

Recombinant human TIMM8A, His-tagged

Cat. No. TIMM8A-2486H **Lot. No.** (See product label)

SPECIFICATION

Product Overview	Recombinant human TIMM8A protein, fused to His-tag at N-terminus, was expressed in E.coli and purified by using conventional chromatography techniques.
Species	Human
Source	E.coli
Description	TIMM8A is involved in the import and insertion of hydrophobic membrane proteins from the cytoplasm into the mitochondrial inner membrane. The gene is mutated in Mohr-Tranebjaerg syndrome/Deafness Dystonia Syndrome (MTS/DDS) and it is postulated that MTS/DDS is a mitochondrial disease caused by a defective mitochondrial protein import system. Defects in this gene also cause Jensen syndrome; an X-linked disease with opticoacoustic nerve atrophy and muscle weakness. This protein, along with TIMM13, forms a 70 kDa heterohexamer. Alternative splicing results in multiple transcript variants encoding distinct isoforms.
Form	Liquid. In 20mM Tris-HCl buffer (pH 8.0) containing 0.15M NaCl, 30% glycerol, 1mM DTT
Molecular Mass	13.4kDa (120aa) confirmed by MALDI-TOF
AA Sequence	MGSSHHHHHH SSGLVPRGSH MGSMDS SSSS SAAGLGAVDP QLQHFIEVET QKQRFQQLVH QMTELCWEKC MDKPGPKLDS RAEACFVNCV ERFIDTSQFI LNRLEQTQKS KPVFSESLSD

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Purity	>90 % by SDS - PAGE
Applications	SDS-PAGE
Storage	Can be stored at +4C short term (1-2 weeks). For long term storage, aliquot and store at -20C or -70C. Avoid repeated freezing and thawing cycles.
Concentration	0.25 mg/ml (determined by Bradford assay)

GENE INFORMATION

Gene Name	TIMM8A translocase of inner mitochondrial membrane 8 homolog A (yeast) [Homo sapiens]
Official Symbol	TIMM8A
Synonyms	TIMM8A; translocase of inner mitochondrial membrane 8 homolog A (yeast); DFN1, translocase of inner mitochondrial membrane 8 (yeast) homolog A; mitochondrial import inner membrane translocase subunit Tim8 A; DDP; MTS; deafness/dystonia peptide; deafness dystonia protein 1; X-linked deafness dystonia protein; DDP1; DFN1; TIM8; MGC12262;
Gene ID	1678
mRNA Refseq	NM_001145951
Protein Refseq	NP_001139423
MIM	300356
UniProt ID	O60220

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Chromosome Location	Xq22
Pathway	Metabolism of proteins, organism-specific biosystem; Mitochondrial Protein Import, organism-specific biosystem;
Function	metal ion binding;

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