

Product name:	FA2H Rabbit Polyclonal Antibody
Cat number:	ABN10744
Conjugate:	Unconjugated
Size:	100µL
Clone:	Polyclonal
Concentration:	1mg/ml
Host:	Rabbit
Isotype:	IgG
Immunogen:	Synthesized peptide derived from human FA2H. at AA range: 101-150
Reactivity:	Human,Mouse,Rat
Applications:	WB 1:500-1:2000,ELISA 1:10000-1:20000
Molecular Weight:	55kDa
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 protein that catalyzes the synthesis of 2-hydroxysphingolipids, a subset of sphingolipids that contain 2-hydroxy fatty acids. Sphingolipids play roles in many cellular processes and their structural diversity arises from modification of the hydrophobic ceramide moiety, such as by 2-hydroxylation of the N-acyl chain, and the existence of many different head groups. Mutations in this gene have been associated with leukodystrophy dysmyelinating with spastic paraparesis with or without dystonia.[provided by RefSeq, Mar 2010],cofactor:Iron.,disease:Defects in FA2H are the cause of leukodystrophy dysmyelinating with spastic paraparesis with or without dystonia (DLDSP) [MIM:612443]. The disorder consists of a progressive neurologic disease manifested by spasticity, disordered tonicity of muscle, and white matter degeneration.,domain:The histidine box domains may contain the active site and/or be involved in metal ion binding.,function:Required for alpha-hydroxylation of free fatty acids and the formation of alpha-hydroxylated sphingolipids.,induction:Up-regulated during keratinocyte differentiation.,similarity:Belongs to the SCS7 family.,similarity:Contains 1 cytochrome b5 heme-binding domain.,tissue specificity:Detected in differentiating cultured keratinocytes (at protein level). Detected in epidermis and cultured keratinocytes. Highly expressed in brain and colon. Detected at lower levels in testis, prostate, pancreas and kidney.,