

## Clinical Research in ALS & Related Disorders for Therapeutic Development

To reference CReATe in your publication please use the following NIH acknowledgement language: “The CReATe consortium (U54NS092091) is part of Rare Diseases Clinical Research Network (RDCRN), an initiative of the Office of Rare Diseases Research (ORDR), NCATS. This consortium is funded through collaboration between NCATS, and the NINDS.”

### Journal Articles

1. Turner MR, Benatar M. Ensuring continued progress in biomarkers for amyotrophic lateral sclerosis. *Muscle Nerve*. Jan 2015;51(1):14-18. PMID: 25288265, PMCID: PMC4270289
2. Rossor AM, Oates EC, Salter HK, et al. Phenotypic and molecular insights into spinal muscular atrophy due to mutations in BICD2. *Brain*. Feb 2015;138(Pt 2):293-310. PMID: 25497877, PMCID: PMC4306822
3. Gendron TF, van Blitterswijk M, Bieniek KF, et al. Cerebellar c9RAN proteins associate with clinical and neuropathological characteristics of C9ORF72 repeat expansion carriers. *Acta Neuropathol*. Oct 2015;130(4):559-573. PMID: 26350237, PMCID: PMC4575385
4. van Blitterswijk M, Gendron TF, Baker MC, et al. Novel clinical associations with specific C9ORF72 transcripts in patients with repeat expansions in C9ORF72. *Acta Neuropathol*. Oct 5 2015. PMID: 26437865, PMCID: PMC4655160
5. van Blitterswijk M, Rademakers R. Neurodegenerative disease: C9orf72 repeats compromise nucleocytoplasmic transport. *Nature reviews. Neurology*. Dec 2015;11(12):670-672. PMID: 26526532
6. Liu Y, Pattamatta A, Zu T, et al. C9orf72 BAC Mouse Model with Motor Deficits and Neurodegenerative Features of ALS/FTD. *Neuron*. 2016;90(3):521-534. PMID: 27112499
7. Benatar M, Boylan K, Jeromin A, et al. ALS biomarkers for therapy development: State of the field and future directions. *Muscle Nerve*. Feb 2016;53(2):169-182. PMID: 26574709, PMCID: PMC4718795
8. Esanov R, Belle KC, van Blitterswijk M, et al. C9orf72 promoter hypermethylation is reduced while hydroxymethylation is acquired during reprogramming of ALS patient cells. *Exp. Neurol*. Mar 2016;277:171-177. PMID: 26746986, PMCID: PMC4761318
9. Capturing clinical data to advance ALS research.  
[https://galaxy.epic.com/Redirect.aspx?DocumentID=3701881&Version=Epic 2017](https://galaxy.epic.com/Redirect.aspx?DocumentID=3701881&Version=Epic%202017), *EpicCare Ambulatory*, 2017
10. DeJesus-Hernandez M, Finch NA, Wang X, et al. In-depth clinico-pathological examination of RNA foci in a large cohort of C9ORF72 expansion carriers. *Acta Neuropathol*. 2017;134(2):255-269. PMID: 28508101, PMCID: PMC5508036

11. Finch NA, Wang X, Baker MC, et al. Abnormal expression of homeobox genes and transthyretin in C9ORF72 expansion carriers. *Neurology Genetics*. 2017;3(4):e161. PMID: 28660252, PMCID: PMC5479438
12. Gendron TF, Chew J, Stankowski JN, et al. Poly(GP) proteins are a useful pharmacodynamic marker for C9ORF72-associated amyotrophic lateral sclerosis. *Sci Transl Med*. 2017;9(383). PMID: 28356511
13. Gendron TF, Daugherty LM, Heckman MG, et al. Phosphorylated neurofilament heavy chain: A biomarker of survival for C9ORF72-associated amyotrophic lateral sclerosis. *Ann Neurol*. 2017;82(1):139-146. PMID: 28628244
14. Jacquier A, Delorme C, Belotti E, et al. Cryptic amyloidogenic elements in mutant NEFH causing Charcot-Marie-Tooth 2 trigger aggregates formation and neuronal death. *Acta neuropathologica communications*. 2017;5(1):55. PMID: 28709447, PMCID: PMC5513089
15. Mackenzie IR, Nicholson AM, Sarkar M, et al. TIA1 Mutations in Amyotrophic Lateral Sclerosis and Frontotemporal Dementia Promote Phase Separation and Alter Stress Granule Dynamics. *Neuron*. 2017;95(4):808-816.e809. PMID: 28817800
16. Murdock BJ, Zhou T, Kashlan SR, Little RJ, Goutman SA, Feldman EL. Correlation of Peripheral Immunity With Rapid Amyotrophic Lateral Sclerosis Progression. *JAMA neurology*. 2017. PMID: 28973548
17. Prudencio M, Gonzales PK, Cook CN, et al. Repetitive element transcripts are elevated in the brain of C9orf72 ALS/FTLD patients. *Hum Mol Genet*. 2017;26(17):3421-3431. PMID: 28637276
18. Sheppard SR, Wu J, Cardoso M, et al. Urinary p75ECD: A prognostic, disease progression, and pharmacodynamic biomarker in ALS. *Neurology*. 2017;88(12):1137-1143. PMID: 28228570, PMCID: PMC5373786
19. Strong MJ, Abrahams S, Goldstein LH, et al. Amyotrophic lateral sclerosis - frontotemporal spectrum disorder (ALS-FTSD): Revised diagnostic criteria. *Amyotrophic lateral sclerosis & frontotemporal degeneration*. 2017;18(3-4):153-174. PMID: 28054827
20. Simone R, Balendra R, Moens TG, et al. G-quadruplex-binding small molecules ameliorate C9orf72 FTD/ALS pathology in vitro and in vivo. *EMBO Mol Med*. 2018;10(1):22-31. PMID: 29113975, PMCID: PMC5760849