

## CLNE0020: Motoneurons, Neuromuscular Junctions and Associated Disease

View Online



Al-Chalabi, Ammar, Leonard H. van den Berg, and Jan Veldink. 'Gene Discovery in Amyotrophic Lateral Sclerosis: Implications for Clinical Management'. *Nature Reviews Neurology* 13.2 (2017): 96–104. Web.

Andreasson, Ulf, Kaj Blennow, and Henrik Zetterberg. 'Update on Ultrasensitive Technologies to Facilitate Research on Blood Biomarkers for Central Nervous System Disorders'. *Alzheimer's & Dementia: Diagnosis, Assessment & Disease Monitoring* 3 (2016): 98–102. Web.

Badders, Nisha M et al. 'Selective Modulation of the Androgen Receptor AF2 Domain Rescues Degeneration in Spinal Bulbar Muscular Atrophy'. *Nature Medicine* 24.4 (2018): 427–437. Web.

Beitel, Lenore K. et al. 'Mechanisms Mediating Spinal and Bulbar Muscular Atrophy: Investigations into Polyglutamine-Expanded Androgen Receptor Function and Dysfunction'. *Frontiers in Neurology* 4 (2013): n. pag. Web.

Belaya, Katsiaryna et al. 'Mutations in [\[redacted\]](#) Cause Congenital Myasthenic Syndrome and Bridge Myasthenic Disorders with Dystroglycanopathies'. *Brain* 138.9 (2015): 2493–2504. Web.

Benatar, Michael et al. 'ALS Biomarkers for Therapy Development: State of the Field and Future Directions'. *Muscle & Nerve* 53.2 (2016): 169–182. Web.

Berlowitz, David J et al. 'Identifying Who Will Benefit from Non-Invasive Ventilation in Amyotrophic Lateral Sclerosis/Motor Neurone Disease in a Clinical Cohort'. *Journal of Neurology, Neurosurgery & Psychiatry* 87.3 (2016): 280–286. Web.

Birnkrant, David J, Katharine Bushby, Carla M Bann, Susan D Apkon, Angela Blackwell, David Brumbaugh, et al. 'Diagnosis and Management of Duchenne Muscular Dystrophy, Part 1: Diagnosis, and Neuromuscular, Rehabilitation, Endocrine, and Gastrointestinal and Nutritional Management'. *The Lancet Neurology* 17.3 (2018): 251–267. Web.

Birnkrant, David J, Katharine Bushby, Carla M Bann, Benjamin A Alman, et al. 'Diagnosis and Management of Duchenne Muscular Dystrophy, Part 2: Respiratory, Cardiac, Bone Health, and Orthopaedic Management'. *The Lancet Neurology* 17.4 (2018): 347–361. Web.

Birnkrant, David J, Katharine Bushby, Carla M Bann, Susan D Apkon, Angela Blackwell, Mary K Colvin, et al. 'Diagnosis and Management of Duchenne Muscular Dystrophy, Part 3:

