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# Rabbit Anti-CTNS antibody

SL12932R

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| **Product Name:** | CTNS |
| **Chinese Name:** | 胱氨酸抗体 |
| **Alias:** | CTNS LSB; Cystinosin; cystinosis, nephropathic; PQLC4; CTNS_HUMAN. |
| **Organism Species:** | Rabbit |
| **Clonality:** | Polyclonal |
| **React Species:** | Human,Mouse,Rat,Rabbit, |
| **Applications:** | ELISA=1:500-1000IHC-P=1:400-800IHC-F=1:400-800ICC=1:100-500IF=1:100-500（Paraffin sections need antigen repair）<br>not yet tested in other applications.<br>optimal dilutions/concentrations should be determined by the end user. |
| **Molecular weight:** | 42kDa |
| **Cellular localization:** | cytoplasmicThe cell membrane |
| **Form:** | Lyophilized or Liquid |
| **Concentration:** | 1mg/ml |
| **immunogen:** | KLH conjugated synthetic peptide derived from human CTNS/Cystinosin:231-330/367 |
| **Lsotype:** | IgG |
| **Purification:** | affinity purified by Protein A |
| **Storage Buffer:** | 0.01M TBS(pH7.4) with 1% BSA, 0.03% Proclin300 and 50% Glycerol. |
| **Storage:** | Store at -20 °C for one year. Avoid repeated freeze/thaw cycles. The lyophilized antibody is stable at room temperature for at least one month and for greater than a year when kept at -20°C. 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. |
| **PubMed:** | PubMed |
| **Product Detail:** | 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.

**Function:**
CTNS (Cystinosin) is thought to transport cystine out of lysosomes. Mutations in the CTNS gene are the cause of cystinosis.

**Subcellular Location:**
Lysosome membrane; Multi-pass membrane protein.

**Tissue Specificity:**
Strongly expressed in pancreas, kidney (adult and fetal) and in skeletal muscle. Expressed at lower levels in placenta and heart. Weakly expressed in lung, liver and brain (adult and fetal).

**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.

**Similarity:**
Belongs to the cystinosin family.
Contains 2 PQ-loop domains.

**SWISS:**
O60931

**Gene ID:**
1497

**Database links:**

Entrez Gene: 1497Human

Entrez Gene: 83429Mouse

Entrez Gene: 287478Rat

Omim: 606272Human

SwissProt: O60931Human

SwissProt: P57757Mouse

Unigene: 187667Human

Unigene: 259852Mouse

**Important Note:**
This product as supplied is intended for research use only, not for use in human, therapeutic or diagnostic applications.