

## CLNE0020: Motoneurons, Neuromuscular Junctions and Associated Disease

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- Al-Chalabi, A., van den Berg, L. H., & Veldink, J. (2017). Gene discovery in amyotrophic lateral sclerosis: implications for clinical management. *Nature Reviews Neurology*, 13(2), 96–104. <https://doi.org/10.1038/nrneurol.2016.182>
- Andreasson, U., Blennow, K., & Zetterberg, H. (2016). Update on ultrasensitive technologies to facilitate research on blood biomarkers for central nervous system disorders. *Alzheimer's & Dementia: Diagnosis, Assessment & Disease Monitoring*, 3, 98–102. <https://doi.org/10.1016/j.dadm.2016.05.005>
- Badders, N. M., Korff, A., Miranda, H. C., Vuppala, P. K., Smith, R. B., Winborn, B. J., Quemain, E. R., Sopher, B. L., Dearman, J., Messing, J., Kim, N. C., Moore, J., Freibaum, B. D., Kanagaraj, A. P., Fan, B., Tillman, H., Chen, P.-C., Wang, Y., III, B. B. F., ... Taylor, J. P. (2018). Selective modulation of the androgen receptor AF2 domain rescues degeneration in spinal bulbar muscular atrophy. *Nature Medicine*, 24(4), 427–437. <https://doi.org/10.1038/nm.4500>
- Beitel, L. K., Alvarado, C., Mokhtar, S., Paliouras, M., & Trifiro, M. (2013). Mechanisms Mediating Spinal and Bulbar Muscular Atrophy: Investigations into Polyglutamine-Expanded Androgen Receptor Function and Dysfunction. *Frontiers in Neurology*, 4. <https://doi.org/10.3389/fneur.2013.00053>
- Belaya, K., Rodríguez Cruz, P. M., Liu, W. W., Maxwell, S., McGowan, S., Farrugia, M. E., Petty, R., Walls, T. J., Sedghi, M., Basiri, K., Yue, W. W., Sarkozy, A., Bertoli, M., Pitt, M., Kennett, R., Schaefer, A., Bushby, K., Parton, M., Lochmüller, H., ... Beeson, D. (2015). Mutations in *CHCHD10* cause congenital myasthenic syndrome and bridge myasthenic disorders with dystroglycanopathies. *Brain*, 138(9), 2493–2504. <https://doi.org/10.1093/brain/awv185>
- Benatar, M., Boylan, K., Jeromin, A., Rutkove, S. B., Berry, J., Atassi, N., & Bruijn, L. (2016). ALS biomarkers for therapy development: State of the field and future directions. *Muscle & Nerve*, 53(2), 169–182. <https://doi.org/10.1002/mus.24979>
- Berlowitz, D. J., Howard, M. E., Fiore, J. F., Vander Hoorn, S., O'Donoghue, F. J., Westlake, J., Smith, A., Beer, F., Mathers, S., & Talman, P. (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), 280–286. <https://doi.org/10.1136/jnnp-2014-310055>
- Birnkrant, D. J., Bushby, K., Bann, C. M., Alman, B. A., Apkon, S. D., Blackwell, A., Case, L. E., Cripe, L., Hadjiyannakis, S., Olson, A. K., Sheehan, D. W., Bolen, J., Weber, D. R., &

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

Birnkrant, D. J., Bushby, K., Bann, C. M., Apkon, S. D., Blackwell, A., Brumbaugh, D., Case, L. E., Clemens, P. R., Hadjiyannakis, S., Pandya, S., Street, N., Tomezsko, J., Wagner, K. R., Ward, L. M., & Weber, D. R. (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), 251–267. [https://doi.org/10.1016/S1474-4422\(18\)30024-3](https://doi.org/10.1016/S1474-4422(18)30024-3)

Birnkrant, D. J., Bushby, K., Bann, C. M., Apkon, S. D., Blackwell, A., Colvin, M. K., Cripe, L., Herron, A. R., Kennedy, A., Kinnett, K., Naprawa, J., Noritz, G., Poysky, J., Street, N., Trout, C. J., Weber, D. R., & Ward, L. M. (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), 445–455. [https://doi.org/10.1016/S1474-4422\(18\)30026-7](https://doi.org/10.1016/S1474-4422(18)30026-7)

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

Brownstone, R. M., & Bui, T. V. (2010). Spinal interneurons providing input to the final common path during locomotion. In *Breathe, Walk and Chew: The Neural Challenge: Part I* (Vol. 187, pp. 81–95). Elsevier. <https://doi.org/10.1016/B978-0-444-53613-6.00006-X>

Carrì, M. T., D'Ambrosi, N., & Cozzolino, M. (2017). Pathways to mitochondrial dysfunction in ALS pathogenesis. *Biochemical and Biophysical Research Communications*, 483(4), 1187–1193. <https://doi.org/10.1016/j.bbrc.2016.07.055>

Cortes, C. J., Ling, S.-C., Guo, L. T., Hung, G., Tsunemi, T., Ly, L., Tokunaga, S., Lopez, E., Sopher, B. L., Bennett, C. F., Shelton, G. D., Cleveland, D. W., & La Spada, A. R. (2014). Muscle Expression of Mutant Androgen Receptor Accounts for Systemic and Motor Neuron Disease Phenotypes in Spinal and Bulbar Muscular Atrophy. *Neuron*, 82(2), 295–307. <https://doi.org/10.1016/j.neuron.2014.03.001>

Couratier, P., Corcia, P., Lautrette, G., Nicol, M., Preux, P.-M., & Marin, B. (2016). Epidemiology of amyotrophic lateral sclerosis: A review of literature. *Revue Neurologique*, 172(1), 37–45. <https://doi.org/10.1016/j.neurol.2015.11.002>

Crisp, S. J., Kullmann, D. M., & Vincent, A. (2016a). Autoimmune synaptopathies. *Nature Reviews Neuroscience*, 17(2), 103–117. <https://doi.org/10.1038/nrn.2015.27>

Crisp, S. J., Kullmann, D. M., & Vincent, A. (2016b). Autoimmune synaptopathies. *Nature Reviews Neuroscience*, 17(2), 103–117. <https://doi.org/10.1038/nrn.2015.27>

Cruz, P. M. R., Palace, J., & Beeson, D. (2014). Congenital myasthenic syndromes and the neuromuscular junction. *Current Opinion in Neurology*, 27(5), 566–575. <https://doi.org/10.1097/WCO.0000000000000134>

Darabid, H., Perez-Gonzalez, A. P., & Robitaille, R. (2014). Neuromuscular synaptogenesis:

coordinating partners with multiple functions. *Nature Reviews Neuroscience*, 15(11), 703–718. <https://doi.org/10.1038/nrn3821>

Dasen, J. S., & Jessell, T. M. (2009). Chapter Six Hox Networks and the Origins of Motor Neuron Diversity. In *Hox Genes* (Vol. 88, pp. 169–200). Elsevier. [https://doi.org/10.1016/S0070-2153\(09\)88006-X](https://doi.org/10.1016/S0070-2153(09)88006-X)

Drory, V. E., Goltsman, E., Goldman Reznik, J., Mosek, A., & Korczyn, A. D. (2001). The value of muscle exercise in patients with amyotrophic lateral sclerosis. *Journal of the Neurological Sciences*, 191(1–2), 133–137. [https://doi.org/10.1016/S0022-510X\(01\)00610-4](https://doi.org/10.1016/S0022-510X(01)00610-4)

Engel, A. G., Shen, X.-M., Selcen, D., & Sine, S. M. (2015). Congenital myasthenic syndromes: pathogenesis, diagnosis, and treatment. *The Lancet Neurology*, 14(4), 420–434. [https://doi.org/10.1016/S1474-4422\(14\)70201-7](https://doi.org/10.1016/S1474-4422(14)70201-7)

Finkel, R. S., Mercuri, E., Meyer, O. H., Simonds, A. K., Schroth, M. K., Graham, R. J., Kirschner, J., Iannaccone, S. T., Crawford, T. O., Woods, S., Muntoni, F., Wirth, B., Montes, J., Main, M., Mazzone, E. S., Vitale, M., Snyder, B., Quijano-Roy, S., Bertini, E., ... Sejersen, T. (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), 197–207. <https://doi.org/10.1016/j.nmd.2017.11.004>

Fratta, P., Nirmalanathan, N., Masset, L., Skorupinska, I., Collins, T., Cortese, A., Pemble, S., Malaspina, A., Fisher, E. M. C., Greensmith, L., & Hanna, M. G. (2014). Correlation of clinical and molecular features in spinal bulbar muscular atrophy. *Neurology*, 82(23), 2077–2084. <https://doi.org/10.1212/WNL.0000000000000507>

