Atlas of Genetics and Cytogenetics in Oncology and Haematology

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Leukaemia Section

Mini Review

inv(11)(p15q22), t(11;11)(p15;q22)

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Identity

Note: Only one case of t(11;11) with NUP98-DDX10 fusion has been described; most cases are cases of inv(11).

Clinics and pathology

Disease

de novo Myelodysplastic syndromes (MDS) and Acute Non Lymphoblastic Leukemias (ANLL), therapy related MDS and ANLL, one case of Chronic Myelogenous Leukemia (CML).

Epidemiology

Thirteen cases reported to date: six adults and seven children (9M/4F).

Cytogenetics

Probes

NUP98: PAC 1173K1; DDX10: RP11-976P22 (centromeric) and RP11-25I9 (telomeric).

Additional anomalies

Sole anomaly in 8 out of 13 cases.

Genes involved and Proteins

NUP98

Location: 11p15.5

Protein

Nucleoporin 98, a 98 kDa component of the nuclear pore complex involved in nucleo-cytoplasmic transport.

DDX10

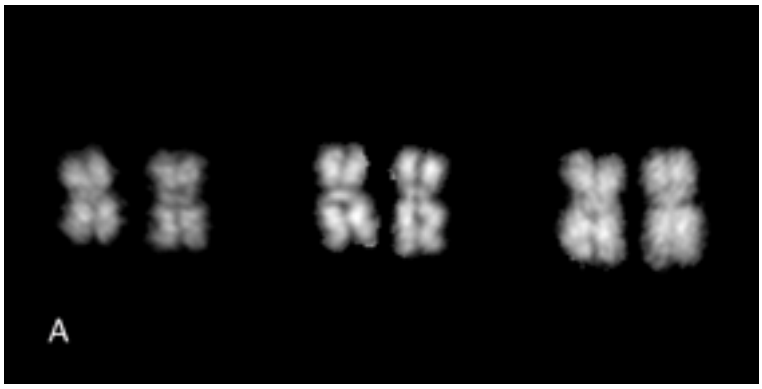
Location: 11q22

DNA / RNA

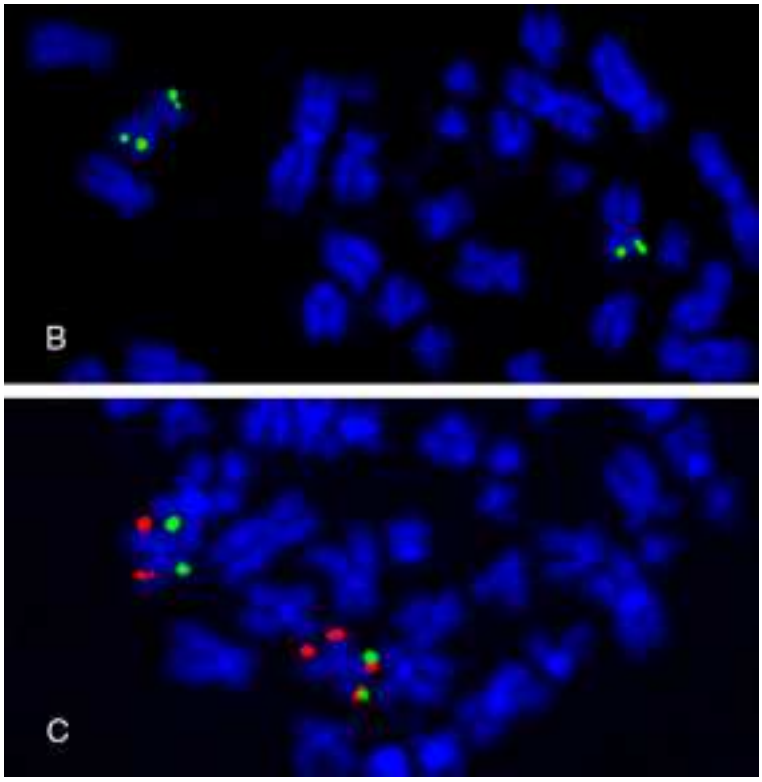
Alternative splicing; 3.2 and 5.0 kb mRNA.

Protein

Putative DEAD-box RNA helicase involved in ribosome biogenesis.



A: inv(11)(p15q22) Q-banding: the derivative chromosomes are on the right of each pair.



B: FISH with NUP98 PAC 1173K1 probe. A split signal indicates pericentric inversion in the der(11). C: Cohybridization of DDX10 centromeric BAC RP11-976P22 (red) and subtelomeric 11p RP11-534I22 probe (green).

Results of the chromosomal anomaly

Hybrid gene

Description

5' NUP98 - 3' DDX10

Twelve out of 13 cases present exon 14 of NUP98 fused in-frame to exon 7 of DDX10, one case with exon 12 of NUP98 fused in-frame to exon 6 of DDX10.

Fusion protein

Description

It fuses the GLFG repeat domains of NUP98 to the acidic domain of DDX10.

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