Recombinant Full Length Human DMD Protein

Cat. No. DMD-127HF **Lot. No.** (See product label)

SPECIFICATION

Product Overview	Recombinant full length Human Dystrophin with N terminal proprietary tag, 95.96kDa inclusive of tag.
Species	Human
Source	In Vitro Cell Free System
ProteinLength	635 amino acids
Description	The dystrophin gene is the largest gene found in nature, measuring 2.4 Mb. The gene was identified through a positional cloning approach, targeted at the isolation of the gene responsible for Duchenne (DMD) and Becker (BMD) Muscular Dystrophies. DMD is a recessive, fatal, X-linked disorder occurring at a frequency of about 1 in 3,500 new-born males. BMD is a milder allelic form. In general, DMD patients carry mutations which cause premature translation termination (nonsense or frame shift mutations), while in BMD patients dystrophin is reduced either in molecular weight (derived from in-frame deletions) or in expression level. The dystrophin gene is highly complex, containing at least eight independent, tissue-specific promoters and two polyA-addition sites. Furthermore, dystrophin RNA is differentially spliced, producing a range of different transcripts, encoding a large set of protein isoforms. Dystrophin (as encoded by the Dp427 transcripts) is a large, rod-like cytoskeletal protein which is found at the inner surface of muscle fibers. Dystrophin is part of the dystrophin-glycoprotein complex (DGC), which bridges the inner cytoskeleton (F-actin) and the extra-cellular matrix.

Tel: 1-631-559-9269 1-516-512-3133
Email: info@creative-biomart.com Fax: 1-631-938-8127
45-1 Ramsey Road, Shirley, NY 11967, USA

Form	Liquid
Molecular Mass	95.960kDa inclusive of tags
AA Sequence	MREQLKGHETQTTCWDHPKMTELYQSLADLNNVRFSAYRT AMKLRRLQKALCLDL LSLSAACDALDQHNLKQNDQPMDIL QIINCLTTIYDRLEQEHNNLVNVPLCVDMCLN WLLNVYDT GRTGRIRVLSFKTGIISLCKAHLEDKYRYLFKQVASSTGF CDQRRLGLL LHDSIQIPRQLGEVASFGGSNIEPSVRSCFQ FANNKPEIEAALFLDWMRLEPQSMV WLPVLHRVAAAETAK HQAKCNICKECPIIGFRYRSLKHFNYDICQSCFFSGRVAK G HKMHYPMVEYCTPTTSGEDVRDFAKVLKNKFRTKRYFAK HPRMGYLPVQTVLEGD NMETPVTLINFWPVDSAPASSPQL SHDDTHSRIEHYASRLAEMENSNGSYLNDSIS PNESIDDE HLLIQHYCQSLNQDSPLSQPRSPAQILISLESEERGELER ILADLEEENR NLQAEYDRLKQQHEHKGLSPLPSPPEMMPT SPQSPRDAELIAEAKLLRQHKGRLEA RMQILEDHNKQLES QLHRLRQLLEQPQAEAKVNGTTVSSPSTSLQRSDSSQPML L RVVGSQTSDSMGEEDLLSPPQDTSTGLEEVMEQLNNSFP SSRGHNVGSLFHMAD DLGRAMESLVSVMTDEEGAE
Purity	Proprietary Purification
Storage	Shipped on dry ice. Upon delivery aliquot and store at -80 centigrade. Avoid freeze / thaw cycles.
Storage Buffer	pH: 8.00. Constituents:0.79% Tris HCl, 0.31% Glutathione.

GENE INFORMATION

Gene Name	DMD dystrophin [Homo sapiens]
Official Symbol	DMD
Synonyms	DMD; dystrophin; dystrophin (muscular dystrophy, Duchenne and Becker types),

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	includes DXS142, DXS164, DXS206, DXS230, DXS239, DXS268, DXS269, DXS270, DXS272; BMD; DXS142; DXS164; DXS206; DXS230; DXS239; DXS268; DXS269; DXS270; DXS272; muscular dystrophy
Gene ID	1756
mRNA Refseq	NM_000109
Protein Refseq	NP_000100
MIM	300377
UniProt ID	P11532

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