## TOR1A

Reactivity: Human

Tested applications:WB

Recommended Dilution: WB 1:500 - 1:2000

Calculated MW:35kDa

Observed MW:Refer to Figures

Immunogen:

A synthetic peptide of human TOR1A

Storage Buffer:

Store at 4. Avoid freeze / thaw cycles. Buffer: PBS with 0.02% sodium azide, 50% glycerol, pH7.3.

Synonym:

DQ2;DYT1;Torsin-1A;Dystonia 1 protein antibody;Torsin family 1 member A antibody;

Monoclonal Antibody

Species: Mouse

Gene ID:1861

Isotype:IgG

Swiss Prot:O14656

Purity: Affinity purification

For research use only.

## Background:

TorsinA is an AAA(+) protein that has been demonstrated as an endoplasmic reticulum (ER) chaperone protein involved in a sensitive reporter system for quantitation of processing through the secretory pathway. It is predominantly located in the lumen of the ER and nuclear envelope. TorsinA is responsible for early onset torsion dystonia (DYT1 dystonia), a dominantly inherited movement disorder and disease of basal ganglia function. DYT1 dystonia is commonly caused by the deletion of a glutamic acid (DeltaE) in the carboxyl terminal region of TorsinA, where the protein then aggregates in perinuclear inclusions instead of the ER. TorsinA is degraded primarily through the macroautophagylysosome pathway, whereas the TorsinA DeltaE mutant protein is degraded by both the proteasome and macroautophagylysosome pathways .

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