

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.
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Species	Human
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Source	In Vitro Cell Free System
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ProteinLength	635 amino acids
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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.
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Form	Liquid
Molecular Mass	95.960kDa inclusive of tags
AA Sequence	MREQLKGHETQTTCDWHPKMTELYQSLADLNNVRFSAAYRT AMKLRLRLQKALCLDL LLSAACDALDQHNLKQNDQPM DIL QIINCLTTIYDRLEQEHNNLVNPLCVD MCLN WLLNVYDT GRTGRIRVLSFKTGIISLCKAHLEDKYRYL FKQVASSTGF CDQRR LGLL LHDSIQIPRQLGEVASFGGSNIEPSVRSCFQ FANNKPEIEAALFLDWMRLEPQSMV WLPVLHRVAAAETAK HQAKCNICKECPIIGFRYRSLKHFN YDICQSCFFSGRVAK G HKMHYPMVEYCTPTTSGEDVRDFAKVLKNKFR TKRYFAK HPRMGYLPVQTVLEGD NMETPVT LINFWPVDSAPASSPQL SHDDTHSRIEHYASRLAEMENSNGSYLND SIS PNESIDDE HLLIQHYCQSLNQDSPLSQPRSPAQILISLESEER GELER ILADLEEENR NLQAEYDRLKQQHEHKGLSPLSPPEMMPT 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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