SunLong Biotech Co.,LTD
Tel: 0086-571- 56623320 Fax:0086-571- 56623318
E-mail:sales@sunlongbiotech.com
www.sunlongbiotech.com

Rabbit Anti-TIMM8A antibody

SL11769R

Product Name:	TIMM8A
Chinese Name:	Mitochondrion内膜转位酶8A/耳聋/肌张力障碍肽抗体
Alias:	DDP 1; DDP; DDP1; Deafness dystonia protein 1; Deafness/dystonia peptide; DFN 1; DFN1; MGC12262; Mitochondrial import inner membrane translocase subunit Tim8 A; MTS; TIM 8A; TIM8A; TIMM 8A; Translocase of inner mitochondrial membrane 8 homolog A; X linked deafness dystonia protein; TIM8A_HUMAN.
Organism Species:	Rabbit
Clonality:	Polyclonal
React Species:	Human,Mouse,Rat,Pig,Cow,Sheep,
Applications:	ELISA=1:500-1000IHC-P=1:400-800IHC-F=1:400-800ICC=1:100-500IF=1:100-500（Paraffin sections need antigen repair） not yet tested in other applications. optimal dilutions/concentrations should be determined by the end user.
Molecular weight:	11kDa
Cellular localization:	cytoplasmicThe cell membraneMitochondrion
Form:	Lyophilized or Liquid
Concentration:	1mg/ml
immunogen:	KLH conjugated synthetic peptide derived from human TIMM8A:31-97/97
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:	The majority of mitochondrial-directed proteins are encoded by the nuclear genome and are transported to the mitochondria via regulated processes involving the mitochondrial Tom and Tim proteins (1). The mitochondrial Tim protein family is comprised of a large group of evolutionarily conserved proteins that are found in most eukaryotes

(1,2). Import of nuclear-encoded precursor proteins into and across the mitochondrial inner membrane is mediated by two distinct complexes, the Tim23 complex and the Tim22 complex, which differ in their substrate specificity (1). Defects in Tim proteins are implicated in several neuro-degenerative diseases, suggesting important roles for Tim proteins in development and health (3,4). Tim8A and Tim8B, which map to human chromosomes Xq22.1 and 11q23.1-q23.2, respectively, are conserved proteins of the mitochondrial intermembrane space, which are organized in hetero-oligomeric complex with Tim13 (5,6,7). Tim8A is highly expressed in fetal and adult brain (5). Tim8A is mutated in deafness dystonia syndrome, a novel type of disease that causes severe neurological defects, thought to be caused by a defective mitochondrial protein transport system (5,8).

Function:
TIMM8A is a mitochondrial intermembrane chaperone that participates in the import and insertion of some multi-pass transmembrane proteins, such as metabolite transporters, from the cytoplasm into the mitochondrial inner membrane. The TIMM8A gene is mutated in Deafness Dystonia Syndrome (MTS/DFN-1) suggesting that it is required for normal neurologic development.

Subunit:
Heterohexamer; composed of 3 copies of TIMM8A and 3 copies of TIMM13, named soluble 70 kDa complex. Associates with the TIM22 complex, whose core is composed of TIMM22.

Subcellular Location:
Mitochondrion inner membrane; Peripheral membrane protein; Intermembrane side.

Tissue Specificity:
Highly expressed in fetal and adult brain, followed by fetal lung, liver and kidney. Also expressed in heart, placenta, lung, liver, kidney, pancreas, skeletal muscle and heart.

DISEASE:
Defects in TIMM8A are the cause of Mohr-Tranebjaerg syndrome (MTS) [MIM:304700]; also known as dystonia-deafness syndrome (DDS) or X-linked progressive deafness type 1 (DFN-1). It is a recessive neurodegenerative syndrome characterized by postlingual progressive sensorineural deafness as the first presenting symptom in early childhood, followed by progressive dystonia, spasticity, dysphagia, mental deterioration, paranoia and cortical blindness.
Defects in TIMM8A are the cause of Jensen syndrome (JENSS) [MIM:311150]; also known as opticoacoustic nerve atrophy with dementia. This X-

Similarity:
Belongs to the small Tim family.

SWISS:
O60220

Gene ID:
1678

Database links:

Entrez Gene: 738690Chimpanzee

Entrez Gene: 101135568Gorilla

Entrez Gene: 1678Human

Entrez Gene: 30058Mouse

Entrez Gene: 100443133Orangutan

Entrez Gene: 702885Rhesus monkey

GenBank: NP_004076Human

Omim: 300356Human

SwissProt: O60220Human

SwissProt: Q9WVA2Mouse

Unigene: 447877Human

Unigene: 214504Mouse

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