t(5;17)(q33;p13)

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Disease

Chronic myelomonocytic leukemia

Epidemiology

Only one case to date, a 29 year old male patient (Magnusson et al., 2001).

Cytogenetics

Additional anomalies

Sole anomaly.

Genes involved and proteins

PDGFRb

Location

5q33

Protein

1106 amino acids. Composed from NH2 to COOH of: Ig-like extracellular domains, a transmembrane domain, and a cytosolic tyrosine kinase domain. Tyrosine kinase membrane receptor.

RABEP1

Location

17p13

Protein

826 amino acids protein with coiled-coil domains (dimerization) and a NH2-term RAB4 binding site, and a COOH-term RAB5 binding site. Role in endocytosis.

Result of the chromosomal anomaly

Hybrid gene

Description

5’ RABEP1- 3’ PDGFRb; no reciprocal transcript.

Fusion protein

Description

1318 amino acids (aa) fusion protein, including most of RABEP1 (the first 739 aa) with 3 and one half of the 4 coiled-coil domains, fused to the transmembrane and intracytosolic tyrosine kinase domains of PDGFRb.

References


This article should be referenced as such: