

## Gene Section

### Mini Review

# WFDC1 (WAP four-disulfide core domain 1)

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### Identity

**Other names:** ps20

**HGNC (Hugo):** WFDC1

**Location:** 16q24.1-24.3

### DNA/RNA

#### Description

The gene encompasses 35 kb of DNA; 7 exons.

#### Transcription

1366 nucleotides mRNA; 660 bp open reading frame.

### Protein

#### Description

220 amino acids; 24 kDa protein. Like rat ps20, human ps20 protein contains a WAP signature domain.

### Expression

Widely expressed. Most abundant in prostate, kidney, testes, stomach, liver and spleen.

### Function

The rat homologue of ps20 was originally identified as a secreted growth inhibitor. These growth regulatory effects and the cell phenotypic properties in vitro, suggest that ps20 may function as a mediator of stromal-epithelial interactions and contribute to the maintenance of tissue homeostasis. The ps20 protein is assumed to function as a protease inhibitor. The ps20 gene, WFDC1, is thus a putative tumour suppressor gene.

### Homology

The human WFDC1 protein shares approximately 86% and 88% identity with the rat and mouse proteins, respectively. The WFDC1 gene organization presents similarities with that of the KAL gene, which extends on 210 kb of DNA, includes 14 exons and is the largest gene in the WAP signature domain family.



Representation of the position of the conserved cysteines for the category of "four-disulfide core" domain and the location of the signature pattern for such a domain in the human WFDC1 amino acid sequence.

## Mutations

### Note

Mutations in WFDC1 gene resulting in Gly9Asp, Pro211Ser and Lys217Arg substitutions have been found at low frequency mainly in the stroma of breast carcinomas. One mutation resulting in a Pro167Ser substitution has been identified in the epithelium of breast carcinoma. Although WFDC1 was mapped to human chromosome 16q24, an area of frequent loss of heterozygosity (LOH) in hepatocellular carcinoma, no mutation of the remaining allele was observed neither in liver tumors nor in hepatocellular carcinoma cell lines.

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