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# Leukaemia Section

## Short Communication

# der(17)t(17;17)(p13;q12-21)

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## 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.Clinics and pathology

### *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with T-ALL after 2 years of lentiviral hematopoietic stem cell gene therapy for Wiskott-Aldrich syndrome (Zamecnikova, unpublished data).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;q2?1) 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.

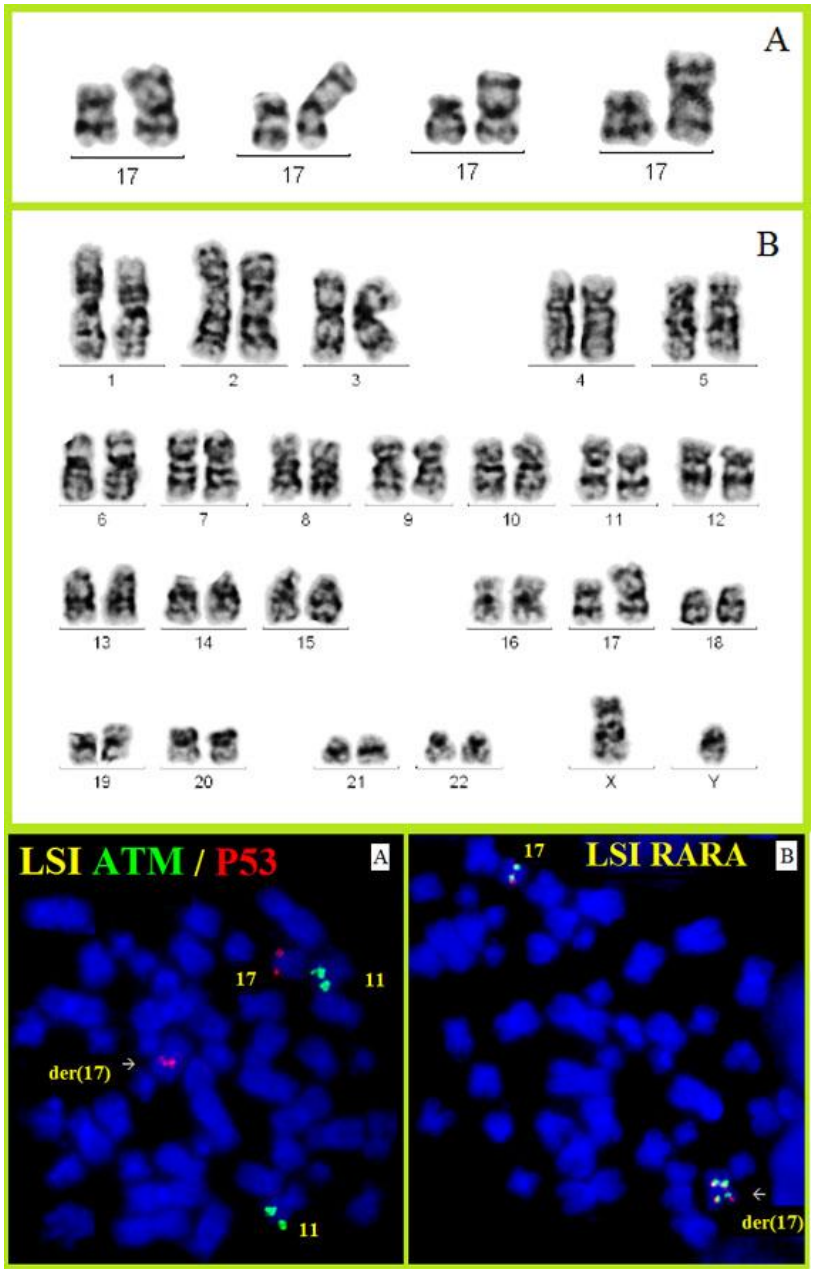


**Figure 1.** Partial karyotypes (A) and karyotype showing the unbalanced rearrangement between chromosomes 17 (B). Fluorescence in situ hybridization with LSI ATM/P53 probe (Abott Molecular/Vysis, US) showing the presence of P53 gene on normal and der(17) chromosomes (A). Hybridization with LSI RARA break-apart probes revealed 3 copies of the gene, confirming the extra 17q on derivative chromosome 17 (B).

# References

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