- Primary Care, Emergency Management, Psychosocial Care, and Transitions of Care across the Lifespan'. *The Lancet Neurology* 17.5 (2018): 445–455. Web.
- Bonanomi, D., and S. L. Pfaff. 'Motor Axon Pathfinding'. *Cold Spring Harbor Perspectives in Biology* 2.3 (2010): a001735–a001735. Web.
- Brownstone, Robert M., and Tuan V. Bui. 'Spinal Interneurons Providing Input to the Final Common Path during Locomotion'. *Breathe, Walk and Chew: The Neural Challenge: Part I*. Vol. 187. Elsevier, 2010. 81–95. Web.  
<<https://linkinghub.elsevier.com/retrieve/pii/B978044453613600006X>>.
- Carrì, Maria Teresa, Nadia D'Ambrosi, and Mauro Cozzolino. 'Pathways to Mitochondrial Dysfunction in ALS Pathogenesis'. *Biochemical and Biophysical Research Communications* 483.4 (2017): 1187–1193. Web.
- Cortes, Constanza J. et al. 'Muscle Expression of Mutant Androgen Receptor Accounts for Systemic and Motor Neuron Disease Phenotypes in Spinal and Bulbar Muscular Atrophy'. *Neuron* 82.2 (2014): 295–307. Web.
- Couratier, P. et al. 'Epidemiology of Amyotrophic Lateral Sclerosis: A Review of Literature'. *Revue Neurologique* 172.1 (2016): 37–45. Web.
- Crisp, Sarah J., Dimitri M. Kullmann, and Angela Vincent. 'Autoimmune Synaptopathies'. *Nature Reviews Neuroscience* 17.2 (2016): 103–117. Web.
- . 'Autoimmune Synaptopathies'. *Nature Reviews Neuroscience* 17.2 (2016): 103–117. Web.
- Cruz, Pedro M. Rodríguez, Jacqueline Palace, and David Beeson. 'Congenital Myasthenic Syndromes and the Neuromuscular Junction'. *Current Opinion in Neurology* 27.5 (2014): 566–575. Web.
- Darabid, Houssam, Anna P. Perez-Gonzalez, and Richard Robitaille. 'Neuromuscular Synaptogenesis: Coordinating Partners with Multiple Functions'. *Nature Reviews Neuroscience* 15.11 (2014): 703–718. Web.
- Dasen, Jeremy S., and Thomas M. Jessell. 'Chapter Six Hox Networks and the Origins of Motor Neuron Diversity'. *Hox Genes*. Vol. 88. Elsevier, 2009. 169–200. Web.  
<<https://linkinghub.elsevier.com/retrieve/pii/S007021530988006X>>.
- Drory, Vivian E. et al. 'The Value of Muscle Exercise in Patients with Amyotrophic Lateral Sclerosis'. *Journal of the Neurological Sciences* 191.1–2 (2001): 133–137. Web.
- Engel, Andrew G et al. 'Congenital Myasthenic Syndromes: Pathogenesis, Diagnosis, and Treatment'. *The Lancet Neurology* 14.4 (2015): 420–434. Web.
- Finkel, Richard S. et al. 'Diagnosis and Management of Spinal Muscular Atrophy: Part 2: Pulmonary and Acute Care; Medications, Supplements and Immunizations; Other Organ Systems; and Ethics'. *Neuromuscular Disorders* 28.3 (2018): 197–207. Web.
- FratTA, P. et al. 'Correlation of Clinical and Molecular Features in Spinal Bulbar Muscular

Atrophy'. *Neurology* 82.23 (2014): 2077–2084. Web.

Fuller, Geraint, and Mark Manford. *Neurology: An Illustrated Colour Text*. 3rd ed. Edinburgh: Churchill Livingstone Elsevier, 2010. Web.  
<[http://ucl.alma.exlibrisgroup.com/view/action/uresolver.do?operation=resolveService&package\\_service\\_id=3669595080004761&institutionId=4761&customerId=4760](http://ucl.alma.exlibrisgroup.com/view/action/uresolver.do?operation=resolveService&package_service_id=3669595080004761&institutionId=4761&customerId=4760)>.

Gendron, Tania F. et al. 'Poly(GP) Proteins Are a Useful Pharmacodynamic Marker for -Associated Amyotrophic Lateral Sclerosis'. *Science Translational Medicine* 9.383 (2017): n. pag. Web.

Gibson, Summer, and Verena Haringer. 'Amyotrophic Lateral Sclerosis: Clinical Perspectives'. *Orphan Drugs: Research and Reviews* (2015): n. pag. Web.

Gilhus, Nils E. 'Myasthenia Gravis'. *New England Journal of Medicine* 375.26 (2016): 2570–2581. Web.

Gordon, Elizabeth, Jonathan D. Rohrer, and Nick C. Fox. 'Advances in Neuroimaging in Frontotemporal Dementia'. *Journal of Neurochemistry* 138 (2016): 193–210. Web.

Harland, Richard. 'Neural Induction'. *Current Opinion in Genetics & Development* 10.4 (2000): 357–362. Web.

