t(1;12)(p36;p13)

Jean-Loup Huret

Genetics, Dept Medical Information, UMR 8125 CNRS, University of Poitiers, CHU Poitiers Hospital, F-86021 Poitiers, France (JLH)

Published in Atlas Database: July 2003


DOI: 10.4267/2042/37992

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Clinics and pathology

**Disease**

Myeloid disorders: one chronic myelogenous leukemia with t(9;22) and one refractory anemia with excess of blasts in transformation.

**Epidemiology**

Only 2 cases so far: 1 male and 1 female patient aged 50 and 66 yrs.

**Prognosis**

Unknown so far.

Genes involved and proteins

**MDS2**

Location

1p36

DNA/RNA

7 exons; alternate splicing.

**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.

Result of the chromosomal anomaly

**Hybrid gene**

*Description*

5′ ETV6 - 3′ MDS2; exons 1 and 2 of ETV6 are fused to exons 6 and 7 of MDS2; fusion is not in frame; the reciprocal fusion gene is not expressed.

**Fusion protein**

*Description*

Truncated ETV6 lacking the PTN domain and the DNA binding domain.

To be noted

**Case Report**

A Case of Myelodysplastic Syndrome with a Translocation t(1;12)(p36;p13)

References


This article should be referenced as such: