Supplementary Table 1

Tissue/Cell Line/Repeat Length	RNA FISH Probe	Mutant Repeat RNA Foci Characteristics	Protein Analyzed by IF and Type of Interaction with Repeat RNA Foci	Ref.
CUG–DM1				
h. muscle biopsy; h. fibroblasts (GM03132-2000 CTG; GM03755-500 CTG)	$(CAG)_{10}$ probe labeled with Cy3 or FITC or DIG; antisense probes to exons of 5'end DMPK labeled with DIG or Cy3	1-3 intense nuclear CUG foci in muscle and up to 13 foci/nucleus (mean of 5 foci) in fibroblasts with Cy3-CAG repeat probe; FITC foci colocalize exactly with red foci of 5'end probes; DIG-labeled CAG probe and 5' end probes generate perinuclear cytoplasmic signals in fibroblasts	Some CUG RNA foci colocalize with a non-snRNP splicing factor SC-35 and some are found in close proximity to SC-35 speckles (3-4 out of 14 RNA foci colocalize with SC-35 in fibroblasts)	Taneja KL 1995
h. fibroblasts; MyoD converted h. fibroblasts (myoblasts); differentiated myoblasts	$(CAG)_{10}$-Cy3	Hundreds of bright nuclear CUG foci (myoblasts); foci in myoblasts brighter than in fibroblasts; no cytoplasmic CUG foci in myotubes; CUG150 forms foci but only in 15% of nuclei and nuclear retention complete with (CUG)>400	NA	Davis BM 1997
h. fibroblasts (GM03132)	$(CAG)_5$-Cy3-PNA $(CAG)_{10}$-FITC	CUG foci detected with both probes but signal intensity about 3.8 times brighter with PNA15 probe; 2-15 foci (average of 5.5) with CAG10 probe and 3-30 (average of 9.6) with PNA15 probe; each RNA focus contains between 15 to 230 RNA molecules measured with a 15-mer probe	NA	Taneja KL 1998
m. myoblasts (C2C12) expressing truncated DMPK 3'UTR with CTG expansion	$(CAG)_{10}$-Cy3	CUG foci are formed in C2C12 cells transiently expressing either CUG100 alone or an entire DMPK 3'UTR with 57-200 CUG repeats; no foci in h. fibroblasts with CUG 50-80	NA	Amack JD 1999
h. myoblasts; MyoD transformed h. fibroblasts (GM03132)	$(CAG)_{10}$-Cy3	Multiple CUG RNA foci in h. myoblasts and converted fibroblasts; bigger, brighter and more abundant RNA foci in transformed fibroblasts than in untransformed; foci are enriched with hEXP42 (MBNL) protein; an average of 15 EXP-enriched foci/nucleus with a range of 6-30 foci /nucleus in myoblasts	Foci enriched with hEXP42 protein are formed in MyoD-uninfected and infected fibroblasts; their number and size increase in MyoD-transfected cells	Miller JW 2000
Skeletal muscle (vastus) from Tg HSA (CTG)250 mouse model	FITC-$(CAG)_6$CA-2-*O*-Me	>50 discrete CUG RNA foci per nucleus in muscle tissue	NA	Mankodi A 2000
h. muscle biopsy (quadriceps) and derived myoblasts (750 CTG); myotubes	$(CAG)_5$-Cy3-PNA	CUG RNA is found in discrete nuclear foci in myoblasts and myotubes but the later are brighter and more abundant; no cytoplasmic foci	NA	Furling D 2001
h. DM1 and DM2 biopsies of quadriceps	FITC-$(CAG)_6$CA-2-*O*-Me	1-3 CUG foci/nucleus in muscle; sparse and infrequent foci with 70-100 CUG and no foci with CUG<70; DM1 foci have round, curvilinear or bean-shaped profile; CAG probe reveals nuclear foci also in DM2 muscle; DM2 foci are larger and intensely fluorescent than DM1; DM1 and DM2 foci are similar in shape and number	MBNL1 is localized precisely to the nuclear DM1 and DM2 RNA foci	Mankodi A 2001
