半乳糖神经酰胺酶抗体

产品货号： mlR20477

英文名称： Galactosylceramidase

中文名称： 半乳糖神经酰胺酶抗体

别　　名： Gacy; Galactocerebrosidase; Galactocerebroside beta galactosidase; Galactosylceramide beta galactosidase; galactosylceraminidase; Galc; GALCERase; Twitcher; GALC_HUMAN.

研究领域： 肿瘤　细胞生物　神经生物学　信号转导　新陈代谢

抗体来源： Rabbit

克隆类型： Polyclonal

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

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： 73kDa

细胞定位： 细胞浆

性　　状： Lyophilized or Liquid

浓　　度： 1mg/ml

免 疫 原： KLH conjugated synthetic peptide derived from human Galactosylceramidase:211-320/685

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： GALC is a lysosomal enzyme that hydrolyzes galactose ester bonds in various galactolipids, including galactosylceramide, galactosylsphingosine, lactosylceramide and monogalactosyldiglyceride. Galactolipids contain glucose and/or galactose, and are found in the brain and other nerve tissue, especially the myelin sheath. Galactosylceramide is a major lipid in myelin, kidney, and epithelial cells of the small intestine and colon. Mutations in the GALC gene that compromise protein function correlate to Krabbe disease (globoid cell leukodystrophy, GLD). GLD is an autosomal recessive condition that affects approximately 1 in 150,000 infants and results in progressive destruction of the nervous system. The “twitcher” mouse is a model system for GLD; the genotype is a premature stop codon (W339X) in the galactosylceramidase (GALC) gene that abolishes enzymatic activity. Alternate transcriptional splice variants, encoding different isoforms, have been characterized.

Function:

Galactosylceramidase hydrolyzes the galactose ester bonds of galactosylceramide, galactosylsphingosine, lactosylceramide, and monogalactosyldiglyceride. It is an enzyme with very low activity responsible for the lysosomal catabolism of galactosylceramide, a major lipid in myelin, kidney and epithelial cells of small intestine and colon. It shows highest level of activity in testes compared to brain, kidney, placenta and liver. It can also be found in urine. Defects in Galactosylceramidase are the cause of globoid cell leukodystrophy (GLD); also known as Krabbe disease. This autosomal recessive disorder results in the insufficient catabolism of several galactolipids that are important in the production of normal myelin. Clinically, the most frequent form is the infantile form. Most patients (90%) present before six months of age with irritability, spasticity, arrest of motor and mental development, and bouts of temperature elevation without infection. This is followed by myoclonic jerks of arms and legs, oposthotonus, hypertonic fits, and mental regression, which progresses to a severe decerebrate condition with no voluntary movements and death from respiratory infections or cerebral hyperpyrexia before 2 years of age. However, a significant number of cases with later onset, presenting with unexplained blindness, weakness and/or progressive motor, and sensory neuropathy that can progress to severe mental incapacity and death, have been identified.

Subcellular Location:

Lysosomal.

Tissue Specificity:

Highest level of activity in testes compared to brain, kidney, placenta and liver. Can also be found in urine.

DISEASE:

Defects in GALC are the cause of leukodystrophy globoid cell (GLD) [MIM:245200]; also known as Krabbe disease. This autosomal recessive disorder results in the insufficient catabolism of several galactolipids that are important in the production of normal myelin. Clinically, the most frequent form is the infantile form. Most patients (90%) present before six months of age with irritability, spasticity, arrest of motor and mental development, and bouts of temperature elevation without infection. This is followed by myoclonic jerks of arms and legs, oposthotonus, hypertonic fits, and mental regression, which progresses to a severe decerebrate condition with no voluntary movements and death from respiratory infections or cerebral hyperpyrexia before 2 years of age. However, a significant number of cases with later onset, presenting with unexplained blindness, weakness and/or progressive motor, and sensory neuropathy that can progress to severe mental incapacity and death, have been identified.

Similarity:

Belongs to the glycosyl hydrolase 59 family.

SWISS:

P54803

Gene ID:

2581

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