

# MSc Neuromuscular Disease

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[1]

A. Caton The branchial arches and HGF are growth-promoting and chemoattractant for cranial motor axons. *Development*. 127, 8, 1751–1766.

[2]

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

[3]

A. M. Gordon 2000. Regulation of Contraction in Striated Muscle. *Physiological Reviews*. 80, 2 (Jan. 2000), 853–924.

[4]

Aartsma-Rus, A. 2012. Overview on AON Design. Exon Skipping. A. Aartsma-Rus, ed. Humana Press. 117–129.

[5]

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

[6]

Ahmed, M. et al. 2016. Targeting protein homeostasis in sporadic inclusion body myositis. *Science Translational Medicine*. 8, 331 (Mar. 2016), 331ra41–331ra41. <https://doi.org/10.1126/scitranslmed.aad4583>.

[7]

Al-Chalabi, A. et al. 2016. Gene discovery in amyotrophic lateral sclerosis: implications for clinical management. *Nature Reviews Neurology*. 13, 2 (Dec. 2016), 96–104.  
<https://doi.org/10.1038/nrneurol.2016.182>.

[8]

AM Rossor 2016. Recent advances in the genetic neuropathies. *Current opinion in neurology*. 29, 5 (2016). <https://doi.org/10.1097/WCO.0000000000000373>.

[9]

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

[10]

Amato, A.A. and Russell, J.A. 2008. *Neuromuscular disorders*. McGraw-Hill.

[11]

Andrew G. Engel 2015. Congenital myasthenic syndromes: pathogenesis, diagnosis, and treatment. *The Lancet. Neurology*. 14, 4 (2015).  
[https://doi.org/10.1016/S1474-4422\(14\)70201-7](https://doi.org/10.1016/S1474-4422(14)70201-7).

[12]

Antoine, J.-C. and Camdessanché, J.-P. 2013. Paraneoplastic disorders of the peripheral nervous system. *La Presse Médicale*. 42, 6 (June 2013), e235–e244.  
<https://doi.org/10.1016/j.lpm.2013.01.059>.

[13]

APCP: <http://apcp.csp.org.uk/>.

[14]

Arthur-Farraj, P.J. et al. 2012. c-Jun Reprograms Schwann Cells of Injured Nerves to Generate a Repair Cell Essential for Regeneration. *Neuron*. 75, 4 (Aug. 2012), 633–647. <https://doi.org/10.1016/j.neuron.2012.06.021>.

[15]

Auer-Grumbach, M. 2013. Hereditary sensory and autonomic neuropathies. *Peripheral Nerve Disorders*. Elsevier. 893–906.

[16]

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>.

[17]

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

[18]

Benson, M.K.D. 2011. *Children's neuromuscular disorders*. Springer.

[19]

Berthelsen, M.P. et al. 2014. Anti-gravity training improves walking capacity and postural balance in patients with muscular dystrophy. *Neuromuscular Disorders*. 24, 6 (June 2014), 492–498. <https://doi.org/10.1016/j.nmd.2014.03.001>.

[20]

Beryl B. Cummings 2017. Improving genetic diagnosis in Mendelian disease with transcriptome sequencing. *Science translational medicine*. 9, 386 (2017). <https://doi.org/10.1126/scitranslmed.aal5209>.

[21]

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

[22]

Boers, M (Boers, M); Brooks, P (Brooks, P); Strand, CV (Strand, CV); Tugwell, P (Tugwell, P) 1998. The OMERACT filter for outcome measures in rheumatology. JOURNAL OF RHEUMATOLOGY JOURNAL OF RHEUMATOLOGY. 25, 2 (1998), 198-199.

[23]

Boldrin, L. et al. 2015. Satellite cells from dystrophic muscle retain regenerative capacity. Stem Cell Research. 14, 1 (Jan. 2015), 20-29. <https://doi.org/10.1016/j.scr.2014.10.007>.

[24]

Boldrin, L. and Morgan, J.E. 2007. Activating muscle stem cells: therapeutic potential in muscle diseases. Current Opinion in Neurology. 20, 5 (Oct. 2007), 577-582. <https://doi.org/10.1097/WCO.0b013e3282ef5919>.

[25]

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

[26]

Bönnemann, C.G. et al. 2014. Diagnostic approach to the congenital muscular dystrophies. Neuromuscular Disorders. 24, 4 (Apr. 2014), 289-311. <https://doi.org/10.1016/j.nmd.2013.12.011>.

[27]

Briggs, D. and Morgan, J.E. 2013. Recent progress in satellite cell/myoblast engraftment -

relevance for therapy. *FEBS Journal*. 280, 17 (Sept. 2013), 4281–4293.  
<https://doi.org/10.1111/febs.12273>.

[28]

Brosius Lutz, A. and Barres, B.A. 2014. Contrasting the Glial Response to Axon Injury in the Central and Peripheral Nervous Systems. *Developmental Cell*. 28, 1 (Jan. 2014), 7–17.  
<https://doi.org/10.1016/j.devcel.2013.12.002>.

[29]

Bushby, K. et al. 2014. Ataluren treatment of patients with nonsense mutation dystrophinopathy. *Muscle & Nerve*. 50, 4 (Oct. 2014), 477–487.  
<https://doi.org/10.1002/mus.24332>.

[30]

Bushby, K. et al. 2010. Diagnosis and management of Duchenne muscular dystrophy, part 1: diagnosis, and pharmacological and psychosocial management. *The Lancet Neurology*. 9, 1 (Jan. 2010), 77–93. [https://doi.org/10.1016/S1474-4422\(09\)70271-6](https://doi.org/10.1016/S1474-4422(09)70271-6).

[31]

Bushby, K. et al. 2010. Diagnosis and management of Duchenne muscular dystrophy, part 2: implementation of multidisciplinary care. *The Lancet Neurology*. 9, 2 (Feb. 2010), 177–189. [https://doi.org/10.1016/S1474-4422\(09\)70272-8](https://doi.org/10.1016/S1474-4422(09)70272-8).

[32]

By: Hunter, S (Hunter, S); White, M (White, M); Thompson, M (Thompson, M) 1998.  
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, 3 (1998).

[33]

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

[34]

C F George 1983. Prescription information leaflets: a pilot study in general practice. *British Medical Journal (Clinical research ed.)*. 287, 6400 (1983).

[35]

Caballero-Hernandez, D. et al. 2016. The 'Omics' of Amyotrophic Lateral Sclerosis. *Trends in Molecular Medicine*. 22, 1 (Jan. 2016), 53-67.  
<https://doi.org/10.1016/j.molmed.2015.11.001>.

[36]

Cannon, S.C. 2011. Channelopathies of Skeletal Muscle Excitability. *Comprehensive Physiology*. R. Terjung, ed. John Wiley & Sons, Inc. 761-790.

[37]

Carr, A.S. et al. 2016. A study of the neuropathy associated with transthyretin amyloidosis (ATTR) in the UK. *Journal of Neurology, Neurosurgery & Psychiatry*. 87, 6 (June 2016), 620-627. <https://doi.org/10.1136/jnnp-2015-310907>.

[38]

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

[39]

Carstens, P.-O. and Schmidt, J. 2014. Diagnosis, pathogenesis and treatment of myositis: recent advances. *Clinical & Experimental Immunology*. 175, 3 (Mar. 2014), 349-358.  
<https://doi.org/10.1111/cei.12194>.

[40]

Chhabra, A. 2014. Peripheral MR Neurography. *Neuroimaging Clinics of North America*. 24, 1 (Feb. 2014), 79-89. <https://doi.org/10.1016/j.nic.2013.03.033>.

[41]

Cirak, S. et al. 2011. 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, 9791 (Aug. 2011), 595–605. [https://doi.org/10.1016/S0140-6736\(11\)60756-3](https://doi.org/10.1016/S0140-6736(11)60756-3).

[42]

Clarke, C. et al. eds 2016. *Neurology*. John Wiley & Sons, Ltd.

[43]

Clarke, C. et al. 2009. *Neurology: a Queen Square textbook*. Wiley-Blackwell.

[44]

Collins, M.P. et al. 2010. 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, 3 (Sept. 2010), 176–184. <https://doi.org/10.1111/j.1529-8027.2010.00281.x>.

[45]

Collins, M.P. and Hadden, R.D. 2017. The nonsystemic vasculitic neuropathies. *Nature Reviews Neurology*. 13, 5 (Apr. 2017), 302–316. <https://doi.org/10.1038/nrneurol.2017.42>.

[46]

Conover, J.C. et al. 1995. Neuronal deficits, not involving motor neurons, in mice lacking BDNF and/or NT4. *Nature*. 375, 6528 (May 1995), 235–238. <https://doi.org/10.1038/375235a0>.

[47]

Conwit, R.A. et al. 2011. Adding more muscle and nerve to clinical trials. *Muscle & Nerve*. 44, 5 (Nov. 2011), 695–702. <https://doi.org/10.1002/mus.22130>.

[48]

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

[49]

Craig, D.M. et al. 2015. Utilizing small nutrient compounds as enhancers of exercise-induced mitochondrial biogenesis. *Frontiers in Physiology*. 6, (Oct. 2015).  
<https://doi.org/10.3389/fphys.2015.00296>.

[50]

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

[51]

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

[52]

Cruz, P.M.R. et al. 2014. Congenital myasthenic syndromes and the neuromuscular junction. *Current Opinion in Neurology*. 27, 5 (Oct. 2014), 566-575.  
<https://doi.org/10.1097/WCO.000000000000134>.

[53]

Cup, E.H. et al. 2007. Exercise Therapy and Other Types of Physical Therapy for Patients With Neuromuscular Diseases: A Systematic Review. *Archives of Physical Medicine and Rehabilitation*. 88, 11 (Nov. 2007), 1452-1464.  
<https://doi.org/10.1016/j.apmr.2007.07.024>.

[54]

Dalakas, M.C. 2015. Inflammatory Muscle Diseases. *New England Journal of Medicine*. 372,

18 (Apr. 2015), 1734–1747. <https://doi.org/10.1056/NEJMra1402225>.

[55]

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

[56]

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

[57]

Davidson, G.L. et al. 2012. Frequency of mutations in the genes associated with hereditary sensory and autonomic neuropathy in a UK cohort. *Journal of Neurology*. 259, 8 (Aug. 2012), 1673–1685. <https://doi.org/10.1007/s00415-011-6397-y>.

[58]

Davies, A.M. 2013. Regulation of Neuronal Survival by Neurotrophins in the Developing Peripheral Nervous System. *Patterning and Cell Type Specification in the Developing CNS and PNS*. Elsevier. 303–311.

[59]

Dimachkie, M.M. and Barohn, R.J. 2013. Guillain-Barré Syndrome and Variants. *Neurologic Clinics*. 31, 2 (May 2013), 491–510. <https://doi.org/10.1016/j.ncl.2013.01.005>.

[60]

Dubrey, S. et al. 2015. The transthyretin amyloidoses: advances in therapy. *Postgraduate Medical Journal*. 91, 1078 (Aug. 2015), 439–448. <https://doi.org/10.1136/postgradmedj-2014-133224>.

[61]

Duncan, J.S. et al. 2016. Brain imaging in the assessment for epilepsy surgery. *The Lancet Neurology*. 15, 4 (Apr. 2016), 420–433. [https://doi.org/10.1016/S1474-4422\(15\)00383-X](https://doi.org/10.1016/S1474-4422(15)00383-X).

[62]

Ebens, A. et al. 1996. Hepatocyte Growth Factor/Scatter Factor Is an Axonal Chemoattractant and a Neurotrophic Factor for Spinal Motor Neurons. *Neuron*. 17, 6 (Dec. 1996), 1157–1172. [https://doi.org/10.1016/S0896-6273\(00\)80247-0](https://doi.org/10.1016/S0896-6273(00)80247-0).

[63]

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

[64]

Evers, M.M. et al. 2015. Antisense oligonucleotides in therapy for neurodegenerative disorders. *Advanced Drug Delivery Reviews*. 87, (June 2015), 90–103. <https://doi.org/10.1016/j.addr.2015.03.008>.

[65]

Ferlini, A. et al. 2013. Biomarkers in Rare Diseases. *Public Health Genomics*. 16, 6 (2013), 313–321. <https://doi.org/10.1159/000355938>.

[66]

Fernández, A. et al. 2017. A history of genome editing in mammals. *Mammalian Genome*. 28, 7–8 (Aug. 2017), 237–246. <https://doi.org/10.1007/s00335-017-9699-2>.

[67]

Forbes, S.C. et al. 2014. 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, 9 (Sept. 2014). <https://doi.org/10.1371/journal.pone.0106435>.

[68]

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

[69]

Fuglsang-Frederiksen, A. 2006. The role of different EMG methods in evaluating myopathy. *Clinical Neurophysiology*. 117, 6 (June 2006), 1173–1189. <https://doi.org/10.1016/j.clinph.2005.12.018>.

[70]

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

[71]

Gabrielle Natalie Samuel 2017. The UK's 100,000 Genomes Project: manifesting policymakers' expectations. *New Genetics and Society*. 36, 4 (2017). <https://doi.org/10.1080/14636778.2017.1370671>.

[72]

Garner, M. et al. 2012. A framework for the evaluation of patient information leaflets. *Health Expectations*. 15, 3 (Sept. 2012), 283–294. <https://doi.org/10.1111/j.1369-7625.2011.00665.x>.

[73]

Ghaoui, R. et al. 2015. Use of Whole-Exome Sequencing for Diagnosis of Limb-Girdle Muscular Dystrophy. *JAMA Neurology*. 72, 12 (Dec. 2015). <https://doi.org/10.1001/jamaneurol.2015.2274>.

[74]

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

[75]

Glover, G.H. and Schneider, E. 1991. Three-point dixon technique for true water/fat decomposition with B0 inhomogeneity correction. *Magnetic Resonance in Medicine*. 18, 2 (Apr. 1991), 371–383. <https://doi.org/10.1002/mrm.1910180211>.

[76]

Goemans, N.M. et al. 2011. Systemic Administration of PRO051 in Duchenne's Muscular Dystrophy. *New England Journal of Medicine*. 364, 16 (Apr. 2011), 1513–1522. <https://doi.org/10.1056/NEJMoa1011367>.

[77]

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

[78]

Gordon, T. and Sulaiman, O.A. 2012. Nerve Regeneration in the Peripheral Nervous System. *Neuroglia*. H. Kettenmann, ed. Oxford University Press. 701–714.

[79]

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.

[80]

Greenfield, A. 2017. Editing mammalian genomes: ethical considerations. *Mammalian Genome*. 28, 7–8 (Aug. 2017), 388–393. <https://doi.org/10.1007/s00335-017-9702-y>.

[81]

Griffin, B.H. et al. 2017. The 100 000 Genomes Project: What it means for paediatrics. *Archives of disease in childhood - Education & practice edition*. 102, 2 (Apr. 2017), 105–107. <https://doi.org/10.1136/archdischild-2016-311029>.

[82]

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

[83]

Hardie, D.G. 2006. AMPK: A Key Sensor of Fuel and Energy Status in Skeletal Muscle. *Physiology*. 21, 1 (Feb. 2006), 48–60. <https://doi.org/10.1152/physiol.00044.2005>.

[84]

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

[85]

Harridge, S.D.R. et al. 1996. Whole-muscle and single-fibre contractile properties and myosin heavy chain isoforms in humans. *Pflügers Archiv - European Journal of Physiology*. 432, 5 (Sept. 1996), 913–920. <https://doi.org/10.1007/s004240050215>.

[86]

Hawkins, P.N. et al. 2015. Evolving landscape in the management of transthyretin amyloidosis. *Annals of Medicine*. 47, 8 (Nov. 2015), 625–638. <https://doi.org/10.3109/07853890.2015.1068949>.

[87]

Hawley, J.A. et al. 2014. Integrative Biology of Exercise. *Cell*. 159, 4 (Nov. 2014), 738–749. <https://doi.org/10.1016/j.cell.2014.10.029>.

[88]

Hilton-Jones, D. and Turner, M.R. eds 2014. *Oxford textbook of neuromuscular disorders*. Oxford University Press.

[89]

Hoch, W. et al. 2001. Auto-antibodies to the receptor tyrosine kinase MuSK in patients with myasthenia gravis without acetylcholine receptor antibodies. *Nature Medicine*. 7, 3 (Mar. 2001), 365–368. <https://doi.org/10.1038/85520>.

[90]

Hoffmann, G.F. et al. 2009. *Inherited metabolic diseases: a clinical approach*. Springer.