Harwood, Ceryl A., Christopher J. McDermott, and Pamela J. Shaw. 'Clinical Aspects of Motor Neurone Disease'. *Medicine* 40.10 (2012): 540–545. Web.

Hughes, JP et al. 'Principles of Early Drug Discovery'. *British Journal of Pharmacology* 162.6 (2011): 1239–1249. Web.

Jacobson, Leslie et al. 'Plasma from Human Mothers of Fetuses with Severe Arthrogryposis Multiplex Congenita Causes Deformities in Mice'. *Journal of Clinical Investigation* 103.7 (1999): 1031–1038. Web.

Jessell, Thomas M. 'Neuronal Specification in the Spinal Cord: Inductive Signals and Transcriptional Codes'. *Nature Reviews Genetics* 1.1 (2000): 20–29. Web.

Jones, Ross A. et al. 'Cellular and Molecular Anatomy of the Human Neuromuscular Junction'. *Cell Reports* 21.9 (2017): 2348–2356. Web.

Kanning, Kevin C., Artem Kaplan, and Christopher E. Henderson. 'Motor Neuron Diversity in Development and Disease'. *Annual Review of Neuroscience* 33.1 (2010): 409–440. Web.

Koneczny, Inga et al. 'MuSK Myasthenia Gravis IgG4 Disrupts the Interaction of LRP4 with MuSK but Both IgG4 and IgG1-3 Can Disperse Preformed Agrin-Independent AChR Clusters'. *PLoS ONE* 8.11 (2013): n. pag. Web.

Koneczny, Inga, Judith Cossins, and Angela Vincent. 'The Role of Muscle-Specific Tyrosine Kinase (MuSK) and Mystery of MuSK Myasthenia Gravis'. *Journal of Anatomy* 224.1 (2014): 29–35. Web.

Kusner, Linda L., and Henry J. Kaminski. 'Myasthenia Gravis'. *Neurobiology of Brain*

Disorders. Elsevier, 2015. 135–150. Web.

<<https://linkinghub.elsevier.com/retrieve/pii/B9780123982704000100>>.

Ladle, David R., Eline Pecho-Vrieseling, and Silvia Arber. 'Assembly of Motor Circuits in the Spinal Cord: Driven to Function by Genetic and Experience-Dependent Mechanisms'. *Neuron* 56.2 (2007): 270–283. Web.

Laurá, Matilde et al. 'Prevalence and Orthopedic Management of Foot and Ankle Deformities in Charcot-Marie-Tooth Disease'. *Muscle & Nerve* 57.2 (2018): 255–259. Web.

Leung, Doris G. Other Proven and Putative Autoimmune Disorders of the Peripheral Nervous System. Vol. 1. Oxford University Press, 2017. Web.

<<http://oxfordmedicine.com/view/10.1093/med/9780199937837.001.0001/med-9780199937837-chapter-98>>.

Li, Lei, Wen-Cheng Xiong, and Lin Mei. 'Neuromuscular Junction Formation, Aging, and Disorders'. *Annual Review of Physiology* 80.1 (2018): 159–188. Web.

Lieberman, Andrew P. et al. 'Peripheral Androgen Receptor Gene Suppression Rescues Disease in Mouse Models of Spinal and Bulbar Muscular Atrophy'. *Cell Reports* 7.3 (2014): 774–784. Web.

Lin, G., D. Mao, and H.J. Bellen. 'Amyotrophic Lateral Sclerosis Pathogenesis Converges on Defects in Protein Homeostasis Associated with TDP-43 Mislocalization and Proteasome-Mediated Degradation Overload'. *Fly Models of Human Diseases*. Vol. 121. Elsevier, 2017. 111–171. Web.

<<https://linkinghub.elsevier.com/retrieve/pii/S0070215316301454>>.

Lu, C.-H. et al. 'Neurofilament Light Chain: A Prognostic Biomarker in Amyotrophic Lateral Sclerosis'. *Neurology* 84.22 (2015): 2247–2257. Web.

