t(6;12)(p21;p13) in lymphoid malignancies

Maria D Odero

Division of Oncology, Center for Applied Medical Research (CIMA), University of Navarra, Pamplona, Spain

Published in Atlas Database: August 2006
Online updated version: http://AtlasGeneticsOncology.org/Anomalies/t0612p21p13ID1424.html
DOI: 10.4267/2042/38382

This work is licensed under a Creative Commons Attribution-Non-commercial-No Derivative Works 2.0 France Licence. © 2007 Atlas of Genetics and Cytogenetics in Oncology and Haematology

Identity

\[
\begin{array}{ccc}
6 & \text{der}(6) & 12 \\
6 & \text{der}(6) & \text{der}(12)
\end{array}
\]

\[t(6;12)(p21;p13)\text{ G-banding.}\]

Clinics and pathology

Disease

t(6;12)(p21;p13) has been described in only 6 cases: chronic lymphocytic leukemia (CLL); acute lymphoblastic leukemia (ALL); diffuse large B-cell lymphoma; acute myeloid leukemia (AML); NOS; myelodysplastic syndrome (MDS), RAEB-2; and breast adenocarcinoma.

Phenotype / cell stem origin

B lineage.

Prognosis

No prognosis value established.

Cytogenetics

Cytogenetics morphological

May be not easy to detect.

Cytogenetics molecular

In CLL, the translocation was detected by FISH with ETV6 probes. The ETV6 gene is rearranged, and the breakpoint is between exon 1 and exon 2.

Additional anomalies

-9 and \(\text{der}(16)t(9;16)(q21;q24)\) in CLL; and \(\text{del}(7)(p13p22)\) in ALL.

Variants

No variants in CLL and ALL.

Genes involved and Proteins

ETV6

Location: 12p13

Note: The gene is known to be involved in a large number of chromosomal rearrangements associated with leukemia and congenital fibrosarcoma.

DNA / RNA

9 exons; alternate splicing.

Protein

The gene encodes an ETS family transcription factor; the product of this gene contains a N-terminal pointed (PNT) domain that is involved in the protein-protein interactions, and a C-terminal ETS DNA-binding domain; wide expression; nuclear localization.

CCND3 (cyclin D3)

Location: 6p21

Note: Could be the putative gene involved on 6p21. No molecular studies on 6p21 are described in cases with t(6;12). In t(6;14)(p21;q32), the breakpoint is centromeric to the CCND3 gene, causing dysregulation and overexpression of CCND3.

Results of the chromosomal anomaly

Fusion protein

Description

In CLL the ETV6 gene is rearranged; the breakpoint in ETV6 is between exon 1 and exon 2.
References


This article should be referenced as such: