

Product name:	Peroxin 7 Rabbit Polyclonal Antibody
Cat number:	ABN15989
Conjugate:	Unconjugated
Size:	100µL
Clone:	Polyclonal
Concentration:	1mg/ml
Host:	Rabbit
Isotype:	IgG
Immunogen:	The antiserum was produced against synthesized peptide derived from human PEX7. AA range:204-253
Reactivity:	Human,Mouse,Rat
Applications:	WB 1:500-1:2000,IHC 1:100-1:300,ICC/IF 1:50-1:200,ELISA 1:20000-1:40000
Molecular Weight:	40kDa
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 the cytosolic receptor for the set of peroxisomal matrix enzymes targeted to the organelle by the peroxisome targeting signal 2 (PTS2). Defects in this gene cause peroxisome biogenesis disorders (PBDs), which are characterized by multiple defects in peroxisome function. There are at least 14 complementation groups for PBDs, with more than one 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 have been associated with PBD complementation group 11 (PBD-CG11) disorders, rhizomelic chondrodysplasia punctata type 1 (RCDP1), and Refsum disease (RD). [provided by RefSeq, Oct 2008],disease:Defects in PEX7 are a cause of Refsum disease (RD) [MIM:266500]; also known as phytanic acid oxidase deficiency. RD is clinically characterized by a tetrad of abnormalities: retinitis pigmentosa, peripheral neuropathy, cerebellar ataxia, and elevated protein levels in the cerebrospinal fluid (CSF). Patients exhibit accumulation of the branched-chain fatty acid, phytanic acid, in blood and tissues. Less constant features are nerve deafness, anosmia, skeletal abnormalities, ichthyosis, cataracts and cardiac impairment. Manifestations of the disease appear in the second or third decade of life.,disease:Defects in PEX7 are the cause of peroxisome biogenesis disorder complementation group 11 (PBD-CG11) [MIM:601757]. 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 13 distinct genetic groups as concluded from complementation studies.,disease:Defects in PEX7 are the cause of rhizomelic chondrodysplasia punctata type 1 (RCDP1) [MIM:215100]. RCDP1 is characterized by rhizomelic shortening of femur and humerus, vertebral disorders, cataract, cutaneous lesions and severe mental retardation.,function:Binds to the N-terminal PTS2-type peroxisomal targeting signal and plays an essential role in peroxisomal protein import.,similarity:Belongs to the WD repeat peroxin-7 family.,similarity:Contains 6 WD repeats.,subunit:Interacts with PEX5.,tissue specificity:Ubiquitous. Highest expression in pancreas, skeletal muscle and heart.,