

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

### 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. 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
7. 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
8. 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
9. Shephard 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
10. 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