[91]

Hoier, B. and Hellsten, Y. 2014. Exercise-Induced Capillary Growth in Human Skeletal Muscle and the Dynamics of VEGF. *Microcirculation*. 21, 4 (May 2014), 301–314. <https://doi.org/10.1111/micc.12117>.

[92]

Hollak, C.E.M. and Lachmann, R. eds 2016. *Inherited metabolic disease in adults: a clinical guide*. Oxford University Press.

[93]

Hollak, C.E.M. and Lachmann, R. eds 2016. *Inherited metabolic disease in adults: a clinical guide*. Oxford University Press.

[94]

Hollingsworth, K.G. et al. 2012. 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, (Oct. 2012), S54–S67. <https://doi.org/10.1016/j.nmd.2012.06.005>.

[95]

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

[96]

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

[97]

Hull, J. et al. 2012. British Thoracic Society guideline for respiratory management of children with neuromuscular weakness. *Thorax*. 67, Suppl 1 (July 2012), i1–i40.  
<https://doi.org/10.1136/thoraxjnl-2012-201964>.

[98]

Irene Colombo 2015. Congenital myopathies: Natural history of a large pediatric cohort. *Neurology*. 84, 1 (2015). <https://doi.org/10.1212/WNL.0000000000001110>.

[99]

Irina Dudanova 2012. Genetic Evidence for a Contribution of EphA:EphrinA Reverse Signaling to Motor Axon Guidance. *Journal of Neuroscience*. 32, 15 (2012), 5209–5215.

[100]

Jain, K.K. ed. 2015. *Applied neurogenomics*. Humana Press.

[101]

Jean-Yves Hogrel 2016. Longitudinal functional and NMR assessment of upper limbs in Duchenne muscular dystrophy. *Neurology*. 86, 11 (2016).  
<https://doi.org/10.1212/WNL.0000000000002464>.

[102]

Jeppesen, T.D. et al. 2006. Aerobic training is safe and improves exercise capacity in patients with mitochondrial myopathy. *Brain*. 129, 12 (June 2006), 3402–3412.  
<https://doi.org/10.1093/brain/awl149>.

[103]

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

[104]

Jessen, K.R. et al. 2015. Schwann Cells: Development and Role in Nerve Repair. *Cold Spring Harbor Perspectives in Biology*. 7, 7 (July 2015).  
<https://doi.org/10.1101/cshperspect.a020487>.

[105]

Jessen, K.R. and Mirsky, R. 2005. The origin and development of glial cells in peripheral nerves. *Nature Reviews Neuroscience*. 6, 9 (Sept. 2005), 671–682.  
<https://doi.org/10.1038/nrn1746>.

[106]

Jessen, K.R. and Mirsky, R. 2016. The repair Schwann cell and its function in regenerating nerves. *The Journal of Physiology*. 594, 13 (July 2016), 3521–3531.  
<https://doi.org/10.1113/JP270874>.

[107]

Jones, D.A. et al. 2004. *Skeletal muscle from molecules to movement: a textbook of muscle physiology for sport, exercise, physiotherapy and medicine*. Churchill Livingstone.

[108]

Kang, J.-H. et al. 2015. The Alzheimer's Disease Neuroimaging Initiative 2 Biomarker Core: A review of progress and plans. *Alzheimer's & Dementia*. 11, 7 (July 2015), 772–791.  
<https://doi.org/10.1016/j.jalz.2015.05.003>.

[109]

Kanning, K.C. et al. 2010. Motor Neuron Diversity in Development and Disease. *Annual Review of Neuroscience*. 33, 1 (June 2010), 409–440.

<https://doi.org/10.1146/annurev.neuro.051508.135722>.

[110]

Kao, T.-J. et al. 2012. Eph and ephrin signaling: Lessons learned from spinal motor neurons. *Seminars in Cell & Developmental Biology*. 23, 1 (Feb. 2012), 83–91. <https://doi.org/10.1016/j.semcd.2011.10.016>.

[111]

Karin E. Lundin 2015. Oligonucleotide Therapies: The Past and the Present. *Human Gene Therapy*. 26, 8 (2015). <https://doi.org/10.1089/hum.2015.070>.

[112]

Keith Baar 2014. Using Molecular Biology to Maximize Concurrent Training. *Sports Medicine (Auckland, N.z.)*. 44, Suppl 2 (2014). <https://doi.org/10.1007/s40279-014-0252-0>.

[113]

Kernell, D. 2006. *The Motoneurone and its Muscle Fibres*. Oxford University Press.

[114]

Khorkova, O. and Wahlestedt, C. 2017. Oligonucleotide therapies for disorders of the nervous system. *Nature Biotechnology*. 35, 3 (Mar. 2017), 249–263. <https://doi.org/10.1038/nbt.3784>.

[115]

Kimberly Amburgey 2017. A natural history study of X-linked myotubular myopathy. *Neurology*. 89, 13 (2017). <https://doi.org/10.1212/WNL.0000000000004415>.

[116]

Kinali, M. et al. 2009. 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, 10 (Oct. 2009),

918–928. [https://doi.org/10.1016/S1474-4422\(09\)70211-X](https://doi.org/10.1016/S1474-4422(09)70211-X).

[117]

Koneczny, I. et al. 2013. MuSK Myasthenia Gravis IgG4 Disrupts the Interaction of LRP4 with MuSK but Both IgG4 and IgG1-3 Can Disperse Preformed Agrin-Independent AChR Clusters. *PLoS ONE*. 8, 11 (Nov. 2013). <https://doi.org/10.1371/journal.pone.0080695>.

