

Total DMPK Cell-Based Colorimetric ELISA Kit

Catalog No :	KA3689C
Reactivity :	Human
Applications :	ELISA
Gene Name :	DMPK
Human Gene Id :	1760
Human Swiss Prot No :	Q09013
Mouse Swiss Prot No :	P54265
Storage Stability :	2-8 °C/6 months
Detection Method :	Colorimetric

Background :	<p>catalytic activity:ATP + a protein = ADP + a phosphoprotein.,cofactor:Magnesium.,disease:Defects in DMPK are the cause of myotonic dystrophy 1 (DM1) [MIM:160900]; also known as Steinert disease. DM is an autosomal dominant neurodegenerative disorder characterized by myotonia, muscle wasting in the distal extremities, cataract, hypogonadism, defective endocrine functions, male baldness, and cardiac arrhythmias. DM patients show decreased levels of kinase expression inversely related to repeat length. The minimum estimated incidence is 1 in 8'000 live births. DM1 is caused by a CTG expansion in the 3'-UTR of the DMPK gene. The repeat length usually increases in successive generations, but not always.,enzyme regulation:Activated in response to G protein second messengers. Maintained in an inactive conformation by the negative autoregulatory C-terminal coiled-coil region. Coiled-coil mediated oligomerization correlated with enhanced catalytic activity as is proteolytical cleavage near the C-terminus.,function:Critical to the modulation of cardiac contractility and to the maintenance of proper cardiac conduction activity. Phosphorylates phospholamban.,similarity:Belongs to the protein kinase superfamily.,similarity:Belongs to the protein kinase superfamily. AGC Ser/Thr protein kinase family. DMPK subfamily.,similarity:Contains 1 AGC-kinase C-terminal domain.,similarity:Contains 1 protein kinase domain.,tissue specificity:Most isoforms are expressed in many tissues including heart, skeletal muscle, liver and brain, except for isoform 2 which is only found in the heart and skeletal muscle, and isoform 14 which is only found in the brain, with high levels in the striatum, cerebellar cortex and pons.,</p>
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Function : regulation of sodium ion transport, protein amino acid phosphorylation, phosphorus metabolic process, phosphate metabolic process, cellular ion homeostasis, regulation of muscle contraction, regulation of striated muscle contraction, cell-cell signaling, synaptic transmission, regulation of heart contraction, regulation of metal ion transport, regulation of skeletal muscle contraction, regulation of skeletal muscle contraction by neural stimulation via neuromuscular junction, regulation of excitatory postsynaptic membrane potential involved in skeletal muscle contraction, phosphorylation, transmission of nerve impulse, cellular homeostasis, regulation of membrane potential, homeostatic process, regulation of ion transport, regulation of system process, chemical homeostasis, ion homeostasis, neurological system process, membrane depolarization, cellular chemical homeostasis, regulation of po

Subcellular Location : Endoplasmic reticulum membrane ; Single-pass type IV membrane protein ; Cytoplasmic side . Nucleus outer membrane ; Single-pass type IV membrane protein ; Cytoplasmic side . Mitochondrion outer membrane ; Single-pass type IV membrane protein . Sarcoplasmic reticulum membrane . Cell membrane . Cytoplasm, cytosol . Localizes to sarcoplasmic reticulum membranes of cardiomyocytes. .; [Isoform 1]: Mitochondrion membrane.; [Isoform 3]: Mitochondrion membrane.

Expression : Most isoforms are expressed in many tissues including heart, skeletal muscle, liver and brain, except for isoform 2 which is only found in the heart and skeletal muscle, and isoform 14 which is only found in the brain, with high levels in the striatum, cerebellar cortex and pons.

Products Images