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

\[ t(1;22)(p13;q13) / \text{acute megakaryocytic leukaemia (M7 ANLL)} \rightarrow OTT - MAL \]

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

This article should be referenced as such: