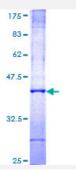


FCMD (Human) Recombinant Protein (Q01)

Catalog # H00002218-Q01 Size 25 ug, 10 ug

Applications



Specification	
Product Description	Human FCMD partial ORF (NP_006722, 29 a.a 138 a.a.) recombinant protein with GST-tag at N-t erminal.
Sequence	KHYLSTKNGAGLSKSKGSRIGFDSTQWRAVKKFIMLTSNQNVPVFLIDPLILELINKNFEQVKNTSH GSTSQCKFFCVPRDFTAFALQYHLWKNEEGWFRIAENMGFQCL
Host	Wheat Germ (in vitro)
Theoretical MW (kDa)	37.84
Interspecies Antigen Sequence	Mouse (85); Rat (85)
Preparation Method	in vitro wheat germ expression system
Purification	Glutathione Sepharose 4 Fast Flow
Quality Control Testing	12.5% SDS-PAGE Stained with Coomassie Blue.
Storage Buffer	50 mM Tris-HCl, 10 mM reduced Glutathione, pH=8.0 in the elution buffer.
Storage Instruction	Store at -80°C. Aliquot to avoid repeated freezing and thawing.
Note	Best use within three months from the date of receipt of this protein.



Applications

- Enzyme-linked Immunoabsorbent Assay
- Western Blot (Recombinant protein)
- Antibody Production
- Protein Array

Gene Info — FKTN	
Entrez GenelD	2218
GeneBank Accession#	NM_006731
Protein Accession#	NP_006722
Gene Name	FKTN
Gene Alias	CMD1X, FCMD, LGMD2M, MGC126857, MGC134944, MGC134945, MGC138243
Gene Description	fukutin
Omim ID	<u>236670</u> <u>253800</u> <u>607440</u>
Gene Ontology	<u>Hyperlink</u>
Gene Summary	The protein encoded by this gene is a putative transmembrane protein that is localized to the cis-Golgi compartment, where it may be involved in the glycosylation of alpha-dystroglycan in skeletal muscle. The encoded protein is thought to be a glycosyltransferase and could play a role in brain development. Defects in this gene are a cause of Fukuyama-type congenital muscular dystrophy (FCMD), Walker-Warburg syndrome (WWS), limb-girdle muscular dystrophy type 2M (LGMD2M), and dilated cardiomyopathy type 1X (CMD1X). Two transcript variants encoding the same protein have been found for this gene. [provided by RefSeq
Other Designations	Fukuyama type congenital muscular dystrophy protein OTTHUMP00000021841

Disease

- Cardiomyopathy
- Muscular Dystrophies



Muscular Dystrophy