

CLNE0022: Skeletal Muscle and Associated Diseases

View Online



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., ... Greensmith, L. (2016). Targeting protein homeostasis in sporadic inclusion body myositis. *Science Translational Medicine*, 8(331), 331ra41-331ra41. <https://doi.org/10.1126/scitranslmed.aad4583>

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

An information leaflet for surgical patients. (1983). *Annals of The Royal College of Surgeons of England*, 65(4). <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2494353/>

Atherton, P. J., & Smith, K. (2012a). Muscle protein synthesis in response to nutrition and exercise. *The Journal of Physiology*, 590(5), 1049–1057. <https://doi.org/10.1113/jphysiol.2011.225003>

Atherton, P. J., & Smith, K. (2012b). Muscle protein synthesis in response to nutrition and exercise. *The Journal of Physiology*, 590(5), 1049–1057. <https://doi.org/10.1113/jphysiol.2011.225003>

Baar, K. (2014a). Using Molecular Biology to Maximize Concurrent Training. *Sports Medicine*, 44(S2), 117–125. <https://doi.org/10.1007/s40279-014-0252-0>

Baar, K. (2014b). Using Molecular Biology to Maximize Concurrent Training. *Sports Medicine*, 44(S2), 117–125. <https://doi.org/10.1007/s40279-014-0252-0>

Baioni, M. T. C., & Ambiel, C. R. (2010). Spinal muscular atrophy: diagnosis, treatment and future prospects. *Jornal de Pediatria*, 86(4), 261–270. <https://doi.org/10.2223/JPED.1988>

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*. (n.d.). <https://www.minervamedica.it/en/journals/europa-medicophysica/article.php?cod=R33Y2013N02A0169>

Barohn, R. J., Dimachkie, M. M., & Jackson, C. E. (2014). A Pattern Recognition Approach to Patients with a Suspected Myopathy. *Neurologic Clinics*, 32(3), 569–593. <https://doi.org/10.1016/j.ncl.2014.04.008>

- Berthelsen, M. P., Husu, E., Christensen, S. B., Prahm, K. P., Vissing, J., & Jensen, B. R. (2014). Anti-gravity training improves walking capacity and postural balance in patients with muscular dystrophy. *Neuromuscular Disorders*, 24(6), 492–498. <https://doi.org/10.1016/j.nmd.2014.03.001>
- Boldrin, L., & Morgan, J. E. (2007). Activating muscle stem cells: therapeutic potential in muscle diseases. *Current Opinion in Neurology*, 20(5), 577–582. <https://doi.org/10.1097/WCO.0b013e3282ef5919>
- Boldrin, L., Zammit, P. S., & Morgan, J. E. (2015). Satellite cells from dystrophic muscle retain regenerative capacity. *Stem Cell Research*, 14(1), 20–29. <https://doi.org/10.1016/j.scr.2014.10.007>
- Briggs, D., & Morgan, J. E. (2013). Recent progress in satellite cell/myoblast engraftment - relevance for therapy. *FEBS Journal*, 280(17), 4281–4293. <https://doi.org/10.1111/febs.12273>
- 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. (2010a). Diagnosis and management of Duchenne muscular dystrophy, part 1: diagnosis, and pharmacological and psychosocial management. *The Lancet Neurology*, 9(1), 77–93. [https://doi.org/10.1016/S1474-4422\(09\)70271-6](https://doi.org/10.1016/S1474-4422(09)70271-6)
- 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. (2010b). Diagnosis and management of Duchenne muscular dystrophy, part 2: implementation of multidisciplinary care. *The Lancet Neurology*, 9(2), 177–189. [https://doi.org/10.1016/S1474-4422\(09\)70272-8](https://doi.org/10.1016/S1474-4422(09)70272-8)
- 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., ... McDonald, C. M. (2014). Ataluren treatment of patients with nonsense mutation dystrophinopathy. *Muscle & Nerve*, 50(4), 477–487. <https://doi.org/10.1002/mus.24332>
- By:Gordon, AM (Gordon, AM); Homsher, E (Homsher, E); Regnier, M (Regnier, M). (2000). Regulation of contraction in striated muscle. *PHYSIOLOGICAL REVIEWS* *PHYSIOLOGICAL REVIEWS*, 80(2), 853–924. http://apps.webofknowledge.com/full_record.do?product=WOS&search_mode=GeneralSearch&qid=13&SID=C49BOGnSGP9s3PqA3ow&page=1&doc=1
- By:Jones, SW (Jones, SW); Hill, RJ (Hill, RJ); Krasney, PA (Krasney, PA); O'Conner, B (O'Conner, B); Peirce, N (Peirce, N); Greenhaff, PL (Greenhaff, PL). (2004). Disuse atrophy and exercise rehabilitation in humans profoundly affects the expression of genes associated with the regulation of skeletal muscle mass. *FASEB JOURNAL* *FASEB JOURNAL*, 18(6). <https://doi.org/10.1096/fj.03-1228fje>
- Carstens, P.-O., & Schmidt, J. (2014). Diagnosis, pathogenesis and treatment of myositis: recent advances. *Clinical & Experimental Immunology*, 175(3), 349–358. <https://doi.org/10.1111/cei.12194>

Cirak, S., Arechavala-Gomez, 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. (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), 595–605. [https://doi.org/10.1016/S0140-6736\(11\)60756-3](https://doi.org/10.1016/S0140-6736(11)60756-3)

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

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. (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), 1452–1464. <https://doi.org/10.1016/j.apmr.2007.07.024>

Dalakas, M. C. (2015). Inflammatory Muscle Diseases. *New England Journal of Medicine*, 372(18), 1734–1747. <https://doi.org/10.1056/NEJMra1402225>

Darras, B. T., De Vivo, D. C., & Jones, H. R. (2003). *Neuromuscular disorders of infancy, childhood, and adolescence: a clinician's approach*. Butterworth-Heinemann.

Department of Health. (2003). *Toolkit for Producing Patient Information, Version 2*. Crown copyright. <https://www.uea.ac.uk/documents/246046/0/Toolkit+for+producing+patient+information.pdf>

Dubowitz, V., Sewry, C. A., & Oldfors, A. (2013). *Muscle biopsy: a practical approach* (Fourth edition). Saunders Elsevier.

Egner, I. M., Bruusgaard, J. C., Eftestøl, E., & Gundersen, K. (2013). A cellular memory mechanism aids overload hypertrophy in muscle long after an episodic exposure to anabolic steroids. *The Journal of Physiology*, 591(24), 6221–6230. <https://doi.org/10.1113/jphysiol.2013.264457>

Feingold, B., Mahle, W. T., Auerbach, S., Clemens, P., Domenighetti, A. A., Jefferies, J. L., Judge, D. P., Lal, A. K., Markham, L. W., Parks, W. J., Tsuda, T., Wang, P. J., & Yoo, S.-J. (2017). Management of Cardiac Involvement Associated With Neuromuscular Diseases: A Scientific Statement From the American Heart Association. *Circulation*, 136(13). <https://doi.org/10.1161/CIR.0000000000000526>

Fitts, R. H., Trappe, S. W., Costill, D. L., Gallagher, P. M., Creer, A. C., Colloton, P. A., Peters, J. R., Romatowski, J. G., Bain, J. L., & Riley, D. A. (2010). Prolonged space flight-induced alterations in the structure and function of human skeletal muscle fibres. *The Journal of Physiology*, 588(18), 3567–3592. <https://doi.org/10.1113/jphysiol.2010.188508>

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. (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). <https://doi.org/10.1371/journal.pone.0106435>

Force-velocity properties of human skeletal muscle fibres: myosin heavy chain isoform and temperature dependence. (1996). *The Journal of Physiology*, 495(Pt 2). <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1160815/>

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

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

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

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

Great Britain. Audit Commission for Local Authorities and the National Health Service in England and Wales. (n.d.). What Seems to Be the Matter Communication (National Health Service Report). Stationery Office. <https://webarchive.nationalarchives.gov.uk/20150410163038/http://archive.audit-commission.gov.uk/auditcommission/aboutus/publications/pages/national-reports-and-studies-archive.aspx.html>

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

Harridge, S. D. R., Bottinelli, R., Canepari, M., Pellegrino, M. A., Reggiani, C., Esbjörnsson, M., & Saltin, B. (1996). Whole-muscle and single-fibre contractile properties and myosin heavy chain isoforms in humans. *Pflügers Archiv - European Journal of Physiology*, 432(5), 913–920. <https://doi.org/10.1007/s004240050215>

Hawley, J. A., Hargreaves, M., Joyner, M. J., & Zierath, J. R. (2014). Integrative Biology of Exercise. *Cell*, 159(4), 738–749. <https://doi.org/10.1016/j.cell.2014.10.029>

Hoffmann, G. F., Zschocke, J., & Nyhan, W. L. (Eds.). (2017). *Inherited metabolic diseases: a clinical approach* (Second edition). Springer.

