t(3;12)(q26;p13)
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Published in Atlas Database: September 1997

Online version is available at: http://AtlasGeneticsOncology.org/Anomalies/0312.html
DOI: 10.4267/2042/32033

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Identity

- t(3;12)(q26;p13) G-banding - Courtesy Jean-Luc Lai and Alain Vanderhaegen.

Clinics and pathology

Disease
Myeloid lineage: MDS in transformation, ANLL, BC-CML.

Phenotype / cell stem origin
Multilineage involvement; RAEB → M2, M7 and others ANLL subtypes.

Epidemiology
Only 8 cases described so far; sex ratio: 3M/1F; age: 3-87 yrs (med: 40 yrs).

Cytology
Megakaryocytes dysplasia

Prognosis
Very poor; survival often below 1 yr.

Cytogenetics, molecular
Heterogenous breakpoints.

Probes
- EVI1: ly2 and 13 E Parganas, St Jude Children's Research Hospital, Memphis, TN;
- MDS1: P856 G Nucifora, Univ. of Chicago, IL;
- TEL: YAC 958b8 CEPHII Mega-YAC library Paris, France;
- TEL: cDNA c50F4, c163E7, c148B6 Lawrence Livermore National Laboratories, Livermore, CA.

Additional anomalies
del(7q) or -7.

Genes involved and Proteins

MDS1

EVI1
Location: 3q26

ETV6
Location: 12p13
DNA / RNA
9 exons; alternate splicing.

Protein
Contains a Helix-Loop-Helix and ETS DNA binding domains; wide expression; nuclear localisation; ETS-related transcription factor.

References


This article should be referenced as such: