t(5;17)(q33;p11.2)

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Published in Atlas Database: September 2004

Online updated version: http://AtlasGeneticsOncology.org/Anomalies/t0517q33p11ID1327.html

DOI: 10.4267/2042/38137

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Clinics and pathology

**Disease**

Juvenile myelomonocytic leukemia.

**Epidemiology**

Only one case available to date: a 18-month-old boy.

**Clinics**

Marked hepatosplenomegaly, trombocytopenia.

**Cytology**

Peripheral blood leukocytosis with eosinophilia, bone marrow fibrosis (grade III), dry tap.

**Treatment**

Allogeneic BMT.

**Evolution**

Alive and free of disease after 4 years.

Cytogenetics

**Probes**

LSI CSF1R probe (Vysis) for PDGFRB (5q33); RP11-7904 (telomeric) and RP11-81D5 (centromeric) for HCMOGT-1 (p11.2).

**Genes involved and proteins**

**PDGFRB**

Location: 5q33

Protein

Ig like, transmembrane and tyrosine kinase domains.

**HCMOGT-1**

Location: 17p11.2

Result of the chromosomal anomaly

**Hybrid gene**

Description

5'HCMOGT-1/3'PDGFRB The HCMOGT1 cDNA sequence was fused in-frame to PDGFRB at nucleotide 1936 and situated 5' to the PDGFRB portion, as in previous studies of PDGFRB fusion partners.

**Transcript**

HCMOGT-1/PDGFRB chimeric RNA present.

**Fusion protein**

Description

Coiled-coil domain from HCMOGT-1 fused to the transmembrane domain and the Tyr kinase domain of PDGFRB; the reciprocal PDGFRB/HCMOGT-1 transcript is not expressed.

References


This article should be referenced as such: