JJAZ1 (joined to JAZF1)

Identity

Other names	**KIAA0160,**
	CHET9
Hugo	**SUZ12**
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

Entity	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

External links

	Nomenclature
Hugo	SUZ12
GDB	SUZ12
Entrez_Gene	SUZ12 23512 suppressor of zeste 12 homolog (Drosophila)
	Cards
Atlas	**JJAZ1ID41039ch17q11**
GeneCards	SUZ12
Ensembl	SUZ12
Genatlas	SUZ12
GeneLynx	SUZ12
eGenome	SUZ12
euGene	23512
	Genomic and cartography
GoldenPath	SUZ12 - 17q11.2 chr17:27288185-27352162 + 17q11.2 (hg18-Mar_2006)
Ensembl	SUZ12 - 17q11.2 [CytoView]
NCBI	Genes Cyto Gene Seq [Map View - NCBI]
OMIM	Disease map [OMIM]
HomoloGene	SUZ12
	Gene and transcription
Genbank	AK074333 [ENTREZ]
Genbank	BC015704 [ENTREZ]
Genbank	BC018583 [ENTREZ]
Genbank	D63881 [ENTREZ]
RefSeq	NM_015355 [SRS] NM_015355 [ENTREZ]

AceView	SUZ12 AceView - NCBI
TRASER	SUZ12 Traser - Stanford
Unigene	Hs.462732 [SRS] Hs.462732 [NCBI] HS462732 [spliceNest]
	Protein : pattern, domain, 3D structure
SwissProt	Q15022 [SRS] Q15022 [EXPASY] Q15022 [INTERPRO]
Prosite	PS00028 ZINC_FINGER_C2H2_1 [SRS] PS00028 ZINC_FINGER_C2H2_1 [Expasy]
Prosite	PS50157 ZINC_FINGER_C2H2_2 [SRS] PS50157 ZINC_FINGER_C2H2_2 [Expasy]
Interpro	IPR007087 Znf_C2H2 [SRS] IPR007087 Znf_C2H2 [EBI]
CluSTr	Q15022
Blocks	Q15022
HPRD	Q15022
	Protein Interaction databases
DIP	Q15022
IntAct	Q15022
	Polymorphism : SNP, mutations, diseases
OMIM	606245 [map]
GENECLINICS	606245
SNP	SUZ12 [dbSNP-NCBI]
SNP	NM_015355 [SNP-NCI]
SNP	SUZ12 [GeneSNPs - Utah] SUZ12] [HGBASE - SRS]
HAPMAP	SUZ12 [HAPMAP]
	General knowledge
Family Browser	SUZ12 [UCSC Family Browser]
SOURCE	NM_015355
SMD	Hs.462732
SAGE	Hs.462732
Amigo	nucleic acid binding
Amigo	intracellular
Amigo	nucleus
Amigo	transcription
Amigo	regulation of transcription, DNA-dependent
Amigo	zinc ion binding
Amigo	chromatin modification
Amigo	metal ion binding
BIOCARTA	The PRC2 Complex Sets Long-term Gene Silencing Through Modification of Histone Tails [Genes]

PubGene	SUZ12
	Other databases
	Probes
Probe	SUZ12 Related clones (RZPD - Berlin)
	PubMed
PubMed	**17 Pubmed reference(s) in LocusLink**

Bibliography

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