

Supplementary Table 2

Tissue/Cell Line/ Repeat Length	FISH RNA Probe	Mutant Repeat RNA Foci Characteristics	Protein Analyzed by IF and Type of Interaction with Repeat RNA Foci	Ref.
CGG-FXTAS				
isolated h. brain nuclei (frontal cortex) (113 CGG)	biotinylated riboprobes targeting coding (~1.1 kb), 5' UTR with CGG repeat (~0.3 kb) and 3'UTR (~2 kb) of FMR1 RNA	A single-spot nuclear signal of enlarged size with either of the antisense probes; 6-11% of probed nuclei had inclusions; 28 out of 507 nuclei were 5'UTR probe positive, 42 out of 466 were positive for coding probe and 50 out of 471 nuclei were 3'UTR probe positive	NA	Tassone F 2004
h. FXTAS brain (hippocampal area); KI mouse brain (98 CGG); kidney COS7, ovarian A172, neuronal PC12 and m. neurons primary cells were transfected with plasmids expressing (CGG) _n (n=20, 40, 60 and 100)	(CGG) _n -Cy3	Large, dynamic CGG nuclear aggregates with endogenously expressed CGG repeats; foci with as few as 40 exogenous CGG; foci expand over time; not all transfected cells support foci formation: foci in PC12, ovarian (SKOV3 and SW626) and COS7; no foci in A172, U-937, THP1, HeLa, HEK293, NG108-15, IMR-32, Neuro-2a, SH-SY5Y, SK-N-MC and in SK-N-SH cells	Sam68 colocalize with CGG foci; hnRNP G, SLM1, SLM2, hnRNP A, Tra2 β and MBNL1 proteins are recruited within CGG aggregates by interaction with Sam68; PML, coilin, SC35 colocalize partially with CGG foci	Sellier C 2010
CUG-SCA8				
HEK293 cells expressing ATXN8OS CR with 23, 88 and 157 combined CTA and CTG repeats	Cy3-(CAG)10 Cy5-ATXN8OS unique sequence	Distinct, mostly perinuclear foci in 88 and 157 ATXN8OS CR cells (results of both probes); no foci in 23 CR cell line	NA	Chen I-Ch 2009
h. brain tissue (cerebellum); SCA8 BAC Tg mouse model (116 CTG) (cerebellum)	Cy5-(CAG)10	CUG foci in h. cerebellar cortical layers; foci varied in distribution, size and number between analyzed cases; in a case with 1000 and 400 CTG single foci are found in the nuclei of molecular layer interneurons and the Bergmann glia surrounding the Purkinje cells in the granule cell layer; in a case with 109 CTG, foci are not reproducibly found and are only detected in a single molecular layer interneuron; CUG foci are readily detected in BAC mice with 116 repeats and have similar distribution in the cerebellar cortex as in SCA8 h. brain with 400 and 1000 repeats; no foci in mice with 11 CTG	CUG foci colocalize with MBNL1 in h. cerebellar sections (molecular layer interneurons); MBNL1 is not found in foci in SCA8 and DM1 Purkinje cells where MBNL1 is predominantly expressed in the cytoplasm, the same results are found in SCA8 Tg BAC mice	Daughters RS 2009
CAG EXPRESSING CELLS				
DM1 fibroblasts (GM3132-2000CTG and GM03987-500CTG); COSM6 expressing 960 interrupted CTG and CAG in exon 15 of truncated <i>DMPK</i>	Cy3-PNA-(CAG)5 Cy3-PNA-(CTG)5	Punctate CUG and CAG RNA foci are formed in COSM6 cells and in C2C12 myoblasts, NIH 3T3 fibroblasts and HeLa cells; similar number of foci/nucleus is formed with CTG and CAG expansions (~11 CAG and 13 CUG RNA foci) in COSM6	Endogenous (41 and 42 kDa) and exogenous MBNL1 (41 kDa) colocalize with CUG and CAG RNA foci in COSM6	Ho TH 2005
