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Galactosylceramidase Rabbit pAb

Catalog # AP99633

Product Information

Application	IHC-P, IHC-F, IF
Primary Accession	P54803
Other Accession	P54803
Reactivity	Rat, Human, Mouse
Predicted	Pig, Rabbit, Chicken, Dog, Horse, Sheep
Host	Rabbit
Clonality	Polyclonal
Calculated MW	77063
Physical State	Liquid
Immunogen	KLH conjugated synthetic peptide derived from human G protein-regulated inducer of neurite outgrowth 2
Epitope Specificity	81-180/685
Isotype	IgG
Purity	affinity purified by Protein A
Buffer	0.01M TBS (pH7.4) with 1% BSA, 0.02% Proclin300 and 50% Glycerol.
SIMILARITY	Belongs to the glycosyl hydrolase 59 family.
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.
Important Note	This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.
Background Descriptions	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.

Additional Information

Gene ID	2581
Other Names	2310068B06Rik; A930008M05Rik; Gacy; twi; twitcher; GALC_CANLF; GALC; GALCERase; Galactocerebroside beta-galactosidase; Galactosylceramidase; Galactosylceramide beta-galactosidase; 3.2.1.46; GALC_HUMAN; GALC_MOUSE; galactosylceramidase (Krabbe disease); Krabbe disease
Dilution	IHC-P=1:100-500,IHC-F=1:100-500,IF=1:100-500
Storage	Store at -20 °C for one year. Avoid repeated freeze/thaw cycles. 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.

Protein Information

Name	GALC (HGNC:4115)
Function	Hydrolyzes the galactose ester bonds of glycolipids such as galactosylceramide and galactosylsphingosine (PubMed:8281145, PubMed:8399327). 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 (PubMed:8281145, PubMed:8399327).
Cellular Location	Lysosome.
Tissue Location	Detected in urine. Detected in testis, brain and placenta (at protein level). Detected in kidney and liver

Please note: All products are 'FOR RESEARCH USE ONLY. NOT FOR USE IN DIAGNOSTIC OR THERAPEUTIC PROCEDURES'.