

Gene Section

Short Communication

PSF (PTB-associated splicing factor)

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Identity

Other names: PSF (PTB-associated splicing factor); SFPQ (splicing factor proline/glutamine rich)

HGNC (Hugo): SFPQ

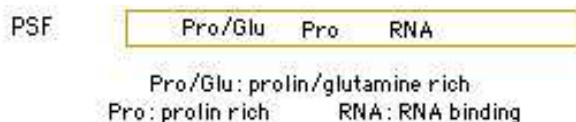
Location: 1p34

DNA/RNA

Transcription

Alternative splicing; 3 kb mRNA complete cds; coding sequence: CDS 86 ... 2209.

Protein



Description

707-712 amino acids; 76 kDa; N-term proline/glutamine rich domain, a proline rich region, 2 tandem RNA binding domains, and C-term.

Localisation

Nucleus.

Function

Splicing factor required early in spliceosome formation; form a complex with the polypyrimidine tract binding protein (PTB, herein the name); involved in pre-m RNA splicing step 2 (step 1: cut after exon n, intron n is still joined to exon n+1; step 2: cut between intron n and exon n+1, join exons n and n+1); forms a heterodimer with p54nrb (nuclear RNA binding) a protein sharing vast homologies, encoded by NONO;

both form complexes with DNA topoisomerase I, which renders this enzyme much more active.

Homology

With the above mentioned NONE product and with other proteins with a DBHS domain (Drosophila behaviour, human splicing) which includes the tandem RNA binding domains.

Implicated in

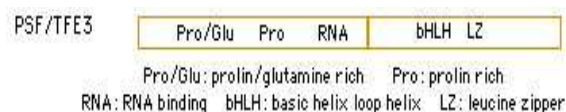
t(X;1)(p11.2;p34) in renal cell carcinoma
→ PSF/TFE3

Disease

t(X;1)(p11.2;p34) has only been found in a handful cases of papillary renal cell carcinoma

Hybrid/Mutated gene

5' PSF - 3' TFE3



Abnormal protein

N-term PSF and most of it fused to the DNA binding domains of TFE3 (excluding the acidic transcriptional activation domain, including the C-term helix-loop-helix, and the leucine zipper); no TFE3-PSF reciprocal transcript, as the der(X) t(X;1) is missing; the normal TFE3 transcript is found.

References

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