

Product name:	Torsin A (2F5) Rabbit Monoclonal Antibody
Cat number:	MABN19137
Conjugate:	Unconjugated
Size:	100µL
Clone:	Monoclonal
Concentration:	1mg/ml
Host:	Rabbit
Isotype:	IgG
Immunogen:	A synthetic peptide of human Torsin A/DYT1
Reactivity:	Human, Mouse, Rat
Applications:	WB 1:500-1:2000, FC 1:50-1:200
Molecular Weight:	38kDa
Purification:	Affinity purification
Form:	Liquid
Buffer:	Rabbit IgG in phosphate buffered saline , pH 7.4, 150mM NaCl, 0.02% New type preservative N and 50% glycerol. Store at +4°C short term. Store at -20°C long term. Avoid freeze / thaw cycle.
Storage:	Store at 4°C short term. Aliquot and store at -20°C for 12 months. Avoid freeze/thaw cycles.

Background:

The neurological condition Dystonia is associated with sustained muscle contractions and abnormal posturing. TorsinA, torsinB, torp2A and torp3A belong to the family of ATPases associated with cellular activities (AAA+) and mutations in torsinA cause early onset dystonia. TorsinA has been shown to suppress intracellular protein aggregation in *C. elegans* and possesses chaperon activity. Interestingly, torsinA is highly expressed in dopaminergic neurons and associates with alpha-synuclein in Lewy bodies, which pathologically characterize Parkinson's Disease. Protein with chaperone functions important for the control of protein folding, processing, stability and localization as well as for the reduction of misfolded protein aggregates. Involved in the regulation of synaptic vesicle recycling, controls STON2 protein stability in collaboration with the COP9 signalosome complex (CSN). In the nucleus, may link the cytoskeleton with the nuclear envelope, this mechanism seems to be crucial for the control of nuclear polarity, cell movement and, specifically in neurons, nuclear envelope integrity. Participates in the cellular trafficking and may regulate the subcellular location of multipass membrane proteins such as the dopamine transporter SLC6A3, leading to the modulation of dopamine neurotransmission. In the endoplasmic reticulum, plays a role in the quality control of protein folding by increasing clearance of misfolded proteins such as SGCE variants or holding them in an intermediate state for proper refolding. May have a redundant function with TOR1B in non- neural tissues.