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Product datasheet

Recombinant Human NF2 / Merlin protein ab131860

1 Image

Description

Product name	Recombinant Human NF2 / Merlin protein
Expression system	Wheat germ
Accession	Q9P1N4
Protein length	Full length protein
Animal free	No
Nature	Recombinant
Species	Human
Sequence	MVVSLRALLLALRQKPHSTGTIHEVTAQLMMSVRLSFWPV AVCSCVAQMA SFILIKAPPQPQQSPQPVLSTLLFMYLPGPAR
Predicted molecular weight	35 kDa including tags
Amino acids	1 to 82

Specifications

Our Abpromise guarantee covers the use of **ab131860** in the following tested applications.

The application notes include recommended starting dilutions; optimal dilutions/concentrations should be determined by the end user.

Applications	SDS-PAGE Western blot ELISA
Form	Liquid

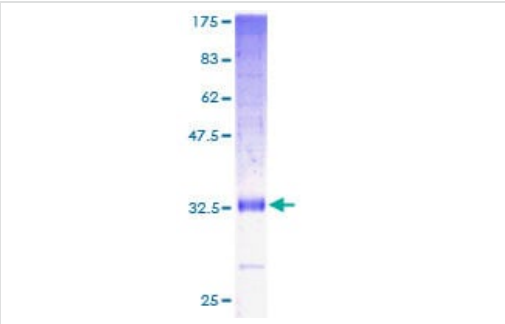
Preparation and Storage

Stability and Storage	Shipped on dry ice. Upon delivery aliquot and store at -80ºC. Avoid freeze / thaw cycles. pH: 8.00 Constituents: 0.31% Glutathione, 0.79% Tris HCl

General Info

Function	Probable regulator of the Hippo/SWH (Sav/Wts/Hpo) signaling pathway, a signaling pathway that plays a pivotal role in tumor suppression by restricting proliferation and promoting apoptosis. Along with WWC1 can synergistically induce the phosphorylation of LATS1 and LATS2 and can probably function in the regulation of the Hippo/SWH (Sav/Wts/Hpo) signaling pathway. May act as a membrane stabilizing protein. May inhibit PI3 kinase by binding to AGAP2 and impairing its stimulating activity. Suppresses cell proliferation and tumorigenesis by inhibiting the CUL4A-RBX1-DDB1-VprBP/DCAF1 E3 ubiquitin-protein ligase complex.
Tissue specificity	Widely expressed. Isoform 1 and isoform 3 are predominant. Isoform 4, isoform 5 and isoform 6 are expressed moderately. Isoform 8 is found at low frequency. Isoform 7, isoform 9 and isoform 10 are not expressed in adult tissues, with the exception of adult retina expressing isoform 10. Isoform 9 is faintly expressed in fetal brain, heart, lung, skeletal muscle and spleen. Fetal thymus expresses isoforms 1, 7, 9 and 10 at similar levels.
Involvement in disease	Defects in NF2 are the cause of neurofibromatosis 2 (NF2) [MIM:101000]; also known as central neurofibromatosis. NF2 is a genetic disorder characterized by bilateral vestibular schwannomas (formerly called acoustic neuromas), schwannomas of other cranial and peripheral nerves, meningiomas, and ependymomas. It is inherited in an autosomal dominant fashion with full penetrance. Affected individuals generally develop symptoms of eighth-nerve dysfunction in early adulthood, including deafness and balance disorder. Although the tumors of NF2 are histologically benign, their anatomic location makes management difficult, and patients suffer great morbidity and mortality. Defects in NF2 are a cause of schwannomatosis (SCHWA) [MIM:162091]; also known as congenital cutaneous neurilemmomatosis. Schwannomas are benign tumors of the peripheral nerve sheath that usually occur singly in otherwise normal individuals. Multiple schwannomas in the same individual suggest an underlying tumor-predisposition syndrome. The most common such syndrome is NF2. The hallmark of NF2 is the development of bilateral vestibular-nerve schwannomas; but two-thirds or more of all NF2-affected individuals develop schwannomas in other locations, and dermal schwannomas may precede vestibular tumors in NF2-affected children. There have been several reports of individuals with multiple schwannomas who do not show evidence of vestibular schwannoma. Clinical report suggests that schwannomatosis is a clinical entity distinct from other forms of neurofibromatosis.
Sequence similarities	Contains 1 FERM domain.
Post-translational modifications	Phosphorylation of Ser-518 inhibits nuclear localization by disrupting the intramolecular association of the FERM domain with the C-terminal tail. Ubiquitinated by the CUL4A-RBX1-DDB1-DCAF1/VprBP E3 ubiquitin-protein ligase complex for ubiquitination and subsequent proteasome-dependent degradation.
Cellular localization	Cytoplasm > perinuclear region. Cytoplasmic granule. Observed in cytoplasmic granules concentrated in a perinuclear location. Isoform 7 is absent from ruffling membranes and filopodia; Cytoplasm > perinuclear region. Cytoplasmic granule. Observed in cytoplasmic granules concentrated in a perinuclear location. Isoform 9 is absent from ruffling membranes and filopodia; Nucleus. Cell projection > filopodium membrane. Cell projection > ruffle membrane. Cytoplasm > perinuclear region. Cytoplasmic granule. Cytoplasm > cytoskeleton. In a fibroblastic cell line, isoform 10 is found homogeneously distributed over the entire cell, with a particularly strong staining in ruffling membranes and filopodia and Cell projection > filopodium membrane. Cell projection > ruffle membrane. Nucleus. In a fibroblastic cell line, isoform 1 is found homogeneously distributed over the entire cell, with a particularly strong staining in ruffling membranes and filopodia. Colocalizes with MPP1 in non-myelin-forming Schwann cells. Binds with VPRBP in the nucleus. The intramolecular association of the FERM domain with the C-terminal tail promotes nuclear accumulation. The unphosphorylated form accumulates predominantly in the nucleus while the phosphorylated form is largely confined to the non-nuclear fractions.

Images



SDS-PAGE - Recombinant Human NF2 / Merlin protein (ab131860)

12.5% SDS-PAGE analysis of ab131860 stained with Coomassie Blue.

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