

## Gene Section

### Mini Review

# ENC1 (ectodermal-neural cortex (with BTB-like domain))

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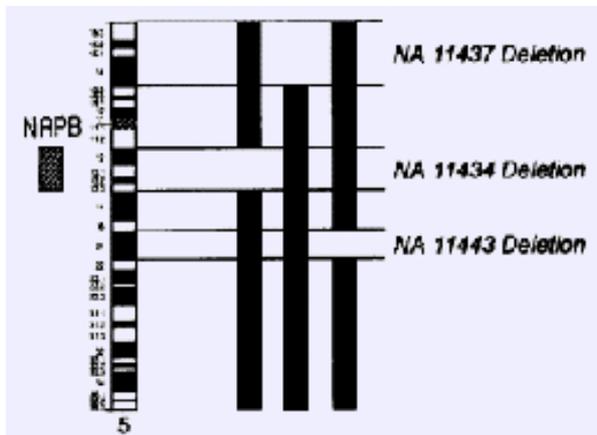
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## Identity

**Hugo:** ENC1

**Other names:** CCL28; ENC-1; NRPB; NRP/B (Nuclear Matrix Restricted Protein/Brain); PIG10; TP53I10

**Location:** 5q12-13



Mapping of human NRP/B/ENC1 to 5q12-13: Chromosome 5 content of each hybrid is shown as a solid bar. Deleted segments of Chromosome 5 are indicated to the right.

## DNA/RNA

### Description

The NRP/B/ENC1 open reading frame is 1770 bp. The murine NRP/B/ENC1 gene consists of four exons interrupted by three introns that span 7.6 kb of DNA. The complete open reading frame is located in exon 3. Transcription is 5.5 kb of RNA.

## Protein

### Description

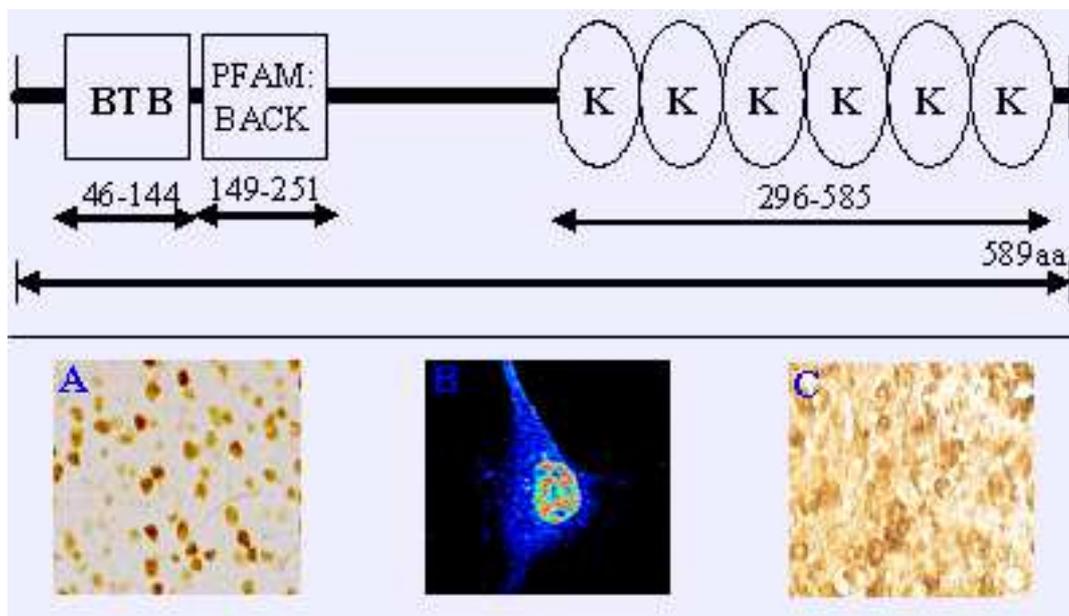
589AA and 66130 Da. NRP/B/ENC1 is a member of a growing family of proteins that contains two major structural elements: A BTB/POZ domain in the N-terminus and kelch motif in the C-terminus [See above]. The BTB/POZ domain, consisting of approximately 115 amino acids is found in several members of the kelch family. It is involved in protein-protein interactions and mediates both dimer and heterodimer formation. The kelch domain includes six repeats (each containing about 50 amino acids), which are implicated in actin binding, protein folding and/or protein-protein interactions. NRP/B/ENC1 encodes an unusually evolutionarily conserved protein between mouse and human.

### Expression

NRP/B/ENC1 is expressed in the nervous system. NRP/B/ENC1 is expressed mostly in primary neurons, but not in primary astrocytes. NRP/B/ENC1 mRNA and protein expression were detected abundantly and observed in human primary brain tumors, including glioblastoma multiformae and astrocytoma, and in human neuroblastoma cell lines (IMR32, SK-N-MC, SK-N-SH), in glioblastoma cell lines (A172, T98G, U87-MG, U118-MG, U138-MG and U373-MG), and in neuroglioma (H4) and astrocytoma cell lines (CCF-STTG1 and SW1088).

### Localisation

Nuclear (Glioblastoma, U87-MG) and Cytosolic (Glioblastoma, GBM).



Nuclear and cytoplasmic expression of NRP/B/ENC1 were demonstrated by immunostaining with specific anti-NRP/B/ENC1 antibody. Nuclear NRP/B/ENC1: (A) Human normal adult brain (frontal cortex), (B) U87-MG human glioblastoma cells. (C) Cytoplasmic NRP/B/ENC1: primary GBM cells.

**Function**

Evidence suggests that NRP/B/ENC1 is involved in multiple cell processes, including nervous system development, neuronal and adipocyte differentiation and brain tumorigenesis. NRP/B/ENC1 functions as an actin-binding protein that may be important in the organization of the actin cytoskeleton during neural fate specification and nervous system development, and plays a crucial role in the regulation of neuronal

process formation via association with the hypophosphorylated form of retinoblastoma protein (p110RB).

**Homology**

NRP/B/ENC1 shares significant homology with the 'kelch' repeats found in several kelch-related genes, including the Drosophila gene which is essential for oogenesis.

**Mutations**

| Mutations of NRP/B in brain tumor cell lines   |                 |         |
|--|-----------------|---------|
| Cell lines                                     | Mutations       | Domains |
| A-172  | V266A           | IVS     |
|  | F333L           | Kelch   |
| CCF-STTG1                                      | A535V           | Kelch   |
| SK-N-SH  | A293T           | IVS     |
| U87  | I549T           | Kelch   |
| Mutations of NRP/B in human brain tumors (GBM) |                 |         |
| Case no.                                       | Mutations       | Domains |
| 1  | L403F; N436T    | Kelch   |
|  | N216S           | IVS     |
| 2  | Q463R           | Kelch   |
|  | E96K            | BTB     |
| 91   | C531R           | Kelch   |
| C178   | C531R           | Kelch   |
| C197   | F23I            | BTB     |
| V564A  | Kelch           |         |
| C400   | H381T           | Kelch   |
| C465   | H381I           | Kelch   |
| C476   | K370 stop codon | Kelch   |

**Note:** p53-induced protein 10 (PIG10) was identified in DLD1 colon cancer cells. PIG10 contains multiple mutations and deletions, and is a variant form of NRP/B/ENC1. It has also been observed that there are a number of mutations and deletions occurred on the BTB, IVS and Kelch domains of NRP/B/ENC1 in human brain tumor cell lines and human brain tumors (GBM) (see tables below). The mutations on the Kelch domain are involved in brain tumor development

## Implicated in

### Brain tumor

**Note:** Little is known.

### Disease

Mutations of NRP/B/ENC1 were reported to be related to brain tumor development. NRP/B/ENC1 has been implicated in nervous system and neuronal differentiation, and these mutations of NRP/B/ENC1 may lead to neuropathology.

### Oncogenesis

The role of NRP/B/ENC1 in oncogenesis has been established. Evidence suggests that both amplification and alteration of NRP/B/ENC1 may lead to brain tumor development.

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