

Product name:	PEX12 Rabbit Polyclonal Antibody
Cat number:	ABN15998
Conjugate:	Unconjugated
Size:	100µL
Clone:	Polyclonal
Concentration:	1mg/ml
Host:	Rabbit
Isotype:	IgG
Immunogen:	Synthesized peptide derived from human protein . at AA range: 180-260
Reactivity:	Human,Rat,Mouse
Applications:	WB 1:500-1:2000,ELISA 1:5000-1:20000
Molecular Weight:	39kDa
Purification:	Affinity purification
Form:	Liquid
Buffer:	Liquid in PBS containing 50% glycerol, 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:

peroxisomal biogenesis factor 12(PEX12) Homo sapiens This gene belongs to the peroxin-12 family. Peroxins (PEXs) are proteins that are essential for the assembly of functional peroxisomes. The peroxisome biogenesis disorders (PBDs) are a group of genetically heterogeneous autosomal recessive, lethal diseases characterized by multiple defects in peroxisome function. The peroxisomal biogenesis disorders are a heterogeneous group with at least 14 complementation groups and with more than 1 phenotype being observed in cases falling into particular complementation groups. Although the clinical features of PBD patients vary, cells from all PBD patients exhibit a defect in the import of one or more classes of peroxisomal matrix proteins into the organelle. Defects in this gene are a cause of Zellweger syndrome (ZWS). [provided by RefSeq, Oct 2008],disease:Defects in PEX12 are a cause of Zellweger syndrome (ZWS) [MIM:214100]. ZWS is a fatal peroxisome biogenesis disorder characterized by dysmorphic facial features, hepatomegaly, ocular abnormalities, renal cysts, hearing impairment, profound psychomotor retardation, severe hypotonia and neonatal seizures. Death occurs within the first year of life.,disease:Defects in PEX12 are the cause of peroxisome biogenesis disorder complementation group 3 (PBD-CG3) [MIM:601758]. PBD refers to a group of peroxisomal disorders arising from a failure of protein import into the peroxisomal membrane or matrix. The PBD group is comprised of four disorders: Zellweger syndrome (ZWS), neonatal adrenoleukodystrophy (NALD), infantile Refsum disease (IRD), and classical rhizomelic chondrodysplasia punctata (RCDP). ZWS, NALD and IRD are distinct from RCDP and constitute a clinical continuum of overlapping phenotypes known as the Zellweger spectrum. The PBD group is genetically heterogeneous with at least 14 distinct genetic groups as concluded from complementation studies.,function:Required for protein import into peroxisomes.,similarity:Belongs to the pex2/pex10/pex12 family.,similarity:Contains 1 RING-type zinc finger.,subunit:Interacts with PEX5 and PEX10. Interacts with PEX19 via its cytoplasmic domain.,