DM1 Tg mouse model (~350 CTG) and derived myoblasts	5' FITC-2'-*O*-Me-$(CAG)_{30}$	CUG RNA foci are formed in myoblasts with greater than 300 repeats when derived from homozygous and heterozygous mice	NA	Seznec H 2001
h. myoblasts (750 CTG) transfected with hammerhead ribozyme	Cy3-$(CAG)_5$-PNA	1-20 CUG foci are formed in myoblasts; reduction in the number and intensity of nuclear CUG foci in functional ribozyme-treated cells versus its mutant form; about 88% of myoblasts have 6-15 nuclear foci (mutant ribozyme) and 0-5 foci (active ribozyme)	NA	Langlois MA 2003
h. DM1 and DM2 biopsy skeletal muscle and derived myoblasts; myotubes; vastus muscle from Tg DM1 HSA mice	FITC-$(CAG)_6$CA-2-*O*-Me FITC-$(CAGG)_5$-2-*O*-Me	1-4 RNA foci/nucleus in h. DM1 and DM2 muscles; DM2 foci are detected with CAG and CCAG probe, and DM1 foci with CAG probe only; in muscle DM2 foci-larger and more intense than DM1: 7.5-fold when detected with CAG probe and 13.4-fold when with CAGG probe; in muscle DM1 foci are spheroidal and DM2 rod-shape; CUG and CCUG foci are found in myoblasts and myotubes but bigger and more intense when formed in myotubes; more numerous CUG foci (>20/nucleus) in myoblasts than in muscle; during differentiation foci coalesce into fewer, larger and more intense in fluorescence	MBNL1, MBNL2 and MBNL3 show strong colocalization with CUG and CCUG RNA foci in human muscle, myoblasts and myotubes and in HSA mouse muscle; no colocalization to DM1 and DM2 foci is found for: PKR, phospho-PKR, ADAR, RNA helicase A, NF90, FLAP, PACT, 2',5'-OAS, CUGBP1 and ETR3	Mankodi A 2003
h. autopsy brain and paired samples of biceps muscle	Texas Red-$(CAG)_6$CA-2-*O*-Me	In cortical neurons CUG RNA foci are distributed throughout all cortical layers and found in >85% of neurons; more than 1 focus is present in ~30% of cortical neurons and occasionally up to 15 small foci/nucleus are found; in patient with 77 CUG repeats, foci are found in 39% neurons of the temporal cortex; CUG foci are also present in the hippocampus, dentate gyrus, thalamus, substantia nigra and brain stem tegmentum; in the cerebellar cortex, small foci are found in some Purkinje cells but absent in neurons of the molecular or granular cell layers; CUG foci are also present in subcortical white matter and the corpus callosum but these foci are smaller and less intense than in cortical neurons–the amount of CUG repeat expansion in frontal cortical neurons is 2.9-fold greater than in Purkinje cells and 18-fold greater than in oligodendrocytes; in paired samples of frontal cortex and bicep muscle 3.1-fold greater foci are found in frontal cortical neurons than in skeletal muscle from the same individual	MBNL1 and MBNL2 show strong colocalization with CUG foci in cortical neurons; hnRNP H and F colocalize with CUG foci in cortical neurons to a limited extent; components of the proteosome (20Sα, 11Sγ, 11Sα subunits) are recruited to CUG foci in cortical neurons; PML bodies, C23 nucleolin, polypyrimidine tract binding protein, hnRNP C; components of exosome (PM/Scl75 or PM/Scl100), CUGBP1, ETR3, CELF4, Staufen, NF90, ADAR1, PACT, PKR, RNA helicase A, hnRNP A1, hnRNP 1, hnRNP M, KSRP, HuR, RARγ and Sp1 do no colocalize with CUG foci in cortical neurons	Jiang H 2004
