

Gene Section

Short Communication

MKL1 (megakaryoblastic leukemia (translocation) 1)

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Identity

Other names: MAL (megakaryocytic acute leukaemia)

HGNC (Hugo): MKL1

Location: 22q13

DNA/RNA

Description

15 exons spanning 226 kb, with 3 non coding exons in 5'; 2 putative promoters.

Transcription

Transcription of 4.5 and 4 kb RNA.

Protein

Description

931 amino acids; contains in the NH₂-term a SAF (scaffold attachment factor) box; SAF box bind the DNA minor groove; supposed to attach DNA to nuclear scaffold.

Expression

Wide.

Function

May have a role in chromatin organization.

Implicated in

t(1;22)(p13;q13) / acute megakaryocytic leukaemia (M7 ANLL) --> OTT - MAL

Disease

Acute megakaryocytic leukaemia found in infants.

Prognosis

Complete remission in only 50% of cases; median survival: 8 months.

Cytogenetics

60% of cases have the t(1;22) as a single anomaly; the remaining cases exhibit complex and hyperploid clones.

Hybrid/Mutated gene

5' OTT- 3' MAL, comprising most of OTT fused to most of MAL; the reciprocal 5' MAL - 3' OTT may or may not be present.

Abnormal protein

Includes most of OTT with the RNA recognition motifs and the SPOC domain in N-term, and most of MAL, with the scaffold attachment factor box in C-term.

Oncogenesis

May play a role in chromatin organization, HOX differentiation pathways, or extracellular signalling.

References

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