

RNA Binding Proteins in Neurodegenerative Diseases

Dysregulation of RNA-binding proteins (RBPs) is a hallmark of neurodegeneration:

- RBP aggregation
- Mutant RBP toxic gain of function
- Sequestration and loss of function of RBPs

ALS/FTD: FUS, EWS, TAF15
Transcription

Alternative Splicing

ALS/FTD: TDP43, FUS, TAF15, EWS
DM: MBNL1
POMA: NOVA
SMA: SMN

POMA: NOVA
OPMD: PABPN1
DM: MBNL1

Alternative Polyadenylation

Localization, Transport & Sequestration

ALS/FTD: TDP43, FUS, TAF15, EWS
ALS/SCA2: ATXN2
FXS: FMR1, MBNL1
SMA: SMN

Degradation & Turnover

Epilepsy, PD: HuR
ALS: MATR3

FXS: FMR1
DM: MBNL1
PD: PARK7

Translation

KEY **RBPs in Neurodegenerative Diseases: Roles in RNA Processing**

Bold | Disease

Italics | Implicated Gene

- ALS | Amyotrophic Lateral Sclerosis
- DM | Myotonic Dystrophy
- FTD | Frontotemporal Dementia
- FXS | Fragile X Syndrome
- POMA | Paraneoplastic Opsoclonus-Myoclonus Ataxia
- PD | Parkinson's Disease
- OPMD | Oculopharyngeal Muscular Dystrophy
- SCA2 | Spinocerebellar Ataxia Type 2
- SMA | Spinal Muscular Atrophy

Image adapted from Nussbacher et al., 2015. PMID: 25765321.

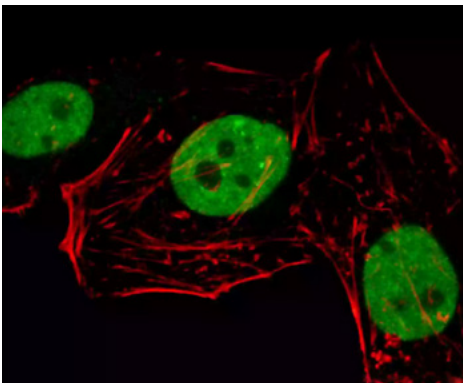
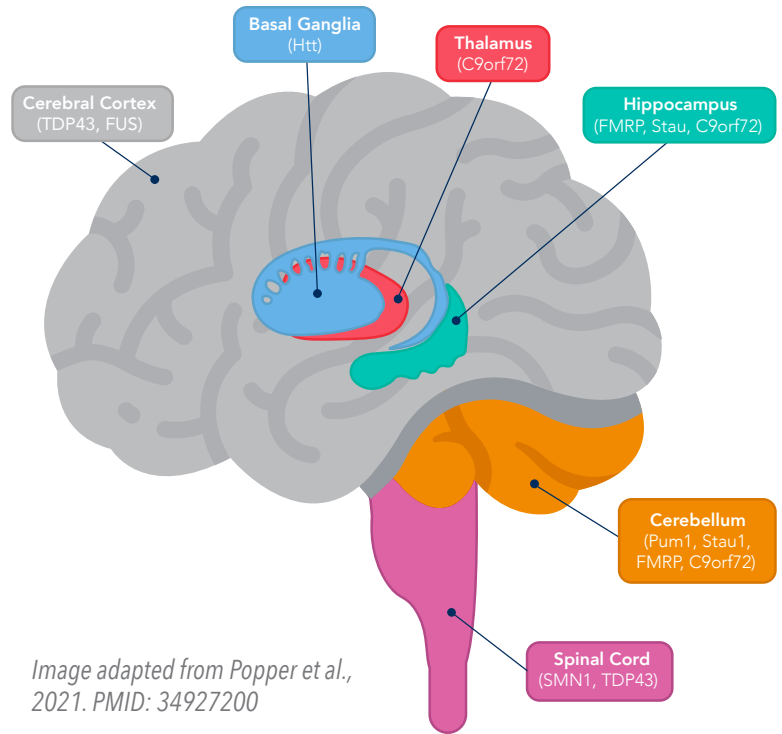
RNA Binding Protein Loss- and Gain-of-Function in Neurodegeneration

Loss-of-function

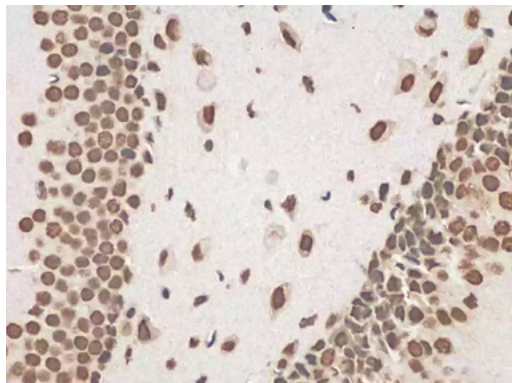
- FMRP
- FUS
- Hu proteins
- PTB1
- PUM1

Toxic gain-of-function

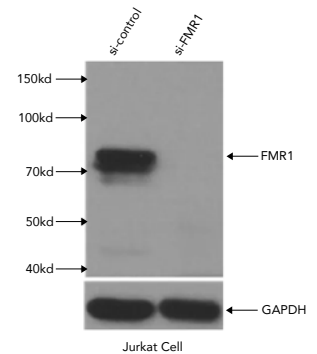
- ATXN2
- C9orf72 Di-peptide repeats
 - Poly-GP
 - Poly-PA
 - Poly-GR
 - Poly-PR
 - Poly-GA
- FUS
- TDP43



▲ **TDP-43 recombinant antibody:** IF of HepG2 cells using 80002-1-RR (green) and CoraLite555-phalloidin (red).



▲ **FUS/TLS monoclonal antibody:** IHC of mouse brain using 68262-1-Ig. Paraffin embedded. Tris-EDTA antigen retrieval.



▲ **FMR1 polyclonal antibody:** WB with 13755-1-AP. Knockdown validation in Jurkat cells.

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