SV40 virus immortalized h. myoblasts	$(CAG)_{10}$-Cy3	Nuclear CUG RNA foci are found in 100% cells; siRNA downregulation of CUGBP1 causes ~20% reduction of cells with foci; MBNL2 downregulation results in ~25% fewer foci-positive cells and MBNL1 in a ~70% reduction; 80% fewer cells with foci are observed when MBNL1 and MBNL2 are downregulated simultaneously	NA	Dansithong W 2005
h. postmortem heart	$(CAG)_7$-Texas Red	Nuclear CUG RNA foci are highly abundant in the left ventricular free wall and in MyBP-H-positive cells of the conduction system in the interventricular septum; foci are not found in the cytoplasm	MBNL1 and MBNL2 are sequestered in nuclear RNA foci; CUGBP1 and ETR3 do not colocalize with foci	Mankodi A 2005
h. DM1 and DM2 biopsies of vastus lateralis muscle; Tg DM1 HSA mouse muscle	2-*O*-Me-$(CAG)_6$CA-Texas Red	Nuclear foci of CUG expansion RNA are formed in DM1 human and mouse muscles and CCUG mutant repeat ribonuclear foci are detected in human DM2 muscle	MBNL1 and MBNL2 are heavily recruited into RNA foci in DM1 and DM2 muscle and the mean intensity of MBNL1 IF is reduced by ≥78%; the amount of MBNL1 is greater in DM2 than in DM1 foci; CUGBP1 does not colocalize with CUG foci in DM1 human and mouse skeletal muscle	Lin X 2006
Inducible Tg DM1 mouse model expressing 200 CTG repeat	$(CAG)_{10}$-Cy3	Nuclear CUG RNA foci are formed in mouse skeletal muscle, smooth muscle in blood vessels and in cardiac muscle after induction of transgene expression	MBNL1 is recruited to CUG RNA foci	Mahadevan MS 2006
h. DM1 and DM2 biceps muscle biopsy	$(CAG)_6$CA-Texas Red $(CAGG)_5$-Texas Red	CUG RNA foci are detected in DM1 muscle sections and CCUG foci are formed in DM2 muscle	MBNL1 is sequestered by DM1 and DM2 RNA nuclear foci; 1-2 MBNL1 foci/nucleus in DM1 and 1-3 in DM2; some DM1 and DM2 RNA foci do not have sequestered MBNL1; MBNL1 foci are bigger in DM2 than in DM1 muscles;	Cardani R 2006

h. myoblasts	$(CAG)_{10}$-Cy3	Nuclear CUG RNA foci are detected in myoblasts	GFP-MBNL1 and GFP-MBNL2 colocalize with CUG RNA foci; no significant localization of endogenous or GFP-hnRNP H to CUG foci; co-expression of GFP-hnRNP H and Flag-MBNL1 and 2 causes striking colocalization of the GFP-hnRNP to CUG foci	Paul S 2006
h. muscle biopsy; h. spinal cord tissue, DM1 Tg HSA mouse skeletal muscle	Texas Red-2-*O*-Me-$(CAG)_6$CA	In human muscle CUG RNA foci are formed in extrajunctional and in subsynaptic nuclei of muscle fibers; the most prominent foci in skeletal muscle occur in subsynaptic nuclei; in mice CUG foci are abundant in extrajunctional myonuclei and sparse or absent in subsynaptic nuclei; in human spinal cord samples CUG foci are found in motor neurons; the most prominent foci are in lumbosacral and cervical cord sections	MBNL1 colocalizes with CUG foci in subsynaptic nuclei of muscle fibers and in motor neurons of the spinal cord	Wheeler TM 2007
Tg DM1 mouse model expressing >350 CTG repeat	FITC-2-*O*-Me-$(CAG)_6$CA	CUG repeat expansion accumulate as foci in soleus where the transgene expression is strongest; rare CUG foci are found in adipose tissue and pancreas, which express the DMPK transgene at very low levels	NA	Guiraud-Dogan C 2007
