

Product name:	ABCD1 Rabbit Polyclonal Antibody
Cat number:	ABN06415
Conjugate:	Unconjugated
Size:	100µL
Clone:	Polyclonal
Concentration:	1mg/ml
Host:	Rabbit
Isotype:	IgG
Immunogen:	The antiserum was produced against synthesized peptide derived from human ABCD1. AA range:531-580
Reactivity:	Human,Rat,Mouse
Applications:	WB 1:500-1:2000,ELISA 1:20000-1:40000
Molecular Weight:	75kDa
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:

The protein encoded by this gene is a member of the superfamily of ATP-binding cassette (ABC) transporters. ABC proteins transport various molecules across extra- and intra-cellular membranes. ABC genes are divided into seven distinct subfamilies (ABC1, MDR/TAP, MRP, ALD, OABP, GCN20, White). This protein is a member of the ALD subfamily, which is involved in peroxisomal import of fatty acids and/or fatty acyl-CoAs in the organelle. All known peroxisomal ABC transporters are half transporters which require a partner half transporter molecule to form a functional homodimeric or heterodimeric transporter. This peroxisomal membrane protein is likely involved in the peroxisomal transport or catabolism of very long chain fatty acids. Defects in this gene have been identified as the underlying cause of adrenoleukodystrophy, an X-chromosome recessively inherited demyelinating disorder. Defects in ABCD1 are the cause of adrenoleukodystrophy X-linked (X-ALD) [MIM:300100]. X-ALD is a peroxisomal metabolic disorder characterized by progressive multifocal demyelination of the central nervous system and by peripheral adrenal insufficiency (Addison disease). It results in mental deterioration, corticospinal tract dysfunction, and cortical blindness. Different clinical manifestations exist like: cerebral childhood ALD (CALD), adult cerebral ALD (ACALD), adrenomyeloneuropathy (AMN) and "Addison disease only" (ADO) phenotype. Microdeletions in ABCD1 are involved in the contiguous ABCD1/DXS1375E deletion syndrome (CADD5) [MIM:300475]. Patients manifest profound neonatal hypotonia, subsequent failure to thrive, and cholestatic liver disease. Probable transporter. The nucleotide-binding fold acts as an ATP-binding subunit with ATPase activity. Belongs to the ABC transporter family. ALD subfamily. Contains 1 ABC transmembrane type-1 domain. Contains 1 ABC transporter domain. Can form homo- and heterodimers with ABCD2/ALDR and ABCD3/PMP70. Dimerization is necessary to form an active transporter. Interacts with PEX19.