10320 Camino Santa Fe, Suite G
San Diego, CA 92121
Tel: 858.875.1900 Fax: 858.875.1999

CTNS Rabbit pAb

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Catalog # AP55022

Product Information

Application	WB
Primary Accession	O60931
Reactivity	Rat, Human, Mouse
Predicted	Rabbit
Host	Rabbit
Clonality	Polyclonal
Calculated MW	41738
Physical State	Liquid
Immunogen	KLH conjugated synthetic peptide derived from human CTNS/Cystinosin
Epitope Specificity	231-330/367
Isotype	IgG
Purity	affinity purified by Protein A
Buffer	0.01M TBS (pH7.4) with 1% BSA, 0.02% Proclin300 and 50% Glycerol.
SUBCELLULAR LOCATION	Lysosome membrane; Multi-pass membrane protein.
DISEASE	Defects in CTNS are the cause of cystinosis nephropathic type (CTNS) [MIM:219800]. It is a form of cystinosis, a lysosomal storage disease due to defective transport of cystine across the lysosomal membrane. This results in cystine accumulation and crystallization in the cells causing widespread tissue damage. The classical nephropathic form has onset in the first year of life and is characterized by a polyuro-polydipsic syndrome, marked height-weight growth delay, generalized impaired proximal tubular reabsorptive capacity, with severe fluid-electrolyte balance alterations, renal failure, ocular symptoms and other systemic complications. Defects in CTNS are the cause of cystinosis adult non-nephropathic type (CTNSANN) [MIM:219750]. It is a form of cystinosis, a lysosomal storage disease due to defective transport of cystine across the lysosomal membrane. This results in cystine accumulation and crystallization in the cells causing widespread tissue damage. Cystinosis adult non-nephropathic type is characterized by ocular features and a benigne course. Patients manifest mild photophobia due to conjunctival and corneal cystine crystals. Defects in CTNS are the cause of cystinosis late-onset juvenile or adolescent nephropathic type (CTNSJAN) [MIM:219900]. It is a form of cystinosis, a lysosomal storage disease due to defective transport of cystine across the lysosomal membrane. This results in cystine accumulation and crystallization in the cells causing widespread tissue damage. Late-onset juvenile or adolescent nephropathic cystinosis manifests itself first at age 10 to 12 years with proteinuria due to glomerular damage rather than with the manifestations of tubular damage that occur first in infantile cystinosis. There is no excess amino aciduria and stature is normal. Photophobia, late development of pigmentary retinopathy, and chronic headaches are features.
Important Note	This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.
Background Descriptions	Cystinosis is an autosomal recessive disorder resulting from defective lysosomal transport of cystine and present at birth as a failure to thrive,

rickets and proximal renal tubular acidosis. The human CTNS gene on chromosome 17p13 encodes the protein Cystinosin, and mutations in CTNS are responsible for nephropathic cystinosis. The CTNS promoter contains an Sp1 binding element. Cystinosin is an integral membrane protein containing 7 transmembrane domains that functions as a H+-driven transporter responsible for cystine export from lysosomes. In humans, Cystinosin is expressed abundantly in pancreas, kidney (mature and fetal), and skeletal muscle. The mouse homolog to CTNS encodes a protein which is expressed in all tissues except skeletal muscle. In the cell, Cystinosin co-localizes with LAMP-2 to lysosomes. A C-terminal GYDQL sorting motif within Cystinosin is critical for lysosomal localization.

Additional Information

Gene ID	1497
Other Names	Cystinosin, CTNS {ECO:0000303|PubMed:9537412, ECO:0000312|HGNC:HGNC:2518}
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.

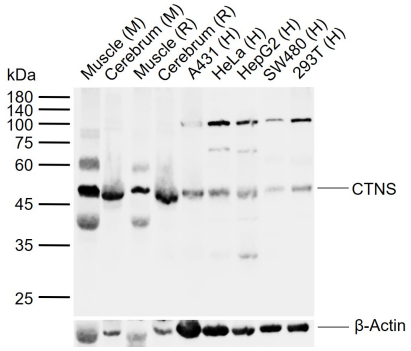
Protein Information

Name	CTNS {ECO:0000303|PubMed:9537412, ECO:0000312|HGNC:HGNC:2518}
Function	Cystine/H(+) symporter that mediates export of cystine, the oxidized dimer of cysteine, from lysosomes (PubMed:11689434, PubMed:15128704, PubMed:18337546, PubMed:22232659, PubMed:29467429, PubMed:33208952, PubMed:36113465). Plays an important role in melanin synthesis by catalyzing cystine export from melanosomes, possibly by inhibiting pheomelanin synthesis (PubMed:22649030). In addition to cystine export, also acts as a positive regulator of mTORC1 signaling in kidney proximal tubular cells, via interactions with components of the v-ATPase and Ragulator complexes (PubMed:36113465). Also involved in small GTPase-regulated vesicle trafficking and lysosomal localization of LAMP2A, independently of cystine transporter activity (By similarity).
Cellular Location	[Isoform 1]: Lysosome membrane; Multi-pass membrane protein. Melanosome membrane; Multi-pass membrane protein. Note=AP-3 complex is required for localization to the lysosome.
Tissue Location	Strongly expressed in pancreas, kidney (adult and fetal), skeletal muscle, melanocytes and keratinocytes (PubMed:22649030). Expressed at lower levels in placenta and heart Weakly expressed in lung, liver and brain (adult and fetal) (PubMed:22649030).

Background

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Images



Sample:
Lane 1: Mouse Muscle tissue lysates
Lane 2: Mouse Cerebrum tissue lysates
Lane 3: Rat Muscle tissue lysates
Lane 4: Rat Cerebrum tissue lysates
Lane 5: Human A431 cell lysates
Lane 6: Human HeLa cell lysates
Lane 7: Human HepG2 cell lysates
Lane 8: Human SW480 cell lysates
Lane 9: Human 293T cell lysates
Primary: Anti-CTNS (AP55022) at 1/1000 dilution
Secondary: IRDye800CW Goat Anti-Rabbit IgG at 1/20000 dilution
Predicted band size: 42 kDa
Observed band size: 48 kDa

Please note: All products are 'FOR RESEARCH USE ONLY. NOT FOR USE IN DIAGNOSTIC OR THERAPEUTIC PROCEDURES'.