

Recombinant Human DMD

Cat. No. DMD-26936TH **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	Wheat Germ
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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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Molecular Weight	95.960kDa inclusive of tags
Tissue specificity	Expressed in muscle fibers accumulating in the costameres of myoplasm at the sarcolemma. Expressed in brain, muscle, kidney, lung and testis. Isoform 5 is expressed in heart, brain, liver, testis and hepatoma cells. Most tissues contain transcripts of mul
Form	Liquid
Purity	Proprietary Purification
Storage buffer	pH: 8.00Constituents:0.3% Glutathione, 0.79% Tris HCl
Storage	Shipped on dry ice. Upon delivery aliquot and store at -80oC. Avoid freeze / thaw cycles.
Sequences of amino acids	MREQLKGHETQTTCWDHPKMTELYQSLADLNNVRFSA YRT AMKLRRRLQKALCLDL LSLSAACDALDQHNLKQNDQPM DIL QIINCLTTIYDRLEQEHNNLVNPLCVD MCLN WLLNVYDT GRTGRIRVLSFKTGIISLCKAHLEDKYRYL FKQVASSTGF CDQRR LGLL LHDSIQIPRQLGEVASFGGSNIEPSVRSCFQ FANNKPEIEAALFLDWMRLEPQSMV WLPVLHRVAAAETAK HQAKCNICKECPIIGFRYRSLKHFN YDICQSCFFSGRVAK G HKMHYPMVEYCTPTTSGEDVRDFAKVLKNKFRTKRYFAK HPRMGYLPVQTVLEGD NMETPVT LINFWPVDSAPASSPQL SHDDTHSRIEHYASRLAEMENSNGSYLND SIS PNESIDDE HLLIQHYCQSLNQDSPLSQPRSPAQILISLESEERGELER ILADLEEENR NLQAEYDRLKQQHEHKGLSPLSPPEMMPT SPQSPRDAELIAEAKLLRQHKGRLEA RMQILEDH NKQLES QLHRLRQLLEQPQAEAKVNGTTVSSPSTSLQRS DSSQPML L RVVGSQTSDSMG EEDLLSPPQDTSTGLEEVMEQLNNSFP SSRGHNVGSLFHMAD DLGRAMESLVSVMTDEEGAE
Sequence Similarities	Contains 2 CH (calponin-homology) domains.Contains 22 spectrin repeats.Contains 1

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WW domain.Contains 1 ZZ-type zinc finger.

GENE INFORMATION

Gene Name DMD dystrophin [Homo sapiens]

Official Symbol DMD

Synonyms DMD; dystrophin; dystrophin (muscular dystrophy, Duchenne and Becker types), 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

Chromosome Location Xp21.2

Pathway Arrhythmogenic right ventricular cardiomyopathy (ARVC), organism-specific biosystem; Arrhythmogenic right ventricular cardiomyopathy (ARVC), conserved biosystem; Dilated cardiomyopathy, organism-specific biosystem; Dilated cardiomyopathy, conserved biosystem; Hypertrophic cardiomyopathy (HCM), organism-specific biosystem;

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Function

PDZ domain binding; actin binding; actin binding; beta-dystroglycan binding; dystroglycan binding;

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