Fuller, G., & Manford, M. (2010). *Neurology: an illustrated colour text* (3rd ed). Churchill Livingstone Elsevier. [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, T. F., Chew, J., Stankowski, J. N., Hayes, L. R., Zhang, Y.-J., Prudencio, M., Carlomagno, Y., Daugherty, L. M., Jansen-West, K., Perkerson, E. A., O'Raw, A., Cook, C., Pregent, L., Belzil, V., van Blitterswijk, M., Tabassian, L. J., Lee, C. W., Yue, M., Tong, J., ... Prelle, A. (2017). Poly(GP) proteins are a useful pharmacodynamic marker for -associated amyotrophic lateral sclerosis. *Science Translational Medicine*, 9(383). <https://doi.org/10.1126/scitranslmed.aai7866>

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

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

Gordon, E., Rohrer, J. D., & Fox, N. C. (2016). Advances in neuroimaging in frontotemporal dementia. *Journal of Neurochemistry*, 138, 193–210. <https://doi.org/10.1111/jnc.13656>

Harland, R. (2000). Neural induction. *Current Opinion in Genetics & Development*, 10(4),

357–362. [https://doi.org/10.1016/S0959-437X\(00\)00096-4](https://doi.org/10.1016/S0959-437X(00)00096-4)

Harwood, C. A., McDermott, C. J., & Shaw, P. J. (2012). Clinical aspects of motor neurone disease. *Medicine*, 40(10), 540–545. <https://doi.org/10.1016/j.mpmed.2012.07.003>

Hughes, J., Rees, S., Kalindjian, S., & Philpott, K. (2011). Principles of early drug discovery. *British Journal of Pharmacology*, 162(6), 1239–1249. <https://doi.org/10.1111/j.1476-5381.2010.01127.x>

Jacobson, L., Polizzi, A., Morriss-Kay, G., & Vincent, A. (1999). Plasma from human mothers of fetuses with severe arthrogryposis multiplex congenita causes deformities in mice. *Journal of Clinical Investigation*, 103(7), 1031–1038. <https://doi.org/10.1172/JCI5943>

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

Jones, R. A., Harrison, C., Eaton, S. L., Llaverro Hurtado, M., Graham, L. C., Alkhamash, L., Oladiran, O. A., Gale, A., Lamont, D. J., Simpson, H., Simmen, M. W., Soeller, C., Wishart, T. M., & Gillingwater, T. H. (2017). Cellular and Molecular Anatomy of the Human Neuromuscular Junction. *Cell Reports*, 21(9), 2348–2356. <https://doi.org/10.1016/j.celrep.2017.11.008>

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

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

Koneczny, I., Cossins, J., Waters, P., Beeson, D., & Vincent, A. (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). <https://doi.org/10.1371/journal.pone.0080695>

Kusner, L. L., & Kaminski, H. J. (2015). Myasthenia Gravis. In *Neurobiology of Brain Disorders* (pp. 135–150). Elsevier. <https://doi.org/10.1016/B978-0-12-398270-4.00010-0>

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

Laurá, M., Singh, D., Ramdharry, G., Morrow, J., Skorupinska, M., Pareyson, D., Burns, J., Lewis, R. A., Scherer, S. S., Herrmann, D. N., Cullen, N., Bradish, C., Gaiani, L., Martinelli, N., Gibbons, P., Pfeffer, G., Phisitkul, P., Wapner, K., Sanders, J., ... Reilly, M. M. (2018). Prevalence and orthopedic management of foot and ankle deformities in Charcot-Marie-Tooth disease. *Muscle & Nerve*, 57(2), 255–259. <https://doi.org/10.1002/mus.25724>

Leung, D. G. (2017). Other Proven and Putative Autoimmune Disorders of the Peripheral

Nervous System (Vol. 1). Oxford University Press.

<https://doi.org/10.1093/med/9780199937837.003.0098>

Li, L., Xiong, W.-C., & Mei, L. (2018). Neuromuscular Junction Formation, Aging, and Disorders. *Annual Review of Physiology*, 80(1), 159–188.

<https://doi.org/10.1146/annurev-physiol-022516-034255>

Lieberman, A. P., Yu, Z., Murray, S., Peralta, R., Low, A., Guo, S., Yu, X. X., Cortes, C. J., Bennett, C. F., Monia, B. P., La Spada, A. R., & Hung, G. (2014). Peripheral Androgen Receptor Gene Suppression Rescues Disease in Mouse Models of Spinal and Bulbar Muscular Atrophy. *Cell Reports*, 7(3), 774–784.

<https://doi.org/10.1016/j.celrep.2014.02.008>

Lin, G., Mao, D., & Bellen, H. J. (2017). Amyotrophic Lateral Sclerosis Pathogenesis Converges on Defects in Protein Homeostasis Associated with TDP-43 Mislocalization and Proteasome-Mediated Degradation Overload. In *Fly Models of Human Diseases* (Vol. 121, pp. 111–171). Elsevier. <https://doi.org/10.1016/bs.ctdb.2016.07.004>

Lu, C.-H., Macdonald-Wallis, C., Gray, E., Pearce, N., Petzold, A., Norgren, N., Giovannoni, G., Fratta, P., Sidle, K., Fish, M., Orrell, R., Howard, R., Talbot, K., Greensmith, L., Kuhle, J., Turner, M. R., & Malaspina, A. (2015). Neurofilament light chain: A prognostic biomarker in amyotrophic lateral sclerosis. *Neurology*, 84(22), 2247–2257.

<https://doi.org/10.1212/WNL.0000000000001642>

Malik, B., Nirmalanathan, N., Bilsland, L. G., La Spada, A. R., Hanna, M. G., Schiavo, G., Gallo, J.-M., & Greensmith, L. (2011). Absence of disturbed axonal transport in spinal and bulbar muscular atrophy. *Human Molecular Genetics*, 20(9), 1776–1786.

<https://doi.org/10.1093/hmg/ddr061>

Malik, B., Nirmalanathan, N., Gray, A. L., La Spada, A. R., Hanna, M. G., & Greensmith, L. (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), 926–943. <https://doi.org/10.1093/brain/aws343>

Manzano, R., Sorarú, G., Grunseich, C., Fratta, P., Zuccaro, E., Pennuto, M., & Rinaldi, C. (2018). Beyond motor neurons: expanding the clinical spectrum in Kennedy's disease. *Journal of Neurology, Neurosurgery & Psychiatry*, 89(8), 808–812.

<https://doi.org/10.1136/jnnp-2017-316961>

Maragakis, N. J. (2017). What can we learn from the edaravone development program for ALS? *Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration*, 18(sup1), 98–103.

<https://doi.org/10.1080/21678421.2017.1361446>

Mazzone, E. S., Mayhew, A., Montes, J., Ramsey, D., Fanelli, L., Young, S. D., Salazar, R., De Sanctis, R., Pasternak, A., Glanzman, A., Coratti, G., Civitello, M., Forcina, N., Gee, R., Duong, T., Pane, M., Scoto, M., Pera, M. C., Messina, S., ... Mercuri, E. (2017). Revised upper limb module for spinal muscular atrophy: Development of a new module. *Muscle & Nerve*, 55(6), 869–874. <https://doi.org/10.1002/mus.25430>

Mercuri, E., Finkel, R. S., Muntoni, F., Wirth, B., Montes, J., Main, M., Mazzone, E. S., Vitale, M., Snyder, B., Quijano-Roy, S., Bertini, E., Davis, R. H., Meyer, O. H., Simonds, A. K.,

- Schroth, M. K., Graham, R. J., Kirschner, J., Iannaccone, S. T., Crawford, T. O., ... Dubousset, J. (2018). Diagnosis and management of spinal muscular atrophy: Part 1: Recommendations for diagnosis, rehabilitation, orthopedic and nutritional care. *Neuromuscular Disorders*, 28(2), 103–115. <https://doi.org/10.1016/j.nmd.2017.11.005>
- Meriggioli, M. N., & Sanders, D. B. (2009). Autoimmune myasthenia gravis: emerging clinical and biological heterogeneity. *The Lancet Neurology*, 8(5), 475–490. [https://doi.org/10.1016/S1474-4422\(09\)70063-8](https://doi.org/10.1016/S1474-4422(09)70063-8)
- Milioto, C., Malena, A., Maino, E., Polanco, M. J., Marchioretta, C., Borgia, D., Pereira, M. G., Blaauw, B., Lieberman, A. P., Venturini, R., Plebani, M., Sambataro, F., Vergani, L., Pegoraro, E., Sorarù, G., & Pennuto, M. (2017). Beta-agonist stimulation ameliorates the phenotype of spinal and bulbar muscular atrophy mice and patient-derived myotubes. *Scientific Reports*, 7(1). <https://doi.org/10.1038/srep41046>
- Mitsumoto, H., Brooks, B. R., & Silani, V. (2014). Clinical trials in amyotrophic lateral sclerosis: why so many negative trials and how can trials be improved? *The Lancet Neurology*, 13(11), 1127–1138. [https://doi.org/10.1016/S1474-4422\(14\)70129-2](https://doi.org/10.1016/S1474-4422(14)70129-2)
- Monahan, Z., Shewmaker, F., & Pandey, U. B. (2016). Stress granules at the intersection of autophagy and ALS. *Brain Research*, 1649, 189–200. <https://doi.org/10.1016/j.brainres.2016.05.022>
- Morgan, S., & Orrell, R. W. (2016). Pathogenesis of amyotrophic lateral sclerosis. *British Medical Bulletin*, 119(1), 87–98. <https://doi.org/10.1093/bmb/ldw026>
- Morren, J. A., & Galvez-Jimenez, N. (2012). Current and prospective disease-modifying therapies for amyotrophic lateral sclerosis. *Expert Opinion on Investigational Drugs*, 21(3), 297–320. <https://doi.org/10.1517/13543784.2012.657303>
- Motor neurone disease: assessment and management | Guidance and guidelines | NICE. (n.d.). <https://www.nice.org.uk/guidance/ng42>
- Nishimune, H., Valdez, G., Jarad, G., Moulson, C. L., Müller, U., Miner, J. H., & Sanes, J. R. (2008). Laminins promote postsynaptic maturation by an autocrine mechanism at the neuromuscular junction. *The Journal of Cell Biology*, 182(6), 1201–1215. <https://doi.org/10.1083/jcb.200805095>
- O'Connor, E., Töpf, A., Zahedi, R. P., Spendiff, S., Cox, D., Roos, A., & Lochmüller, H. (2018). Clinical and research strategies for limb-girdle congenital myasthenic syndromes. *Annals of the New York Academy of Sciences*, 1412(1), 102–112. <https://doi.org/10.1111/nyas.13520>
- Orrell, Richard WBarclay, Chris. (n.d.). Diagnosis and management of motor neurone disease. *Practitioner*, 260, 17–21. <https://search.proquest.com/docview/1844334383/64C39DCAF3D346C0PQ/1?accountid=14511>
- Otto, M., Bowser, R., Turner, M., Berry, J., Brettschneider, J., Connor, J., Costa, J., Cudkowicz, M., Glass, J., Jahn, O., Lehnert, S., Malaspina, A., Parnetti, L., Petzold, A., Shaw, P., Sherman, A., Steinacker, P., Süßmuth, S., Teunissen, C., ... Ludolph, A. (2012).

- Roadmap and standard operating procedures for biobanking and discovery of neurochemical markers in ALS. *Amyotrophic Lateral Sclerosis*, 13(1), 1–10. <https://doi.org/10.3109/17482968.2011.627589>
- Peragallo, J. H. (2017). Pediatric Myasthenia Gravis. *Seminars in Pediatric Neurology*, 24(2), 116–121. <https://doi.org/10.1016/j.spen.2017.04.003>
- Ramdharry, G. M., Pollard, A., Anderson, C., Laurá, M., Murphy, S. M., Dudziec, M., Dewar, E. L., Hutton, E., Grant, R., & Reilly, M. M. (2014). A pilot study of proximal strength training in Charcot-Marie-Tooth disease. *Journal of the Peripheral Nervous System*, 19(4), 328–332. <https://doi.org/10.1111/jns.12100>
- Ramsey, D., Scoto, M., Mayhew, A., Main, M., Mazzone, E. S., Montes, J., de Sanctis, R., Dunaway Young, S., Salazar, R., Glanzman, A. M., Pasternak, A., Quigley, J., Mirek, E., Duong, T., Gee, R., Civitello, M., Tennekoon, G., Pane, M., Pera, M. C., ... Muntoni, F. (2017). Revised Hammersmith Scale for spinal muscular atrophy: A SMA specific clinical outcome assessment tool. *PLOS ONE*, 12(2). <https://doi.org/10.1371/journal.pone.0172346>
- Reilly, M. M., Pareyson, D., Burns, J., Laurá, M., Shy, M. E., Singh, D., Agren, P. H., Altmann, V., Baets, J., Briggs, P., Burns, J., Butcher, K., Gaiani, L., Genovese, F., Gibbons, P., Laurá, M., Louwerens, J. W., Manzur, A., Moroni, I., ... Wenz, W. (2017). 221st ENMC International Workshop: Neuromuscular Disorders, 27(12), 1138–1142. <https://doi.org/10.1016/j.nmd.2017.09.005>
- Renton, A. E., Chiò, A., & Traynor, B. J. (2014). State of play in amyotrophic lateral sclerosis genetics. *Nature Neuroscience*, 17(1), 17–23. <https://doi.org/10.1038/nn.3584>
- Rodríguez Cruz, P. M., Palace, J., & Beeson, D. (2014). Inherited disorders of the neuromuscular junction: an update. *Journal of Neurology*, 261(11), 2234–2243. <https://doi.org/10.1007/s00415-014-7520-7>
- Rodríguez Cruz, P. M., Sewry, C., Beeson, D., Jayawant, S., Squier, W., McWilliam, R., & Palace, J. (2014). Congenital myopathies with secondary neuromuscular transmission defects; A case report and review of the literature. *Neuromuscular Disorders*, 24(12), 1103–1110. <https://doi.org/10.1016/j.nmd.2014.07.005>
- Rudolf, R., Khan, M. M., Labeit, S., & Deschenes, M. R. (2014). Degeneration of Neuromuscular Junction in Age and Dystrophy. *Frontiers in Aging Neuroscience*, 6. <https://doi.org/10.3389/fnagi.2014.00099>
- Ruegsegger, C., & Saxena, S. (2016). Proteostasis impairment in ALS. *Brain Research*, 1648, 571–579. <https://doi.org/10.1016/j.brainres.2016.03.032>
- Scoto, M., Finkel, R. S., Mercuri, E., & Muntoni, F. (2017). Therapeutic approaches for spinal muscular atrophy (SMA). *Gene Therapy*, 24(9), 514–519. <https://doi.org/10.1038/gt.2017.45>
- Singhal, N., & Martin, P. T. (2011). Role of extracellular matrix proteins and their receptors in the development of the vertebrate neuromuscular junction. *Developmental Neurobiology*, 71(11), 982–1005. <https://doi.org/10.1002/dneu.20953>