[118]

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

[119]

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

[120]

Leslie Jacobson 1999. Plasma from human mothers of fetuses with severe arthrogyrosis multiplex congenita causes deformities in mice. *Journal of Clinical Investigation*. 103, 7 (1999). <https://doi.org/10.1172/JCI5943>.

[121]

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

[122]

Lin, G. et al. 2017. Amyotrophic Lateral Sclerosis Pathogenesis Converges on Defects in Protein Homeostasis Associated with TDP-43 Mislocalization and Proteasome-Mediated Degradation Overload. *Fly Models of Human Diseases*. Elsevier. 111–171.

[123]

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

[124]

Machado, P. et al. 2013. Update in inclusion body myositis. *Current Opinion in Rheumatology*. 25, 6 (Nov. 2013), 763–771.  
<https://doi.org/10.1097/01.bor.0000434671.77891.9a>.

[125]

Machado, P.M. et al. 2014. Ongoing Developments in Sporadic Inclusion Body Myositis. *Current Rheumatology Reports*. 16, 12 (Dec. 2014).  
<https://doi.org/10.1007/s11926-014-0477-9>.

[126]

Machado, P.M. et al. 2014. Sporadic inclusion body myositis. *Current Opinion in Neurology*. 27, 5 (Oct. 2014), 591–598. <https://doi.org/10.1097/WCO.0000000000000129>.

[127]

Marie-Christine Birling 2017. Modeling human disease in rodents by CRISPR/Cas9 genome editing. *Mammalian Genome*. 28, 7 (2017). <https://doi.org/10.1007/s00335-017-9703-x>.

[128]

Matthew N Meriggioli 2009. Autoimmune myasthenia gravis: emerging clinical and biological heterogeneity. *Lancet neurology*. 8, 5 (2009).  
[https://doi.org/10.1016/S1474-4422\(09\)70063-8](https://doi.org/10.1016/S1474-4422(09)70063-8).

[129]

Matthews, E. et al. 2010. The non-dystrophic myotonias: molecular pathogenesis, diagnosis and treatment. *Brain*. 133, 1 (Jan. 2010), 9–22.  
<https://doi.org/10.1093/brain/awp294>.

[130]

Mendell, J.R. et al. 2013. Eteplirsen for the treatment of Duchenne muscular dystrophy. *Annals of Neurology*. 74, 5 (Nov. 2013), 637–647. <https://doi.org/10.1002/ana.23982>.

[131]

Mendell, J.R. et al. 2016. Longitudinal effect of eteplirsen versus historical control on ambulation in Duchenne muscular dystrophy. *Annals of Neurology*. 79, 2 (Feb. 2016), 257–271. <https://doi.org/10.1002/ana.24555>.

[132]

Menezes, M.P. and North, K.N. 2012. Inherited neuromuscular disorders: Pathway to diagnosis. *Journal of Paediatrics and Child Health*. 48, 6 (June 2012), 458–465. <https://doi.org/10.1111/j.1440-1754.2011.02210.x>.

[133]

Metzker, M.L. 2010. Sequencing technologies — the next generation. *Nature Reviews Genetics*. 11, 1 (Jan. 2010), 31–46. <https://doi.org/10.1038/nrg2626>.

[134]

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

[135]

Mhoriam Ahmed 2016. Targeting Protein Homeostasis in Sporadic Inclusion Body Myositis. *Science translational medicine*. 8, 331 (2016). <https://doi.org/10.1126/scitranslmed.aad4583>.

[136]

Michael Benatar 2016. ALS Biomarkers for Therapy Development: State of the Field & Future Directions. *Muscle & nerve*. 53, 2 (2016). <https://doi.org/10.1002/mus.24979>.

[137]

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

[138]

Michell, A. 2013. *Understanding EMG*. Oxford University Press.

[139]

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

[140]

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

[141]

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

[142]

Monk, K.R. et al. 2015. New insights on schwann cell development. *Glia*. 63, 8 (Aug. 2015), 1376–1393. <https://doi.org/10.1002/glia.22852>.

[143]

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

[144]

Morrow, J.M. et al. 2016. MRI biomarker assessment of neuromuscular disease progression: a prospective observational cohort study. *The Lancet Neurology*. 15, 1 (Jan. 2016), 65–77. [https://doi.org/10.1016/S1474-4422\(15\)00242-2](https://doi.org/10.1016/S1474-4422(15)00242-2).

[145]

Muntoni, F. et al. 2003. Dystrophin and mutations: one gene, several proteins, multiple phenotypes. *The Lancet Neurology*. 2, 12 (Dec. 2003), 731–740.  
[https://doi.org/10.1016/S1474-4422\(03\)00585-4](https://doi.org/10.1016/S1474-4422(03)00585-4).

[146]

Muscular Dystrophy UK: <http://www.muscardystrophyuk.org/>.

[147]

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

[148]

Nancy Leslie 2017. Pompe Disease. (2017).

[149]

Needham, M. and Mastaglia, F.L. 2016. Sporadic inclusion body myositis: A review of recent clinical advances and current approaches to diagnosis and treatment. *Clinical Neurophysiology*. 127, 3 (Mar. 2016), 1764–1773.  
<https://doi.org/10.1016/j.clinph.2015.12.011>.

[150]

Nerve Signaling:  
[https://www.nobelprize.org/educational/medicine/nerve\\_signaling/index.html](https://www.nobelprize.org/educational/medicine/nerve_signaling/index.html).

[151]

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

[152]

Neuromuscular Disease Centre: <http://neuromuscular.wustl.edu/>.

