CASC5 (cancer sensitivity candidate 5)
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Identity

Hugo: CASC5
Other names: AF15q14 (ALL1 fused gene from 15q14); KIAA1570; D40
Location: 15q14

DNA/RNA

Note: Whole genomic size is not determined, but consists of at least 10 exons.

Transcription

D40/CASC5 mRNA expression is dominant in normal human testis and slight expression are observed in other organs, such as placenta. At least two alternative isoforms of cDNA were identified. Northern blotting analysis on testis shows two bands with size of approximately 6 and 8.5 kb which are probably derived from the two isoforms. Analysis on cancer cell lines, such as HeLa, gave single band with 8.5 kb. There is another alternative splicing site at the 5' side of this gene that generates a short exon with 78 bp in cDNA. There are potential other alternative splicing at cancer cell lines.

Protein

Description

Encodes 1833 amino acids and 2342 amino acids.

Expression

In human testis D40/CASC5 protein expression with molecular weight of approximately 300 kDa and 250 kDa are observed in germ cell. The significant high expressions are observed in nucleus of spermatocytes and Pre-acrosome of spermatids. As D40/CASC5 protein has no hydrophobic signal peptide in its amino terminal.

Localisation

It localizes outer surface of Pre-acrosome membrane. Kinetochore proteins in C. elegans and yeast have sequence homology to D40 and it was shown that D40 is localized in kinetochore in a human cancer cell line.

Implicated in

t(11;15)(q23;q14)/acute non lymphocytic leukemia (ANLL) → MLL-CASC5

Note: It is reported that MLL gene and D40 (AF15q14) gene are translocated each other in three cases of leukemias.

Lung cancer

Note: In primary lung cancer, clinicopathological findings correlates with D40 expression. D40 mRNA expression is more frequent in the tumors with low differentiation than the ones with moderate and high differentiation. Further, the tumors derived from smoker express higher incidence of D40 mRNA than the ones from non-smoker. D40 is a member of cancer/testis gene family.

References


This article should be referenced as such: