

CLNE0020: Motoneurons, Neuromuscular Junctions and Associated Disease

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1.

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

2.

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

3.

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

4.

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

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.: Diagnosis and management of Duchenne muscular dystrophy, part 1: diagnosis, and neuromuscular, rehabilitation, endocrine, and gastrointestinal and

nutritional management. *The Lancet Neurology*. 17, 251–267 (2018).
[https://doi.org/10.1016/S1474-4422\(18\)30024-3](https://doi.org/10.1016/S1474-4422(18)30024-3).

6.

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

7.

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.: 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, 445–455 (2018).
[https://doi.org/10.1016/S1474-4422\(18\)30026-7](https://doi.org/10.1016/S1474-4422(18)30026-7).

8.

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., Woods, S., Qian, Y., Sejersen, T., Muntoni, F., Wirth, B., Tiziano, F.D., Kirschner, J., Tizzano, E., Topaloglu, H., Swoboda, K., Laing, N., Kayoko, S., Prior, T., Chung, W.K., Wu, S.-M., Montes, J., Mazzone, E., Main, M., Coleman, C., Gee, R., Glanzman, A., Kroksmark, A.-K., Krosschell, K., Nelson, L., Rose, K., Stępień, A., Vuillerot, C., Vitale, M., Snyder, B., Quijano-Roy, S., Dubousset, J.: Diagnosis and management of spinal muscular atrophy: Part 1: Recommendations for diagnosis, rehabilitation, orthopedic and nutritional care. *Neuromuscular Disorders*. 28, 103–115 (2018). <https://doi.org/10.1016/j.nmd.2017.11.005>.

9.

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., Davis, R.H., Qian, Y., Sejersen, T.: 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, 197–207 (2018).
<https://doi.org/10.1016/j.nmd.2017.11.004>.

10.

Scoto, M., Finkel, R.S., Mercuri, E., Muntoni, F.: Therapeutic approaches for spinal muscular atrophy (SMA). *Gene Therapy*. 24, 514–519 (2017). <https://doi.org/10.1038/gt.2017.45>.

11.

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., Bushby, K., Day, J., Darras, B.T., De Vivo, D., Finkel, R., Mercuri, E., Muntoni, F.: Revised Hammersmith Scale for spinal muscular atrophy: A SMA specific clinical outcome assessment tool. *PLOS ONE*. 12, (2017). <https://doi.org/10.1371/journal.pone.0172346>.

12.

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., Tennekoon, G., Day, J.W., Darras, B.T., De Vivo, D.C., Finkel, R., Muntoni, F., Mercuri, E.: Revised upper limb module for spinal muscular atrophy: Development of a new module. *Muscle & Nerve*. 55, 869–874 (2017). <https://doi.org/10.1002/mus.25430>.

13.

Westerberg, E., Molin, C.J., Spörndly Nees, S., Widenfalk, J., Punga, A.R.: The impact of physical exercise on neuromuscular function in Myasthenia gravis patients. *Medicine*. 97, (2018). <https://doi.org/10.1097/MD.00000000000011510>.

14.

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

15.

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., Flemister, S., Shy, M.E., Reilly,

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

16.

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., Martinelli, N., Pareyson, D., Pfeffer, G., Ramdharry, G., Reilly, M.M., Shy, M., Singh, D., van der Linden, M., Wenz, W.: 221st ENMC International Workshop: Neuromuscular Disorders. 27, 1138–1142 (2017).
<https://doi.org/10.1016/j.nmd.2017.09.005>.

17.

Ramdharry, G.M., Pollard, A., Anderson, C., Laurá, M., Murphy, S.M., Dudzic, M., Dewar, E.L., Hutton, E., Grant, R., Reilly, M.M.: A pilot study of proximal strength training in Charcot-Marie-Tooth disease. *Journal of the Peripheral Nervous System*. 19, 328–332 (2014). <https://doi.org/10.1111/jns.12100>.

18.

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

19.

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.: 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, 280–286 (2016).
<https://doi.org/10.1136/jnnp-2014-310055>.

20.

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

21.

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

22.

Al-Chalabi, A., van den Berg, L.H., Veldink, J.: Gene discovery in amyotrophic lateral sclerosis: implications for clinical management. *Nature Reviews Neurology*. 13, 96–104 (2017). <https://doi.org/10.1038/nrneurol.2016.182>.

23.

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

24.

Lin, G., Mao, D., Bellen, H.J.: 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*. pp. 111–171. Elsevier (2017). <https://doi.org/10.1016/bs.ctdb.2016.07.004>.

25.

Monahan, Z., Shewmaker, F., Pandey, U.B.: Stress granules at the intersection of autophagy and ALS. *Brain Research*. 1649, 189–200 (2016). <https://doi.org/10.1016/j.brainres.2016.05.022>.

26.

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

27.

Renton, A.E., Chiò, A., Traynor, B.J.: State of play in amyotrophic lateral sclerosis genetics. *Nature Neuroscience*. 17, 17–23 (2014). <https://doi.org/10.1038/nn.3584>.

