# MSc Neuromuscular Disease

View Online

1.

Blottner, D., Salanova, M.: The neuromuscular system: from earth to space life science : neuromuscular cell signalling in disuse and exercise. Springer, Cham (2015).

2.

Benson, M.K.D.: Children's neuromuscular disorders. Springer, London (2011).

3.

Shaibani, A.: A Video Atlas of Neuromuscular Disorders. Oxford University Press (2014). https://doi.org/10.1093/med/9780199898152.001.0001.

4.

Hilton-Jones, D., Turner, M.R. eds: Oxford textbook of neuromuscular disorders. Oxford University Press, [Oxford] (2014).

5.

Kernell, D.: The Motoneurone and its Muscle Fibres. Oxford University Press (2006). https://doi.org/10.1093/acprof:oso/9780198526551.001.0001.

6.

Amato, A.A., Russell, J.A.: Neuromuscular disorders. McGraw-Hill, New York (2008).

7.

Jain, K.K. ed: Applied neurogenomics. Humana Press, New York, NY (2015).

8.

Davies, A.M.: Regulation of Neuronal Survival by Neurotrophins in the Developing Peripheral Nervous System. In: Patterning and Cell Type Specification in the Developing CNS and PNS. pp. 303–311. Elsevier (2013). https://doi.org/10.1016/B978-0-12-397265-1.00095-2.

9.

Gordon, T., Sulaiman, O.A.: Nerve Regeneration in the Peripheral Nervous System. In: Kettenmann, H. (ed.) Neuroglia. pp. 701–714. Oxford University Press (2012). https://doi.org/10.1093/med/9780199794591.003.0055.

10.

Welch, M.B., Brummett, C.M.: Peripheral Nervous SystemAnatomy and Function. In: Mashour, G.A. and Lydic, R. (eds) Neuroscientific Foundations of Anesthesiology. pp. 133–140. Oxford University Press (2011). https://doi.org/10.1093/med/9780195398243.003.0067.

11.

Rossor, A.M., Tomaselli, P.J., Reilly, M.M.: Recent advances in the genetic neuropathies. Current Opinion in Neurology. (2016). https://doi.org/10.1097/WCO.0000000000000373.

12.

Rossor, A.M., Evans, M.R.B., Reilly, M.M.: A practical approach to the genetic neuropathies. Practical Neurology. 15, 187–198 (2015). https://doi.org/10.1136/practneurol-2015-001095.

13.

AM Rossor: Recent advances in the genetic neuropathies. Current opinion in neurology. 29, (2016). https://doi.org/10.1097/WCO.0000000000000373.

14.

Reilly, M., Fridman, V.: Inherited Neuropathies. Seminars in Neurology. 35, 407–423 (2015). https://doi.org/10.1055/s-0035-1558981.

15.

Rossor, A.M., Evans, M.R.B., Reilly, M.M.: A practical approach to the genetic neuropathies. Practical Neurology. 15, 187–198 (2015). https://doi.org/10.1136/practneurol-2015-001095.

16.

Pasterkamp, R.J.: Getting neural circuits into shape with semaphorins. Nature Reviews Neuroscience. 13, 605–618 (2012). https://doi.org/10.1038/nrn3302.

17.

R Klein: Role of neurotrophins in mouse neuronal development. The FASEB Journal. 8, 738–744 (1994).

18.

Schlosser, G.: Induction and specification of cranial placodes. Developmental Biology. 294, 303–351 (2006). https://doi.org/10.1016/j.ydbio.2006.03.009.

19.

Thiede-Stan, N.K., Schwab, M.E.: Attractive and repulsive factors act through multi-subunit receptor complexes to regulate nerve fiber growth. Journal of Cell Science. 128, 2403–2414 (2015). https://doi.org/10.1242/jcs.165555.

20.

Irina Dudanova: Genetic Evidence for a Contribution of EphA:EphrinA Reverse Signaling to Motor Axon Guidance. Journal of Neuroscience. 32, 5209–5215 (2012).

21.

Wang, L., Klein, R., Zheng, B., Marquardt, T.: Anatomical Coupling of Sensory and Motor Nerve Trajectory via Axon Tracking. Neuron. 71, 263–277 (2011). https://doi.org/10.1016/j.neuron.2011.06.021.

22.

Van Battum, E.Y., Brignani, S., Pasterkamp, R.J.: Axon guidance proteins in neurological disorders. The Lancet Neurology. 14, 532–546 (2015). https://doi.org/10.1016/S1474-4422(14)70257-1.

23.

Kao, T.-J., Law, C., Kania, A.: Eph and ephrin signaling: Lessons learned from spinal motor neurons. Seminars in Cell & Developmental Biology. 23, 83–91 (2012). https://doi.org/10.1016/j.semcdb.2011.10.016.

24.

Byung-Yong Park: Induction and Segregation of the Vertebrate Cranial Placodes. (2010).

25.

Taniguchi, M., Yuasa, S., Fujisawa, H., Naruse, I., Saga, S., Mishina, M., Yagi, T.: Disruption of Semaphorin III/D Gene Causes Severe Abnormality in Peripheral Nerve Projection. Neuron. 19, 519–530 (1997). https://doi.org/10.1016/S0896-6273(00)80368-2.

26.

Ebens, A., Brose, K., Leonardo, E.D., Jr, M.G.H., Bladt, F., Birchmeier, C., Barres, B.A., Tessier-Lavigne, M.: Hepatocyte Growth Factor/Scatter Factor Is an Axonal Chemoattractant and a Neurotrophic Factor for Spinal Motor Neurons. Neuron. 17, 1157–1172 (1996). https://doi.org/10.1016/S0896-6273(00)80247-0.

27.

A. Caton: The branchial arches and HGF are growth-promoting and chemoattractant for cranial motor axons. Development. 127, 1751–1766.

28.

Conover, J.C., Erickson, J.T., Katz, D.M., Bianchi, L.M., Poueymirou, W.T., McClain, J., Pan, L., Helgren, M., Ip, N.Y., Boland, P., Friedman, B., Wiegand, S., Vejsada, R., Kato, A.C., DeChiara, T.M., Yancopoulos, G.D.: Neuronal deficits, not involving motor neurons, in mice lacking BDNF and/or NT4. Nature. 375, 235–238 (1995). https://doi.org/10.1038/375235a0.

29.

Jessen, K.R., Mirsky, R.: The origin and development of glial cells in peripheral nerves. Nature Reviews Neuroscience. 6, 671–682 (2005). https://doi.org/10.1038/nrn1746.

30.

Monk, K.R., Feltri, M.L., Taveggia, C.: New insights on schwann cell development. Glia. 63, 1376–1393 (2015). https://doi.org/10.1002/glia.22852.

31.

Jessen, K.R., Mirsky, R., Lloyd, A.C.: Schwann Cells: Development and Role in Nerve Repair. Cold Spring Harbor Perspectives in Biology. 7, (2015). https://doi.org/10.1101/cshperspect.a020487.

32.

Salzer, J.L.: Schwann Cell Myelination. Cold Spring Harbor Perspectives in Biology. 7, (2015). https://doi.org/10.1101/cshperspect.a020529.

33.

Arthur-Farraj, P.J., Latouche, M., Wilton, D.K., Quintes, S., Chabrol, E., Banerjee, A., Woodhoo, A., Jenkins, B., Rahman, M., Turmaine, M., Wicher, G.K., Mitter, R., Greensmith, L., Behrens, A., Raivich, G., Mirsky, R., Jessen, K.R.: c-Jun Reprograms Schwann Cells of Injured Nerves to Generate a Repair Cell Essential for Regeneration. Neuron. 75, 633–647 (2012). https://doi.org/10.1016/j.neuron.2012.06.021.

34.

Brosius Lutz, A., Barres, B.A.: Contrasting the Glial Response to Axon Injury in the Central and Peripheral Nervous Systems. Developmental Cell. 28, 7–17 (2014). https://doi.org/10.1016/j.devcel.2013.12.002.

35.

Jessen, K.R., Mirsky, R.: The repair Schwann cell and its function in regenerating nerves. The Journal of Physiology. 594, 3521–3531 (2016). https://doi.org/10.1113/JP270874.

36.

Auer-Grumbach, M.: Hereditary sensory and autonomic neuropathies. In: Peripheral Nerve Disorders. pp. 893–906. Elsevier (2013). https://doi.org/10.1016/B978-0-444-52902-2.00050-3.

37.

Davidson, G.L., Murphy, S.M., Polke, J.M., Laura, M., Salih, M.A.M., Muntoni, F., Blake, J., Brandner, S., Davies, N., Horvath, R., Price, S., Donaghy, M., Roberts, M., Foulds, N., Ramdharry, G., Soler, D., Lunn, M.P., Manji, H., Davis, M.B., Houlden, H., Reilly, M.M.: Frequency of mutations in the genes associated with hereditary sensory and autonomic neuropathy in a UK cohort. Journal of Neurology. 259, 1673–1685 (2012). https://doi.org/10.1007/s00415-011-6397-y.

38.

Rossor, A.M., Evans, M.R.B., Reilly, M.M.: A practical approach to the genetic neuropathies. Practical Neurology. 15, 187–198 (2015). https://doi.org/10.1136/practneurol-2015-001095.

39.

Chhabra, A.: Peripheral MR Neurography. Neuroimaging Clinics of North America. 24, 79–89 (2014). https://doi.org/10.1016/j.nic.2013.03.033.

40.

Purves, D.: Neuroscience. National Library of Medicine, Bethesda (2001).

41.

Effects of axon diameter and myelination (video) | Khan Academy, https://www.khanacademy.org/science/health-and-medicine/nervous-system-and-sensory-infor/neuron-membrane-potentials-2014-03-27T17:58:17.207Z/v/effects-of-axon-diameter-and-myelination.

42.

Nerve Signaling, https://www.nobelprize.org/educational/medicine/nerve_signaling/index.html.

43.

Duncan, J.S., Winston, G.P., Koepp, M.J., Ourselin, S.: Brain imaging in the assessment for epilepsy surgery. The Lancet Neurology. 15, 420–433 (2016). https://doi.org/10.1016/S1474-4422(15)00383-X.

44.

Nowell, M., Sparks, R., Zombori, G., Miserocchi, A., Rodionov, R., Diehl, B., Wehner, T., White, M., Ourselin, S., McEvoy, A., Duncan, J.: Resection planning in extratemporal epilepsy surgery using 3D multimodality imaging and intraoperative MRI. British Journal of Neurosurgery. 31, 468–470 (2017). https://doi.org/10.1080/02688697.2016.1265086.

45.

Vakharia, V.N., Sparks, R., O'Keeffe, A.G., Rodionov, R., Miserocchi, A., McEvoy, A., Ourselin, S., Duncan, J.: Accuracy of intracranial electrode placement for stereoelectroencephalography: A systematic review and meta-analysis. Epilepsia. 58, 921–932 (2017). https://doi.org/10.1111/epi.13713.

46.

Michell, A.: Understanding EMG. Oxford University Press (2013). https://doi.org/10.1093/med/9780199595501.001.0001.

47.

Preston, D.C., Shapiro, B.E.: Electromyography and neuromuscular disorders:

clinical-electrophysiologic correlations. Elsevier Saunders, London (2013).

48.

Top tips for writing a lay summary | The Academy of Medical Sciences, https://acmedsci.ac.uk/more/news/10-tips-for-writing-a-lay-summary.

49.

How to Write a Lay Summary | Digital Curation Centre, http://www.dcc.ac.uk/resources/how-guides/write-lay-summary.

50.

Free guides, http://www.plainenglish.co.uk/free-guides.html.

51.

Part two - The specifics - Access to Understanding, http://www.access2understanding.org/guidance/part-two-the-specifics/.

52.

Readable | Free Readability Test Tool, https://www.webpagefx.com/tools/read-able/.

53.

Rees, J.H.: Paraneoplastic syndromes: when to suspect, how to confirm, and how to manage. Journal of Neurology, Neurosurgery & Psychiatry. 75, ii43–ii50 (2004). https://doi.org/10.1136/jnnp.2004.040378.

54.

Antoine, J.-C., Camdessanché, J.-P.: Paraneoplastic disorders of the peripheral nervous system. La Presse Médicale. 42, e235–e244 (2013). https://doi.org/10.1016/j.lpm.2013.01.059.

55.

How to Write a Lay Summary | DCC How-to Guides, http://www.dcc.ac.uk/resources/how-guides/.

56.

Rossor, A.M., Evans, M.R.B., Reilly, M.M.: A practical approach to the genetic neuropathies. Practical Neurology. 15, 187–198 (2015). https://doi.org/10.1136/practneurol-2015-001095.

57.

Rossor, A.M., Kalmar, B., Greensmith, L., Reilly, M.M.: The distal hereditary motor neuropathies. Journal of Neurology, Neurosurgery & Psychiatry. 83, 6–14 (2012). https://doi.org/10.1136/jnnp-2011-300952.

58.

Rossor, A.M., Carr, A.S., Devine, H., Chandrashekar, H., Pelayo-Negro, A.L., Pareyson, D., Shy, M.E., Scherer, S.S., Reilly, M.M.: Peripheral neuropathy in complex inherited diseases: an approach to diagnosis. Journal of Neurology, Neurosurgery & Psychiatry. 88, 846–863 (2017). https://doi.org/10.1136/jnnp-2016-313960.

59.

Carr, A.S., Pelayo-Negro, A.L., Evans, M.R., Laurà, M., Blake, J., Stancanelli, C., Iodice, V., Wechalekar, A.D., Whelan, C.J., Gillmore, J.D., Hawkins, P.N., Reilly, M.M.: A study of the neuropathy associated with transthyretin amyloidosis (ATTR) in the UK. Journal of Neurology, Neurosurgery & Psychiatry. 87, 620–627 (2016). https://doi.org/10.1136/jnnp-2015-310907.

60.

Hawkins, P.N., Ando, Y., Dispenzeri, A., Gonzalez-Duarte, A., Adams, D., Suhr, O.B.: Evolving landscape in the management of transthyretin amyloidosis. Annals of Medicine. 47, 625–638 (2015). https://doi.org/10.3109/07853890.2015.1068949.

61.

Plante-Bordeneuve, V., Ferreira, A., Lalu, T., Zaros, C., Lacroix, C., Adams, D., Said, G.: Diagnostic pitfalls in sporadic transthyretin familial amyloid polyneuropathy (TTR-FAP). Neurology. 69, 693–698 (2007). https://doi.org/10.1212/01.wnl.0000267338.45673.f4.

62.

Dubrey, S., Ackermann, E., Gillmore, J.: The transthyretin amyloidoses: advances in therapy. Postgraduate Medical Journal. 91, 439–448 (2015). https://doi.org/10.1136/postgradmedj-2014-133224.

63.

Dimachkie, M.M., Barohn, R.J.: Guillain-Barré Syndrome and Variants. Neurologic Clinics. 31, 491–510 (2013). https://doi.org/10.1016/j.ncl.2013.01.005.

64.

Willison, H.J., Jacobs, B.C., van Doorn, P.A.: Guillain-Barré syndrome. The Lancet. 388, 717–727 (2016). https://doi.org/10.1016/S0140-6736(16)00339-1.

65.

Collins, M.P., Hadden, R.D.: The nonsystemic vasculitic neuropathies. Nature Reviews Neurology. 13, 302–316 (2017). https://doi.org/10.1038/nrneurol.2017.42.

66.

Yates, M., Watts, R.A., Bajema, I.M., Cid, M.C., Crestani, B., Hauser, T., Hellmich, B., Holle, J.U., Laudien, M., Little, M.A., Luqmani, R.A., Mahr, A., Merkel, P.A., Mills, J., Mooney, J., Segelmark, M., Tesar, V., Westman, K., Vaglio, A., Yalçındağ, N., Jayne, D.R., Mukhtyar, C.: EULAR/ERA-EDTA recommendations for the management of ANCA-associated vasculitis. Annals of the Rheumatic Diseases. 75, 1583–1594 (2016). https://doi.org/10.1136/annrheumdis-2016-209133.

67.

Collins, M.P., Dyck, P.J.B., Gronseth, G.S., Guillevin, L., Hadden, R.D.M., Heuss, D., Léger, J.-M., Notermans, N.C., Pollard, J.D., Said, G., Sobue, G., Vrancken, A.F.J.E., Kissel, J.T.: Peripheral Nerve Society Guideline* on the classification, diagnosis, investigation, and immunosuppressive therapy of non-systemic vasculitic neuropathy: executive summary.

Journal of the Peripheral Nervous System. 15, 176–184 (2010). https://doi.org/10.1111/j.1529-8027.2010.00281.x.

68.

Berthelsen, M.P., Husu, E., Christensen, S.B., Prahm, K.P., Vissing, J., Jensen, B.R.: Anti-gravity training improves walking capacity and postural balance in patients with muscular dystrophy. Neuromuscular Disorders. 24, 492–498 (2014). https://doi.org/10.1016/j.nmd.2014.03.001.

69.

Cup, E.H., Pieterse, A.J., ten Broek-Pastoor, J.M., Munneke, M., van Engelen, B.G., Hendricks, H.T., van der Wilt, G.J., Oostendorp, R.A.: Exercise Therapy and Other Types of Physical Therapy for Patients With Neuromuscular Diseases: A Systematic Review. Archives of Physical Medicine and Rehabilitation. 88, 1452–1464 (2007). https://doi.org/10.1016/j.apmr.2007.07.024.

70.

Strength training and aerobic exercise training for muscle disease - van der Kooi - 2005 - The Cochrane Library - Wiley Online Library.

71.

Sveen, M.-L., Andersen, S.P., Ingelsrud, L.H., Blichter, S., Olsen, N.E., Jønck, S., Krag, T.O., Vissing, J.: Resistance training in patients with limb-girdle and becker muscular dystrophies. Muscle & Nerve. 47, 163–169 (2013). https://doi.org/10.1002/mus.23491.

72.

Jeppesen, T.D., Schwartz, M., Olsen, D.B., Wibrand, F., Krag, T., Duno, M., Hauerslev, S., Vissing, J.: Aerobic training is safe and improves exercise capacity in patients with mitochondrial myopathy. Brain. 129, 3402–3412 (2006). https://doi.org/10.1093/brain/awl149.

73.

Balance and walking involvement in facioscapulohumeral dystrophy: a pilot study on the effects of custom lower limb orthoses - European Journal of Physical and Rehabilitation

Medicine 2013 April;49(2):169-78 - Minerva Medica - Journals, https://www.minervamedica.it/en/journals/europa-medicophysica/article.php?cod=R33Y2013N02A0169.

74.

Michael P. Wiggs: Can endurance exercise preconditioning prevention disuse muscle atrophy? Frontiers in Physiology. 6, (2015). https://doi.org/10.3389/fphys.2015.00063.

75.

Keith Baar: Using Molecular Biology to Maximize Concurrent Training. Sports Medicine (Auckland, N.z.). 44, (2014). https://doi.org/10.1007/s40279-014-0252-0.

76.

Craig, D.M., Ashcroft, S.P., Belew, M.Y., Stocks, B., Currell, K., Baar, K., Philp, A.: Utilizing small nutrient compounds as enhancers of exercise-induced mitochondrial biogenesis. Frontiers in Physiology. 6, (2015). https://doi.org/10.3389/fphys.2015.00296.

77.

Hoier, B., Hellsten, Y.: Exercise-Induced Capillary Growth in Human Skeletal Muscle and the Dynamics of VEGF. Microcirculation. 21, 301–314 (2014). https://doi.org/10.1111/micc.12117.

78.

Hardie, D.G.: AMPK: A Key Sensor of Fuel and Energy Status in Skeletal Muscle. Physiology. 21, 48–60 (2006). https://doi.org/10.1152/physiol.00044.2005.

79.

Hawley, J.A., Hargreaves, M., Joyner, M.J., Zierath, J.R.: Integrative Biology of Exercise. Cell. 159, 738–749 (2014). https://doi.org/10.1016/j.cell.2014.10.029.

80.

Jones, D.A., Haan, A. de, Round, J.M.: Skeletal muscle from molecules to movement: a textbook of muscle physiology for sport, exercise, physiotherapy and medicine. Churchill Livingstone, Edinburgh (2004).

81.

A. M. Gordon: Regulation of Contraction in Striated Muscle. Physiological Reviews. 80, 853–924 (2000).

82.

R Bottinelli: Force-velocity properties of human skeletal muscle fibres: myosin heavy chain isoform and temperature dependence. The Journal of Physiology. 495, (1996).

83.

Harridge, S.D.R., Bottinelli, R., Canepari, M., Pellegrino, M.A., Reggiani, C., Esbjörnsson, M., Saltin, B.: Whole-muscle and single-fibre contractile properties and myosin heavy chain isoforms in humans. Pflügers Archiv - European Journal of Physiology. 432, 913–920 (1996). https://doi.org/10.1007/s004240050215.

84.

By:Hunter, S (Hunter, S); White, M (White, M); Thompson, M (Thompson, M): Techniques to evaluate elderly human muscle function: A physiological basis. JOURNALS OF GERONTOLOGY SERIES A-BIOLOGICAL SCIENCES AND MEDICAL SCIENCES JOURNALS OF GERONTOLOGY SERIES A-BIOLOGICAL SCIENCES AND MEDICAL SCIENCES. 53, (1998).

85.

O'Brien, T.D., Reeves, N.D., Baltzopoulos, V., Jones, D.A., Maganaris, C.N.: In vivo measurements of muscle specific tension in adults and children. Experimental Physiology. 95, 202–210 (2010). https://doi.org/10.1113/expphysiol.2009.048967.

86.

