

Recombinant Human MBNL1 Protein, Myc/DDK-tagged, C13 and N15-labeled

Cat. No. MBNL1-2139H **Lot. No.** (See product label)

SPECIFICATION

Product Overview	MBNL1 MS Standard C13 and N15-labeled recombinant protein (NP_997180) with a C-terminal MYC/DDK tag, was expressed in HEK293 cells.
Species	Human
Source	HEK293
Description	This gene encodes a member of the muscleblind protein family which was initially described in <i>Drosophila melanogaster</i> . The encoded protein is a C3H-type zinc finger protein that modulates alternative splicing of pre-mRNAs. Muscleblind proteins bind specifically to expanded dsCUG RNA but not to normal size CUG repeats and may thereby play a role in the pathophysiology of myotonic dystrophy. Mice lacking this gene exhibited muscle abnormalities and cataracts. Several alternatively spliced transcript variants have been described but the full-length natures of only some have been determined. The different isoforms are thought to have different binding specificities and/or splicing activities.
Molecular Mass	36.9 kDa
AA Sequence	MAVSVTPIRDTKWLTLEVCREFQRGTCSPDTECKFAHPSKSCQVENGRVIACFDS LKGRCsRENCKYLHPPPHLKTQLEINGRNLIQKQNMAMLAQQMQLANAMMPGAP LQPVPMFSVAPSLATNASAAAFNPYLGPVSPSLVPAEILPTAPMLVTGNPGVPVPA AAAAAQKLMRTDRLEVCREYQRGNCNRGENDCRFAHPADSTMIDTNDNTVTVCMD

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YIKGRCSREKCKYFHPPAHLQAKIKAAQYQVNQAAAAQAAATAAAMGIPQAVLPPLP
KRPALKTNGATAVENTGIFQYQQALANMQLQQHTAFLPPGSILCMTPATSVVDTHNI
CRTSDTRTRPLEQKLISEEDLAANDILDYKDDDDKV

Purity > 80% as determined by SDS-PAGE and Coomassie blue staining

Stability Stable for 3 months from receipt of products under proper storage and handling conditions.

Storage Store at -80 centigrade. Avoid repeated freeze-thaw cycles.

Concentration 50 µg/mL as determined by BCA

Storage Buffer 100 mM glycine, 25 mM Tris-HCl, pH 7.3.

GENE INFORMATION

Gene Name MBNL1 muscleblind-like splicing regulator 1 [Homo sapiens (human)]

Official Symbol MBNL1

Synonyms MBNL1; muscleblind-like splicing regulator 1; MBNL, muscleblind (Drosophila) like, muscleblind like (Drosophila); muscleblind-like protein 1; EXP; EXP35; EXP40; EXP42; KIAA0428; triplet-expansion RNA-binding protein; MBNL; DKFZp686P06174;

Gene ID 4154

mRNA Refseq NM_207297

Protein Refseq NP_997180

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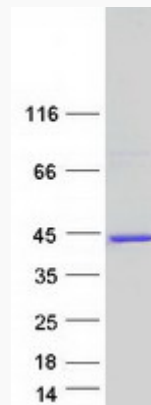
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MIM 606516

UniProt ID Q9NR56

SDS-PAGE



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