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P5CS Rabbit pAb

Catalog # AP98939

Product Information

Application	IHC-P, IHC-F, IF
Primary Accession	P54886
Other Accession	P54886
Reactivity	Mouse
Predicted	Pig, Human, Dog, Horse, Sheep
Host	Rabbit
Clonality	Polyclonal
Calculated MW	87302
Physical State	Liquid
Immunogen	KLH conjugated synthetic peptide derived from human P5CS
Epitope Specificity	721-820/795
Isotype	IgG
Purity	affinity purified by Protein A
Buffer	0.01M TBS (pH7.4) with 1% BSA, 0.02% Proclin300 and 50% Glycerol.
SIMILARITY	In the N-terminal section; belongs to the glutamate 5-kinase family. In the C-terminal section; belongs to the gamma-glutamyl phosphate reductase family.
DISEASE	Defects in ALDH18A1 are the cause of mental retardation-joint hypermobility-skin laxity with or without metabolic abnormalities (MRJHSL) [MIM:612652]. Clinical manifestations include microcephaly, progressive neurologic dysfunction, mental retardation, progeroid appearance, joint hypermobility, skin laxity and hyperelasticity, cataracts. Some patients manifest metabolic disturbances such as hyperammonemia, hypoornithinemia, hypocitrullinemia, hypoargininemia and hypoprolinemia.
Important Note	This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.
Background Descriptions	This gene is a member of the aldehyde dehydrogenase family and encodes a bifunctional ATP- and NADPH-dependent mitochondrial enzyme with both gamma-glutamyl kinase and gamma-glutamyl phosphate reductase activities. The encoded protein catalyzes the reduction of glutamate to delta1-pyrroline-5-carboxylate, a critical step in the de novo biosynthesis of proline, ornithine and arginine. Mutations in this gene lead to hyperammonemia, hypoornithinemia, hypocitrullinemia, hypoargininemia and hypoprolinemia and may be associated with neurodegeneration, cataracts and connective tissue diseases. Alternatively spliced transcript variants, encoding different isoforms, have been described for this gene. [provided by RefSeq, Jul 2008]

Additional Information

Gene ID	5832
Other Names	ADCL3; ARCL3A; GSAS; P5CS; PYCS; SPG9; SPG9A; SPG9B; 2810433K04Rik;

	P5CS_HUMAN; ALDH18A1; Aldehyde dehydrogenase family 18 member A1; P5CS_MOUSE; aldehyde dehydrogenase 18 family member A1; pyrroline-5-carboxylate synthetase (glutamate gamma-semialdehyde synthetase); spastic paraplegia 9 (autosomal dominant); delta-1-pyrroline-5-carboxylate synthase
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	ALDH18A1
Synonyms	GSAS, P5CS, PYCS
Function	Bifunctional enzyme that converts glutamate to glutamate 5- semialdehyde, an intermediate in the biosynthesis of proline, ornithine and arginine.
Cellular Location	Mitochondrion. Mitochondrion matrix. Note=Exhibits puncta-like structures inside mitochondria under nutrient stress (PubMed:32770108). When cellular dependence on oxidative phosphorylation increases, forms filaments, which may increase its activity, and is sequestered in a subset of atypical mitochondria that lack cristae and ATP synthase (PubMed:39506109).

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