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

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Dear Editor,

A 65-year-old man presented with general fatigue and unexplained fever. Blood tests revealed a slightly elevated white cell count (12.17×10^9/L), low hemoglobin (47g/L), and normal platelet levels (123×10^9/L). The bone marrow smear showed a significant increase in immature granulocytes (33.5%) (Fig 1A), while flow cytometry detected three abnormal myeloid precursor cell populations accounting for 33.86% of the nucleated cells (Fig 1B, C, D). The karyotype analysis yielded the following results: 45,XY,dup(3)(q21q26.2)×2,-7[13]/45,XY,t(3;3)(q21;q26.2),-7[7] (Fig 2A). Both interphase and metaphase FISH revealed the fusion of *MECOM* and GATA2 in both the dup(3q) and t(3;3) clones (Fig 2B, C, D). Consequently, the patient was diagnosed with AML with *MECOM* rearrangement. He underwent treatment with the DA (Daunorubicin + Cytarabine) regimen and was monitored using flow cytometry over a five-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 the *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 fuse 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 both chromosome 3, which revealing a novel mechanism of chromosomal alteration contributing to *MECOM* rearrangement, simultaneously, the clones presented with *MECOM* rearrangement caused by dup(3q) is sensitive to DA regimen.

AUTHOR CONTRIBUTIONS

Shaobin Yang, Shiyang Ma collected and analyzed the data and wrote the manuscript. Xiaoyan Yan and Jingya Yao prepared the images. Yani Lin organized the study and edited the manuscript.

CONFLICT OF INTEREST STATEMENT

The authors declare no conflicts of interest.

DATA AVAILABILITY STATEMENT

Data sharing is not applicable to this article as no new data were created or analyzed in this study.

ETHICS STATEMENT

Not applicable (single case report).

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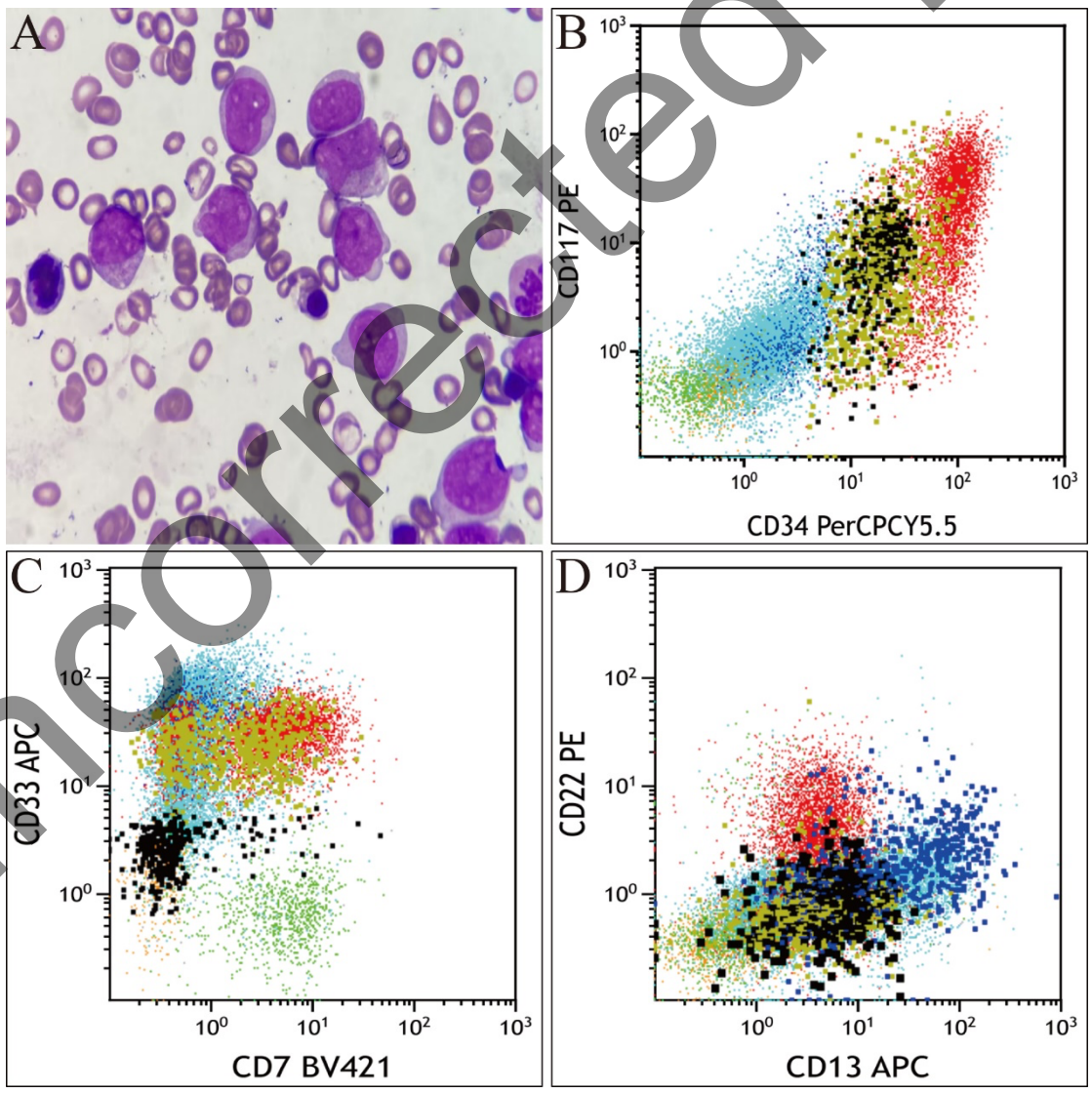

Figure 1 A: Bone marrow smear revealed increased blasts. B, C, D: Flow cytometry configured the gating strategy with 34/SSC to output all blast cells. Then, use CD33/HLA-DR to distinguish the CD33-negative and weakly positive HLA-DR black cell population. For another cell group, use CD38 for gating: strongly positive CD38 cells are the military green group, and CD38-positive cells are the red group. Three abnormal myeloid precursor cell populations were identified. The first group (28.48%) (red) expressed myeloid (CD33, CD13, CD117, with minor weak expression of MPO), lymphoid (CD7, CD22, CD56, CD2, CD4) antigens ,as well as HLA-DR and CD38. The second group (3.46%) （military green） expressed myeloid (CD33, CD13, CD117,with minor weak expression of MPO) and lymphoid (CD7, 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, and was also negative for MPO.

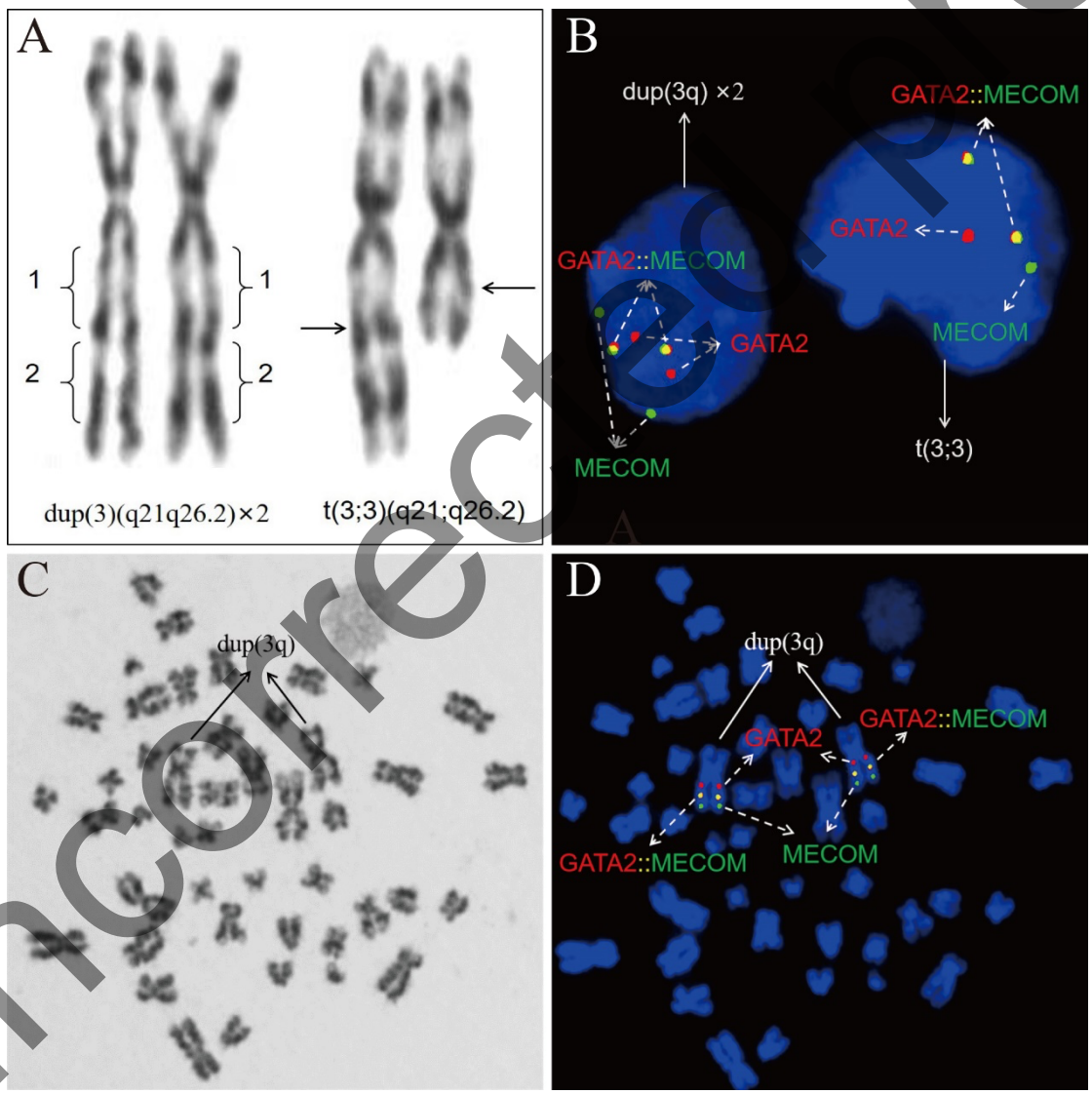

Figure 2 A: Bone marrow karyotype analysis showed dup(3)(q21q26.2)×2 and t(3;3)(q21;q26.2) in two different clones. B, C, D: Interphase and metaphase FISH proved co-localization of GATA2 and *MECOM* gene in dup(3q) and t(3;3) clones.