

## t(9;12)(q34;p13)

### Clinics and Pathology

**Disease** described in only 6 cases; acute lymphoblastic leukemia (ALL), acute non lymphocytic leukemia (ANLL) and [chronic myeloid leukemia](#) (CML)

**Prognosis** numbers small, but one CML case had allogeneic BMT and is in complete remission, the remaining cases had rapid disease progression and died of shortly after diagnosis

### Cytogenetics

**Cytogenetics** t(9;12)(q34;p13), cryptic at the cytogenetic level

**Morphological Variants** t(9;12;14)(q34;p13;q22) and complex insertions of ETV6 into ABL

### Genes involved and Proteins

**Gene Name** [ABL](#)

**Location** 9q34

**Dna / Rna** ETV6 is fused to exon 2 of ABL in the three cases described

**Protein** tyrosine kinase, localized primarily to the nucleus

**Gene Name** [ETV6](#)

**Location** 12p13

**Dna / Rna** 9 exons; alternate splicing

**Protein** contains Helix-Loop-Helix (HLH) at N-terminal end and ETS DNA binding domain at C-terminal end; wide expression; nuclear localization; ETS-related transcription factor

### Result of the chromosomal anomaly

**Hybrid gene** 5' ETV6 - 3' ABL; two different fusion breakpoints have been described; ETV6 exon 4 fused in frame to ABL exon 2 (Type A) and ETV6 exon 5 fused in frame to ABL exon 2 (Type B); ETV6 maintains the HLH domain and ABL the tyrosine kinase domain

**Fusion Protein** a 155 kDa protein in Type A, 180 kDa protein in Type B; has elevated tyrosine kinase activity, localized in the cytoplasm and co-localizes with the actin filaments of the cells

**Description** the HLH domain of ETV6 induces oligomerization, which results in the constitutive activation of the kinase domain of ABL; this is thought to result in phosphorylation of [JAK2](#) and activation of the ONCOGENESIS biological activity very similar to BCR-ABL

### External links

**Other database** [t\(9;12\)\(q34;p13\)](#) [Mitelman database \(CGAP - NCBI\)](#)

**Other** [t\(9;12\)\(q34;p13\)](#) [CancerChromosomes \(NCBI\)](#)

database

## 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

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