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Gene Section
Mini Review

JAZF1 (JAZF zinc finger 1)

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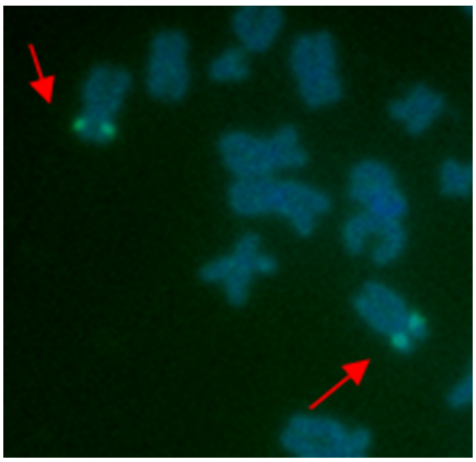
Identity

Other names: TIP27; ZNF802; DKFZp761K2222

HGNC (Hugo): JAZF1

Location: 7p15.2



Metaphase FISH using as probe YAC908B12, which encompasses the entire JAZF1 at 7p15.2.

DNA/RNA

Description

5 exons; spans 350kb.

Transcription

Major transcript: 2,980bp; coding sequence: 52-783.

Protein

Description

243 amino acids.

Expression

Expressed in all the tissues tested with variable level. The tissues or organs that express JAZF1 include cerebellum, lung, thymus, liver, kidney, stomach/esophagus, skeleton muscle, skin and eye.

Localisation

Mostly nucleus.

Function

JAZF1 has three C2H2-type zinc fingers. It is mostly detected within the nucleus, with lesser amounts found in the cytoplasm. JAZF1 copurifies with chromatin, and presumably has DNA-binding properties. It has been reported to interact with TAK1 and function as a transcriptional repressor of the TAK1 gene.
SNPs in intron 1 of JAZF1 has been reported to be associated with type 2 diabetes and body height.
SNPs in intron 2 of JAZF1 have been reported to be associated with reduced prevalence of prostate cancer.
Chimeric JAZF1-JJAZ1 protein (amino acid sequence of the first three exons of JAZF1 joined to sequence of the last 15 exons of JJAZ1) resulting from trans-splicing of precursor mRNAs and identical to a product generated from the JAZF1-JJAZ1 gene fusion in endometrial tumors has been found in normal endometrium.

Homology

Unkown.

Mutations

Somatic

JAZF1 has been identified at the breakpoints of a recurrent chromosomal translocation, the

t(7;17)(p15;q21), in endometrial stromal tumors (benign nodules and sarcomas). The translocation leads to a JAZF1-JJAZ1 fusion gene. This gene fusion is detected in about 50% of endometrial stromal sarcomas and most endometrial stromal nodules.
Another common chromosomal translocation in endometrial stroma sarcomas, the t(6;7)(p21;p15), results in a JAZF1-PHF1 fusion. About 25-30% of endometrial stromal sarcomas are reported to contain this fusion. The sites of fusion within JAZF1 RNA to JJAZ1 and PHF1 RNA sequence are the same. Both JJAZ1(also called SUZ12) and PHF1 belong to the Polycomb group (PcG) gene family.

Implicated in

t(7;17)(p15;q21) / endometrial stromal nodule and endometrial sarcoma

Disease
Endometrial stroma nodule and sarcoma.

Cytogenetics
t(7;17)(p15;q21)

Hybrid/Mutated gene
JAZF1-JJAZ1

Abnormal protein
JAZF1-JJAZ1

Oncogenesis
The fusion protein protects cells from hypoxia-induced apoptosis, and also promotes proliferation when the wild-type allele of JJAZ1 is silenced (as it is in endometrial stromal sarcomas carrying the t(7;17)(p15;q21)).

t(6;7)(p21;p15)/ endometrial stroma sarcoma

Disease
Endometrial stroma sarcoma.

Cytogenetics
t(6;7)(p21;p15)

Hybrid/Mutated gene
JAZF1-PHF1

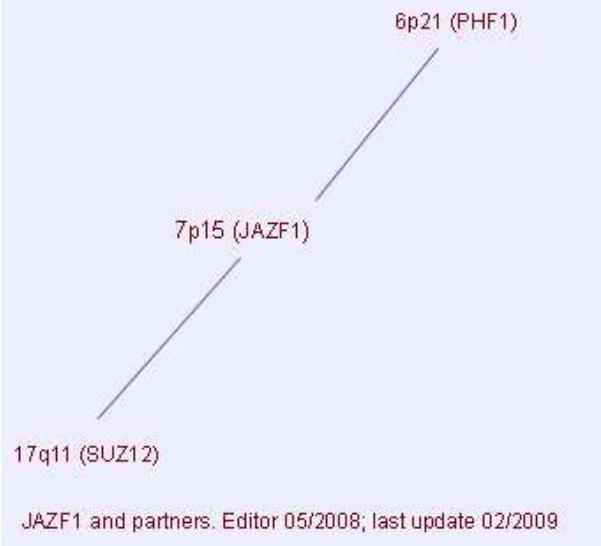
Abnormal protein
JAZF1-PHF1

Oncogenesis
The function of the JAZF1-PHF1 fusion is not currently known.

Prostate carcinoma

Oncogenesis
A SNIP in intron 2 of JAZF1 is associated with a somewhat decreased risk of prostate cancer, especially cancers that have been classified as being less aggressive. The mechanism by which polymer-phisms alter the susceptibility toward prostate cancer is not currently known.

Breakpoints



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