Clarke, C., Howard, R., Rossor, M., Shorvon, S.D., National Hospital for Neurology and Neurosurgery (London, England), Institute of Neurology, Queen Square: Neurology: a Queen Square textbook. Wiley-Blackwell, Chichester (2009).

87.

Neuromuscular Disease Centre, http://neuromuscular.wustl.edu/.

88.

Richard J. Barohn: A PATTERN RECOGNITION APPROACH TO THE PATIENT WITH A SUSPECTED MYOPATHY. Neurologic clinics. 32, (2014). https://doi.org/10.1016/j.ncl.2014.04.008.

89.

Wattjes, M.P., Kley, R.A., Fischer, D.: Neuromuscular imaging in inherited muscle diseases. European Radiology. 20, 2447–2460 (2010). https://doi.org/10.1007/s00330-010-1799-2.

90.

Morrow, J.M., Sinclair, C.D.J., Fischmann, A., Machado, P.M., Reilly, M.M., Yousry, T.A., Thornton, J.S., Hanna, M.G.: MRI biomarker assessment of neuromuscular disease progression: a prospective observational cohort study. The Lancet Neurology. 15, 65–77 (2016). https://doi.org/10.1016/S1474-4422(15)00242-2.

91.

Briggs, D., Morgan, J.E.: Recent progress in satellite cell/myoblast engraftment - relevance for therapy. FEBS Journal. 280, 4281–4293 (2013). https://doi.org/10.1111/febs.12273.

92.

Peter S. Zammit: Muscle satellite cells adopt divergent fates: a mechanism for self-renewal? The Journal of Cell Biology. 166, (2004). https://doi.org/10.1083/jcb.200312007.

93.

Boldrin, L., Morgan, J.E.: Activating muscle stem cells: therapeutic potential in muscle diseases. Current Opinion in Neurology. 20, 577–582 (2007).

https://doi.org/10.1097/WCO.0b013e3282ef5919.

94.

Boldrin, L., Zammit, P.S., Morgan, J.E.: Satellite cells from dystrophic muscle retain regenerative capacity. Stem Cell Research. 14, 20–29 (2015). https://doi.org/10.1016/j.scr.2014.10.007.

95.

Ross, J., Benn, A., Jonuschies, J., Boldrin, L., Muntoni, F., Hewitt, J.E., Brown, S.C., Morgan, J.E.: Defects in Glycosylation Impair Satellite Stem Cell Function and Niche Composition in the Muscles of the Dystrophic Large Mouse. STEM CELLS. 30, 2330–2341 (2012). https://doi.org/10.1002/stem.1197.

96.

Ravenscroft, G., Davis, M.R., Lamont, P., Forrest, A., Laing, N.G.: New era in genetics of early-onset muscle disease: Breakthroughs and challenges. Seminars in Cell & Developmental Biology. 64, 160–170 (2017). https://doi.org/10.1016/j.semcdb.2016.08.002.

97.

Ravenscroft, G., Laing, N.G., Bönnemann, C.G.: Pathophysiological concepts in the congenital myopathies: blurring the boundaries, sharpening the focus. Brain. 138, 246–268 (2015). https://doi.org/10.1093/brain/awu368.

98.

North, K.N., Wang, C.H., Clarke, N., Jungbluth, H., Vainzof, M., Dowling, J.J., Amburgey, K., Quijano-Roy, S., Beggs, A.H., Sewry, C., Laing, N.G., Bönnemann, C.G.: Approach to the diagnosis of congenital myopathies. Neuromuscular Disorders. 24, 97–116 (2014). https://doi.org/10.1016/j.nmd.2013.11.003.

99.

Quijano-Roy, S., Carlier, R.Y., Fischer, D.: Muscle Imaging in Congenital Myopathies. Seminars in Pediatric Neurology. 18, 221–229 (2011). https://doi.org/10.1016/j.spen.2011.10.003.

100.

Kimberly Amburgey: A natural history study of X-linked myotubular myopathy. Neurology. 89, (2017). https://doi.org/10.1212/WNL.0000000000004415.

101.

Irene Colombo: Congenital myopathies: Natural history of a large pediatric cohort. Neurology. 84, (2015). https://doi.org/10.1212/WNL.0000000000001110.

102.

Guidance for Paediatric Physiotherapists - Managing Neuromuscular Disorders, http://apcp.csp.org.uk/publications/guidance-paediatric-physiotherapists-managing-neuromuscular-disorders.

103.

APCP, http://apcp.csp.org.uk/.

104.

Neuromuscular | Department of Neurology, https://neuro.wustl.edu/education/fellowships/neuromuscular/.

105.

Muscular Dystrophy UK, http://www.musculardystrophyuk.org/.

106.

Hollak, C.E.M., Lachmann, R. eds: Inherited metabolic disease in adults: a clinical guide. Oxford University Press, [New York] (2016).

107.

Saudubray, J.M., Baumgartner, M.R., Walter, J. eds: Inborn metabolic diseases: diagnosis and treatment. Springer, Berlin (2016).

108.

Hoffmann, G.F., Zschocke, J., Nyhan, W.L.: Inherited metabolic diseases: a clinical approach. Springer, Heidelberg (2009).

109.

Machado, P., Brady, S., Hanna, M.G.: Update in inclusion body myositis. Current Opinion in Rheumatology. 25, 763–771 (2013). https://doi.org/10.1097/01.bor.0000434671.77891.9a.

110.

Machado, P.M., Dimachkie, M.M., Barohn, R.J.: Sporadic inclusion body myositis. Current Opinion in Neurology. 27, 591–598 (2014). https://doi.org/10.1097/WCO.0000000000000129.

111.

Machado, P.M., Ahmed, M., Brady, S., Gang, Q., Healy, E., Morrow, J.M., Wallace, A.C., Dewar, L., Ramdharry, G., Parton, M., Holton, J.L., Houlden, H., Greensmith, L., Hanna, M.G.: Ongoing Developments in Sporadic Inclusion Body Myositis. Current Rheumatology Reports. 16, (2014). https://doi.org/10.1007/s11926-014-0477-9.

112.

Mhoriam Ahmed: Targeting Protein Homeostasis in Sporadic Inclusion Body Myositis. Science translational medicine. 8, (2016). https://doi.org/10.1126/scitranslmed.aad4583.

113.

Needham, M., Mastaglia, F.L.: Sporadic inclusion body myositis: A review of recent clinical advances and current approaches to diagnosis and treatment. Clinical Neurophysiology. 127, 1764–1773 (2016). https://doi.org/10.1016/j.clinph.2015.12.011.

114.

Schröder, R., Schoser, B.: Myofibrillar Myopathies: A Clinical and Myopathological Guide. Brain Pathology. 19, 483–492 (2009). https://doi.org/10.1111/j.1750-3639.2009.00289.x.

115.

Amato, A.A., Greenberg, S.A.: Inflammatory Myopathies. CONTINUUM: Lifelong Learning in Neurology. 19, 1615–1633 (2013). https://doi.org/10.1212/01.CON.0000440662.26427.bd.

116.

Olivé, M., Kley, R.A., Goldfarb, L.G.: Myofibrillar myopathies. Current Opinion in Neurology. 26, 527–535 (2013). https://doi.org/10.1097/WCO.0b013e328364d6b1.

117.

Carstens, P.-O., Schmidt, J.: Diagnosis, pathogenesis and treatment of myositis: recent advances. Clinical & Experimental Immunology. 175, 349–358 (2014). https://doi.org/10.1111/cei.12194.

118.

Dalakas, M.C.: Inflammatory Muscle Diseases. New England Journal of Medicine. 372, 1734–1747 (2015). https://doi.org/10.1056/NEJMra1402225.

119.

Great Britain. Audit Commission for Local Authorities and the National Health Service in England and Wales.: What Seems to Be the Matter Communication (National Health Service Report). Stationery Office.

120.

T. D. Bunker: An information leaflet for surgical patients. Annals of The Royal College of Surgeons of England. 65, (1983).

121.

Toolkit for producing patient information.pdf, https://www.uea.ac.uk/documents/246046/0/Toolkit+for+producing+patient+information.pdf.

122.

Garner, M., Ning, Z., Francis, J.: A framework for the evaluation of patient information leaflets. Health Expectations. 15, 283–294 (2012). https://doi.org/10.1111/j.1369-7625.2011.00665.x.

123.

C F George: Prescription information leaflets: a pilot study in general practice. British Medical Journal (Clinical research ed.). 287, (1983).

124.

Hollingsworth, K.G., de Sousa, P.L., Straub, V., Carlier, P.G.: Towards harmonization of protocols for MRI outcome measures in skeletal muscle studies: Consensus recommendations from two TREAT-NMD NMR workshops, 2 May 2010, Stockholm, Sweden, 1–2 October 2009, Paris, France. Neuromuscular Disorders. 22, S54–S67 (2012). https://doi.org/10.1016/j.nmd.2012.06.005.

125.

Wattjes, M.P., Fischer, D.: Neuromuscular imaging. Springer, New York (2013).

126.

Forbes, S.C., Willcocks, R.J., Triplett, W.T., Rooney, W.D., Lott, D.J., Wang, D.-J., Pollaro, J., Senesac, C.R., Daniels, M.J., Finkel, R.S., Russman, B.S., Byrne, B.J., Finanger, E.L., Tennekoon, G.I., Walter, G.A., Sweeney, H.L., Vandenborne, K.: Magnetic Resonance Imaging and Spectroscopy Assessment of Lower Extremity Skeletal Muscles in Boys with Duchenne Muscular Dystrophy: A Multicenter Cross Sectional Study. PLoS ONE. 9, (2014). https://doi.org/10.1371/journal.pone.0106435.

127.

Glover, G.H., Schneider, E.: Three-point dixon technique for true water/fat decomposition withB0 inhomogeneity correction. Magnetic Resonance in Medicine. 18, 371–383 (1991). https://doi.org/10.1002/mrm.1910180211.

128.

Willcocks, R.J., Rooney, W.D., Triplett, W.T., Forbes, S.C., Lott, D.J., Senesac, C.R., Daniels, M.J., Wang, D.-J., Harrington, A.T., Tennekoon, G.I., Russman, B.S., Finanger, E.L., Byrne, B.J., Finkel, R.S., Walter, G.A., Sweeney, H.L., Vandenborne, K.: Multicenter prospective longitudinal study of magnetic resonance biomarkers in a large duchenne muscular dystrophy cohort. Annals of Neurology. 79, 535–547 (2016). https://doi.org/10.1002/ana.24599.

129.

Jean-Yves Hogrel: Longitudinal functional and NMR assessment of upper limbs in Duchenne muscular dystrophy. Neurology. 86, (2016). https://doi.org/10.1212/WNL.0000000000002464.

130.

American Journal of Roentgenology.

131.

Hollak, C.E.M., Lachmann, R. eds: Inherited metabolic disease in adults: a clinical guide. Oxford University Press, [New York] (2016).

132.

Nancy D Leslie: Very Long-Chain Acyl-Coenzyme A Dehydrogenase Deficiency. (2014).

133.

Thomas Wieser: Carnitine Palmitoyltransferase II Deficiency. (2017).

134.

Miguel A Martín: Glycogen Storage Disease Type V. (2014).

135.

Nancy Leslie: Pompe Disease. (2017).

136.

Ørngreen, M.C., Vissing, J.: Treatment Opportunities in Patients With Metabolic Myopathies. Current Treatment Options in Neurology. 19, (2017). https://doi.org/10.1007/s11940-017-0473-2.

137.

Olpin, S.E., Murphy, E., Kirk, R.J., Taylor, R.W., Quinlivan, R.: The investigation and management of metabolic myopathies. Journal of Clinical Pathology. 68, 410–417 (2015). https://doi.org/10.1136/jclinpath-2014-202808.

138.

Muntoni, F., Torelli, S., Ferlini, A.: Dystrophin and mutations: one gene, several proteins, multiple phenotypes. The Lancet Neurology. 2, 731–740 (2003). https://doi.org/10.1016/S1474-4422(03)00585-4.

139.

Bushby, K., Finkel, R., Birnkrant, D.J., Case, L.E., Clemens, P.R., Cripe, L., Kaul, A., Kinnett, K., McDonald, C., Pandya, S., Poysky, J., Shapiro, F., Tomezsko, J., Constantin, C.: Diagnosis and management of Duchenne muscular dystrophy, part 1: diagnosis, and pharmacological and psychosocial management. The Lancet Neurology. 9, 77–93 (2010). https://doi.org/10.1016/S1474-4422(09)70271-6.

140.

Bushby, K., Finkel, R., Birnkrant, D.J., Case, L.E., Clemens, P.R., Cripe, L., Kaul, A., Kinnett, K., McDonald, C., Pandya, S., Poysky, J., Shapiro, F., Tomezsko, J., Constantin, C.: Diagnosis and management of Duchenne muscular dystrophy, part 2: implementation of multidisciplinary care. The Lancet Neurology. 9, 177–189 (2010). https://doi.org/10.1016/S1474-4422(09)70272-8.

141.

Ricotti, V., Ridout, D.A., Scott, E., Quinlivan, R., Robb, S.A., Manzur, A.Y., Muntoni, F., Manzur, A., Muntoni, F., Robb, S., Quinlivan, R., Ricotti, V., Main, M., Bushby, K., Straub, V., Sarkozy, A., Guglieri, M., Strehle, E., Eagle, M., Mayhew, A., Roper, H., McMurchie, H., Childs, A., Pysden, K., Pallant, L., Spinty, S., Peachey, G., Shillington, A., Wraige, E., Jungbluth, H., Sheehan, J., Spahr, R., Hughes, I., Bateman, E., Cammiss, C., Willis, T., Groves, L., Emery, N., Baxter, P., Senior, M., Hartley, L., Parsons, B., Majumdar, A., Jenkins, L., Naismith, K., Keddie, A., Horrocks, I., Di Marco, M., Chow, G., Miah, A.: Long-term benefits and adverse effects of intermittent versus daily glucocorticoids in boys with Duchenne muscular dystrophy. Journal of Neurology, Neurosurgery & Psychiatry. 84, 698–705 (2013). https://doi.org/10.1136/jnnp-2012-303902.

142.

Goemans, N.M., Tulinius, M., van den Akker, J.T., Burm, B.E., Ekhart, P.F., Heuvelmans, N., Holling, T., Janson, A.A., Platenburg, G.J., Sipkens, J.A., Sitsen, J.M.A., Aartsma-Rus, A., van Ommen, G.-J.B., Buyse, G., Darin, N., Verschuuren, J.J., Campion, G.V., de Kimpe, S.J., van Deutekom, J.C.: Systemic Administration of PRO051 in Duchenne's Muscular Dystrophy. New England Journal of Medicine. 364, 1513–1522 (2011). https://doi.org/10.1056/NEJMoa1011367.

