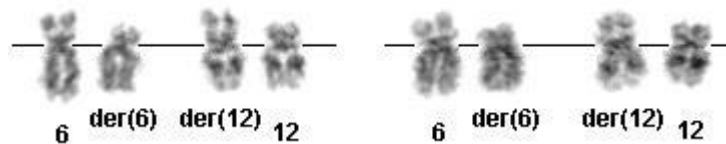


## t(6;12)(p21;p13) in lymphoid malignancies

### Identity



t(6;12)(p21;p13) 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](#); (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 der(16)t(9;16)(q21;q24) in CLL; and del(7)(p13p22) in ALL.

**Variants** No variants in CLL and ALL.

### Genes involved and Proteins

**Gene Name** [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.

**Gene Name** [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.

Dna / Rna .

Protein .

## Result 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.

## External links

Other database [t\(6;12\)\(p21;p13\) in lymphoid malignancies](#) [Mitelman database \(CGAP - NCBI\)](#)

Other database [t\(6;12\)\(p21;p13\) in lymphoid malignancies](#) [CancerChromosomes \(NCBI\)](#)

## To be noted

Additional cases are needed to delineate the epidemiology of this rare entity:  
**you are welcome to submit a paper to our new [Case Report](#) section.**

## Bibliography

### Immunophenotype-karyotype associations in human acute lymphoblastic leukemia.

Uckun FM, Gajl-Peczalska KJ, Provisor AJ, Heerema NA.

Blood 1989; 73: 271-280.

Medline [2910365](#)

### Identification of new translocations involving ETV6 in hematologic malignancies by fluorescence in situ hybridization and spectral karyotyping.

Odero MD, Carlson K, Calasanz MJ, Lahortiga I, Chinwalla V, Rowley JD.

Cancer 2001; 31: 134-142

Medline [11319801](#)

## Contributor(s)

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