

Gene Section

Short Communication

SEPT6 (septin 6)

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Identity

Other names: KIAA0128

HGNC (Hugo): SEPT6

Location: Xq24



Probe(s) - Courtesy Mariano Rocchi, Resources for Molecular Cytogenetics.

DNA/RNA

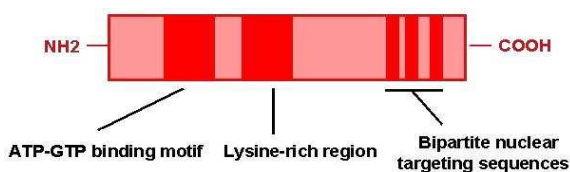
Description

12 exons.

Transcription

Four types of transcripts: 2,3 kb, 2,7 kb, 3,1 kb and 4,6 kb coding for three isoforms.

Protein



Description

Isoform A: 427 amino acids; 46,9 kDa.

Isoform B: 434 amino acids; 49,7 kDa.

Isoform D: 429 amino acids; 47,2 kDa.

Expression

Ubiquitously expressed; highest levels in placenta, lung, kidney and testes, the 2,7kb transcript can be found only in fetal heart and adult brain tissue.

Localisation

Cytoplasmatic.

Function

The conserved septin protein family was first identified in yeast and subsequently shown to play an important role in cytoskeletal organization and cytokinesis.

Implicated in

Acute myloid leukemia

Disease

AML-M2 and M4.

Cytogenetics

ins(X;11)(q24;q23)

ins(X;11)(q22-24;q23)

t(X;3)(q22;p21)

ins(X;11)(q22;q13q25).

Hybrid/Mutated gene

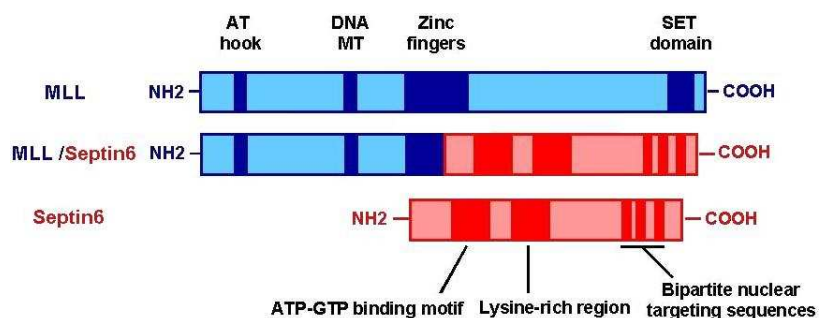
MLL-Septin 6

Abnormal protein

MLL exons 1 to 8 - Septin 6 exon 2 to end.

Oncogenesis

Basically unknown.



References

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