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Case Report Section

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Reciprocal translocation t(2;12)(q31;p13) in a case of CMML

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Clinics

Age and sex: 78 years old male patient
Previous History : no preleukemia; no previous malignant disease; no inborn condition of note.
Organomegaly : no hepatomegaly; splenomegaly; no enlarged lymph nodes; no central nervous system involvement.

Blood

WBC: 3.14. x 10^9/l; Hb: 5.7 g/dl; platelets: 86x 10^9/l;
Bone marrow: increased cellularity, hyperplastic granulocytic series with dysgranulopoiesis, polymorphous and dysplastic megakaryocytes, numerous micromegakaryocytes, depressed erythroid series. Blasts: 5%, Monocytes: 17%. Note: WBC Differential: Neu: 23, Lymph: 31, Mono: 41, Myelo-Metamyelocytes: 5%, granulocytic dysplasia. Absolute monocyte count: 1.3 x 10^9l

Cytopathology classification

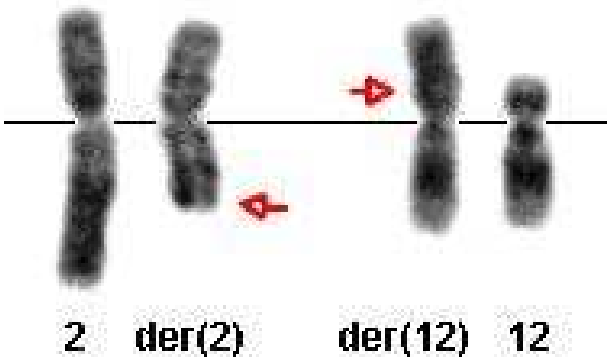
Pathology: Increased cellularity, F:C ratio 5:95 mainly due to hyperplasia of myelocytic series with dysgranulopoiesis, CD34(+) cells approx. 13%, significant depression of erythroid series, abundant micromegacaryocytes.
Precise diagnosis: Chronic myelomonocytic leukemia (CMML).

Survival

Date of diagnosis: 02-2003.
Treatment: Supportive; blood transfusions, steroids, platelets transfusions.
Complete remission: None
Status: Dead 11-2005.
Survival: 33 months.

Karyotype

Sample: Bone marrow; Culture time: Direct preparations (after 1 h in culture) and 24 h; Banding: G-banding with Wright stain.
Results: 46,XY,t(2;12)(q31;p13)[22]/47,idem,+21[3].



Partial karyogram of the cytogenetically abnormal clone with the translocation t(2;12)(q31;p13). The arrows indicate the breakpoint in the abnormal chromosomes 2 and 12.

Comments

Among the haematological malignancies with clonal chromosome aberrations reported in the world literature, there is only one case with the same cytogenetic clonal evolution, since a second abnormal translocation t(2;12)(q31;p13), a non Hodgkin lymphoma published by Sato et al., 1997. The karyotypic findings of the present case indicates a clone with the translocation t(2;12)(q31;p13) and trisomy of chromosome 21 was identified together with the clone displaying the t(2;12)(q31;p13) as the sole change.

References

Sato Y, Bohlander SK, Kobayashi H, Reshmi S, Suto Y, Davis EM, Espinosa III R, Hoopes R, Montgomery KT, Kucherlapati RS, Le Beau MM, Rowley JD. Heterogeneity in the breakpoints in balanced rearrangements involving band 12p13 in hematologic malignancies identified by fluorescence in situ hybridization: TEL (ETV6) is involved in only one half. Blood 1997;90:4886-4893.

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