- Spillane, J., Beeson, D. J., & Kullmann, D. M. (2010). Myasthenia and related disorders of the neuromuscular junction. *Journal of Neurology, Neurosurgery & Psychiatry*, 81(8), 850–857. <https://doi.org/10.1136/jnnp.2008.169367>
- Spillane, J., Ermolyuk, Y., Cano-Jaimez, M., Lang, B., Vincent, A., Volynski, K. E., & Kullmann, D. M. (2015). Lambert-Eaton syndrome IgG inhibits transmitter release via P/Q Ca<sup>2+</sup> channels. *Neurology*, 84(6), 575–579. <https://doi.org/10.1212/WNL.0000000000001225>
- Viegas, S., Jacobson, L., Waters, P., Cossins, J., Jacob, S., Leite, M. I., Webster, R., & Vincent, A. (2012). Passive and active immunization models of MuSK-Ab positive myasthenia: Electrophysiological evidence for pre and postsynaptic defects. *Experimental Neurology*, 234(2), 506–512. <https://doi.org/10.1016/j.expneurol.2012.01.025>
- Vincent, A. (2002). Unravelling the pathogenesis of myasthenia gravis. *Nature Reviews Immunology*, 2(10), 797–804. <https://doi.org/10.1038/nri916>
- Volume 58, Issue 3, March 2016. (n.d.). Volume 58, Issue 3, March 2016. <https://link.springer.com/journal/12031/58/3>
- Westerberg, E., Molin, C. J., Spöndly Nees, S., Widenfalk, J., & Punga, A. R. (2018). The impact of physical exercise on neuromuscular function in Myasthenia gravis patients. *Medicine*, 97(31). <https://doi.org/10.1097/MD.00000000000011510>
- Wolfe, G. I., Kaminski, H. J., Aban, I. B., Minisman, G., Kuo, H.-C., Marx, A., Ströbel, P., Mazia, C., Oger, J., Cea, J. G., Heckmann, J. M., Evoli, A., Nix, W., Ciafaloni, E., Antonini, G., Witoonpanich, R., King, J. O., Beydoun, S. R., Chalk, C. H., ... Cutter, G. R. (2016). Randomized Trial of Thymectomy in Myasthenia Gravis. *New England Journal of Medicine*, 375(6), 511–522. <https://doi.org/10.1056/NEJMoa1602489>
- Woollacott, I. O. C., & Rohrer, J. D. (2016). The clinical spectrum of sporadic and familial forms of frontotemporal dementia. *Journal of Neurochemistry*, 138, 6–31. <https://doi.org/10.1111/jnc.13654>