

## Gene Section

### Review

# SMARCB1 (SW1/SNF related, matrix associated, actin dependent regulator of chromatin B1)

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### Identity

**Other names:** hSNF5; SNF5L1; INI1 (integrase interactor 1); BAF47 (BRG1-associated factor)

**HGNC (Hugo):** SMARCB1

**Location:** 22q11.2

**Local order:** Distal to BCR.

### DNA/RNA

#### Description

Spans over 50 kb; 9 exons.

#### Transcription

Alternative splicing including/excluding exon 2.

### Protein

#### Description

385 amino acids; 47 kDa; a DNA binding domain and 2 repeat motifs.

#### Localisation

Nucleus; associated with the chromatin and with the nuclear matrix.

#### Function

Binds and activates human immunodeficiency virus integrase; member of the SWI/SNF complex, thought to facilitate the transcriptional activation of inducible genes through the remodelling of the chromatin; could be involved in the chromatin organization associated with the nuclear matrix attachment; could also have a role in the cell cycle control, through binding to P105-Rb.

### Homology

with SNF5 (yeast transcription factor)

### Mutations

#### Germinal

Found in the rhabdoid tumor predisposition syndrome.

#### Somatic

Mutation and allele loss events in sporadic rhabdoid tumors are consistent with the two-hit model of Knutson; deletion of the entire gene on one allele (sometimes due to translocations involving 22q11), and mutation (frameshift mutations, widely dispersed through the entire gene and leading to stop codons) on the other allele.

### Implicated in

#### Rhabdoid tumor

##### Disease

Tumor of uncertain origin, occurring in early childhood.

##### Prognosis

Highly aggressive; 80% mortality rate.

##### Cytogenetics

Normal karyotype or t(Var; 22)(-;q11.2).

##### Hybrid/Mutated gene

No hybrid gene but inactivation of both alleles.

#### Rhabdoid tumor predisposition syndrome

##### Hybrid/Mutated gene

Germline mutation on one allele, predisposing to a

rhabdoid tumor (and perhaps to tumors of the central nervous system) when the other allele is also inactivated.

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