Duplication of der(21)t(8;21)(q22;q22) in Acute Myeloid Leukemia

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The t(8;21)(q22;q22) results in the formation of RUNXI/RUNX1T1 (alias AML1/ETO) and RUNX1T1/RUNX1 fusion genes on der(8)t(8;21)(q22;q22) and der(21)t(8;21)(q22;q22), respectively. An 18-year-old woman was diagnosed as having acute myeloid leukemia (AML) M2, as her bone marrow was infiltrated with 42.2% myeloblasts. Auer rods were found in myeloblasts and metamyelocytes (Picture A, B, arrows). A chromosome analysis showed 46,XX,t(8;21)(q22;q22)[8]/47,sl,+der(21)t(8;21)[2]/46,XX[10] (Picture C, the arrows indicate rearranged chromosomes). Fluorescence in situ hybridization (FISH) revealed one RUNX1/RUNX1T1 fusion (green/red) signal on the der(8)t(8;21) (Picture D, arrowhead) and two RUNX1T1/RUNX1 fusion signals on the two der(21)t(8;21) (Picture D, arrows). FISH of the interphase nuclei confirmed three fusion signals (Picture D, inset, arrowheads). Reverse-transcription polymerase chain reaction also detected the RUNX1/RUNX1T1 fusion transcript.

Despite the essential role of RUNX1/RUNX1T1 in leukemogenesis, +der(8)t(8;21) has never been found in an AML patient with t(8;21). Conversely, +der(21)t(8;21) has been sporadically described in 23 AML cases, indicating that +der(21)t(8;21) is a rare but recurrent secondary abnormality (1, 2). The reciprocal RUNX1T1/RUNX1 fusion gene may therefore play a role in the progression of AML, although its fusion transcript is not usually identified (3).

The authors state that they have no Conflict of Interest (COI).
References


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