

Leukaemia Section

Short Communication

t(12;13)(p12;q12-14) ETV6/CDX2

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Published in Atlas Database: April 2000

Online updated version : <http://AtlasGeneticsOncology.org/Anomalies/t1213ID1204.html>

DOI: 10.4267/2042/37623

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Clinics and pathology

Disease

Chronic myelogenous leukemia (CML) in transformation, myelodysplastic syndrome (MDS), acute non lymphocytic leukemia (ANLL), B and T-acute lymphocytic leukemias (ALL).

Cytogenetics

Probes

The chromosome 13 breakpoint in most myeloid cases falls within YAC 875a8. Other cases are further proximal and may involve CDX2, contained in BAC RP11-328P22. In most lymphoid cases the chromosome 13 breakpoint falls within YAC 879f5.

Additional anomalies

No consistent additional abnormalities.

Genes involved and proteins

ETV6

Location

12p13

CDX2

Location

13q12.3

To be noted

Note

The t(12;13) is heterogeneous at the molecular level. A single case of ANLL has been reported with an ETV6-CDX2 fusion and ectopic expression of normal CDX2 but in the same study two other t(12;13) positive ANLL

patients did not have CDX2 involvement. FISH analysis has suggested that the chromosome 13 breakpoints are different in myeloid and lymphoid disease. ETV6 seems to be involved in some cases but not others.

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This article should be referenced as such:

Cross NCP. t(12;13)(p12;q12-14) ETV6/CDX2. *Atlas Genet Cytogenet Oncol Haematol*. 2000; 4(2):78-79.
