Leukaemia Section
Short Communication

t(11;18)(p15;q12)

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Published in Atlas Database: October 2011
Online updated version: http://AtlasGeneticsOncology.org/Anomalies/t1118p15q12ID1466.html
DOI: 10.4267/2042/47286

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Clinics and pathology

Disease
T-cell acute lymphoid leukemia (T-ALL)

Epidemiology
One case to date, a 9-year-old boy.

Evolution
Remission was obtained and the patient remains in complete remission 28 months after diagnosis.

Cytogenetics

Cytogenetics morphological
The translocation was accompanied with a del(12p).

Genes involved and proteins

NUP98
Location
11p15.4
Protein
Nucleoporin: associated with the nuclear pore complex. Role in nucleocytoplasmic transport processes.

SETBP1
Location
18q12.3
Protein
Contains 3 DNA binding domains (A.T hooks). SETBP1 protects SET from protease cleavage. SETBP1 forms a complex with SET and PP2A (protein phosphatase 2A). SETBP1 impairs PP2A activity via SET and promotes proliferation of acute myeloid leukemia cells (Cristóbal et al., 2010).

Germinal mutations
In Schinzel-Giedion midface retraction syndrome.

Result of the chromosomal anomaly

Hybrid gene
Description
5’ NUP98 - 3’ SETBP1
Transcript
Exon 12 of NUP98 (nucleotide (nt) 1552) fused in-frame with exon 5 of SETBP1 (nt 4015).

References


This article should be referenced as such: