Abstract

Rare translocation. Because of its rarity, the clinical significance of der(17)t(17;17)(p13;q12-21) is unknown.

Keywords
Unbalanced rearrangements, der(17)t(17;17)(p13;q12-21), myeloid malignancies, genomic unbalances.

Disease
Chronic myeloid leukemia (CML) and acute myeloid leukemia (AML)

Phenotype/cell stem origin
1 acute myeloid leukemia (Huh et al., 2016), 1 chronic myeloid leukemia (Barbouti et al., 2002) and the present patient diagnosed with T-ALL after 2 years of lentiviral hematopoietic stem cell gene therapy for Wiskott-Aldrich syndrome (Zamecnikova, unpublished data). In addition, there was an acute myelomonocytic leukemia (AML-M4) patient with der(17)t(17;17)(p13;q21) breakpoints (Larson et al., 1986).

Epidemiology
Only 4 cases, to date, 3 male patients, aged 60, 76 and years and the present 14 years old female patient with Wiskott-Aldrich syndrome.

Cytogenetics

Cytogenetics morphological
Presents as 1 normal chromosome 17 and a der(17)t(17;17) chromosome.

Additional anomalies

Sole anomaly in the present patient; found in association with +6 and 15-100 dmin in AML, as an additional anomaly to -Y, t(9;22)(q34;q11) in CML and to inv(16)(p13q22),+22 in CC: TXT: AML-M4 ID: 1506>.

Genes involved and proteins

The involvement of RARA and the presence/deletion of TP53 from 17p13.1 was not tested in described cases.

Result of the chromosomal anomaly

Fusion protein

Oncogenesis
The unbalanced der(17)t(17;17)(p13;q12-21) has been found mainly as an additional aberration to known primary anomalies, therefore it is probably involved in disease evolution.

The formation of the unbalanced der(17)t(17;17)(p13;q12-21) results in partial trisomy of the long arm of chromosome 17 leading to trisomies of genes located on 17q.

Potential candidate genes on 17q include RARA, NF1, CSF3 (G-CSF), MPO, ERBB2 and miRNAs. Extra copies of these genes may lead to alterations of gene expression that may play roles during disease development or progression.
Figure 1. Partial karyotypes (A) and karyotype showing the unbalanced rearrangement between chromosomes 17 (B). Fluorescence in situ hybridization with LSI ATM/ P53 probe (Abott Molecular/Vysis, US) showing the presence of P53 gene on normal and der(17) chromosomes (A). Hybridization with LSI RARA break-apart probes revealed 3 copies of the gene, confirming the extra 17q on derivative chromosome 17 (B).

References


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