Inducible heart-specific Tg DM1 mouse model expressing 960 CTG repeat in the context of DMPK 3'UTR	$(CAG)_5$-Cy3-PNA	Nuclear CUG RNA foci are formed as early as 3 hr after tamoxifen administration in cardiac tissue; higher fraction of nuclei contains foci in the highest-expressing line rather than in moderate-expressing line; multiple foci/nucleus are formed in higher-expressing line and single RNA foci in moderate-expressing line	Weak MBNL1 colocalization with CUG ribonuclear inclusions is detected 3 hr after transgene expression and become clearly detectable after 6 hr; CUGBP1 and CUGBP2 do not colocalize with CUG foci but their levels are higher in foci-positive nuclei	Wang GS 2007
h. DM1 homozygous and heterozygous myoblasts; h. DM2 fibroblasts	$(CAG)_6$-FITC $(CAGG)_7$-FITC	In nuclei from homozygous DM1 myoblasts numerous round discrete spots of CUG RNA foci are found; intact but rather diffused CUG RNA foci are found in differentiated myonuclei after siRNA MBNL1 downregulation	In DM1 myonuclei ~92% of CUG foci associates with the edge of SC-35-defined domains; similar positioning is found in DM1 myotubes; in DM2 51% of CCUG foci is associated with SC-35 but not colocalized; MBNL1 is highly concentrated with mutant CUG RNA foci in myoblasts and myotubes	Smith KP 2007
h. DM1 myoblasts from quadriceps (1800 CTG); h. DM1 and DM2 fibroblasts	$(CAG)_{10}$-Cy3 $(CCAG)_{10}$-Cy3	Numerous CUG foci/nucleus are detected in DM1 myoblasts and fibroblasts with a CAG probe, and the same foci are detected with an anti-MBNL1 antibody; actinomycin A treatment has no effect on foci number; in DM2 fibroblasts nuclear CCUG RNA foci are more abundant than in DM1 cells	MBNL1 colocalizes with DM1 foci; MBNL1-positive foci do not colocalize with YT521-B, coilin p80 and PsP proteins but overlap with MB1a; significant association between MBNL1-positive CUG RNA foci and Y12 (~90%) and 9G8 proteins in DM1 myoblasts and fibroblasts; 9G8 staining colocalizes with SC35 speckles; Y14 is associated with nuclear DM1 foci but not preferentially concentrated at the foci; most of hnRNP H speckles is not associated with DM1 RNA foci; Y12, 9G8 and exosome are not associated with MBNL1 foci or CCUG foci in DM2 fibroblasts	Holt I 2007
h. muscle biopsy (vastus lateralis) (150-2000 CTG repeat)	Cy3-$(CAG)_{10}$	CUG RNA accumulate in muscle sections from each of the 12 patients used; 0-5 foci/nucleus (average of 1.18) in muscle with 165-430 CTG and 39% nuclei had no foci; 0-18 foci/nucleus (average of 2.92) in muscle with 1250-1900 CTG and 8% nuclei had no foci; CUG foci tend to be greater and more fluorescent with the increase of the CTG length but number of nuclei without foci decreases; no cytoplasmic foci are detected in muscle sections	NA	Botta A 2008
Cardiac-specific Tg DM1 mouse model expressing 400 CTG; primary and immortalized h. myoblasts (CTG=2800, 2860, 2100 and 2200); h. fibroblasts (CTG=333 and 667)	Cy3-$(CAG)_{10}$	Nuclear and cytoplasmic CUG RNA foci are detected in h. myoblasts and fibroblasts; ~30% h. fibroblasts have both the foci; in m. cardiac cells only cytoplasmic CUG RNA foci are formed; cytoplasmic foci are more diffused in m. cardiomyocytes than h. myoblasts but still effectively sequester MBNL1	Mbnl1 colocalizes with nuclear and cytoplasmic CUG RNA foci in mouse and human cells	Dansithong W 2008
