

Desmin protein

Catalog No :	YD0025
Reactivity :	Human
Applications :	WB;SDS-PAGE
Gene Name :	DES
Protein Name :	Desmin protein
Sequence :	Amino acid: 242-270, with his-MBP tag.
Human Gene Id :	1674
Human Swiss Prot No :	P17661
Mouse Swiss Prot No :	P31001
Formulation :	Liquid in PBS
Source :	E.coli
Dilution :	WB 1:500-2000
Concentration :	SDS-PAGE >90%
Storage Stability :	-20°C/6 month,-80°C for long storage
Background :	<p>disease:Defects in DES are the cause of cardiomyopathy dilated type 1I (CMD1I) [MIM:604765]. Dilated cardiomyopathy is a disorder characterized by ventricular dilation and impaired systolic function, resulting in congestive heart failure and arrhythmia. Patients are at risk of premature death.,disease:Defects in DES are the cause of desmin-related cardio-skeletal myopathy (CSM) [MIM:601419]; also known as desmin-related myopathy (DRM). CSM is characterized by skeletal muscle weakness associated with cardiac conduction blocks, arrhythmias, restrictive heart failure, and by intracytoplasmic accumulation of desmin-reactive deposits in cardiac and skeletal muscle cells. A desmin-related myopathy can have a distal onset, it is then known as hereditary distal myopathy (HDM).,disease:Defects in DES are the cause of neurogenic</p>

scapuloperoneal syndrome Kaeser type (Kaeser syndrome) [MIM:181400].
Kaeser syndrome is an autosomal dominant disorder with a peculiar scapuloperoneal distribution of weakness and atrophy. A large clinical variability is observed ranging from scapuloperoneal, limb grindle and distal phenotypes with variable cardiac or respiratory involvement. Facial weakness, dysphagia and gynaecomastia are frequent additional symptoms. Affected men seemingly bear a higher risk of sudden, cardiac death as compared to affected women. Histological and immunohistochemical examination of muscle biopsy specimens reveal a wide spectrum of findings ranging from near normal or unspecific pathology to typical, myofibrillar changes with accumulation of desmin.,function:Desmin are class-III intermediate filaments found in muscle cells. In adult striated muscle they form a fibrous network connecting myofibrils to each other and to the plasma membrane from the periphery of the Z-line structures.,online information:Desmin entry,similarity:Belongs to the intermediate filament family.,subunit:Homopolymer.,

Function :

muscle system process, muscle contraction, cytoskeleton organization, regulation of heart contraction, regulation of system process,

Subcellular Location :

Cytoplasm, myofibril, sarcomere, Z line . Cytoplasm . Cell membrane, sarcolemma . Nucleus . Localizes in the intercalated disks which occur at the Z line of cardiomyocytes (PubMed:24200904, PubMed:26724190). Localizes in the nucleus exclusively in differentiating cardiac progenitor cells and premature cardiomyocytes (By similarity). .

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