143.

Mendell, J.R., Rodino-Klapac, L.R., Sahenk, Z., Roush, K., Bird, L., Lowes, L.P., Alfano, L., Gomez, A.M., Lewis, S., Kota, J., Malik, V., Shontz, K., Walker, C.M., Flanigan, K.M., Corridore, M., Kean, J.R., Allen, H.D., Shilling, C., Melia, K.R., Sazani, P., Saoud, J.B., Kaye, E.M.: Eteplirsen for the treatment of Duchenne muscular dystrophy. Annals of Neurology. 74, 637–647 (2013). https://doi.org/10.1002/ana.23982.

144.

Mendell, J.R., Goemans, N., Lowes, L.P., Alfano, L.N., Berry, K., Shao, J., Kaye, E.M., Mercuri, E.: Longitudinal effect of eteplirsen versus historical control on ambulation in Duchenne muscular dystrophy. Annals of Neurology. 79, 257–271 (2016). https://doi.org/10.1002/ana.24555.

145.

Kinali, M., Arechavala-Gomeza, V., Feng, L., Cirak, S., Hunt, D., Adkin, C., Guglieri, M., Ashton, E., Abbs, S., Nihoyannopoulos, P., Garralda, M.E., Rutherford, M., Mcculley, C., Popplewell, L., Graham, I.R., Dickson, G., Wood, M.J., Wells, D.J., Wilton, S.D., Kole, R.,

Straub, V., Bushby, K., Sewry, C., Morgan, J.E., Muntoni, F.: Local restoration of dystrophin expression with the morpholino oligomer AVI-4658 in Duchenne muscular dystrophy: a single-blind, placebo-controlled, dose-escalation, proof-of-concept study. The Lancet Neurology. 8, 918–928 (2009). https://doi.org/10.1016/S1474-4422(09)70211-X.

146.

Cirak, S., Arechavala-Gomeza, V., Guglieri, M., Feng, L., Torelli, S., Anthony, K., Abbs, S., Garralda, M.E., Bourke, J., Wells, D.J., Dickson, G., Wood, M.J., Wilton, S.D., Straub, V., Kole, R., Shrewsbury, S.B., Sewry, C., Morgan, J.E., Bushby, K., Muntoni, F.: Exon skipping and dystrophin restoration in patients with Duchenne muscular dystrophy after systemic phosphorodiamidate morpholino oligomer treatment: an open-label, phase 2, dose-escalation study. The Lancet. 378, 595–605 (2011). https://doi.org/10.1016/S0140-6736(11)60756-3.

147.

Bushby, K., Finkel, R., Wong, B., Barohn, R., Campbell, C., Comi, G.P., Connolly, A.M., Day, J.W., Flanigan, K.M., Goemans, N., Jones, K.J., Mercuri, E., Quinlivan, R., Renfroe, J.B., Russman, B., Ryan, M.M., Tulinius, M., Voit, T., Moore, S.A., Lee Sweeney, H., Abresch, R.T., Coleman, K.L., Eagle, M., Florence, J., Gappmaier, E., Glanzman, A.M., Henricson, E., Barth, J., Elfring, G.L., Reha, A., Spiegel, R.J., O'donnell, M.W., Peltz, S.W., Mcdonald, C.M.: Ataluren treatment of patients with nonsense mutation dystrophinopathy. Muscle & Nerve. 50, 477–487 (2014). https://doi.org/10.1002/mus.24332.

148.

Victorian Department of Health / University of Melbourne, https://www2.health.vic.gov.au/.

149.

Best Practice in Memory Services: Learning from across England, https://www.england.nhs.uk/wp-content/uploads/2014/12/memory-clinics-final.pdf.

150.

Matthews, E., Fialho, D., Tan, S.V., Venance, S.L., Cannon, S.C., Sternberg, D., Fontaine, B., Amato, A.A., Barohn, R.J., Griggs, R.C., Hanna, M.G.: The non-dystrophic myotonias: molecular pathogenesis, diagnosis and treatment. Brain. 133, 9–22 (2010). https://doi.org/10.1093/brain/awp294.

151.

Venance, S.L., Cannon, S.C., Fialho, D., Fontaine, B., Hanna, M.G., Ptacek, L.J., Tristani-Firouzi, M., Tawil, R., Griggs, R.C.: The primary periodic paralyses: diagnosis, pathogenesis and treatment. Brain. 129, 8–17 (2006). https://doi.org/10.1093/brain/awh639.

152.

Cannon, S.C.: Channelopathies of Skeletal Muscle Excitability. In: Terjung, R. (ed.) Comprehensive Physiology. pp. 761–790. John Wiley & Sons, Inc., Hoboken, NJ, USA (2011). https://doi.org/10.1002/cphy.c140062.

153.

Tan, S.V., Matthews, E., Barber, M., Burge, J.A., Rajakulendran, S., Fialho, D., Sud, R., Haworth, A., Koltzenburg, M., Hanna, M.G.: Refined exercise testing can aid dna-based diagnosis in muscle channelopathies. Annals of Neurology. 69, 328–340 (2011). https://doi.org/10.1002/ana.22238.

154.

Sharp, L., Trivedi, J.R.: Treatment and Management of Neuromuscular Channelopathies. Current Treatment Options in Neurology. 16, (2014). https://doi.org/10.1007/s11940-014-0313-6.

155.

Paganoni, S., Amato, A.: Electrodiagnostic Evaluation of Myopathies. Physical Medicine and Rehabilitation Clinics of North America. 24, 193–207 (2013). https://doi.org/10.1016/j.pmr.2012.08.017.

156.

Fuglsang-Frederiksen, A.: The role of different EMG methods in evaluating myopathy. Clinical Neurophysiology. 117, 1173–1189 (2006). https://doi.org/10.1016/j.clinph.2005.12.018.

157.

Ferlini, A., Scotton, C., Novelli, G.: Biomarkers in Rare Diseases. Public Health Genomics. 16, 313–321 (2013). https://doi.org/10.1159/000355938.

158.

Boers, M (Boers, M); Brooks, P (Brooks, P); Strand, CV (Strand, CV); Tugwell, P (Tugwell, P): The OMERACT filter for outcome measures in rheumatology. JOURNAL OF RHEUMATOLOGY JOURNAL OF RHEUMATOLOGY. 25, 198–199 (1998).

159.

Conwit, R.A., Bhanushali, M.J., Porter, J.D., Kaufmann, P., Gutmann, L.: Adding more muscle and nerve to clinical trials. Muscle & Nerve. 44, 695–702 (2011). https://doi.org/10.1002/mus.22130.

160.

Qualification Process for Drug Development Tools, https://www.fda.gov/downloads/drugs/guidances/ucm230597.pdf.

161.

Ahmed, M., Machado, P.M., Miller, A., Spicer, C., Herbelin, L., He, J., Noel, J., Wang, Y., McVey, A.L., Pasnoor, M., Gallagher, P., Statland, J., Lu, C.-H., Kalmar, B., Brady, S., Sethi, H., Samandouras, G., Parton, M., Holton, J.L., Weston, A., Collinson, L., Taylor, J.P., Schiavo, G., Hanna, M.G., Barohn, R.J., Dimachkie, M.M., Greensmith, L.: Targeting protein homeostasis in sporadic inclusion body myositis. Science Translational Medicine. 8, 331ra41-331ra41 (2016). https://doi.org/10.1126/scitranslmed.aad4583.

