

Product name:	SPTLC1 Rabbit Polyclonal Antibody
Cat number:	ABN18230
Conjugate:	Unconjugated
Size:	100µL
Clone:	Polyclonal
Concentration:	1mg/ml
Host:	Rabbit
Isotype:	IgG
Immunogen:	Synthesized peptide derived from SPTLC1 . at AA range: 411-460
Reactivity:	Human,Mouse,Rat
Applications:	WB 1:500-1:2000,IHC 1:100-1:300,ICC/IF 1:50-1:200,ELISA 1:10000-1:20000
Molecular Weight:	52kDa
Purification:	Affinity purification
Form:	Liquid
Buffer:	Liquid in PBS containing 50% glycerol, 0.5% BSA and 0.02% New type preservative N.
Storage:	Store at 4°C short term. Aliquot and store at -20°C for 12 months. Avoid freeze/thaw cycles.

Background:

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], catalytic activity: Palmitoyl-CoA + L-serine = CoA + 3-dehydro-D-sphinganine + CO(2), cofactor: Pyridoxal phosphate, disease: Defects in SPTLC1 are the cause of hereditary sensory and autonomic neuropathy type 1 (HSAN1) [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. HSAN1 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., pathway: Lipid metabolism; sphingolipid metabolism., similarity: Belongs to the class-II pyridoxal-phosphate-dependent aminotransferase family., subunit: SPTLC1, SPTLC2 and SPTLC3 may encode subunits of the enzyme., tissue specificity: Widely expressed. Not detected in small intestine.,