[153]

Nishimune, H. 2012. Active zones of mammalian neuromuscular junctions: formation, density, and aging. *Annals of the New York Academy of Sciences*. 1274, 1 (Dec. 2012), 24–32. <https://doi.org/10.1111/j.1749-6632.2012.06836.x>.

[154]

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

[155]

North, K.N. et al. 2014. Approach to the diagnosis of congenital myopathies. *Neuromuscular Disorders*. 24, 2 (Feb. 2014), 97–116. <https://doi.org/10.1016/j.nmd.2013.11.003>.

[156]

North, K.N. et al. 2014. Approach to the diagnosis of congenital myopathies. *Neuromuscular Disorders*. 24, 2 (Feb. 2014), 97–116. <https://doi.org/10.1016/j.nmd.2013.11.003>.

[157]

Nowell, M. et al. 2017. Resection planning in extratemporal epilepsy surgery using 3D multimodality imaging and intraoperative MRI. *British Journal of Neurosurgery*. 31, 4 (July 2017), 468–470. <https://doi.org/10.1080/02688697.2016.1265086>.

[158]

O'Brien, T.D. et al. 2010. In vivo measurements of muscle specific tension in adults and children. *Experimental Physiology*. 95, 1 (Jan. 2010), 202–210. <https://doi.org/10.1113/expphysiol.2009.048967>.

[159]

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

[160]

O'Grady, G.L. et al. 2016. Diagnosis and etiology of congenital muscular dystrophy: We are halfway there. *Annals of Neurology*. 80, 1 (July 2016), 101-111. <https://doi.org/10.1002/ana.24687>.

[161]

Olivé, M. et al. 2013. Myofibrillar myopathies. *Current Opinion in Neurology*. 26, 5 (Oct. 2013), 527-535. <https://doi.org/10.1097/WCO.0b013e328364d6b1>.

[162]

Olpin, S.E. et al. 2015. The investigation and management of metabolic myopathies. *Journal of Clinical Pathology*. 68, 6 (June 2015), 410-417. <https://doi.org/10.1136/jclinpath-2014-202808>.

[163]

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

[164]

Ørngreen, M.C. and Vissing, J. 2017. Treatment Opportunities in Patients With Metabolic Myopathies. *Current Treatment Options in Neurology*. 19, 11 (Nov. 2017). <https://doi.org/10.1007/s11940-017-0473-2>.

[165]

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

[166]

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

[167]

Paganoni, S. and Amato, A. 2013. Electrodiagnostic Evaluation of Myopathies. *Physical Medicine and Rehabilitation Clinics of North America*. 24, 1 (Feb. 2013), 193–207. <https://doi.org/10.1016/j.pmr.2012.08.017>.

[168]

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

[169]

Pasterkamp, R.J. 2012. Getting neural circuits into shape with semaphorins. *Nature Reviews Neuroscience*. 13, 9 (Aug. 2012), 605–618. <https://doi.org/10.1038/nrn3302>.

[170]

Peter S. Zammit 2004. Muscle satellite cells adopt divergent fates: a mechanism for self-renewal? *The Journal of Cell Biology*. 166, 3 (2004). <https://doi.org/10.1083/jcb.200312007>.

[171]

Plante-Bordeneuve, V. et al. 2007. Diagnostic pitfalls in sporadic transthyretin familial amyloid polyneuropathy (TTR-FAP). *Neurology*. 69, 7 (Aug. 2007), 693–698. <https://doi.org/10.1212/01.wnl.0000267338.45673.f4>.

[172]

Preston, D.C. and Shapiro, B.E. 2013. *Electromyography and neuromuscular disorders: clinical-electrophysiologic correlations*. Elsevier Saunders.

[173]

Purves, D. 2001. Neuroscience. National Library of Medicine.

[174]

Quijano-Roy, S. et al. 2011. Muscle Imaging in Congenital Myopathies. *Seminars in Pediatric Neurology*. 18, 4 (Dec. 2011), 221-229.  
<https://doi.org/10.1016/j.spen.2011.10.003>.

[175]

R Bottinelli 1996. Force-velocity properties of human skeletal muscle fibres: myosin heavy chain isoform and temperature dependence. *The Journal of Physiology*. 495, Pt 2 (1996).

[176]

R Klein 1994. Role of neurotrophins in mouse neuronal development. *The FASEB Journal*. 8, 10 (1994), 738-744.

[177]

Ravenscroft, G. et al. 2017. New era in genetics of early-onset muscle disease: Breakthroughs and challenges. *Seminars in Cell & Developmental Biology*. 64, (Apr. 2017), 160-170. <https://doi.org/10.1016/j.semcdb.2016.08.002>.

[178]

Ravenscroft, G. et al. 2015. Pathophysiological concepts in the congenital myopathies: blurring the boundaries, sharpening the focus. *Brain*. 138, 2 (Feb. 2015), 246-268.  
<https://doi.org/10.1093/brain/awu368>.

[179]

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

[180]

Rees, J.H. 2004. Paraneoplastic syndromes: when to suspect, how to confirm, and how to manage. *Journal of Neurology, Neurosurgery & Psychiatry*. 75, suppl\_2 (June 2004), ii43-ii50. <https://doi.org/10.1136/jnnp.2004.040378>.

[181]

Reilly, M. and Fridman, V. 2015. Inherited Neuropathies. *Seminars in Neurology*. 35, 04 (Oct. 2015), 407-423. <https://doi.org/10.1055/s-0035-1558981>.

[182]

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

[183]

Richard J. Barohn 2014. A PATTERN RECOGNITION APPROACH TO THE PATIENT WITH A SUSPECTED MYOPATHY. *Neurologic clinics*. 32, 3 (2014). <https://doi.org/10.1016/j.ncl.2014.04.008>.

[184]

Richards, S. et al. 2015. 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, 5 (May 2015), 405-423. <https://doi.org/10.1038/gim.2015.30>.

[185]

Ricotti, V. et al. 2013. Long-term benefits and adverse effects of intermittent versus daily glucocorticoids in boys with Duchenne muscular dystrophy. *Journal of Neurology, Neurosurgery & Psychiatry*. 84, 6 (June 2013), 698-705. <https://doi.org/10.1136/jnnp-2012-303902>.

[186]

Robert M. Brownstone 2010. 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>.

[187]

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

[188]

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

[189]

Ross, J. et al. 2012. Defects in Glycosylation Impair Satellite Stem Cell Function and Niche Composition in the Muscles of the Dystrophic Large Mouse. *STEM CELLS*. 30, 10 (Oct. 2012), 2330–2341. <https://doi.org/10.1002/stem.1197>.

[190]

Rossor, A.M. et al. 2015. A practical approach to the genetic neuropathies. *Practical Neurology*. 15, 3 (June 2015), 187–198. <https://doi.org/10.1136/practneurol-2015-001095>.

[191]

Rossor, A.M. et al. 2015. A practical approach to the genetic neuropathies. *Practical Neurology*. 15, 3 (June 2015), 187–198. <https://doi.org/10.1136/practneurol-2015-001095>.

[192]

Rossor, A.M. et al. 2015. A practical approach to the genetic neuropathies. *Practical Neurology*. 15, 3 (June 2015), 187–198. <https://doi.org/10.1136/practneurol-2015-001095>.

[193]

Rossor, A.M. et al. 2015. A practical approach to the genetic neuropathies. *Practical Neurology*. 15, 3 (June 2015), 187–198. <https://doi.org/10.1136/practneurol-2015-001095>.

[194]

Rossor, A.M. et al. 2015. A practical approach to the genetic neuropathies. *Practical Neurology*. 15, 3 (June 2015), 187–198. <https://doi.org/10.1136/practneurol-2015-001095>.

[195]

Rossor, A.M. et al. 2018. Antisense oligonucleotides and other genetic therapies made simple. *Practical Neurology*. (Feb. 2018). <https://doi.org/10.1136/practneurol-2017-001764>.

[196]

Rossor, A.M. et al. 2017. Peripheral neuropathy in complex inherited diseases: an approach to diagnosis. *Journal of Neurology, Neurosurgery & Psychiatry*. 88, 10 (Oct. 2017), 846–863. <https://doi.org/10.1136/jnnp-2016-313960>.

[197]

Rossor, A.M. et al. 2016. Recent advances in the genetic neuropathies. *Current Opinion in Neurology*. (Sept. 2016). <https://doi.org/10.1097/WCO.0000000000000373>.

[198]

Rossor, A.M. et al. 2016. Recent advances in the genetic neuropathies. *Current Opinion in Neurology*. (Sept. 2016). <https://doi.org/10.1097/WCO.0000000000000373>.

[199]

Rossor, A.M. et al. 2012. The distal hereditary motor neuropathies. *Journal of Neurology, Neurosurgery & Psychiatry*. 83, 1 (Jan. 2012), 6–14. <https://doi.org/10.1136/jnnp-2011-300952>.

[200]

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

[201]

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

[202]

Salzer, J.L. 2015. Schwann Cell Myelination. *Cold Spring Harbor Perspectives in Biology*. 7, 8 (Aug. 2015). <https://doi.org/10.1101/cshperspect.a020529>.

[203]

Samuel, G.N. and Farsides, B. 2017. 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*. (Oct. 2017). <https://doi.org/10.1007/s11019-017-9810-1>.

[204]

Saudubray, J.M. et al. eds 2016. *Inborn metabolic diseases: diagnosis and treatment*. Springer.

[205]

Schlosser, G. 2006. Induction and specification of cranial placodes. *Developmental Biology*. 294, 2 (June 2006), 303–351. <https://doi.org/10.1016/j.ydbio.2006.03.009>.

[206]

Schofield, D. et al. 2017. Cost-effectiveness of massively parallel sequencing for diagnosis of paediatric muscle diseases. *npj Genomic Medicine*. 2, 1 (Dec. 2017). <https://doi.org/10.1038/s41525-017-0006-7>.

[207]

Schröder, R. and Schoser, B. 2009. Myofibrillar Myopathies: A Clinical and Myopathological Guide. *Brain Pathology*. 19, 3 (July 2009), 483–492. <https://doi.org/10.1111/j.1750-3639.2009.00289.x>.

[208]

Shaibani, A. 2014. *A Video Atlas of Neuromuscular Disorders*. Oxford University Press.

[209]

Sharp, L. and Trivedi, J.R. 2014. Treatment and Management of Neuromuscular Channelopathies. *Current Treatment Options in Neurology*. 16, 10 (Oct. 2014). <https://doi.org/10.1007/s11940-014-0313-6>.

[210]

Simon, N.G. et al. 2014. Quantifying disease progression in amyotrophic lateral sclerosis. *Annals of Neurology*. 76, 5 (Nov. 2014), 643–657. <https://doi.org/10.1002/ana.24273>.

[211]

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

[212]

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

[213]

Sun, Y. et al. 2015. Next-Generation Diagnostics: Gene Panel, Exome, or Whole Genome? *Human Mutation*. 36, 6 (June 2015), 648–655. <https://doi.org/10.1002/humu.22783>.

[214]

Sveen, M.-L. et al. 2013. Resistance training in patients with limb-girdle and becker muscular dystrophies. *Muscle & Nerve*. 47, 2 (Feb. 2013), 163–169.  
<https://doi.org/10.1002/mus.23491>.

