

Rabbit Anti-DOK7 antibody

SL13633R

Product Name:	DOK7
Chinese Name:	接头蛋白DOK7抗体
Alias:	Docking protein 7; DOK 7; DOK7; DOK7_HUMAN; Downstream of tyrosine kinase 7; Protein Dok-7.
Organism Species:	Rabbit
Clonality:	Polyclonal
React Species:	Human,Mouse,Rat,Dog,Pig,Cow,Horse,Sheep,
Applications:	WB=1:500-2000ELISA=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:	53kDa
Cellular localization:	The cell membrane
Form:	Lyophilized or Liquid
Concentration:	1mg/ml
immunogen:	KLH conjugated synthetic peptide derived from human DOK7:21-120/504
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 downstream of kinase family (Dok1-7) are members of a class of "docking" proteins that include the tyrosine kinase substrates IRS-1 and Cas, which contain multiple tyrosine residues and putative SH2 binding sites. Based on their similarities, the Dok family of proteins can be divided into three subgroups: Dok-1/2/3, Dok-4/5/6 and Dok-7. Through its interaction with muscle-specific receptor kinase (MuSK), Dok-7 is crucial for neuromuscular synaptogenesis and for MuSK activation. Mice lacking

Dok-7 do not form neuromuscular synapses nor acetylcholine receptor clusters. Mutations in the Dok-7 gene can cause congenital myasthenic syndromes (CMA) — recessively inherited disorders characterized by muscle weakness.

Function:

Probable muscle-intrinsic activator of MUSK that plays an essential role in neuromuscular synaptogenesis. Acts in aneural activation of MUSK and subsequent acetylcholine receptor (AchR) clustering in myotubes. Induces autophosphorylation of MUSK.

Subcellular Location:

Cell membrane. Cell junction > synapse. Accumulates at neuromuscular junctions.

Tissue Specificity:

Preferentially expressed in skeletal muscle and heart Present in thigh muscle, diaphragm and heart but not in the liver or spleen (at protein level).

DISEASE:

Defects in DOK7 are the cause of familial limb-girdle myasthenia autosomal recessive (LGM) [MIM:254300]; also called congenital myasthenic syndrome type 1B or CMS1B. LGM is a congenital myasthenic syndrome characterized by a typical 'limb girdle' pattern of muscle weakness with small, simplified neuromuscular junctions but normal acetylcholine receptor and acetylcholinesterase function.

Similarity:

Contains 1 IRS-type PTB domain. Contains 1 PH domain.

SWISS: Q18PE1

Gene ID: 285489

Database links:

Entrez Gene: 285489 Human

Entrez Gene: 231134 Mouse

<u>Omim: 610285</u> Human

SwissProt: Q18PE1 Human

SwissProt: Q18PE0 Mouse

