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Leukaemia Section
Short Communication

der(3)t(3;3)(p25-26;q12-21)

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Abstract

Partial or complete chromosome gains are frequently found in hematological malignancies, but the unbalanced der(3)t(3;3) is a relatively rare chromosome anomaly.

Keywords
chromosome 3; hematological malignancies

Clinics and pathology

Disease

Myeloid malignancies and lymphomas.

Phenotype/cell stem origin

Myeloid malignancies in 3 cases: 1 refractory anemia with excess blasts-1 (RAEB) (Yamamoto et al., 2004), 1 acute erythroleukemia (Olopade et al, 1992) and the present patient diagnosed with acute myeloid leukemia (AML). In addition, there was a 66- years old female patient with 3q23 breakpoint diagnosed with idiopathic myelofibrosis (Reilly et al., 1997). 4 patients had various forms of lymphomas: 1 angioimmunoblastic T-cell lymphoma (Levine et al., 1985), 1 follicular lymphoma (Schlegelberger et al., 1990) 1 diffuse large B-cell lymphoma (Goyns et al., 1993) and 1 mantle cell lymphoma (Wlodarska et al., 19991).

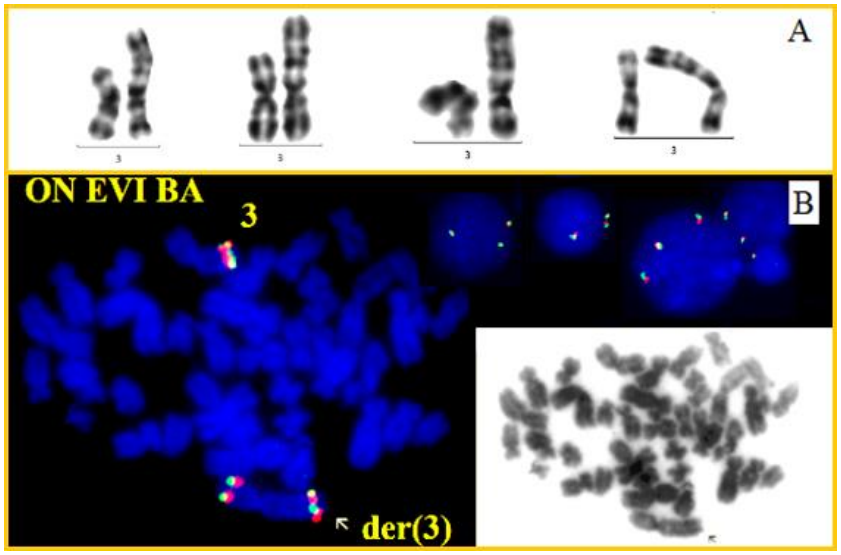

Figure 1. Karyotype of the patient showing the unbalanced translocation of chromosome 3 and associated with 13q deletion. Partial karyotypes showing the rearranged chromosome 3 (A). Fluorescence in situ hybridization with Keratech MECOM t(3;3); inv(3)(3q26) also known as EVI t(3;3); inv(3)(3q26) break-apart probe (Kreatech Biotechnology B.V., NL) showing 3 copies of the gene located on 3q26 as a result of the unbalanced translocation (B).

Epidemiology

Only 6 reported patients (3M/3F) aged 56, 66, 60 and 75 years (2 unknown) and the present 46-years old female patient (unpublished data).

Clinics

The present patient was diagnosed with AML, NOS in 2006 and achieved complete hematological remission after chemotherapy but relapsed 4 years later. After bone marrow transplantation she maintained her remission status until November 2017 when she relapsed with 26% blasts in the blood.

Prognosis

Found as part of highly complex karyotypes, therefore it may be associated with advanced-stage disease.

Cytogenetics

Cytogenetics morphological

Presents as 1 normal chromosome 3 and a der(3)t(3;3) chromosome in 6 and as 2 normal chromosomes 3 and +der(3) in 1 patient.

Additional anomalies

Highly complex karyotypes in both myeloid and lymphoid malignancies, found in a sideline with del(13q) as a sole additional anomaly in the idiopathic myelofibrosis patient with 3q23 breakpoint (Reilly et al., 1997). Found at relapse after bone marrow transplantation as a sole anomaly in 10 and in association with del(13)(q22?) in 5 out of the 25 examined metaphases in the present AML patient.

Result of the chromosomal anomaly

Fusion protein

Oncogenesis

der(3)t(3;3)(p25-26;q12-21) is a rare cytogenetic abnormality that has been observed in sporadic cases of myeloid malignancies and lymphomas. Mainly found as part of complex karyotypes with multiple numerical and structural anomalies, reflecting stepwise development of chromosomal abnormalities. The result of this unbalanced translocation is partial trisomy of the long arm of chromosome 3 causing deregulation of proto-oncogenes via gene dosage effect that may lead to their overexpression.

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