Gene Section
Short Communication

SEPT6 (septin 6)
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Identity
Other names: KIAA0128
HGNC (Hugo): SEPT6
Location: Xq24

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


Slater DJ, Hilgenfeldt E, Rappaport EF, Shah N, Meek RG, Williams WR, Lovett BD, Osheroff N, Autor RS, Ried T, Felix CA. MLL-SEPTIN6 fusion recurs in novel translocation of chromosomes 3, X, and 11 in infant acute myelomonocytic leukaemia and in t(X;11) in infant acute myeloid leukaemia, and MLL genomic breakpoint in complex MLL-SEPTIN6 rearrangement is a DNA topoisomerase II cleavage site. Oncogene. 2002 Jul 11;21(30):4706-14

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