Tg SCA3 <i>Drosophila</i> expressing pure (CAG)100 and (CAG)250 and interrupted (CAG)78	FITC-(CAG)7 FITC-(CUG)7	Robust CUG RNA foci with 250 repeats; smaller foci in a limited number of nuclei with 250 CAG repeats	NA	Li L-B 2008
Tg <i>Caenorhabditis elegans</i> expressing 5,30,83,125 and 200 CAG and CTG repeat	Cy3-(CAG)10 Cy3-(CTG)10	CAG125 and CUG125 repeats form nuclear foci in the worm muscle	CeMBL colocalize with CAG and CUG foci and 92% of CAG RNA foci and 60% of CUG RNA foci are enriched with CeMBL	Wang LC 2011
h. HD fibroblasts GM04281(68 CAG) and GM09197 (151 CAG)	Cy3-LNA-(CTG)6-CA	Numerous nuclear CAG RNA aggregates with CAG 68 and 151 repeats	MBNL1 colocalize with CAG RNA foci	de Mezer M 2011
Tg mouse model expressing 23 and 200 CAG repeat; C2C12 expressing 58 and 200 CTG and CAG repeats	Cy3-(CTG)13 Cy3-(CAG)13	Punctate nuclear foci in m. skeletal muscle expressing 200 CAG but not 13 repeats; CUG and CAG RNA foci in C2C12 expressing 58 and 200 repeats and the proportion of nuclei with foci similar among CAG58, CAG200 and CUG200, 13.6%, 17.4% and 13.1%; respectively, number of foci/nucleus higher in CAG200 and CUG200 than in CAG58, 6.7, 6.5 and 3.4, respectively	Endogenous Mbn1 colocalize with CAG RNA foci in mice and with CAG and CUG RNA foci in m. myoblasts	Hsu RJ 2011
AUUCU-SCA10				
h. fibroblasts (1000 and 2000 ATTCT); Tg mouse brain (500 ATTCT); h. Sy5y cells and h. fibroblasts (ectopically expressing 500 ATTCT)	Cy3-(AGAAU) ₁₀	Large number of nuclear and cytoplasmic AUUCU RNA aggregates (fibroblasts; m. brain, Sy5y cells); aggregates are more abundant in 6-month-old than in 3-month-old mice; no aggregates in Sy5y expressing 12-25 AUUCU repeats	hnRNP K colocalizes with cytoplasmic and nuclear AUUCU RNA aggregates	White MC 2010
UGGAA-SCA31				
h. brain (cerebellar cortex)	DIG-LNA-(TTTTATTCTA) _{2,5} and (TAGAATAAAA) _{2,5}	RNA foci in Purkinje cells with (TTTTATTCTA) _{2,5} probe targeting sense (UAAAAUAGAA) _n transcript; 30-50% nuclei have foci (Purkinje cells); antisense transcript searched with (TAGAATAAAA) _{2,5} probe does not appear as foci	NA	Sato N 2009
CUG-HDL2				
h. brain tissue (44-55 CTG); HEK293 and HT22 cells ectopically expressing (CTG)53 from <i>JPH3</i> gene	T.Red-2-O-Me-(CAG) ₂₀ biotin-O-Me RNA 43mer targeting 3' to the <i>JPH3</i> CUG repeat (exon 2A) biotin-RNA 25mer targeting exon 2B of the <i>JPH3</i> DIG-2-O-Me-RNA 26mer targeting intron 1 of the <i>JPH3</i>	CUG RNA foci are detected with CAG probe; signals of CAG probe colocalize with 43mer probe and 25mer probe; less consistent colocalization is found with 26mer probe; 30% neurons (frontal cortex) have 1-13 CUG foci/nucleus; 15-20% neurons (striatum) have 5-20 foci/nucleus, and foci are bigger and brighter than in cortex; in hippocampus and dentate nuclei of cerebellum foci in <5% neurons; cells with 44 CTG fewer foci than with longer repeats; foci in HEK293 expressing 53 CUG but not 11 repeats	MBNL1 colocalizes with CUG RNA foci in frontal cortex and in HEK293 expressing 53 CUG repeats; CUGBP1 (no colocalization)	Rudnicki DD 2007