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HADHSC Rabbit pAb

Catalog # AP99987

Product Information

Application	WB, IHC-P, IHC-F, IF
Primary Accession	Q16836
Other Accession	Q16836
Reactivity	Rat, Mouse
Predicted	Pig, Human
Host	Rabbit
Clonality	Polyclonal
Calculated MW	34294
Physical State	Liquid
Immunogen	KLH conjugated synthetic peptide derived from human HADHSC
Epitope Specificity	241-314/314
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 3-hydroxyacyl-CoA dehydrogenase family.
SUBUNIT	Homodimer.
DISEASE	Defects in HADH are the cause of 3-alpha-hydroxyacyl-CoA dehydrogenase deficiency (HADH deficiency) [MIM:231530]. HADH deficiency is a metabolic disorder with various clinical presentations including hypoglycemia, hepatoencephalopathy, myopathy or cardiomyopathy, and in some cases sudden death. Defects in HADH are the cause of familial hyperinsulinemic hypoglycemia type 4 (HHF4) [MIM:609975]; also known as persistent hyperinsulinemic hypoglycemia of infancy (PHHI) or congenital hyperinsulinism. HHF is the most common cause of persistent hypoglycemia in infancy and is due to defective negative feedback regulation of insulin secretion by low glucose levels. It causes nesidioblastosis, a diffuse abnormality of the pancreas in which there is extensive, often disorganized formation of new islets. Unless early and aggressive intervention is undertaken, brain damage from recurrent episodes of hypoglycemia may occur. HHF4 should be easily recognizable by analysis of acylcarnitine species and that this disorder responds well to treatment with diazoxide. It provides the first 'experiment of nature' that links impaired fatty acid oxidation to hyperinsulinism and that provides support for the concept that a lipid signaling pathway is implicated in the control of insulin secretion.
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 3-hydroxyacyl-CoA dehydrogenase gene family. The encoded protein functions in the mitochondrial matrix to catalyze the oxidation of straight-chain3-hydroxyacyl-CoAs as part of the beta-oxidation pathway. Its enzymatic activity is highest with medium-chain-length fatty acids. Mutations in this gene cause one form of familial hyperinsulinemic hypoglycemia. The human genome contains a related pseudogene of this gene on chromosome 15. [provided by RefSeq.]

Additional Information

Gene ID	3033
Other Names	HAD; HADH1; HADHSC; HCDH; HHF4; MSCHAD; SCHAD; HCDH_HUMAN; HADH; Medium and short-chain L-3-hydroxyacyl-coenzyme A dehydrogenase; Short-chain 3-hydroxyacyl-CoA dehydrogenase; 1.1.1.35; HAD1; HCDH_MOUSE; HCDH_RAT; hydroxyacyl-CoA dehydrogenase; L-3-hydroxyacyl-Coenzyme A dehydrogenase, short chain; hydroxyacyl-Coenzyme A dehydrogenase
Dilution	WB=1:500-2000,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	HADH
Function	Mitochondrial fatty acid beta-oxidation enzyme that catalyzes the third step of the beta-oxidation cycle for medium and short-chain 3-hydroxy fatty acyl-CoAs (C4 to C10) (PubMed:10231530, PubMed:11489939, PubMed:16725361). Plays a role in the control of insulin secretion by inhibiting the activation of glutamate dehydrogenase 1 (GLUD1), an enzyme that has an important role in regulating amino acid-induced insulin secretion (By similarity). Plays a role in the maintenance of normal spermatogenesis through the reduction of fatty acid accumulation in the testes (By similarity).
Cellular Location	Mitochondrion matrix
Tissue Location	Expressed in liver, kidney, pancreas, heart and skeletal muscle.

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