Malik, B. et al. 'Absence of Disturbed Axonal Transport in Spinal and Bulbar Muscular Atrophy'. *Human Molecular Genetics* 20.9 (2011): 1776–1786. Web.

Malik, Bilal et al. 'Co-Induction of the Heat Shock Response Ameliorates Disease Progression in a Mouse Model of Human Spinal and Bulbar Muscular Atrophy: Implications for Therapy'. *Brain* 136.3 (2013): 926–943. Web.

Manzano, Raquel et al. 'Beyond Motor Neurons: Expanding the Clinical Spectrum in Kennedy's Disease'. *Journal of Neurology, Neurosurgery & Psychiatry* 89.8 (2018): 808–812. Web.

Maragakis, Nicholas J. 'What Can We Learn from the Edaravone Development Program for ALS?' *Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration* 18.sup1 (2017): 98–103. Web.

Mazzone, Elena S. et al. 'Revised Upper Limb Module for Spinal Muscular Atrophy: Development of a New Module'. *Muscle & Nerve* 55.6 (2017): 869–874. Web.

Mercuri, Eugenio et al. 'Diagnosis and Management of Spinal Muscular Atrophy: Part 1:

Recommendations for Diagnosis, Rehabilitation, Orthopedic and Nutritional Care'. *Neuromuscular Disorders* 28.2 (2018): 103–115. Web.

Meriggioli, Matthew N, and Donald B Sanders. 'Autoimmune Myasthenia Gravis: Emerging Clinical and Biological Heterogeneity'. *The Lancet Neurology* 8.5 (2009): 475–490. Web.

Milioto, Carmelo et al. 'Beta-Agonist Stimulation Ameliorates the Phenotype of Spinal and Bulbar Muscular Atrophy Mice and Patient-Derived Myotubes'. *Scientific Reports* 7.1 (2017): n. pag. Web.

Mitsumoto, Hiroshi, Benjamin R Brooks, and Vincenzo Silani. 'Clinical Trials in Amyotrophic Lateral Sclerosis: Why so Many Negative Trials and How Can Trials Be Improved?' *The Lancet Neurology* 13.11 (2014): 1127–1138. Web.

Monahan, Zachary, Frank Shewmaker, and Udai Bhan Pandey. 'Stress Granules at the Intersection of Autophagy and ALS'. *Brain Research* 1649 (2016): 189–200. Web.

Morgan, Sarah, and Richard W. Orrell. 'Pathogenesis of Amyotrophic Lateral Sclerosis'. *British Medical Bulletin* 119.1 (2016): 87–98. Web.

Morren, John A, and Nestor Galvez-Jimenez. 'Current and Prospective Disease-Modifying Therapies for Amyotrophic Lateral Sclerosis'. *Expert Opinion on Investigational Drugs* 21.3 (2012): 297–320. Web.

'Motor Neurone Disease: Assessment and Management | Guidance and Guidelines | NICE'. n. pag. Web. <<https://www.nice.org.uk/guidance/ng42>>.

Nishimune, Hiroshi et al. 'Laminins Promote Postsynaptic Maturation by an Autocrine Mechanism at the Neuromuscular Junction'. *The Journal of Cell Biology* 182.6 (2008): 1201–1215. Web.

O'Connor, Emily et al. 'Clinical and Research Strategies for Limb-Girdle Congenital Myasthenic Syndromes'. *Annals of the New York Academy of Sciences* 1412.1 (2018): 102–112. Web.

Orrell, Richard WBarclay, Chris. 'Diagnosis and Management of Motor Neurone Disease'. *Practitioner* 260 17–21. Web.  
<<https://search.proquest.com/docview/1844334383/64C39DCAF3D346C0PQ/1?accountid=14511>>.

Otto, Markus et al. 'Roadmap and Standard Operating Procedures for Biobanking and Discovery of Neurochemical Markers in ALS'. *Amyotrophic Lateral Sclerosis* 13.1 (2012): 1–10. Web.

Peragallo, Jason H. 'Pediatric Myasthenia Gravis'. *Seminars in Pediatric Neurology* 24.2 (2017): 116–121. Web.

Ramdharry, Gita M. et al. 'A Pilot Study of Proximal Strength Training in Charcot-Marie-Tooth Disease'. *Journal of the Peripheral Nervous System* 19.4 (2014): 328–332. Web.

- Ramsey, Danielle et al. 'Revised Hammersmith Scale for Spinal Muscular Atrophy: A SMA Specific Clinical Outcome Assessment Tool'. *PLOS ONE* 12.2 (2017): n. pag. Web.
- Reilly, Mary M. et al. '221st ENMC International Workshop': *Neuromuscular Disorders* 27.12 (2017): 1138–1142. Web.
- Renton, Alan E, Adriano Chiò, and Bryan J Traynor. 'State of Play in Amyotrophic Lateral Sclerosis Genetics'. *Nature Neuroscience* 17.1 (2014): 17–23. Web.
- Rodríguez Cruz, Pedro M. et al. 'Congenital Myopathies with Secondary Neuromuscular Transmission Defects; A Case Report and Review of the Literature'. *Neuromuscular Disorders* 24.12 (2014): 1103–1110. Web.
- Rodríguez Cruz, Pedro M., Jacqueline Palace, and David Beeson. 'Inherited Disorders of the Neuromuscular Junction: An Update'. *Journal of Neurology* 261.11 (2014): 2234–2243. Web.
- Rudolf, RÃ¼diger et al. 'Degeneration of Neuromuscular Junction in Age and Dystrophy'. *Frontiers in Aging Neuroscience* 6 (2014): n. pag. Web.
- Ruegsegger, Céline, and Smita Saxena. 'Proteostasis Impairment in ALS'. *Brain Research* 1648 (2016): 571–579. Web.
- Scoto, M et al. 'Therapeutic Approaches for Spinal Muscular Atrophy (SMA)'. *Gene Therapy* 24.9 (2017): 514–519. Web.
- Singhal, Neha, and Paul T. Martin. 'Role of Extracellular Matrix Proteins and Their Receptors in the Development of the Vertebrate Neuromuscular Junction'. *Developmental Neurobiology* 71.11 (2011): 982–1005. Web.
- Spillane, J. et al. 'Lambert-Eaton Syndrome IgG Inhibits Transmitter Release via P/Q Ca<sup>2+</sup> Channels'. *Neurology* 84.6 (2015): 575–579. Web.
- Spillane, J., D. J. Beeson, and D. M. Kullmann. 'Myasthenia and Related Disorders of the Neuromuscular Junction'. *Journal of Neurology, Neurosurgery & Psychiatry* 81.8 (2010): 850–857. Web.
- Viegas, Stuart et al. 'Passive and Active Immunization Models of MuSK-Ab Positive Myasthenia: Electrophysiological Evidence for Pre and Postsynaptic Defects'. *Experimental Neurology* 234.2 (2012): 506–512. Web.
- Vincent, Angela. 'Unravelling the Pathogenesis of Myasthenia Gravis'. *Nature Reviews Immunology* 2.10 (2002): 797–804. Web.
- 'Volume 58, Issue 3, March 2016'. Volume 58, Issue 3, March 2016 n. pag. Web. <<https://link.springer.com/journal/12031/58/3>>.
- Westerberg, Elisabet et al. 'The Impact of Physical Exercise on Neuromuscular Function in Myasthenia Gravis Patients'. *Medicine* 97.31 (2018): n. pag. Web.
- Wolfe, Gil I. et al. 'Randomized Trial of Thymectomy in Myasthenia Gravis'. *New England*

Journal of Medicine 375.6 (2016): 511–522. Web.

Woollacott, Ione O. C., and Jonathan D. Rohrer. 'The Clinical Spectrum of Sporadic and Familial Forms of Frontotemporal Dementia'. Journal of Neurochemistry 138 (2016): 6–31. Web.