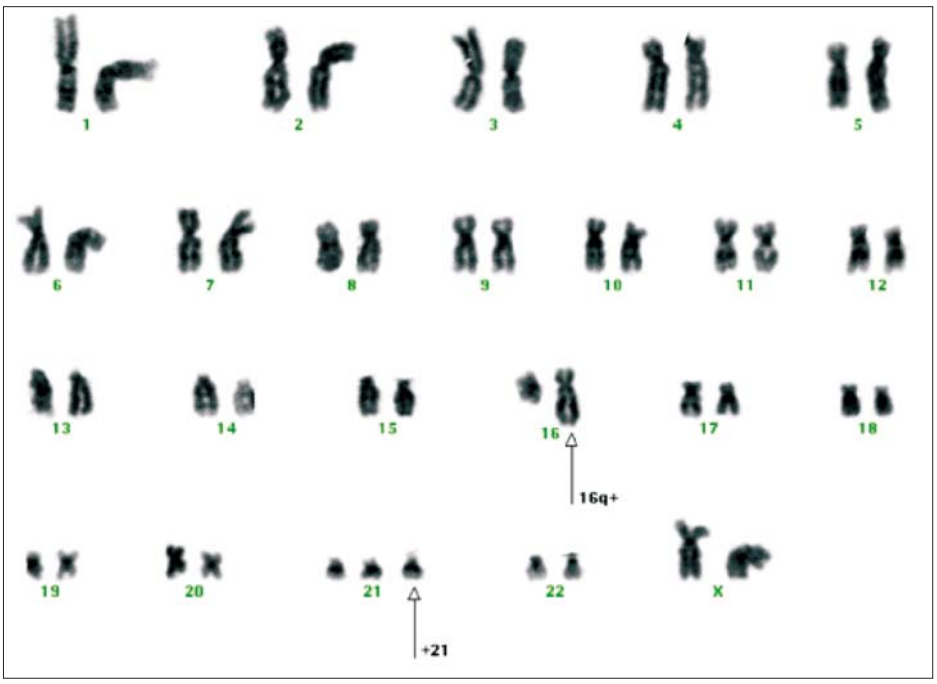

Figura 1 – G-banded karyotype showing 47,XX, der (16) (16q22+), +21 in a case of chronic eosinophilic leukaemia

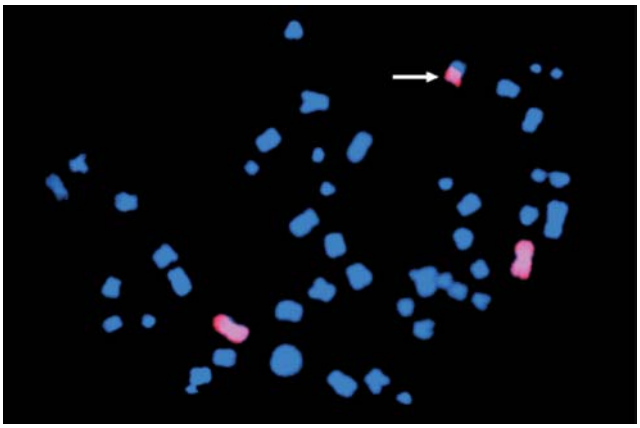

Figura 2 – FISH analysis in the CEL patient, using a specific DNA probe for chromosome 1 (•), showing an abnormal metaphase with a portion of chromosome 1 inserted on the long arm of chromosome 16 (arrow)

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Avaliação: Editor e dois revisores externos.
Conflito de interesse: não declarado

Recebido: 12/01/2005
Aceito: 02/02/2005