

Rabbit Anti-FRMD7 antibody

SL8239R

Product Name:	FRMD7
Chinese Name:	FRMD7蛋白抗体
Alias:	FERM domain-containing protein 7; FRMD7; FRMD7 HUMAN.
Organism Species:	Rabbit
Clonality:	Polyclonal
React Species:	Human, Mouse, Rat, Dog, Pig, Cow, Horse,
Applications:	ELISA=1:500-1000IHC-P=1:400-800IHC-F=1:400-800IF=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:	82kDa
Cellular localization:	The nucleuscytoplasmicExtracellular matrixSecretory protein
Form:	Lyophilized or Liquid
Concentration:	1mg/ml
immunogen:	KLH conjugated synthetic peptide derived from human FRMD7:551-660/714
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:	<u>PubMed</u>
	Mutations in this gene are associated with X-linked congenital nystagmus. [provided by RefSeq, Dec 2008]
Product Detail:	Function: Plays a role in neurite development (By similarity). May play a specific role in the control of eye movement and gaze stability.

Subcellular Location:

Cell projection. Cell projection, growth cone. Note=In undifferentiated neurons, located in the actin-rich regions of the cell body. In differentiated neurons, located in the actin-rich regions of the cell body and primary neurite processes but is almost absent from secondary extensions arising from the primary neurite. Also found at the actin-rich distal end of growth cones.

Tissue Specificity:

Expressed in liver, kidney, pancreas and at low levels in brain and heart. Expressed in embryonic brain and developing neural retina.

DISEASE:

Defects in FRMD7 are the cause of nystagmus congenital X-linked type 1 (NYS1) [MIM:310700]. NYS1 is a condition defined as conjugated, spontaneous and involuntary ocular oscillations that appear at birth or during the first three months of life. Other associated features may include mildly decreased visual acuity, strabismus, astigmatism, and occasionally head nodding.

Similarity:

Contains 1 FERM domain.

SWISS:

Q6ZUT3

Gene ID:

90167

Database links:

Entrez Gene: 90167 Human

Omim: 300628 Human

SwissProt: Q6ZUT3 Human

Unigene: 170776 Human

Important Note:

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