28.

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

29.

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

30.

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

31.

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

32.

Darabid, H., Perez-Gonzalez, A.P., Robitaille, R.: Neuromuscular synaptogenesis: coordinating partners with multiple functions. *Nature Reviews Neuroscience*. 15, 703–718 (2014). <https://doi.org/10.1038/nrn3821>.

33.

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

34.

Ladle, D.R., Pecho-Vrieseling, E., Arber, S.: Assembly of Motor Circuits in the Spinal Cord: Driven to Function by Genetic and Experience-Dependent Mechanisms. *Neuron*. 56,

270–283 (2007). <https://doi.org/10.1016/j.neuron.2007.09.026>.

35.

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

36.

Li, L., Xiong, W.-C., Mei, L.: Neuromuscular Junction Formation, Aging, and Disorders. *Annual Review of Physiology*. 80, 159–188 (2018). <https://doi.org/10.1146/annurev-physiol-022516-034255>.

37.

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

38.

Nishimune, H., Valdez, G., Jarad, G., Moulson, C.L., Müller, U., Miner, J.H., Sanes, J.R.: Laminins promote postsynaptic maturation by an autocrine mechanism at the neuromuscular junction. *The Journal of Cell Biology*. 182, 1201–1215 (2008). <https://doi.org/10.1083/jcb.200805095>.

39.

Rudolf, R., Khan, M.M., Labeit, S., Deschenes, M.R.: Degeneration of Neuromuscular Junction in Age and Dystrophy. *Frontiers in Aging Neuroscience*. 6, (2014). <https://doi.org/10.3389/fnagi.2014.00099>.

40.

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

41.

O'Connor, E., Töpf, A., Zahedi, R.P., Spendiff, S., Cox, D., Roos, A., Lochmüller, H.: Clinical and research strategies for limb-girdle congenital myasthenic syndromes. *Annals of the New York Academy of Sciences*. 1412, 102–112 (2018).
<https://doi.org/10.1111/nyas.13520>.

42.

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

43.

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

44.

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

45.

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., Palace, J., Muntoni, F., Beeson, D.: Mutations in *CHST3* cause congenital myasthenic syndrome and bridge myasthenic disorders with dystroglycanopathies. *Brain*. 138, 2493–2504 (2015).
<https://doi.org/10.1093/brain/awv185>.

46.

Rodríguez Cruz, P.M., Sewry, C., Beeson, D., Jayawant, S., Squier, W., McWilliam, R.,

Palace, J.: Congenital myopathies with secondary neuromuscular transmission defects; A case report and review of the literature. *Neuromuscular Disorders*. 24, 1103–1110 (2014). <https://doi.org/10.1016/j.nmd.2014.07.005>.

47.

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

48.

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

49.

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

50.

Leung, D.G.: *Other Proven and Putative Autoimmune Disorders of the Peripheral Nervous System*. Oxford University Press (2017). <https://doi.org/10.1093/med/9780199937837.003.0098>.

51.

Spillane, J., Beeson, D.J., Kullmann, D.M.: Myasthenia and related disorders of the neuromuscular junction. *Journal of Neurology, Neurosurgery & Psychiatry*. 81, 850–857 (2010). <https://doi.org/10.1136/jnnp.2008.169367>.

52.

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

53.

Spillane, J., Ermolyuk, Y., Cano-Jaimez, M., Lang, B., Vincent, A., Volynski, K.E., Kullmann, D.M.: Lambert-Eaton syndrome IgG inhibits transmitter release via P/Q Ca²⁺ channels. *Neurology*. 84, 575–579 (2015). <https://doi.org/10.1212/WNL.0000000000001225>.

54.

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., Barboi, A.C., Amato, A.A., Shaibani, A.I., Katirji, B., Lecky, B.R.F., Buckley, C., Vincent, A., Dias-Tosta, E., Yoshikawa, H., Waddington-Cruz, M., Pulley, M.T., Rivner, M.H., Kostera-Pruszczyk, A., Pascuzzi, R.M., Jackson, C.E., Garcia Ramos, G.S., Verschuuren, J.J.G.M., Massey, J.M., Kissel, J.T., Werneck, L.C., Benatar, M., Barohn, R.J., Tandan, R., Mozaffar, T., Conwit, R., Odenkirchen, J., Sonett, J.R., Jaretzki, A., Newsom-Davis, J., Cutter, G.R.: Randomized Trial of Thymectomy in Myasthenia Gravis. *New England Journal of Medicine*. 375, 511–522 (2016). <https://doi.org/10.1056/NEJMoa1602489>.

55.

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

56.

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

57.

Fuller, G., Manford, M.: *Neurology: an illustrated colour text*. Churchill Livingstone Elsevier, Edinburgh (2010).

58.

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59.

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

60.

Otto, M., Bowser, R., Turner, M., Berry, J., Brettschneider, J., Connor, J., Costa, J., Cudkovicz, M., Glass, J., Jahn, O., Lehnert, S., Malaspina, A., Parnetti, L., Petzold, A., Shaw, P., Sherman, A., Steinacker, P., Süßmuth, S., Teunissen, C., Tumani, H., Wuolikainen, A., Ludolph, A.: Roadmap and standard operating procedures for biobanking and discovery of neurochemical markers in ALS. *Amyotrophic Lateral Sclerosis*. 13, 1–10 (2012). <https://doi.org/10.3109/17482968.2011.627589>.

61.

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.: Neurofilament light chain: A prognostic biomarker in amyotrophic lateral sclerosis. *Neurology*. 84, 2247–2257 (2015). <https://doi.org/10.1212/WNL.0000000000001642>.

62.

Benatar, M., Boylan, K., Jeromin, A., Rutkove, S.B., Berry, J., Atassi, N., Bruijn, L.: ALS biomarkers for therapy development: State of the field and future directions. *Muscle & Nerve*. 53, 169–182 (2016). <https://doi.org/10.1002/mus.24979>.

63.

Andreasson, U., Blennow, K., Zetterberg, H.: 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 (2016). <https://doi.org/10.1016/j.dadm.2016.05.005>.

64.

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., Song,

Y., Castanedes-Casey, M., Rousseau, L., Phillips, V., Dickson, D.W., Rademakers, R., Fryer, J.D., Rush, B.K., Pedraza, O., Caputo, A.M., Desaro, P., Palmucci, C., Robertson, A., Heckman, M.G., Diehl, N.N., Wiggs, E., Tierney, M., Braun, L., Farren, J., Lacomis, D., Ladha, S., Fournier, C.N., McCluskey, L.F., Elman, L.B., Toledo, J.B., McBride, J.D., Tiloca, C., Morelli, C., Poletti, B., Solca, F., Prella, A.: Poly(GP) proteins are a useful pharmacodynamic marker for -associated amyotrophic lateral sclerosis. *Science Translational Medicine*. 9, (2017). <https://doi.org/10.1126/scitranslmed.aai7866>.

65.

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

66.

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

67.

Viegas, S., Jacobson, L., Waters, P., Cossins, J., Jacob, S., Leite, M.I., Webster, R., Vincent, A.: Passive and active immunization models of MuSK-Ab positive myasthenia: Electrophysiological evidence for pre and postsynaptic defects. *Experimental Neurology*. 234, 506–512 (2012). <https://doi.org/10.1016/j.expneurol.2012.01.025>.

68.

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

69.

Koneczny, I., Cossins, J., Waters, P., Beeson, D., Vincent, A.: 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, (2013). <https://doi.org/10.1371/journal.pone.0080695>.

70.

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

71.

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

72.

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

73.

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74.

Badders, N.M., Korff, A., Miranda, H.C., Vuppala, P.K., Smith, R.B., Winborn, B.J., Quemim, 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., Li, Y., Kim, H.J., La Spada, A.R., Taylor, J.P.: Selective modulation of the androgen receptor AF2 domain rescues degeneration in spinal bulbar muscular atrophy. *Nature Medicine*. 24, 427–437 (2018). <https://doi.org/10.1038/nm.4500>.

75.

Beitel, L.K., Alvarado, C., Mokhtar, S., Paliouras, M., Trifiro, M.: Mechanisms Mediating Spinal and Bulbar Muscular Atrophy: Investigations into Polyglutamine-Expanded Androgen Receptor Function and Dysfunction. *Frontiers in Neurology*. 4, (2013). <https://doi.org/10.3389/fneur.2013.00053>.

76.

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

77.

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

78.

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

79.

Malik, B., Nirmalanathan, N., Bilisland, L.G., La Spada, A.R., Hanna, M.G., Schiavo, G., Gallo, J.-M., Greensmith, L.: Absence of disturbed axonal transport in spinal and bulbar muscular atrophy. *Human Molecular Genetics*. 20, 1776–1786 (2011). <https://doi.org/10.1093/hmg/ddr061>.

80.

Malik, B., Nirmalanathan, N., Gray, A.L., La Spada, A.R., Hanna, M.G., Greensmith, L.: 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, 926–943 (2013). <https://doi.org/10.1093/brain/aws343>.

81.

Manzano, R., Sorarú, G., Grunseich, C., Fratta, P., Zuccaro, E., Pennuto, M., Rinaldi, C.: Beyond motor neurons: expanding the clinical spectrum in Kennedy's disease. *Journal of Neurology, Neurosurgery & Psychiatry*. 89, 808–812 (2018). <https://doi.org/10.1136/jnnp-2017-316961>.

82.

Milioto, C., Malena, A., Maino, E., Polanco, M.J., Marchioretti, 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.: Beta-agonist stimulation ameliorates the phenotype of spinal and bulbar muscular atrophy mice and patient-derived myotubes. *Scientific Reports*. 7, (2017). <https://doi.org/10.1038/srep41046>.