

PEX7 Rabbit pAb

Catalog # AP97075

Product Information

Application	WB
Primary Accession	O00628
Other Accession	O00628
Host	Rabbit
Clonality	Polyclonal
Calculated MW	35892
Physical State	Liquid
Immunogen	KLH conjugated synthetic peptide derived from human PEX7
Epitope Specificity	61-160/323
Isotype	IgG
Purity	affinity purified by Protein A
Buffer	0.01M TBS (pH7.4) with 1% BSA, 0.02% Proclin300 and 50% Glycerol.
SIMILARITY	Belongs to the WD repeat peroxin-7 family. Contains 6 WD repeats.
SUBUNIT	nteracts with PEX5.
DISEASE	Defects in PEX7 are the cause of peroxisome biogenesis disorder complementation group 11 (PBD-CG11) [MIM:614879]. 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. 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. Defects in PEX7 are the cause of peroxisome biogenesis disorder 9B (PBD9B) [MIM:614879]. A peroxisome biogenesis disorder with unusually mild clinical and biochemical manifestations. Affected individuals manifest a variable phenotype similar to, and in some cases indistinguishable from, classic Refsum disease. Variable features include ocular abnormalities, sensorimotor neuropathy, ichthyosis, deafness, chondrodysplasia punctata without rhizomelia or growth failure.
Important Note	This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.
Background Descriptions	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]

Additional Information

Gene ID	5191
Other Names	PEX7_HUMAN; PEX7; PTS2 receptor; Peroxin-7; PTS2R; PEX7_MOUSE; peroxisomal biogenesis factor 7; Refsum disease
Dilution	WB=1:500-2000
Storage	Store at -20 °C for one year. Avoid repeated freeze/thaw cycles. When reconstituted in sterile pH 7.4 0.01M PBS or diluent of antibody the antibody is stable for at least two weeks at 2-4 °C.

Protein Information

Name	PEX7 {ECO:0000303 PubMed:9090383, ECO:0000312 HGNC:HGNC:8860}
Function	Receptor required for the peroxisomal import of proteins containing an N-terminal PTS2-type peroxisomal targeting signal (PubMed: 11931631 , PubMed: 22057399 , PubMed: 25538232 , PubMed: 9090381 , PubMed: 40739340). Specifically binds to cargo proteins containing a PTS2 peroxisomal targeting signal in the cytosol (PubMed: 11931631 , PubMed: 22057399 , PubMed: 25538232). Cargo protein-binding triggers interaction with PEX5 and formation of a ternary complex composed of PEX5 and PEX7 along with PTS2-containing cargo proteins, which is translocated into peroxisomes by passing through the PEX13-PEX14 docking complex (PubMed: 11546814 , PubMed: 25538232).
Cellular Location	Cytoplasm, cytosol. Peroxisome matrix. Note=Translocated into the peroxisome matrix together with PTS2-containing cargo proteins and PEX5
Tissue Location	Ubiquitous (PubMed:9090381). Highest expression in pancreas, skeletal muscle and heart (PubMed:9090381)

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