blood work
images in hematology

Double insertion in normal karyotype CML

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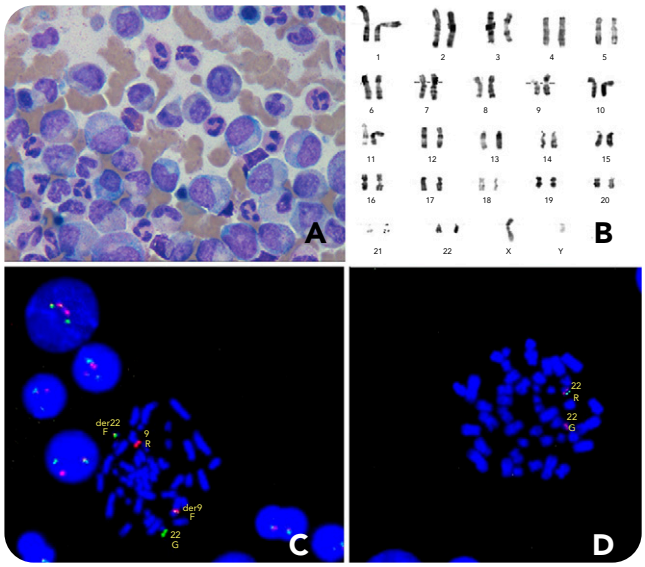

A 67-year-old man consulted for the diagnosis of chronic myeloid leukemia, a myeloproliferative neoplasm revealed by myeloid hyperplasia on the bone marrow aspirate with granulocytes present at all stages of maturation with basophilia and eosinophilia (panel A, May-Grünwald-Giemsa stain, original magnification ×500). Medullar karyotype was normal (panel B, R-banding karyotype, 46,XY[20]). A typical e13a2 (b2a2, *M-BCR*) fusion transcript was detected by real-time polymerase chain reaction. Fluorescence in situ hybridization (FISH) analysis using dual-color, dual-fusion probes for *BCR/ABL1* showed 2 *BCR/ABL1* fusions (panel C, dual color (D)-FISH *BCR/ABL1* probes; *ABL1*[9q34.12], red [R]; *BCR*[22q11.23], green [G]; and fusion [F]). Dual-color probes targeting a region upstream (*TBX1*) and downstream (D22S1254, distal to the ARSA gene) of *BCR* showed these 2 probes located on chromosomes 22 (panel D, D-FISH *DG* probes; *TBX11*[22q11.21], red; subtelomeric probe[22q13.3], green; G, normal green signal; R, normal red signal). The rearrangement mechanism involves probably reciprocal 4 break translocations of 2 interstitial segments leading to insertion of ABL into BCR and insertion of BCR into ABL. Karyotype was 46,XY[20].isht(9;22)(q34q34;q11q11)(ABL1+,BCR+;TBX1+, BCR+,ABL1+,D22S1254+)[40].nuc ish(ABL1,BCR)x3(ABL1 con BCR1x2)[98/100].

This rare case of t(9;22) translocation masked with 2 BCR-ABL1 fusions shows the importance of FISH studies to understand the chromosomal mechanism of cryptic and complex rearrangements.

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