h. smooth muscle (gallbladder) (500-1000 CTG)	Texas Red-$(CAG)_6$CA-2-*O*-Me	CUG foci are found in nuclei of smooth muscle from gallbladder, in epithelial layer and connective tissue; 1-2 foci/nucleus are found in wall cells of gallbladder; 1 focus/nucleus is found in some epithelial cells; CUG foci are of similar size in skeletal and smooth muscles	Mbnl1 colocalizes with CUG foci in smooth muscle, epithelial layer and connective tissue	Cardani R 2008
HeLa and Hek293 expressing minigene with DMPK 960 interrupted CTG repeat; pentamidine treatment	$(CAG)_{10}$-Cy3	CUG foci are formed in HeLa and HEK293 cells upon transfection with CUG960 expressing plasmid; foci formation is reduced by 21% upon pentamidine treatment (HeLa) and by 28% (HEK293)	Nearly all of the nuclear MBNL1 is sequestered by CUG960 foci in HEK293 and HeLa cells; pentamidine relieves MBNL1 sequestration which is found to be diffuse throughout the nucleus	Warf MB 2009
Tg HSA DM1 mouse model (250 CTG); h. fibroblasts (GM03132); morpholino AON CAG25 treatment	2-*O*-Me-$(CAG)_6$CA-Texas Red	Numerous CUG RNA foci accumulate in nuclei of m. skeletal muscle; intramuscular injection with CAG25 causes ~50% reduction in the overall mutant CUG RNA, nuclear CUG foci become dispersed and their number reduced; morpholino reduces RNA foci also in h. fibroblasts	Mbnl1 colocalizes with CUG RNA foci in m. skeletal muscle, AON CAG25 blocks this interaction and Mbnl1 shifts from punctate to diffuse nuclear distribution with morpholino	Wheeler TM 2009
DM1 Tg mouse (DM300)- derived myoblasts and myotubes (~500 CTG); DM1 Tg HSA mouse model (250 CTG); PS58 2'-OMe PT modified (CAG)7 AON treatment	Cy3-2'-OMe PT $(CAG)_7$ FAM-2'-OMe PT $(CAG)_7$ Cy3-$(CAG)_7$	5-10 mainly nuclear CUG RNA foci are found in proliferating m. myoblasts and myotubes; 4-fold reduction in the percentage of cells with ribonuclear foci after (CAG)7 treatment; In gastrocnemius muscle of the HSA model every nucleus is filled with large aggregates of CUG foci and with individual foci; (CAG)7 reduces the number of CUG foci and causes a shift from nuclei packed with foci toward nuclei with smaller discrete foci	In HSA mice Mbnl1 colocalizes with CUG RNA foci and (CAG)7 treatment causes diffused distribution of Mbnl1	Mulders S.A.M. 2009
h. DM1 muscle biopsy; h. DM1 (800 and 2300 CTG) and DM2 myoblasts and fibroblasts; myotubes	Cy3-$(CAG)_{10}$ Cy3-$(CAGG)_{10}$	Expanded CUG and CCUG repeats are identified in nuclear foci in myoblasts and fibroblasts; the mean number of foci/nucleus is greater in myotubes than in myoblasts, 10.7±3.6 and 5.6±1.9, respectively	Endogenous MBNL1 and MBNL2 colocalize with CUG and CCUG foci; nucleocytoplasmic MBNL1 is partially sequestered into nuclear CUG foci in myoblasts and muscle tissue	Holt I 2009
COSM6 cells expressing 960 interrupted CTG repeat from DMPK 3'UTR minigene	Cy3-$(CAG)_5$-LNA	Multiple CUG ribonuclear foci in COSM6 expressing 960 CUG repeat	Exogenous MBNL1 and MBNL3 colocalize with CUG foci; at least one pair of zinc fingers of MBNL1 and MBNL3 is required for binding to CUG RNA in vivo and for colocalization with the foci	Grammatikakis I 2010
h. DM1 and DM2 muscle biopsies	FAM-$(CAG)_6$CA-2-*O*-Me FAM-$(CAGG)_5$-2-*O*-Me	Nuclear repeat RNA foci are formed in muscle fibers from DM1 and DM2 and the amount of nuclear foci between NCAM-positive and negative fibers is similar	MBNL1 colocalizes with CUG and CCUG RNA foci in DM1 and DM2 muscles but no colocalization is detected for CUGBP1	Santoro M 2010
