

Recombinant Human FKTN, His-tagged

Cat. No. FKTN-12921H **Lot. No.** (See product label)

SPECIFICATION

Product Overview	Recombinant Human FKTN protein, fused to His-tag, was expressed in E.coli and purified by Ni-sepharose.
Species	Human
Source	E.coli
ProteinLength	35-461a.a.
Description	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). Alternatively spliced transcript variants have been found for this gene.
Storage	The protein is stored in PBS buffer at -20°C. Avoid repeated freezing and thawing cycles.
Storage Buffer	1M PBS (58mM Na ₂ HPO ₄ , 17mM NaH ₂ PO ₄ , 68mM NaCl, pH8.) added with 300mM Imidazole and 0.7% Sarcosyl, 15%glycerol.

GENE INFORMATION

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Gene Name	FKTN fukutin [Homo sapiens]
Official Symbol	FKTN
Synonyms	FKTN; fukutin; FCMD, Fukuyama type congenital muscular dystrophy (fukutin); LGMD2M; patient fukutin; Fukuyama type congenital muscular dystrophy protein; FCMD; CMD1X; MDDGA4; MDDGB4; MDDGC4; MGC126857; MGC134944; MGC134945; MGC138243;
Gene ID	2218
mRNA Refseq	NM_001079802
Protein Refseq	NP_001073270
MIM	607440
UniProt ID	O75072
Chromosome Location	9q31-q33
Function	transferase activity;

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