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Leukaemia Section
Short Communication

t(17;17)(q21;q24), del(17)(q21q24)

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Clinics and pathology

Disease

Acute myeloid leukaemia, M3 subtype (M3-AML)

Epidemiology

Only one case to date, a 66-year-old male patient (Catalano et al., 2007).

Cytology

Auer rods and fagot cells were absent.

Evolution

Complete remission was obtained with ATRA, and the patient remains healthy 2 years after the diagnosis.

Cytogenetics

Cytogenetics morphological

Cryptic deletion, FISH studies are needed to uncover the rearrangement.

Genes involved and proteins

RARA

Location
17q21.1

Protein
Contains Zn fingers and a ligand binding region. Receptor for retinoic acid. Forms heterodimers with RXR. At the DNA level, binds to retinoic acid response elements (RARE). Ligand-dependent transcription factor specifically involved in hematopoietic cells differentiation and maturation.

PRKAR1A

Location
17q24.2

Protein
Contains two tandem cAMP-binding domains. Forms heterotetramers with PRKACA (protein kinase, cAMP-dependent, catalytic, alpha), also called PKA. Interacts with RARA, and regulates RARA transcriptional activity.

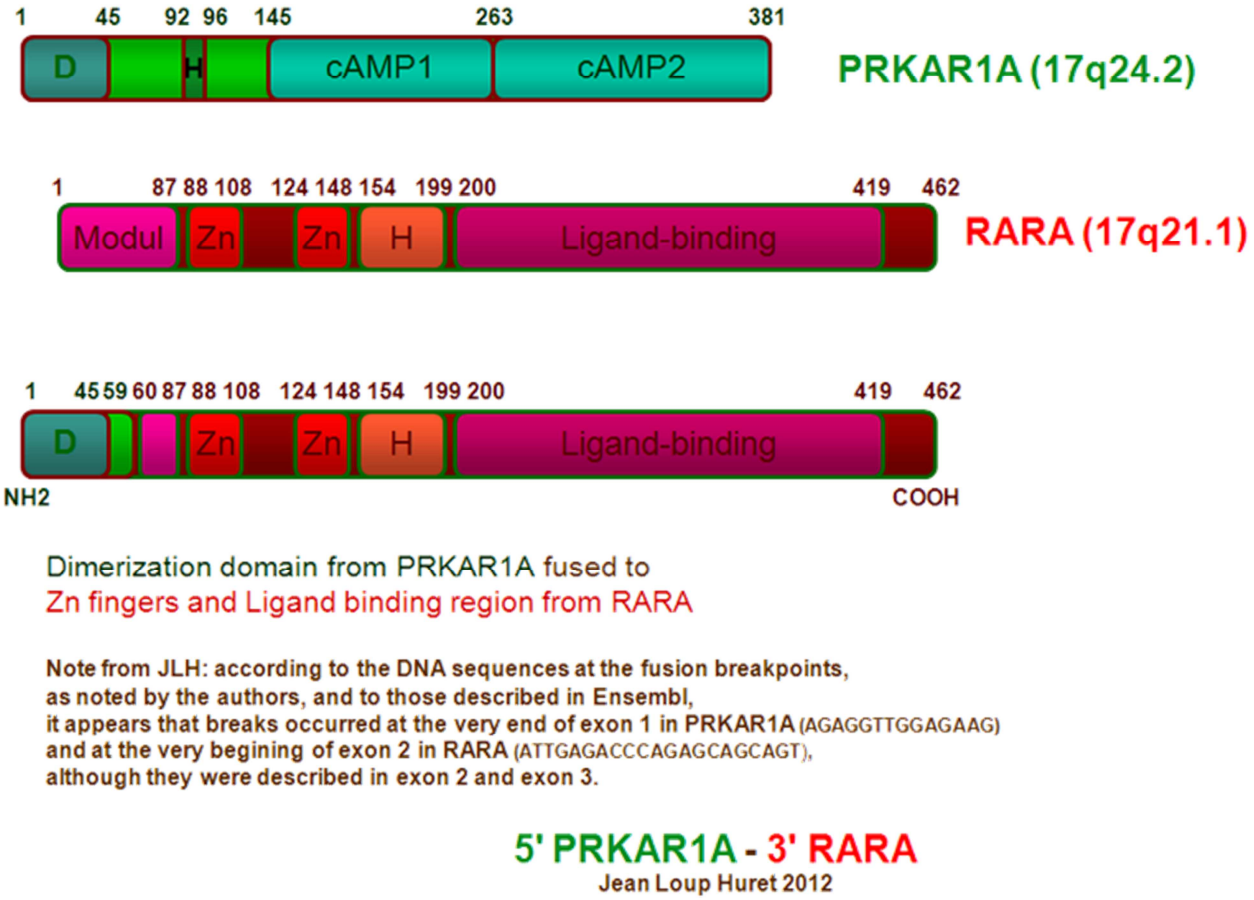
Result of the chromosomal anomaly

Hybrid gene

Description
5' PRKAR1A - 3' RARA. When we look closely to the DNA sequences at the fusion breakpoints, they correspond to the very end of exon 1 in PRKAR1A (AGAGGTTGGAGAAG) and the very begining of exon 2 in RARA (ATTGAGACCCAGAGCAGCAGT, see sequences in Ensembl), although they were described in exon 2 and exon 3 in the first and only report of this rearrangement (Catalano et al., 2007).

Fusion protein

See figure 5' PRKAR1A - 3' RARA.



Description

The fusion protein contains the dimerization domain from PRKAR1A fused to the Zn fingers and ligand binding regions from RARA.

References

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