[215]

T. D. Bunker 1983. An information leaflet for surgical patients. *Annals of The Royal College of Surgeons of England*. 65, 4 (1983).

[216]

Tan, S.V. et al. 2011. Refined exercise testing can aid dna-based diagnosis in muscle channelopathies. *Annals of Neurology*. 69, 2 (Feb. 2011), 328–340.  
<https://doi.org/10.1002/ana.22238>.

[217]

Taniguchi, M. et al. 1997. Disruption of Semaphorin III/D Gene Causes Severe Abnormality in Peripheral Nerve Projection. *Neuron*. 19, 3 (Sept. 1997), 519–530.  
[https://doi.org/10.1016/S0896-6273\(00\)80368-2](https://doi.org/10.1016/S0896-6273(00)80368-2).

[218]

Teboul, L. et al. 2017. Introduction to Mammalian Genome Special Issue: Genome Editing. *Mammalian Genome*. 28, 7–8 (Aug. 2017), 235–236.  
<https://doi.org/10.1007/s00335-017-9708-5>.

[219]

Thiede-Stan, N.K. and Schwab, M.E. 2015. Attractive and repulsive factors act through multi-subunit receptor complexes to regulate nerve fiber growth. *Journal of Cell Science*. 128, 14 (July 2015), 2403–2414. <https://doi.org/10.1242/jcs.165555>.

[220]

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

[221]

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>.

[222]

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

[223]

Vaithinathan, A.G. and Asokan, V. 2017. Public health and precision medicine share a goal. *Journal of Evidence-Based Medicine*. 10, 2 (May 2017), 76-80.  
<https://doi.org/10.1111/jebm.12239>.

[224]

Vakharia, V.N. et al. 2017. Accuracy of intracranial electrode placement for stereoelectroencephalography: A systematic review and meta-analysis. *Epilepsia*. 58, 6 (June 2017), 921-932. <https://doi.org/10.1111/epi.13713>.

[225]

Van Battum, E.Y. et al. 2015. Axon guidance proteins in neurological disorders. *The Lancet Neurology*. 14, 5 (May 2015), 532-546. [https://doi.org/10.1016/S1474-4422\(14\)70257-1](https://doi.org/10.1016/S1474-4422(14)70257-1).

[226]

Venance, S.L. et al. 2006. The primary periodic paralyses: diagnosis, pathogenesis and treatment. *Brain*. 129, 1 (Jan. 2006), 8-17. <https://doi.org/10.1093/brain/awh639>.

[227]

Victorian Department of Health / University of Melbourne:

<https://www2.health.vic.gov.au/>.

[228]

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

[229]

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

[230]

Wang, L. et al. 2011. Anatomical Coupling of Sensory and Motor Nerve Trajectory via Axon Tracking. *Neuron*. 71, 2 (July 2011), 263–277. <https://doi.org/10.1016/j.neuron.2011.06.021>.

[231]

Ward, S. 2005. Randomised controlled trial of non-invasive ventilation (NIV) for nocturnal hypoventilation in neuromuscular and chest wall disease patients with daytime normocapnia. *Thorax*. 60, 12 (Dec. 2005), 1019–1024. <https://doi.org/10.1136/thx.2004.037424>.

[232]

Wattjes, M.P. et al. 2010. Neuromuscular imaging in inherited muscle diseases. *European Radiology*. 20, 10 (Oct. 2010), 2447–2460. <https://doi.org/10.1007/s00330-010-1799-2>.

[233]

Wattjes, M.P. and Fischer, D. 2013. *Neuromuscular imaging*. Springer.

[234]

Welch, M.B. and Brummett, C.M. 2011. *Peripheral Nervous System Anatomy and Function*.

Neuroscientific Foundations of Anesthesiology. G.A. Mashour and R. Lydic, eds. Oxford University Press. 133–140.

[235]

Willcocks, R.J. et al. 2016. Multicenter prospective longitudinal study of magnetic resonance biomarkers in a large duchenne muscular dystrophy cohort. *Annals of Neurology*. 79, 4 (Apr. 2016), 535–547. <https://doi.org/10.1002/ana.24599>.

[236]

Willison, H.J. et al. 2016. Guillain-Barré syndrome. *The Lancet*. 388, 10045 (Aug. 2016), 717–727. [https://doi.org/10.1016/S0140-6736\(16\)00339-1](https://doi.org/10.1016/S0140-6736(16)00339-1).

[237]

Wood, N. 2012. *Neurogenetics*. Cambridge University Press.

[238]

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

[239]

Yang, Y. et al. 2013. Clinical Whole-Exome Sequencing for the Diagnosis of Mendelian Disorders. *New England Journal of Medicine*. 369, 16 (Oct. 2013), 1502–1511. <https://doi.org/10.1056/NEJMoa1306555>.

[240]

Yates, M. et al. 2016. EULAR/ERA-EDTA recommendations for the management of ANCA-associated vasculitis. *Annals of the Rheumatic Diseases*. 75, 9 (Sept. 2016), 1583–1594. <https://doi.org/10.1136/annrheumdis-2016-209133>.

[241]

American Journal of Respiratory and Critical Care Medicine.

[242]

American Journal of Roentgenology.

[243]

Best Practice in Memory Services: Learning from across England.

[244]

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

[245]

Qualification Process for Drug Development Tools.

[246]

2015. 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, 9 (Sept. 2015), 883–892.  
[https://doi.org/10.1016/S1474-4422\(15\)00152-0](https://doi.org/10.1016/S1474-4422(15)00152-0).

[247]

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

[248]

Toolkit for producing patient information.pdf.