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Gene Section
Mini Review

ELL (eleven nineteen lysin rich leukemia gene)

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Identity

Other names: MEN (myeloid eleven nineteen translocation: WARNING: unrelated to MEN1 and MEN2); ELL-PEN

HGNC (Hugo): ELL

Location: 19p13.1

Local order: proximal from LYL1 in 19p13.2-p13.1; ENL and E2A are more distal in 19p13.3.

DNA/RNA

Transcription

Alternate splicing; 4.4 and 2.8 kb mRNA; coding sequence: 1.9 kb.

Protein

Description

621 amino acids; 68 kDa; contains a Lysin rich domain (basic motif).

Expression

Wide; especially in leukocytes, muscle, testis, placenta.

Localisation

Nuclear, except the nucleolus.

Function

RNA polymerase II elongation factor, promotes transcription by suppressing transient pausings. In Drosophila ELL is associated with active sites of transcription in vivo. Overexpression of ELL is toxic, suggesting the normal protein may play a role in the regulation of cell growth and survival.

Homology

ELL2, ELL3

Implicated in

t(11.19)(q23;p13.1) /ANLL --> MLL-ELL

Disease
Mainly M4/M5; treatment related leukemia; all ages.

Prognosis
Very poor.

Cytogenetics
Detected with R banding.

Hybrid/Mutated gene
5' MLL - 3' ELL

Abnormal protein
Similar to other MLL fusion proteins. The amino terminal AT hook and DNA methyltransferase homology regions from from MLL are fused to most of ELL.

Oncogenesis
The carboxyl terminal region of ELL is required for transformation by MLL-ELL in murine bone marrow transformation assays. This region has potent transcriptional activating activity, and interacts with EAF1, a protein that shares homology with AF4, LAF4, and AF5q31. Interestingly the EAF1 interacting domain, but not the ELL elongation domain is required for transformation. ELL has also been reported to interact withp53 and inhibit its transcriptional activating activity.

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