巨轴索神经病蛋白 GAN 抗体

产品货号： mlR11025

英文名称： Gigaxonin

中文名称： 巨轴索神经病蛋白 GAN 抗体

别　　名： FLJ38059; GAN; GAN1; Kelch-like protein 16; giant axonal neuropathy; KLHL16; GAN_HUMAN.

研究领域： 神经生物学　信号转导　细胞粘附分子　细胞骨架

抗体来源： Rabbit

克隆类型： Polyclonal

交叉反应： Human, Mouse, Rat, Pig, Cow, Horse, Rabbit, Sheep,

产品应用： ELISA=1:500-1000 IHC-P=1:400-800 IHC-F=1:400-800 ICC=1:100-500 IF=1:100-500 （石蜡切片需做抗原修复）

not yet tested in other applications.

optimal dilutions/concentrations should be determined by the end user.

分 子 量： 68kDa

细胞定位： 细胞浆

性　　状： **L**yophilized or Liquid

浓　　度： 1mg/ml

免 疫 原： KLH conjugated synthetic peptide derived from human Gigaxonin:351-450/597

亚　　型： IgG

纯化方法： affinity purified by Protein A

储 存 液： 0.01M TBS(pH7.4) with 1% BSA, 0.03% Proclin300 and 50% Glycerol.

保存条件： Store at -20 °C for one year. Avoid repeated freeze/thaw cycles. The lyophilized antibody is stable at room temperature for at least one month and for greater than a year when kept at -20°C. When reconstituted in sterile pH 7.4 0.01M PBS or diluent of antibody the antibody is stable for at least two weeks at 2-4 °C.

PubMed： PubMed

产品介绍： Gigaxonin, also refered to as giant axonal neuropathy, GAN1, or KLHL16, controls protein degradation and is essential for neuronal function and survival. Gigaxonin is a member of the cytoskeletal BTB/kelch repeat family and influences cytoskeletal organization and dynamics, playing a large role in neurofilament architecture. The amino terminal BTB domain of gigaxonin binds to the ubiquitin-activating enzyme E1, while the carboxy-terminal kelch repeat domain interacts directly with the light chain of microtubule-associated protein 1B (MAP1B), and tags it for degredation. Overexpression of MAP1B may lead to neuronal cell death, whereas a reduction of MAP1B significantly improves the survival rate of neurons. Mutations in the Gigaxonin gene result in human giant axonal neuropathy (GAN), an autosomal recessive neurodegenerative disorder characterized by axonal degeneration caused by cytoskeletal abnormalities, including accumulated intermediate filaments.

Function:

Mutations in gigaxonin result in a sensory and motor neuropathy called Giant Axonal Neuropathy (GAN). Giant axonal neuropathy, a severe autosomal recessive sensorineural neuropathy affecting both the peripheral nerves and the central nervous system, is characterized by neurofilament accumulation, leading to segmental distention of axons. Gigaxonin is a member of the cytoskeletal BTB/kelch (Broad-Complex, Tramtrack and Bric a brac) repeat family. Gigaxonin contains an N-terminal BTB domain followed by 6 kelch repeats, which were predicted to adopt a beta-propeller shape. Gigaxonin controls protein degradation and is essential for neuronal function and survival. Substrate-specific adapter of an E3 ubiquitin-protein ligase complex which mediates the ubiquitination and subsequent proteasomal degradation of target proteins. Controls degradation of TBCB. Controls degradation of MAP1B and MAP1S, and is critical for neuronal maintenance and survival

Subunit:

Interacts with TBCB. Interacts with CUL3. Part of a complex that contains CUL3, RBX1 and GAN. Interacts (via BTB domain) with UBA1. Interacts (via Kelch domains) with MAP1B (via C-terminus) and MAP1S (via C-terminus).

Subcellular Location:

Cytoplasmic; Cytoskeleton.

Tissue Specificity:

Expressed in brain, heart and muscle.

Post-translational modifications:

Ubiquitinated by E3 ubiquitin ligase complex formed by CUL3 and RBX1 and probably targeted for proteasome-independent degradation.

DISEASE:

Defects in GAN are the cause of giant axonal neuropathy (GAN) [MIM:256850]. GAN is a severe autosomal recessive sensorimotor neuropathy affecting both the peripheral nerves and the central nervous system. It is characterized by neurofilament accumulation, leading to segmental distention of axons.

Similarity:

Contains 1 BACK (BTB/Kelch associated) domain.

Contains 1 BTB (POZ) domain.

Contains 6 Kelch repeats.

SWISS:

Q9H2C0

Gene ID:

8139

Important Note:

This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.

产品图片