Leukaemia Section
Mini Review

**t(5;12)(q33;p13)**

Jean-Loup Huret

Genetics, Dept Medical Information, University of Poitiers, CHU Poitiers Hospital, F-86021 Poitiers, France

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**Identity**

*Note:* this translocation must not be confused with the t(5;12)(q31;p13) found in an ALL cell line with IL3 and ETV6 involvements.

![G-banding and R-banding images](image)

**Clinics and pathology**

**Disease**

Myeloid lineage.

**Phenotype / cell stem origin**

Myeloproliferative/myelodysplastic syndrome (intermediate between CML and CMML) with eosinophilia; appear to be a specific entity.

**Epidemiology**

Rarely described; mostly in adult male patients (13 of 14 cases herein reviewed).

**Clinics**

Organomegaly in 9 of 12 cases; blood data: median WBC: 40 X 10^9/l; numerous eosinophils: median 2.8 X 10^9/l, range 0.8-128 X 10^9/l, n=9 (normal range is 0.02-0.45 X 10^9/l), and, at times, of monocytes; no blast cells.

**Treatment**

Hydroxyurea alone or polychemotherapy have been essayed.

**Prognosis**

Yet partly undetermined; median survival < 20 mths (n=11).
Cytogenetics

Additional anomalies
+8.

Genes involved and Proteins

**PDGFRB**

Location: 5q33

Protein

PDGFRB is the receptor for PDGFB (platelet-derived growth factor-b); membrane protein; belongs to the immunoglobulin superfamily.

**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.

Results of the chromosomal anomaly

Hybrid gene

Description

5' ETV6 - 3' PDGFRB.

Fusion protein

Description

N-term HLH domain of ETV6 fused to the transmembrane domain and the Tyr kinase domain of PDGFRb in C-term; the reciprocal transcript is not expressed.

References


This article should be referenced as such: