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in Oncology and Haematology

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Leukaemia Section
Mini Review

t(X;11)(q21;q23)

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Identity

Other names

Originally, this translocation had been published as t(X;11)(q13;23), before BRWD3 was re-mapped to Xq21.1.

Clinics and pathology

Disease

B-cell chronic lymphocytic leukemia (B-CLL)

Epidemiology

Only one case reported to date. In a second B-CLL case, a variant translocation t(11;13)(q23;q14) rearranged the same gene on 11q23 with another partner.

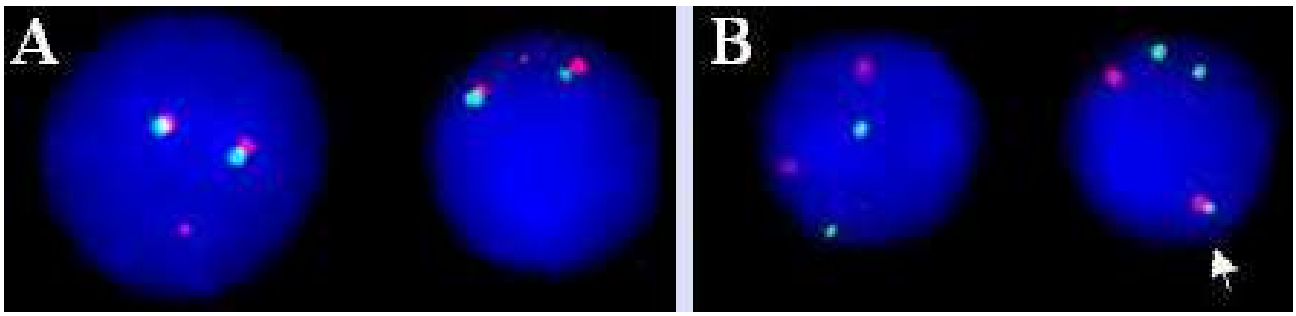
Cytogenetics

Probes

BAC clones covering the Xq21 breakpoint region: RP11-325E14, RP11-625B7.
BAC clones covering the 11q23 breakpoint region: RP11-468P24, RP11-264L21, RP11-285P16.

Variants

t(11;13)(q23;q14) is a variant translocation that rearranged ARHGAP20 with a novel gene on 13q14 (unpublished data).



Dual-color interphase FISH analysis of the 11q23 and Xq21 breakpoints with BAC clones.
(A) FISH analysis with 11q23 specific BACs: RP11-468P24 (red signals) and RP11-206G12 (green signals). Three red signals indicate translocation within the genomic region represented by RP11-468P24.
(B) FISH analysis with the 11q23 specific BAC RP11-264L21 (green signals) and the Xq21 BAC RP11-325E14 (red signals). In the right cell, colocalization of one red and one of the three green signals indicates transfer of 11q23 sequences to Xq21 (white arrow).

Genes involved and Proteins

BRWD3

Location: Xq21.1
Note: BRWD3 had been originally mapped to Xq13.3.

DNA / RNA

5' telomeric → 3' centromeric orientation; 44 exons spanning 132.7 kb genomic DNA; mRNA coding sequence: 4.2 - 5.4 kb.

Protein

Contains eight tandem WD40 repeats and two bromodomains; involved in the JAK/STAT signalling cascade.

ARHGAP20

Location: 11q23.1

DNA / RNA

5' telomeric → 3' centromeric orientation; 19 exons spanning 136.1 kb genomic DNA; mRNA coding sequence: 3.5-3.6 kb.

Protein

Contains a RhoGAP domain in combination with PH and RA modules; involved in the regulation of Rho-family GTPases (e.g. regulating the neurite outhgrowth); cytoplasmic localisation.

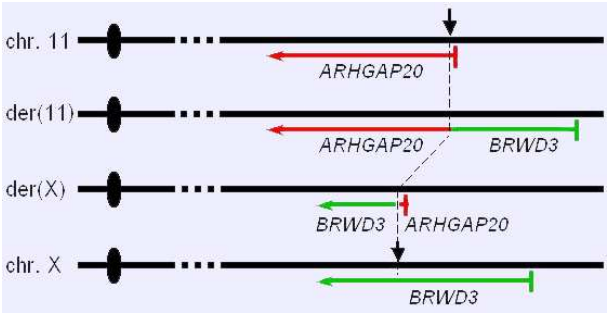
Results of the chromosomal anomaly

Hybrid gene

Description

der(11): 5' BRWD3 (exons 1-22) - 3' ARHGAP20 (exons 1e-15);
der(X): 5' ARHGAP20 (promoter-part of exon 1e) - 3' BRWD3 (exons 23-41);

BRWD3 and ARHGAP20 are in the same transcriptional directions, fusion transcripts, however, were not detected.



Schematic representation of the ARHGAP20-BRWD3 gene rearrangement. Black ovals represent the centromeres. The gene loci and orientation of ARHGAP20 and BRWD3 and their promoter regions are indicated by red and green arrows and boxes, respectively. Black arrows and the dashed line indicate the position of the breakpoints.

Fusion protein

Note: No fusion transcript expressed.

To be noted

Both t(X;11)(q21;q23) and the variant translocation t(11;13)(q23;q14) affect ARHGAP20, which resides within the critical 11q22-q23 deletion region in B-CLL. Deletion of this genomic region is associated with an aggressive course of B-CLL.

References

Kalla C, Nentwich H, Schlotter M, Mertens D, Wildenberger K, Döhner H, Stilgenbauer S, Lichter P. Translocation t(X;11)(q13;q23) in B-cell chronic lymphocytic leukemia disrupts two novel genes. Genes Chromosomes Cancer 2005;42:128-143.

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