t(5;7)(q33;q11)

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Disease

Only one case available to date: a 54 yr old man with chronic myelomonocytic leukaemia.

Prognosis

Yet unknown.

Genes involved and Proteins

PDGFRB

Location: 5q33

Protein

Contains from N-term to C-term: a ligand binding domain, a transmembrane domain, and a Tyrosine kinase domain; PDGFRB is the receptor for PDGFB (platelet-derived growth factor-b); membrane protein; belongs to the immunoglobulin superfamily.

HIP1

Location: 7q11

Protein

Contains a leucine zipper and, in C-term, a region with homology to a protein of the cytoskeleton; membrane associated protein.

Results of the chromosomal anomaly

Hybrid gene

Description

5' HIP1- 3' PDGFRb; breakpoint in PDGFRb similar to what is found in the t(5;12)(q33;p12).

Fusion protein

Description

180 kDa; contained nearly all of the HIP1 coding sequence, including the leucine zipper and talin homology domains, fused in frame to the transmembrane and tyrosine kinase domain of the PDGFB; the reciprocal PDGFB-HIP1 transcript is not expressed.

Oncogenesis

Constitutive activation of PDGFb (by tyrosine autophosphorylation).

References


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