162.

A Cruz-Martínez: Single fiber electromyography (SFEMG) in mitochondrial diseases (MD). Electromyography and clinical neurophysiology. (2004).

163.

Hull, J., Aniapravan, R., Chan, E., Chatwin, M., Forton, J., Gallagher, J., Gibson, N., Gordon, J., Hughes, I., McCulloch, R., Russell, R.R., Simonds, A.: British Thoracic Society guideline

for respiratory management of children with neuromuscular weakness. Thorax. 67, i1–i40 (2012). https://doi.org/10.1136/thoraxjnl-2012-201964.

164.

American Journal of Respiratory and Critical Care Medicine.

165.

Ward, S.: Randomised controlled trial of non-invasive ventilation (NIV) for nocturnal hypoventilation in neuromuscular and chest wall disease patients with daytime normocapnia. Thorax. 60, 1019–1024 (2005). https://doi.org/10.1136/thx.2004.037424.

166.

Safety and efficacy of diaphragm pacing in patients with respiratory insufficiency due to amyotrophic lateral sclerosis (DiPALS): a multicentre, open-label, randomised controlled trial. The Lancet Neurology. 14, 883–892 (2015). https://doi.org/10.1016/S1474-4422(15)00152-0.

167.

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.

168.

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.

169.

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

170.

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.

171.

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.

172.

Nishimune, H.: Active zones of mammalian neuromuscular junctions: formation, density, and aging. Annals of the New York Academy of Sciences. 1274, 24–32 (2012). https://doi.org/10.1111/j.1749-6632.2012.06836.x.

173.

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.

174.

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., 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.

175.

Simon, N.G., Turner, M.R., Vucic, S., Al-Chalabi, A., Shefner, J., Lomen-Hoerth, C., Kiernan, M.C.: Quantifying disease progression in amyotrophic lateral sclerosis. Annals of Neurology. 76, 643–657 (2014). https://doi.org/10.1002/ana.24273.

176.

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.

177.

Caballero-Hernandez, D., Toscano, M.G., Cejudo-Guillen, M., Garcia-Martin, M.L., Lopez, S., Franco, J.M., Quintana, F.J., Roodveldt, C., Pozo, D.: The 'Omics' of Amyotrophic Lateral Sclerosis. Trends in Molecular Medicine. 22, 53–67 (2016). https://doi.org/10.1016/j.molmed.2015.11.001.

178.

Michael Benatar: ALS Biomarkers for Therapy Development: State of the Field & Future Directions. Muscle & nerve. 53, (2016). https://doi.org/10.1002/mus.24979.

179.

Ulf Andreasson: Update on ultrasensitive technologies to facilitate research on blood biomarkers for central nervous system disorders. Alzheimer's & Dementia : Diagnosis, Assessment & Disease Monitoring. 3, (2016). https://doi.org/10.1016/j.dadm.2016.05.005.

180.

Kang, J.-H., Korecka, M., Figurski, M.J., Toledo, J.B., Blennow, K., Zetterberg, H., Waligorska, T., Brylska, M., Fields, L., Shah, N., Soares, H., Dean, R.A., Vanderstichele, H., Petersen, R.C., Aisen, P.S., Saykin, A.J., Weiner, M.W., Trojanowski, J.Q., Shaw, L.M.: The Alzheimer's Disease Neuroimaging Initiative 2 Biomarker Core: A review of progress and plans. Alzheimer's & Dementia. 11, 772–791 (2015). https://doi.org/10.1016/j.jalz.2015.05.003.

181.

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 (2016). https://doi.org/10.1038/nrneurol.2016.182.

182.

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.

183.

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.

184.

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.

185.

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

186.

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.

187.

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.

188.

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

189.

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.

190.

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.

191.

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.

192.

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.

193.

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.

194.

Robert M. Brownstone: Spinal interneurons providing input to the final common path during locomotion. Progress in brain research. 187, (2010). https://doi.org/10.1016/B978-0-444-53613-6.00006-X.

195.

O'Connor, E., Töpf, A., Zahedi, R., 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. (2018). https://doi.org/10.1111/nyas.13520.

196.

Andrew G. Engel: Congenital myasthenic syndromes: pathogenesis, diagnosis, and treatment. The Lancet. Neurology. 14, (2015). https://doi.org/10.1016/S1474-4422(14)70201-7.

197.

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.

198.

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.

199.

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 cause congenital myasthenic syndrome and bridge myasthenic disorders with dystroglycanopathies. Brain. 138, 2493–2504 (2015). https://doi.org/10.1093/brain/awv185.

200.

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.

201.

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.

202.

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

203.

Matthew N Meriggioli: Autoimmune myasthenia gravis: emerging clinical and biological heterogeneity. Lancet neurology. 8, (2009). https://doi.org/10.1016/S1474-4422(09)70063-8.

204.

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.

205.

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

206.

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

207.

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

208.

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.

209.

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

210.

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

211.

Leslie Jacobson: Plasma from human mothers of fetuses with severe arthrogryposis multiplex congenita causes deformities in mice. Journal of Clinical Investigation. 103, (1999). https://doi.org/10.1172/JCI5943.

212.

Hoch, W., McConville, J., Helms, S., Newsom-Davis, J., Melms, A., Vincent, A.: Auto-antibodies to the receptor tyrosine kinase MuSK in patients with myasthenia gravis without acetylcholine receptor antibodies. Nature Medicine. 7, 365–368 (2001). https://doi.org/10.1038/85520.

213.

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.

214.

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.

215.

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.

216.

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.

217.

Beryl B. Cummings: Improving genetic diagnosis in Mendelian disease with transcriptome sequencing. Science translational medicine. 9, (2017). https://doi.org/10.1126/scitranslmed.aal5209.

218.

Schofield, D., Alam, K., Douglas, L., Shrestha, R., MacArthur, D.G., Davis, M., Laing, N.G., Clarke, N.F., Burns, J., Cooper, S.T., North, K.N., Sandaradura, S.A., O'Grady, G.L.: Cost-effectiveness of massively parallel sequencing for diagnosis of paediatric muscle diseases. npj Genomic Medicine. 2, (2017). https://doi.org/10.1038/s41525-017-0006-7.

219.

O'Grady, G.L., Lek, M., Lamande, S.R., Waddell, L., Oates, E.C., Punetha, J., Ghaoui, R., Sandaradura, S.A., Best, H., Kaur, S., Davis, M., Laing, N.G., Muntoni, F., Hoffman, E., MacArthur, D.G., Clarke, N.F., Cooper, S., North, K.: Diagnosis and etiology of congenital muscular dystrophy: We are halfway there. Annals of Neurology. 80, 101–111 (2016). https://doi.org/10.1002/ana.24687.

220.

Bönnemann, C.G., Wang, C.H., Quijano-Roy, S., Deconinck, N., Bertini, E., Ferreiro, A., Muntoni, F., Sewry, C., Béroud, C., Mathews, K.D., Moore, S.A., Bellini, J., Rutkowski, A., North, K.N.: Diagnostic approach to the congenital muscular dystrophies. Neuromuscular Disorders. 24, 289–311 (2014). https://doi.org/10.1016/j.nmd.2013.12.011.

221.

North, K.N., Wang, C.H., Clarke, N., Jungbluth, H., Vainzof, M., Dowling, J.J., Amburgey, K., Quijano-Roy, S., Beggs, A.H., Sewry, C., Laing, N.G., Bönnemann, C.G.: Approach to the diagnosis of congenital myopathies. Neuromuscular Disorders. 24, 97–116 (2014).

https://doi.org/10.1016/j.nmd.2013.11.003.

222.

Menezes, M.P., North, K.N.: Inherited neuromuscular disorders: Pathway to diagnosis. Journal of Paediatrics and Child Health. 48, 458–465 (2012). https://doi.org/10.1111/j.1440-1754.2011.02210.x.

223.

Milestones timeline : Nature Milestones in DNA, https://www.nature.com/milestones/miledna/timeline.html.

224.

Metzker, M.L.: Sequencing technologies — the next generation. Nature Reviews Genetics. 11, 31–46 (2010). https://doi.org/10.1038/nrg2626.

225.

Yang, Y., Muzny, D.M., Reid, J.G., Bainbridge, M.N., Willis, A., Ward, P.A., Braxton, A., Beuten, J., Xia, F., Niu, Z., Hardison, M., Person, R., Bekheirnia, M.R., Leduc, M.S., Kirby, A., Pham, P., Scull, J., Wang, M., Ding, Y., Plon, S.E., Lupski, J.R., Beaudet, A.L., Gibbs, R.A., Eng, C.M.: Clinical Whole-Exome Sequencing for the Diagnosis of Mendelian Disorders. New England Journal of Medicine. 369, 1502–1511 (2013). https://doi.org/10.1056/NEJMoa1306555.

226.

Sun, Y., Ruivenkamp, C.A.L., Hoffer, M.J.V., Vrijenhoek, T., Kriek, M., van Asperen, C.J., den Dunnen, J.T., Santen, G.W.E.: Next-Generation Diagnostics: Gene Panel, Exome, or Whole Genome? Human Mutation. 36, 648–655 (2015). https://doi.org/10.1002/humu.22783.

227.

Ghaoui, R., Cooper, S.T., Lek, M., Jones, K., Corbett, A., Reddel, S.W., Needham, M., Liang, C., Waddell, L.B., Nicholson, G., O'Grady, G., Kaur, S., Ong, R., Davis, M., Sue, C.M., Laing, N.G., North, K.N., MacArthur, D.G., Clarke, N.F.: Use of Whole-Exome Sequencing for Diagnosis of Limb-Girdle Muscular Dystrophy. JAMA Neurology. 72, (2015). https://doi.org/10.1001/jamaneurol.2015.2274.

228.

Richards, S., Aziz, N., Bale, S., Bick, D., Das, S., Gastier-Foster, J., Grody, W.W., Hegde, M., Lyon, E., Spector, E., Voelkerding, K., Rehm, H.L.: Standards and guidelines for the interpretation of sequence variants: a joint consensus recommendation of the American College of Medical Genetics and Genomics and the Association for Molecular Pathology. Genetics in Medicine. 17, 405–423 (2015). https://doi.org/10.1038/gim.2015.30.

229.

Gabrielle Natalie Samuel: The UK's 100,000 Genomes Project: manifesting policymakers' expectations. New Genetics and Society. 36, (2017). https://doi.org/10.1080/14636778.2017.1370671.

230.

Samuel, G.N., Farsides, B.: Public trust and 'ethics review' as a commodity: the case of Genomics England Limited and the UK's 100,000 genomes project. Medicine, Health Care and Philosophy. (2017). https://doi.org/10.1007/s11019-017-9810-1.

231.

Vaithinathan, A.G., Asokan, V.: Public health and precision medicine share a goal. Journal of Evidence-Based Medicine. 10, 76–80 (2017). https://doi.org/10.1111/jebm.12239.

232.

Griffin, B.H., Chitty, L.S., Bitner-Glindzicz, M.: The 100 000 Genomes Project: What it means for paediatrics. Archives of disease in childhood - Education & practice edition. 102, 105–107 (2017). https://doi.org/10.1136/archdischild-2016-311029.

233.

Evers, M.M., Toonen, L.J.A., van Roon-Mom, W.M.C.: Antisense oligonucleotides in therapy for neurodegenerative disorders. Advanced Drug Delivery Reviews. 87, 90–103 (2015). https://doi.org/10.1016/j.addr.2015.03.008.

234.

Karin E. Lundin: Oligonucleotide Therapies: The Past and the Present. Human Gene Therapy. 26, (2015). https://doi.org/10.1089/hum.2015.070.

235.

Khorkova, O., Wahlestedt, C.: Oligonucleotide therapies for disorders of the nervous system. Nature Biotechnology. 35, 249–263 (2017). https://doi.org/10.1038/nbt.3784.

236.

Aartsma-Rus, A.: Overview on AON Design. In: Aartsma-Rus, A. (ed.) Exon Skipping. pp. 117–129. Humana Press, Totowa, NJ (2012). https://doi.org/10.1007/978-1-61779-767-5_8.

237.

Rossor, A.M., Reilly, M.M., Sleigh, J.N.: Antisense oligonucleotides and other genetic therapies made simple. Practical Neurology. (2018). https://doi.org/10.1136/practneurol-2017-001764.

238.

Wood, N.: Neurogenetics. Cambridge University Press, Cambridge (2012). https://doi.org/10.1017/CBO9781139087711.

239.

Clarke, C., Howard, R., Rossor, M., Shorvon, S. eds: Neurology. John Wiley &;#38; Sons, Ltd, Chichester, UK (2016). https://doi.org/10.1002/9781118486160.

240.

OMIM - Online Mendelian Inheritance in Man, https://www.omim.org/.

241.

Teboul, L., Hérault, Y., Smith, C., Whitelaw, B.: Introduction to Mammalian Genome Special Issue: Genome Editing. Mammalian Genome. 28, 235–236 (2017).

https://doi.org/10.1007/s00335-017-9708-5.

242.

Fernández, A., Josa, S., Montoliu, L.: A history of genome editing in mammals. Mammalian Genome. 28, 237–246 (2017). https://doi.org/10.1007/s00335-017-9699-2.

243.

Marie-Christine Birling: Modeling human disease in rodents by CRISPR/Cas9 genome editing. Mammalian Genome. 28, (2017). https://doi.org/10.1007/s00335-017-9703-x.

244.

Greenfield, A.: Editing mammalian genomes: ethical considerations. Mammalian Genome. 28, 388–393 (2017). https://doi.org/10.1007/s00335-017-9702-y.

245.

Addgene: CRISPR Guide, https://www.addgene.org/crispr/guide/.

246.

MGI-Mouse Genome Informatics -The international database resource for the laboratory mouse, http://www.informatics.jax.org/.

247.

Rossor, A.M., Tomaselli, P.J., Reilly, M.M.: Recent advances in the genetic neuropathies. Current Opinion in Neurology. (2016). https://doi.org/10.1097/WCO.0000000000000373.

248.

Rossor, A.M., Evans, M.R.B., Reilly, M.M.: A practical approach to the genetic neuropathies. Practical Neurology. 15, 187–198 (2015). https://doi.org/10.1136/practneurol-2015-001095.