h. myoblasts (800-2000 CTG); lentiviral vector hU7-(CAG)15 treatment	Cy3-PNA-(CAG)7	In myoblasts expressing 800 CUG over 50% nuclei have ≥5 foci, ~20% have a single focus and ~3% have no foci; in hU7-(CAG)15 treated myoblasts up to 60% myonuclei bear no foci, 25% nuclei have single faint focus	MBNL1 colocalizes with nuclear CUG foci; in hU7-(CAG)15 treated cells MBNL1 is released from sequestration by CUG foci and is redistributed; number of MBNL1 foci is reduced after the treatment	Francois V 2011
CCUG–DM2				
h. muscle biopsy (DM2 and DM1)	Cy3-2'-O-Me-$(CAGG)_{10}$ Cy3-2'-O-Me-$(CAG)_{10}$	1-5 intense CCUG foci/nucleus, no cytoplasmic foci; more foci/nucleus in DM2 than in DM1	NA	Liquori CL 2001

h. muscle biopsies (DM2 and DM1)	T.Red-2'-O-Me-$(CAGG)_5$ T. Red-2-O-Me-$(CAG)_6$-CA	1-3 intense foci/nucleus (DM2), no cytoplasmic foci; DM2 foci are smaller and less fluorescent with a CAG probe than with a CCAG probe; the CAG probe detects DM1 and DM2 foci and the CAGG probe identifies foci only in DM2; DM2 foci are present irrespective of severity of clinical involvement	NA	Cardani R 2004
h. muscle biopsies	Cy3-2'-O-Me-$(CAGG)_{10}$	Foci are present in muscle with histological symptoms of pathology and in muscle samples without abnormalities; no foci characteristics	NA	Savkur RS 2004
h. cardiac muscle	Cy3-$(CAGG)_5$	CCUG foci in cardiomyocytes; no foci characteristics	NA	Schoser BGH 2004
h. muscle biopsy (vastus lateralis) (>11,000 CCTG); heterozygous and homozygous myoblasts (2000-11000 CCTG); haploid cell lines established from fibroblasts (1000 CCTG)	FITC-$(CAGG)_{10}$ Cy3-$(CAGG)_{10}$ FITC-exon1 probe FITC-exon 5 probe Cy3-intron 1 probes	Intense nuclear CCUG foci do not contain other parts of intron 1 of *Znf9*; exon 1 or exon 5 sequences not found in nuclear foci; cy3-CAGG probe more sensitive than FITC-CCAG, which labels only large foci	NA	Margolis JM 2006
h. muscle biopsies (100-600 CCTG)	T. Red-2-O-Me-$(CAGG)_5$	>70% of nuclei with foci and >90% foci colocalize with MBNL1; expansion of 100 CCUG is long enough for nuclear retention and foci formation	MBNL1 colocalizes with CCUG foci	Lucchiari S 2008
h. muscle biopsies (biceps brachii) (1000-2500 CCTG); myoblasts and myotubes	T. Red-2-O-Me-$(CAGG)_5$	Myoblasts (multiple small foci, average 14 foci/nucleus with a range of 3-25/nucleus, low percentage of cells with more than 25 small foci/nucleus); myotubes (less foci but larger than in myoblasts); multinucleated syncytia (nuclei with different number and size of foci); elongated myotubes (fewer, larger and more fluorescent foci than in myoblasts); no cytoplasmic foci	MBNL1 colocalizes with CCUG foci in myoblasts, multinucleated syncytia, and elongated myotubes; each MBNL1 focus colocalizes with CCUG inclusion, but the opposite is not always true	Cardani R 2009
h. muscle biopsies (biceps brachii); myoblasts	T. Red-2-O-Me-$(CAGG)_5$	No characteristics	MBNL1 colocalizes with nuclear CCUG in myoblasts; some MBNL1-positive foci colocalize with snRNP Sm antigen and with hnRNP core proteins; SC-35, RNA polymerase II, CStF and PML proteins (no colocalization with MBNL1)	Perdoni F 2009
h. muscle biopsies (biceps brachii)	T. Red-2-O-Me-$(CAGG)_5$	CCUG foci in myonuclei of thawed and properly stored muscle samples	NA	Cardani R 2009