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Duplication of the Long Arm of Chromosome 3 Leads to *MECOM* Rearrangement in Acute Myeloid Leukemia

Akut Lenfoblastik Lösemide Kromozom 3 Uzun Kolunda Duplikasyon *MECOM* Yeniden Düzenlenmesine Yol Açar

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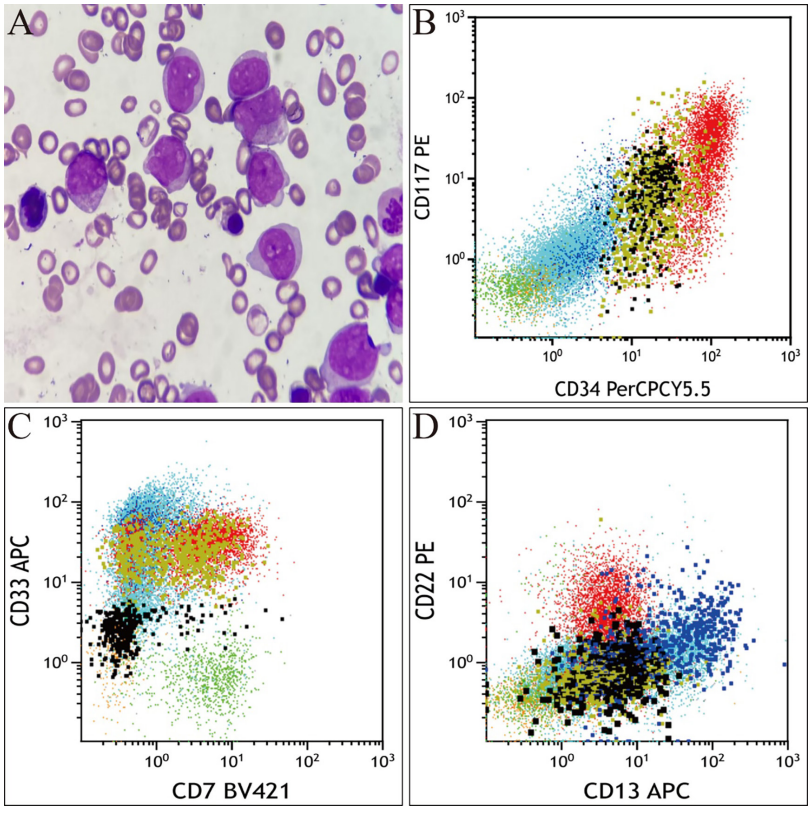

Figure 1. A) Bone marrow smear revealed increased blasts. B, C, D) Flow cytometry was conducted with a CD34/SSC gating strategy to output all blast cells. Subsequently, CD33/HLA-DR was used to distinguish the CD33-negative and weakly HLA-DR-positive cell population (shown in black). CD38 was also used for gating; strongly CD38-positive cells are dark green and CD38-positive cells are red. Three abnormal myeloid precursor cell populations were identified. The first group (28.48%; red) expressed myeloid (CD33, CD13, and CD117, with minor weak expression of MPO) and lymphoid (CD7, CD22, CD56, CD2, and CD4) antigens as well as HLA-DR and CD38. The second group (3.46%; dark green) expressed myeloid (CD33, CD13, and CD117, with minor weak expression of MPO) and lymphoid (CD7 and CD4) antigens, along with HLA-DR and CD38. The third group (1.92%; black) expressed myeloid antigens CD13 and CD117, was negative for CD33, and lacked expression of lymphoid antigens; it was also negative for MPO.

