Gene Section
Short Communication

JJAZ1 (joined to JAZF1)
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Identity
Hugo: SUZ12
Other names: KIAA0160; CHET9
Location: 17q11.2

DNA/RNA
Description
16 exons; spans 64 kb.
Transcription
4,441 kb cDNA.
Pseudogene
Yes, also located in 17q11.2, contains exons 1-9.

Protein
Description
739 amino acids.
Expression
Tissue and stage specifically expressed; expression is noted in embryonic, juvenile and adult tissues. The tissues or organs that express SUZ12 are: bladder, blood, bone, bone marrow, brain, cervix, colon, eye, heart, kidney, liver, lung, lymph node, mammary gland, muscle, ovary, pancreas, peripheral nervous system, placenta, prostate, skin, soft tissue, stomach, tongue, testis, uterus, and the vascular system.
Localisation
Nucleus.
Function
SUZ12 is a zinc finger protein and member of the polycomb group (PcG) protein family. They act by forming multiprotein complexes, which are required to maintain the transcriptionally repressive state of homeotic genes throughout development. PcG proteins are required to maintain the repression during later stages of development. They probably act via the methylation of histones, rendering chromatin heritably changed in its expressibility. SUZ12 is a component of the PRC2 complex, which methylates Lys-9 and Lys-27 residues of histone H3. SUZ12 is induced by E2F1 transcription factor.

Homology
Polycomb group of proteins.

Mutations
Germinal
Deleted in patients with Neurofibromatosis type 1 and large deletions in the NF1 gene region type-1 (spanning 1.4 Mb).
Somatic
Disrupted by deletion breakpoints of Neurofibromatosis type 1 patients with deletions that span 1.2 Mb (type-2 deletions). JJAZ1/SUZ12 has been identified at the breakpoints of a recurrent chromosomal translocation reported in endometrial stromal sarcoma and the translocation mediated recombination of both leads to a JJAZ1/JAZF1 fusion gene.

Implicated in
Endometrial stromal neoplasms with classic histology
Cytogenetics
Nonrandom t(7;17)(p15;q21) in endometrial stromal neoplasms.
Hybrid/Mutated Gene
JJAZ1/JAZF1 fusion gene.
Abnormal Protein
Unknown.
Oncogenesis
Unknown.

References


This article should be referenced as such: