

CLNE0020: Motoneurons, Neuromuscular Junctions and Associated Disease

View Online



[1]

Al-Chalabi, A. et al. 2017. Gene discovery in amyotrophic lateral sclerosis: implications for clinical management. *Nature Reviews Neurology*. 13, 2 (Feb. 2017), 96–104. DOI:<https://doi.org/10.1038/nrneurol.2016.182>.

[2]

Andreasson, U. et al. 2016. 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. DOI:<https://doi.org/10.1016/j.dadm.2016.05.005>.

[3]

Badders, N.M. et al. 2018. Selective modulation of the androgen receptor AF2 domain rescues degeneration in spinal bulbar muscular atrophy. *Nature Medicine*. 24, 4 (Mar. 2018), 427–437. DOI:<https://doi.org/10.1038/nm.4500>.

[4]

Beitel, L.K. et al. 2013. Mechanisms Mediating Spinal and Bulbar Muscular Atrophy: Investigations into Polyglutamine-Expanded Androgen Receptor Function and Dysfunction. *Frontiers in Neurology*. 4, (2013). DOI:<https://doi.org/10.3389/fneur.2013.00053>.

[5]

Belaya, K. et al. 2015. Mutations in [\[redacted\]](#) cause congenital myasthenic syndrome and bridge myasthenic disorders with dystroglycanopathies. *Brain*. 138, 9 (Sep. 2015), 2493–2504. DOI:<https://doi.org/10.1093/brain/awv185>.

[6]

Benatar, M. et al. 2016. ALS biomarkers for therapy development: State of the field and future directions. *Muscle & Nerve*. 53, 2 (Feb. 2016), 169–182.
DOI:<https://doi.org/10.1002/mus.24979>.

[7]

Berlowitz, D.J. et al. 2016. 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 (Mar. 2016), 280–286.
DOI:<https://doi.org/10.1136/jnnp-2014-310055>.

[8]

Birnkrant, D.J. et al. 2018. Diagnosis and management of Duchenne muscular dystrophy, part 1: diagnosis, and neuromuscular, rehabilitation, endocrine, and gastrointestinal and nutritional management. *The Lancet Neurology*. 17, 3 (Mar. 2018), 251–267.
DOI:[https://doi.org/10.1016/S1474-4422\(18\)30024-3](https://doi.org/10.1016/S1474-4422(18)30024-3).

[9]

Birnkrant, D.J. et al. 2018. Diagnosis and management of Duchenne muscular dystrophy, part 2: respiratory, cardiac, bone health, and orthopaedic management. *The Lancet Neurology*. 17, 4 (Apr. 2018), 347–361.
DOI:[https://doi.org/10.1016/S1474-4422\(18\)30025-5](https://doi.org/10.1016/S1474-4422(18)30025-5).

[10]

Birnkrant, D.J. et al. 2018. 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 (May 2018), 445–455.
DOI:[https://doi.org/10.1016/S1474-4422\(18\)30026-7](https://doi.org/10.1016/S1474-4422(18)30026-7).

[11]

Bonanomi, D. and Pfaff, S.L. 2010. Motor Axon Pathfinding. *Cold Spring Harbor Perspectives in Biology*. 2, 3 (Mar. 2010), a001735–a001735.
DOI:<https://doi.org/10.1101/cshperspect.a001735>.

[12]

Brownstone, R.M. and Bui, T.V. 2010. Spinal interneurons providing input to the final common path during locomotion. *Breathe, Walk and Chew: The Neural Challenge: Part I*. Elsevier. 81–95.

[13]

Carrì, M.T. et al. 2017. Pathways to mitochondrial dysfunction in ALS pathogenesis. *Biochemical and Biophysical Research Communications*. 483, 4 (Feb. 2017), 1187–1193. DOI:<https://doi.org/10.1016/j.bbrc.2016.07.055>.

[14]

Cortes, C.J. et al. 2014. Muscle Expression of Mutant Androgen Receptor Accounts for Systemic and Motor Neuron Disease Phenotypes in Spinal and Bulbar Muscular Atrophy. *Neuron*. 82, 2 (Apr. 2014), 295–307. DOI:<https://doi.org/10.1016/j.neuron.2014.03.001>.

[15]

Couratier, P. et al. 2016. Epidemiology of amyotrophic lateral sclerosis: A review of literature. *Revue Neurologique*. 172, 1 (Jan. 2016), 37–45. DOI:<https://doi.org/10.1016/j.neurol.2015.11.002>.

[16]

Crisp, S.J. et al. 2016. Autoimmune synaptopathies. *Nature Reviews Neuroscience*. 17, 2 (Feb. 2016), 103–117. DOI:<https://doi.org/10.1038/nrn.2015.27>.

[17]

Crisp, S.J. et al. 2016. Autoimmune synaptopathies. *Nature Reviews Neuroscience*. 17, 2 (Feb. 2016), 103–117. DOI:<https://doi.org/10.1038/nrn.2015.27>.

[18]

Cruz, P.M.R. et al. 2014. Congenital myasthenic syndromes and the neuromuscular

junction. *Current Opinion in Neurology*. 27, 5 (Oct. 2014), 566–575.
DOI:<https://doi.org/10.1097/WCO.0000000000000134>.

[19]

Darabid, H. et al. 2014. Neuromuscular synaptogenesis: coordinating partners with multiple functions. *Nature Reviews Neuroscience*. 15, 11 (Nov. 2014), 703–718.
DOI:<https://doi.org/10.1038/nrn3821>.

[20]

Dasen, J.S. and Jessell, T.M. 2009. Chapter Six Hox Networks and the Origins of Motor Neuron Diversity. *Hox Genes*. Elsevier. 169–200.

[21]

Drory, V.E. et al. 2001. The value of muscle exercise in patients with amyotrophic lateral sclerosis. *Journal of the Neurological Sciences*. 191, 1–2 (Oct. 2001), 133–137.
DOI:[https://doi.org/10.1016/S0022-510X\(01\)00610-4](https://doi.org/10.1016/S0022-510X(01)00610-4).

[22]

Engel, A.G. et al. 2015. Congenital myasthenic syndromes: pathogenesis, diagnosis, and treatment. *The Lancet Neurology*. 14, 4 (Apr. 2015), 420–434.
DOI:[https://doi.org/10.1016/S1474-4422\(14\)70201-7](https://doi.org/10.1016/S1474-4422(14)70201-7).

[23]

Finkel, R.S. et al. 2018. 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 (Mar. 2018), 197–207.
DOI:<https://doi.org/10.1016/j.nmd.2017.11.004>.

[24]

Fratta, P. et al. 2014. Correlation of clinical and molecular features in spinal bulbar muscular atrophy. *Neurology*. 82, 23 (Jun. 2014), 2077–2084.
DOI:<https://doi.org/10.1212/WNL.0000000000000507>.

[25]

Fuller, G. and Manford, M. 2010. Neurology: an illustrated colour text. Churchill Livingstone Elsevier.

[26]

Gendron, T.F. et al. 2017. Poly(GP) proteins are a useful pharmacodynamic marker for -associated amyotrophic lateral sclerosis. *Science Translational Medicine*. 9, 383 (Mar. 2017). DOI:<https://doi.org/10.1126/scitranslmed.aai7866>.

[27]

Gibson, S. and Haringer, V. 2015. Amyotrophic lateral sclerosis: clinical perspectives. *Orphan Drugs: Research and Reviews*. (Apr. 2015). DOI:<https://doi.org/10.2147/ODRR.S63585>.

[28]

Gilhus, N.E. 2016. Myasthenia Gravis. *New England Journal of Medicine*. 375, 26 (Dec. 2016), 2570–2581. DOI:<https://doi.org/10.1056/NEJMra1602678>.

[29]

Gordon, E. et al. 2016. Advances in neuroimaging in frontotemporal dementia. *Journal of Neurochemistry*. 138, (Aug. 2016), 193–210. DOI:<https://doi.org/10.1111/jnc.13656>.

[30]

Harland, R. 2000. Neural induction. *Current Opinion in Genetics & Development*. 10, 4 (Aug. 2000), 357–362. DOI:[https://doi.org/10.1016/S0959-437X\(00\)00096-4](https://doi.org/10.1016/S0959-437X(00)00096-4).

[31]

Harwood, C.A. et al. 2012. Clinical aspects of motor neurone disease. *Medicine*. 40, 10 (Oct. 2012), 540–545. DOI:<https://doi.org/10.1016/j.mpmed.2012.07.003>.

[32]

Hughes, J. et al. 2011. Principles of early drug discovery. *British Journal of Pharmacology*. 162, 6 (Mar. 2011), 1239–1249. DOI:<https://doi.org/10.1111/j.1476-5381.2010.01127.x>.

[33]

Jacobson, L. et al. 1999. Plasma from human mothers of fetuses with severe arthrogryposis multiplex congenita causes deformities in mice. *Journal of Clinical Investigation*. 103, 7 (Apr. 1999), 1031–1038. DOI:<https://doi.org/10.1172/JCI5943>.

[34]

Jessell, T.M. 2000. Neuronal specification in the spinal cord: inductive signals and transcriptional codes. *Nature Reviews Genetics*. 1, 1 (Oct. 2000), 20–29. DOI:<https://doi.org/10.1038/35049541>.

[35]

Jones, R.A. et al. 2017. Cellular and Molecular Anatomy of the Human Neuromuscular Junction. *Cell Reports*. 21, 9 (Nov. 2017), 2348–2356. DOI:<https://doi.org/10.1016/j.celrep.2017.11.008>.

[36]

Kanning, K.C. et al. 2010. Motor Neuron Diversity in Development and Disease. *Annual Review of Neuroscience*. 33, 1 (Jun. 2010), 409–440. DOI:<https://doi.org/10.1146/annurev.neuro.051508.135722>.

[37]

Koneczny, I. et al. 2013. 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 (Nov. 2013). DOI:<https://doi.org/10.1371/journal.pone.0080695>.

[38]

Koneczny, I. et al. 2014. The role of muscle-specific tyrosine kinase (MuSK) and mystery of MuSK myasthenia gravis. *Journal of Anatomy*. 224, 1 (Jan. 2014), 29–35.
DOI:<https://doi.org/10.1111/joa.12034>.

[39]

Kusner, L.L. and Kaminski, H.J. 2015. Myasthenia Gravis. *Neurobiology of Brain Disorders*. Elsevier. 135–150.

[40]

Ladle, D.R. et al. 2007. Assembly of Motor Circuits in the Spinal Cord: Driven to Function by Genetic and Experience-Dependent Mechanisms. *Neuron*. 56, 2 (Oct. 2007), 270–283.
DOI:<https://doi.org/10.1016/j.neuron.2007.09.026>.

[41]

Laurá, M. et al. 2018. Prevalence and orthopedic management of foot and ankle deformities in Charcot-Marie-Tooth disease. *Muscle & Nerve*. 57, 2 (Feb. 2018), 255–259.
DOI:<https://doi.org/10.1002/mus.25724>.

[42]

Leung, D.G. 2017. Other Proven and Putative Autoimmune Disorders of the Peripheral Nervous System. Oxford University Press.

[43]

Li, L. et al. 2018. Neuromuscular Junction Formation, Aging, and Disorders. *Annual Review of Physiology*. 80, 1 (Feb. 2018), 159–188.
DOI:<https://doi.org/10.1146/annurev-physiol-022516-034255>.

[44]

Lieberman, A.P. et al. 2014. Peripheral Androgen Receptor Gene Suppression Rescues Disease in Mouse Models of Spinal and Bulbar Muscular Atrophy. *Cell Reports*. 7, 3 (May 2014), 774–784. DOI:<https://doi.org/10.1016/j.celrep.2014.02.008>.

[45]

Lin, G. et al. 2017. 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*. Elsevier. 111–171.

[46]

Lu, C.-H. et al. 2015. Neurofilament light chain: A prognostic biomarker in amyotrophic lateral sclerosis. *Neurology*. 84, 22 (Jun. 2015), 2247–2257.
DOI:<https://doi.org/10.1212/WNL.0000000000001642>.

[47]

Malik, B. et al. 2011. Absence of disturbed axonal transport in spinal and bulbar muscular atrophy. *Human Molecular Genetics*. 20, 9 (May 2011), 1776–1786.
DOI:<https://doi.org/10.1093/hmg/ddr061>.

[48]

Malik, B. et al. 2013. 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 (Mar. 2013), 926–943. DOI:<https://doi.org/10.1093/brain/aws343>.

[49]

Manzano, R. et al. 2018. Beyond motor neurons: expanding the clinical spectrum in Kennedy's disease. *Journal of Neurology, Neurosurgery & Psychiatry*. 89, 8 (Aug. 2018), 808–812. DOI:<https://doi.org/10.1136/jnnp-2017-316961>.

[50]

Maragakis, N.J. 2017. What can we learn from the edaravone development program for ALS? *Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration*. 18, sup1 (Oct. 2017), 98–103. DOI:<https://doi.org/10.1080/21678421.2017.1361446>.

[51]

Mazzone, E.S. et al. 2017. Revised upper limb module for spinal muscular atrophy: Development of a new module. *Muscle & Nerve*. 55, 6 (Jun. 2017), 869–874. DOI:<https://doi.org/10.1002/mus.25430>.

[52]

Mercuri, E. et al. 2018. Diagnosis and management of spinal muscular atrophy: Part 1: Recommendations for diagnosis, rehabilitation, orthopedic and nutritional care. *Neuromuscular Disorders*. 28, 2 (Feb. 2018), 103–115. DOI:<https://doi.org/10.1016/j.nmd.2017.11.005>.

[53]

Meriggioli, M.N. and Sanders, D.B. 2009. Autoimmune myasthenia gravis: emerging clinical and biological heterogeneity. *The Lancet Neurology*. 8, 5 (May 2009), 475–490. DOI:[https://doi.org/10.1016/S1474-4422\(09\)70063-8](https://doi.org/10.1016/S1474-4422(09)70063-8).

[54]

Milioto, C. et al. 2017. Beta-agonist stimulation ameliorates the phenotype of spinal and bulbar muscular atrophy mice and patient-derived myotubes. *Scientific Reports*. 7, 1 (Dec. 2017). DOI:<https://doi.org/10.1038/srep41046>.

[55]

Mitsumoto, H. et al. 2014. Clinical trials in amyotrophic lateral sclerosis: why so many negative trials and how can trials be improved? *The Lancet Neurology*. 13, 11 (Nov. 2014), 1127–1138. DOI:[https://doi.org/10.1016/S1474-4422\(14\)70129-2](https://doi.org/10.1016/S1474-4422(14)70129-2).

[56]

Monahan, Z. et al. 2016. Stress granules at the intersection of autophagy and ALS. *Brain Research*. 1649, (Oct. 2016), 189–200. DOI:<https://doi.org/10.1016/j.brainres.2016.05.022>.

[57]

Morgan, S. and Orrell, R.W. 2016. Pathogenesis of amyotrophic lateral sclerosis. *British Medical Bulletin*. 119, 1 (Sep. 2016), 87–98. DOI:<https://doi.org/10.1093/bmb/ldw026>.

[58]

Morren, J.A. and Galvez-Jimenez, N. 2012. Current and prospective disease-modifying therapies for amyotrophic lateral sclerosis. *Expert Opinion on Investigational Drugs*. 21, 3 (Mar. 2012), 297–320. DOI:<https://doi.org/10.1517/13543784.2012.657303>.

[59]

Nishimune, H. et al. 2008. Laminins promote postsynaptic maturation by an autocrine mechanism at the neuromuscular junction. *The Journal of Cell Biology*. 182, 6 (Sep. 2008), 1201–1215. DOI:<https://doi.org/10.1083/jcb.200805095>.

[60]

O'Connor, E. et al. 2018. Clinical and research strategies for limb-girdle congenital myasthenic syndromes. *Annals of the New York Academy of Sciences*. 1412, 1 (Jan. 2018), 102–112. DOI:<https://doi.org/10.1111/nyas.13520>.

[61]

Orrell, Richard WBarclay, Chris Diagnosis and management of motor neurone disease. *Practitioner*. 260, 17–21.

[62]

Otto, M. et al. 2012. Roadmap and standard operating procedures for biobanking and discovery of neurochemical markers in ALS. *Amyotrophic Lateral Sclerosis*. 13, 1 (Jan. 2012), 1–10. DOI:<https://doi.org/10.3109/17482968.2011.627589>.

[63]

Peragallo, J.H. 2017. Pediatric Myasthenia Gravis. *Seminars in Pediatric Neurology*. 24, 2 (May 2017), 116–121. DOI:<https://doi.org/10.1016/j.spen.2017.04.003>.

[64]

Ramdharry, G.M. et al. 2014. A pilot study of proximal strength training in

Charcot-Marie-Tooth disease. *Journal of the Peripheral Nervous System*. 19, 4 (Dec. 2014), 328–332. DOI:<https://doi.org/10.1111/jns.12100>.

[65]

Ramsey, D. et al. 2017. Revised Hammersmith Scale for spinal muscular atrophy: A SMA specific clinical outcome assessment tool. *PLOS ONE*. 12, 2 (Feb. 2017). DOI:<https://doi.org/10.1371/journal.pone.0172346>.

[66]

Reilly, M.M. et al. 2017. 221st ENMC International Workshop: Neuromuscular Disorders. 27, 12 (Dec. 2017), 1138–1142. DOI:<https://doi.org/10.1016/j.nmd.2017.09.005>.

[67]

Renton, A.E. et al. 2014. State of play in amyotrophic lateral sclerosis genetics. *Nature Neuroscience*. 17, 1 (Jan. 2014), 17–23. DOI:<https://doi.org/10.1038/nn.3584>.

[68]

Rodríguez Cruz, P.M. et al. 2014. Congenital myopathies with secondary neuromuscular transmission defects; A case report and review of the literature. *Neuromuscular Disorders*. 24, 12 (Dec. 2014), 1103–1110. DOI:<https://doi.org/10.1016/j.nmd.2014.07.005>.

[69]

Rodríguez Cruz, P.M. et al. 2014. Inherited disorders of the neuromuscular junction: an update. *Journal of Neurology*. 261, 11 (Nov. 2014), 2234–2243. DOI:<https://doi.org/10.1007/s00415-014-7520-7>.

[70]

Rudolf, R. et al. 2014. Degeneration of Neuromuscular Junction in Age and Dystrophy. *Frontiers in Aging Neuroscience*. 6, (May 2014). DOI:<https://doi.org/10.3389/fnagi.2014.00099>.

[71]

Ruegsegger, C. and Saxena, S. 2016. Proteostasis impairment in ALS. *Brain Research*. 1648, (Oct. 2016), 571–579. DOI:<https://doi.org/10.1016/j.brainres.2016.03.032>.

[72]

Scoto, M. et al. 2017. Therapeutic approaches for spinal muscular atrophy (SMA). *Gene Therapy*. 24, 9 (Sep. 2017), 514–519. DOI:<https://doi.org/10.1038/gt.2017.45>.

[73]

Singhal, N. and Martin, P.T. 2011. Role of extracellular matrix proteins and their receptors in the development of the vertebrate neuromuscular junction. *Developmental Neurobiology*. 71, 11 (Nov. 2011), 982–1005. DOI:<https://doi.org/10.1002/dneu.20953>.

[74]

Spillane, J. et al. 2015. Lambert-Eaton syndrome IgG inhibits transmitter release via P/Q Ca²⁺ channels. *Neurology*. 84, 6 (Feb. 2015), 575–579. DOI:<https://doi.org/10.1212/WNL.0000000000001225>.

[75]

Spillane, J. et al. 2010. Myasthenia and related disorders of the neuromuscular junction. *Journal of Neurology, Neurosurgery & Psychiatry*. 81, 8 (Aug. 2010), 850–857. DOI:<https://doi.org/10.1136/jnnp.2008.169367>.

[76]

Viegas, S. et al. 2012. Passive and active immunization models of MuSK-Ab positive myasthenia: Electrophysiological evidence for pre and postsynaptic defects. *Experimental Neurology*. 234, 2 (Apr. 2012), 506–512. DOI:<https://doi.org/10.1016/j.expneurol.2012.01.025>.

[77]

Vincent, A. 2002. Unravelling the pathogenesis of myasthenia gravis. *Nature Reviews Immunology*. 2, 10 (Oct. 2002), 797–804. DOI:<https://doi.org/10.1038/nri916>.

[78]

Westerberg, E. et al. 2018. The impact of physical exercise on neuromuscular function in Myasthenia gravis patients. *Medicine*. 97, 31 (Aug. 2018).
DOI:<https://doi.org/10.1097/MD.00000000000011510>.

[79]

Wolfe, G.I. et al. 2016. Randomized Trial of Thymectomy in Myasthenia Gravis. *New England Journal of Medicine*. 375, 6 (Aug. 2016), 511-522.
DOI:<https://doi.org/10.1056/NEJMoa1602489>.

[80]

Woollacott, I.O.C. and Rohrer, J.D. 2016. The clinical spectrum of sporadic and familial forms of frontotemporal dementia. *Journal of Neurochemistry*. 138, (Aug. 2016), 6-31.
DOI:<https://doi.org/10.1111/jnc.13654>.

[81]

Motor neurone disease: assessment and management | Guidance and guidelines | NICE.

[82]

Volume 58, Issue 3, March 2016. Volume 58, Issue 3, March 2016.