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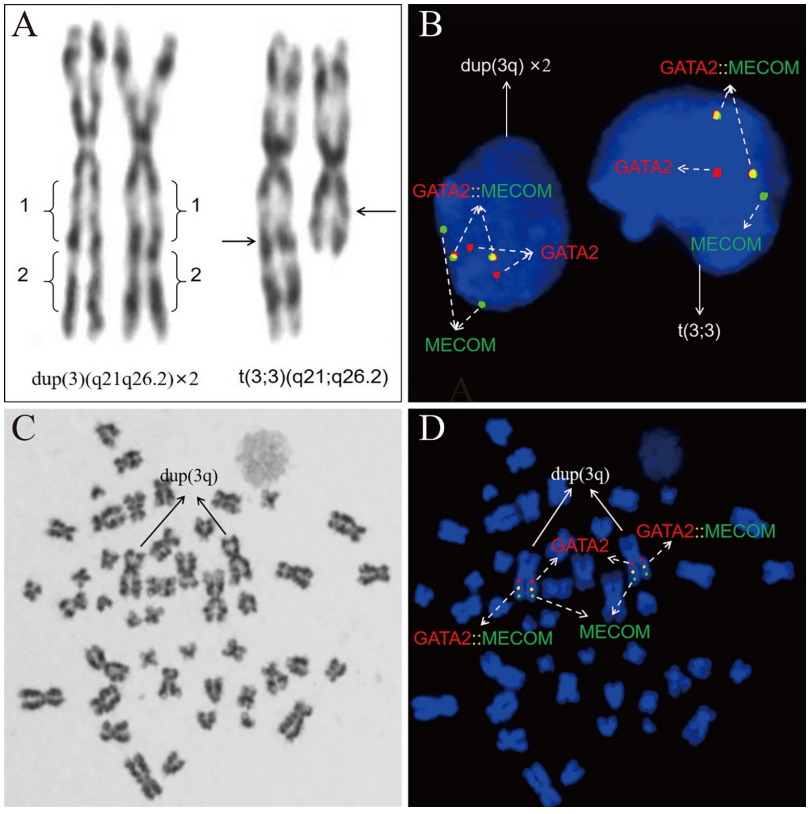

Figure 2. A) Bone marrow karyotype analysis showed dup(3)(q21q26.2)x2 and t(3;3)(q21;q26.2) in two different clones. B, C, D) Interphase and metaphase fluorescence in situ hybridization proved the co-localization of *GATA2* and *MECOM* in the dup(3q) and t(3;3) clones.

A 65-year-old man presented with general fatigue and unexplained fever. Blood tests revealed a slightly elevated white cell count (12.17x10^9/L), low hemoglobin (47 g/L), and normal platelet levels (123x10^9/L). A bone marrow smear showed a significant increase in immature granulocytes (33.5%) (Figure 1A), while flow cytometry revealed three abnormal myeloid precursor cell populations accounting for 33.86% of the nucleated cells (Figures 1B-1D). Karyotype analysis yielded the following result: 45,XY,dup(3)(q21q26.2)x2,-7[13]/45,XY,t(3;3)(q21;q26.2),-7[7] (Figure 2A). Both interphase and metaphase fluorescence in situ hybridization (FISH) revealed the fusion of *MECOM* and *GATA2* in both the dup(3q) and t(3;3) clones (Figures 2B-2D). Consequently, the patient was diagnosed with acute myeloid leukemia with *MECOM* rearrangement. He underwent treatment with the DA regime (daunorubicin + cytarabine) and was monitored using flow cytometry over a 5-month period, during which abnormal blast cell percentages fluctuated between 0.4% and 3.4%. In the fifth month, FISH testing indicated the disappearance of the dup(3q) clone; however, 90% of the cells displayed the t(3;3) clone.

Overexpression of *MECOM* results in compromised differentiation, apoptosis, and cell cycle arrest of hematopoietic stem cells [1]. It is mainly driven by inv(3)/t(3;3), which fuses the *GATA2* enhancer at 3q21.3 with *MECOM* at 3q26.2. Rare chromosomal alterations can also lead to *MECOM* rearrangement [2]. This patient exhibited a chromosomal duplication on the long arm of chromosome 3, revealing a novel mechanism of chromosomal alteration contributing to *MECOM* rearrangement. Simultaneously, the clones presented with *MECOM* rearrangement caused by dup(3q) with sensitivity to the DA regimen.

Keywords: *MECOM*, dup(3)(q21;q26.2), AML

Anahtar Sözcükler: *MECOM*, dup(3)(q21;q26.2), AML

Ethics

Informed Consent: Informed consent was obtained from the patient.

Footnotes

Authorship Contributions

Surgical and Medical Practices: X.Y.; Concept: S.Y.; Design: Y.L.; Data Collection or Processing: S.M.; Analysis or Interpretation: S.M., J.Y.; Literature Search: S.Y.; Writing: S.Y., S.M.

Conflict of Interest: No conflict of interest was declared by the authors.

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