神经细胞蜡样质脂褐质沉积病蛋白 CLN8 抗体

产品货号： mlR11715

英文名称： CLN8

中文名称： 神经细胞蜡样质脂褐质沉积病蛋白 CLN8 抗体

别　　名： Ceroid-lipofuscinosis, neuronal 8 (epilepsy, progressive with mental retardation); Cln8; CLN8_HUMAN; EPMR; Protein CLN8.

研究领域： 肿瘤　细胞生物　神经生物学　信号转导

抗体来源： Rabbit

克隆类型： Polyclonal

交叉反应： Human, Mouse, Rat, Dog, Pig, Horse, Rabbit,

产品应用： WB=1:500-2000 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： 33kDa

细胞定位： 细胞浆 细胞膜

性　　状： Lyophilized or Liquid

浓　　度： 1mg/ml

免 疫 原： KLH conjugated synthetic peptide derived from human CLN8:201-286/286

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： CLN8, a 286 amino acid transmembrane protein, localizes mainly to the endoplasmic reticulum, but also partially to the ER-Golgi intermediate compartment (ERGIC). Mutations in the CLN8 gene cause neuronal ceroid lipofuscinosis 8 and progressive epilepsy with mental retardation (EPMR). Both disorders are forms of neuronal ceroid-lipofuscinose (NCL), a group of progressive neurodegenerative diseases found in children, characterized by failure of psychomotor development, impaired vision, seizures and premature death. The CLN8 protein is one of eight proteins in the CLN family, including CLN1-CLN7, which are associated with NCL.

Function:

Could play a role in cell proliferation during neuronal differentiation and in protection against cell death.

Subcellular Location:

Endoplasmic reticulum membrane. Endoplasmic reticulum-Golgi intermediate compartment membrane.

Post-translational modifications:

Does not seem to be N-glycosylated.

DISEASE:

Defects in CLN8 are the cause of neuronal ceroid lipofuscinosis type 8 (CLN8) [MIM:600143]. A form of neuronal ceroid lipofuscinosis with onset in childhood. Neuronal ceroid lipofuscinoses are progressive neurodegenerative, lysosomal storage diseases characterized by intracellular accumulation of autofluorescent liposomal material, and clinically by seizures, dementia, visual loss, and/or cerebral atrophy. The lipopigment patterns observed most often in neuronal ceroid lipofuscinosis type 8 comprise mixed combinations of granular, curvilinear, and fingerprint profiles.

Defects in CLN8 are the cause of neuronal ceroid lipofuscinosis type 8 Northern epilepsy variant (CLN8NE) [MIM:610003]. A form of neuronal ceroid lipofuscinosis clinically characterized by epilepsy that presents between 5 and 10 years of age with frequent tonic-clonic seizures followed by progressive mental retardation. Visual loss is not a prominent feature. Intracellular accumulation of autofluorescent material results in curvilinear and granular profiles on ultrastructural analysis.

Similarity:

Contains 1 TLC (TRAM/LAG1/CLN8) domain.

SWISS:

Q9UBY8

Gene ID:

2055

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

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