

Rabbit Anti-SPTLC1 antibody

SL4087R

Product Name:	SPTLC1
Chinese Name:	丝氨酸棕榈酰转移酶1抗体
Alias:	HSAN; HSAN1; HSN1; LBC1; LCB 1; LCB1; Long chain base biosynthesis protein 1; Serine C palmitoyltransferase; Serine palmitoyl CoA transferase 1; Serine palmitoyltransferase 1; Serine palmitoyltransferase long chain base subunit 1; Serine palmitoyltransferase subunit 1; SPT 1; SPT1; SPTLC 1; SPTC1 HUMAN.
Organism Species:	Rabbit
Clonality:	Polyclonal
React Species:	Human, Mouse, Rat, Dog, Pig, Cow,
Applications:	WB=1:500-2000ELISA=1:500-1000IHC-P=1:400-800IHC-F=1:400-800IF=1:100- 500 (Paraffin sections need antigen repair) not yet tested in other applications. optimal dilutions/concentrations should be determined by the end user.
Molecular weight:	53kDa
Cellular localization:	cytoplasmicThe cell membrane
Form:	Lyophilized or Liquid
Concentration:	1mg/ml
immunogen:	KLH conjugated synthetic peptide derived from human SPTLC1:121-220/473
Lsotype:	IgG
Purification:	affinity purified by Protein A
Storage Buffer:	0.01M TBS(pH7.4) with 1% BSA, 0.03% Proclin300 and 50% Glycerol.
Storage:	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
Product Detail:	This gene encodes a member of the class-II pyridoxal-phosphate-dependent aminotransferase family. The encoded protein is the long chain base subunit 1 of serine palmitoyltransferase. Serine palmitoyltransferase converts L-serine and palmitoyl-CoA to 3-oxosphinganine with pyridoxal 5'-phosphate and is the key enzyme in sphingolipid

biosynthesis. Mutations in this gene were identified in patients with hereditary sensory neuropathy type 1. Alternatively spliced variants encoding different isoforms have been identified. Pseudogenes of this gene have been defined on chromosomes 1, 6, 10, and 13. [provided by RefSeq, Jul 2013]

Function:

Serine palmitoyltransferase (SPT). The heterodimer formed with SPTLC2 or SPTLC3 constitutes the catalytic core. The composition of the serine palmitoyltransferase (SPT) complex determines the substrate preference. The SPTLC1-SPTLC2-SPTSSA complex shows a strong preference for C16-CoA substrate, while the SPTLC1-SPTLC3-SPTSSA isozyme uses both C14-CoA and C16-CoA as substrates, with a slight preference for C14-CoA. The SPTLC1-SPTLC2-SPTSSB complex shows a strong preference for C18-CoA substrate, while the SPTLC1-SPTLC3-SPTSSB isozyme displays an ability to use a broader range of acyl-CoAs, without apparent preference.

Subunit:

Heterodimer with SPTLC2 or SPTLC3. Component of the serine palmitoyltransferase (SPT) complex, composed of SPTLC1, either SPTLC2 or SPTLC3, and either SSSPTA or SSSPTB. Interacts with SPTSSA and SPTSSB; the interaction is direct. Interacts with ORMDL3.

Subcellular Location:

Endoplasmic reticulum membrane; Single-pass membrane protein

Tissue Specificity:

Widely expressed. Not detected in small intestine.

DISEASE:

Defects in SPTLC1 are the cause of hereditary sensory and autonomic neuropathy type 1A (HSAN1A) [MIM:162400]. The hereditary sensory and autonomic neuropathies are a genetically and clinically heterogeneous group of disorders characterized by degeneration of dorsal root and autonomic ganglion cells, and by sensory and/or autonomic abnormalities. HSAN1A is an autosomal dominant axonal neuropathy with onset in the second or third decades. Initial symptoms are loss of pain, touch, heat, and cold sensation over the feet, followed by distal muscle wasting and weakness. Loss of pain sensation leads to chronic skin ulcers and distal amputations.

Similarity:

Belongs to the class-II pyridoxal-phosphate-dependent aminotransferase family.

SWISS:

O15269

Gene ID: 10558

Database links:

Entrez Gene: 426145 Chicken

Entrez Gene: 739412 Chimpanzee

Entrez Gene: 614165 Cow

Entrez Gene: 100726468 Guinea pig

Entrez Gene: 10558 Human

Entrez Gene: 268656 Mouse

Entrez Gene: 100344536 Rabbit

Entrez Gene: 705324 Rhesus monkey

<u>Omim: 605712</u> Human

SwissProt: Q3MHG1 Cow

SwissProt: Q60HD1 Cynomolgus Monkey

SwissProt: O15269 Human

SwissProt: O35704 Mouse

Unigene: 90458 Human

Unigene: 240336 Mouse

Important Note:

This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.

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