

Leukaemia Section

Short Communication

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

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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

Laguange LM, Lin S, Samara RN, Salesiotis AN, Jessup JM. Nitrosative stress in rotated three-dimensional colorectal carcinoma cell cultures induces microtubule depolymerization and apoptosis. *Cancer Res.* 2004 Apr 15;64(8):2643-8

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