Hogrel, J.-Y., Wary, C., Moraux, A., Azzabou, N., Decostre, V., Ollivier, G., Canal, A., Lilien, C., Ledoux, I., Annoussamy, M., Reguiba, N., Gidaro, T., Le Moing, A. G., Cardas, R., Voit, T., Carlier, P. G., & Servais, L. (2016). Longitudinal functional and NMR assessment of upper

limbs in Duchenne muscular dystrophy. *Neurology*, 86(11), 1022–1030.
<https://doi.org/10.1212/WNL.0000000000002464>

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

Hollak, C. E. M., & Lachmann, R. (Eds.). (2016a). *Inherited Metabolic Disease in Adults* (Vol. 1). Oxford University Press. <https://doi.org/10.1093/med/9780199972135.001.0001>

Hollak, C. E. M., & Lachmann, R. (Eds.). (2016b). *Inherited Metabolic Disease in Adults* (Vol. 1). Oxford University Press. <https://doi.org/10.1093/med/9780199972135.001.0001>

Hollingsworth, K. G., de Sousa, P. L., Straub, V., & Carlier, P. G. (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, S54–S67. <https://doi.org/10.1016/j.nmd.2012.06.005>

Hunter, S., White, M., & Thompson, M. (1998). Techniques to Evaluate Elderly Human Muscle Function: A Physiological Basis. *The Journals of Gerontology Series A: Biological Sciences and Medical Sciences*, 53A(3), B204–B216.
<https://doi.org/10.1093/gerona/53A.3.B204>

Institute of Neurology, Queen Square & National Hospital for Neurology and Neurosurgery (London, England). (2016). *Neurology: a Queen Square textbook* (C. Clarke, R. Howard, M. Rossor, & S. D. Shorvon, Eds.; Second edition). John Wiley & Sons, Inc.
<https://onlinelibrary.wiley.com/doi/book/10.1002/9781118486160>

Jeppesen, T. D., Schwartz, M., Olsen, D. B., Wibrand, F., Krag, T., Duno, M., Hauerslev, S., & Vissing, J. (2006). Aerobic training is safe and improves exercise capacity in patients with mitochondrial myopathy. *Brain*, 129(12), 3402–3412. <https://doi.org/10.1093/brain/awl149>

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

Kadi, F. (2008). Cellular and molecular mechanisms responsible for the action of testosterone on human skeletal muscle. A basis for illegal performance enhancement. *British Journal of Pharmacology*, 154(3), 522–528. <https://doi.org/10.1038/bjp.2008.118>

Kim, H. K., Serai, S., Lindquist, D., Merrow, A. C., Horn, P. S., Kim, D. H., & Wong, B. L. (2015). Quantitative Skeletal Muscle MRI: Part 2, MR Spectroscopy and T2 Relaxation Time Mapping—Comparison Between Boys With Duchenne Muscular Dystrophy and Healthy Boys. *American Journal of Roentgenology*, 205(2), W216–W223.
<https://doi.org/10.2214/AJR.14.13755>

Kinali, M., Arechavala-Gomez, 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., ... Muntoni,

- F. (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), 918–928. [https://doi.org/10.1016/S1474-4422\(09\)70211-X](https://doi.org/10.1016/S1474-4422(09)70211-X)
- Machado, P., Brady, S., & Hanna, M. G. (2013). Update in inclusion body myositis. *Current Opinion in Rheumatology*, 25(6), 763–771. <https://doi.org/10.1097/01.bor.0000434671.77891.9a>
- 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. (2014). Ongoing Developments in Sporadic Inclusion Body Myositis. *Current Rheumatology Reports*, 16(12). <https://doi.org/10.1007/s11926-014-0477-9>
- Machado, P. M., Dimachkie, M. M., & Barohn, R. J. (2014). Sporadic inclusion body myositis. *Current Opinion in Neurology*, 27(5), 591–598. <https://doi.org/10.1097/WCO.0000000000000129>
- Marcotte, G. R., West, D. W. D., & Baar, K. (2015). The Molecular Basis for Load-Induced Skeletal Muscle Hypertrophy. *Calcified Tissue International*, 96(3), 196–210. <https://doi.org/10.1007/s00223-014-9925-9>
- Matthews, E., & Hanna, M. G. (2014). Skeletal muscle channelopathies. In D. Hilton-Jones & M. R. Turner (Eds.), *Oxford Textbook of Neuromuscular Disorders* (pp. 316–325). Oxford University Press. <https://doi.org/10.1093/med/9780199698073.003.0031>
- Mendell, J. R., Goemans, N., Lowes, L. P., Alfano, L. N., Berry, K., Shao, J., Kaye, E. M., & Mercuri, E. (2016). Longitudinal effect of eteplirsen versus historical control on ambulation in Duchenne muscular dystrophy. *Annals of Neurology*, 79(2), 257–271. <https://doi.org/10.1002/ana.24555>
- 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., ... Kaye, E. M. (2013). Eteplirsen for the treatment of Duchenne muscular dystrophy. *Annals of Neurology*, 74(5), 637–647. <https://doi.org/10.1002/ana.23982>
- Mercuri, E., Bertini, E., & Iannaccone, S. T. (2012). Childhood spinal muscular atrophy: controversies and challenges. *The Lancet Neurology*, 11(5), 443–452. [https://doi.org/10.1016/S1474-4422\(12\)70061-3](https://doi.org/10.1016/S1474-4422(12)70061-3)
- Miguel A Martín. (2014). Glycogen Storage Disease Type V. *Gene Reviews*. <https://www.ncbi.nlm.nih.gov/books/NBK1344/>
- Muntoni, F., Torelli, S., & Ferlini, A. (2003). Dystrophin and mutations: one gene, several proteins, multiple phenotypes. *The Lancet Neurology*, 2(12), 731–740. [https://doi.org/10.1016/S1474-4422\(03\)00585-4](https://doi.org/10.1016/S1474-4422(03)00585-4)
- Nancy D Leslie. (2018). Very Long-Chain Acyl-Coenzyme A Dehydrogenase Deficiency. *Gene Reviews*. <https://www.ncbi.nlm.nih.gov/books/NBK6816/>

Nancy Leslie. (2017). Pompe Disease. Gene Reviews.
<https://www.ncbi.nlm.nih.gov/books/NBK1261/>

Needham, M., & 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), 1764–1773. <https://doi.org/10.1016/j.clinph.2015.12.011>

Neuromuscular Disease Center. (n.d.). <https://neuromuscular.wustl.edu/>

Neuropathology and Applied Neurobiology. (n.d.). Volume 43, Issue 1.
<https://onlinelibrary.wiley.com/toc/13652990/2017/43/1>

O'Brien, T. D., Reeves, N. D., Baltzopoulos, V., Jones, D. A., & Maganaris, C. N. (2010). In vivo measurements of muscle specific tension in adults and children. *Experimental Physiology*, 95(1), 202–210. <https://doi.org/10.1113/expphysiol.2009.048967>

Olivé, M., Kley, R. A., & Goldfarb, L. G. (2013). Myofibrillar myopathies. *Current Opinion in Neurology*, 26(5), 527–535. <https://doi.org/10.1097/WCO.0b013e328364d6b1>

Olpin, S. E., Murphy, E., Kirk, R. J., Taylor, R. W., & Quinlivan, R. (2015). The investigation and management of metabolic myopathies. *Journal of Clinical Pathology*, 68(6), 410–417. <https://doi.org/10.1136/jclinpath-2014-202808>

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

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

Prescription information leaflets: a pilot study in general practice. (1983). *British Medical Journal (Clinical Research Ed.)*, 287(6400).
<https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1549423/>

Puthucheary, Z. A., Rawal, J., McPhail, M., Connolly, B., Ratnayake, G., Chan, P., Hopkinson, N. S., Padhke, R., Dew, T., Sidhu, P. S., Velloso, C., Seymour, J., Agle, C. C., Selby, A., Limb, M., Edwards, L. M., Smith, K., Rowlerson, A., Rennie, M. J., ... Montgomery, H. E. (2013). Acute Skeletal Muscle Wasting in Critical Illness. *JAMA*, 310(15).
<https://doi.org/10.1001/jama.2013.278481>

Rennie, M. J., Wackerhage, H., Spangenburg, E. E., & Booth, F. W. (2004a). Control of the Size of the Human Muscle Mass. *Annual Review of Physiology*, 66(1), 799–828.
<https://doi.org/10.1146/annurev.physiol.66.052102.134444>

Rennie, M. J., Wackerhage, H., Spangenburg, E. E., & Booth, F. W. (2004b). Control of the Size of the Human Muscle Mass. *Annual Review of Physiology*, 66(1), 799–828.
<https://doi.org/10.1146/annurev.physiol.66.052102.134444>

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., ... Miah, A. (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), 698–705. <https://doi.org/10.1136/jnnp-2012-303902>
- Ross, J., Benn, A., Jonuschies, J., Boldrin, L., Muntoni, F., Hewitt, J. E., Brown, S. C., & Morgan, J. E. (2012). Defects in Glycosylation Impair Satellite Stem Cell Function and Niche Composition in the Muscles of the Dystrophic Large Mouse. *STEM CELLS*, 30(10), 2330–2341. <https://doi.org/10.1002/stem.1197>
- Rudnik-Schöneborn, S., Hausmanowa-Petrusewicz, I., Borkowska, J., & Zerres, K. (2001). The Predictive Value of Achieved Motor Milestones Assessed in 441 Patients with Infantile Spinal Muscular Atrophy Types II and III. *European Neurology*, 45(3), 174–181. <https://doi.org/10.1159/000052118>
- Russman, B. S., Buncher, C. R., White, M., Samaha, F. J., & Iannaccone, S. T. (1996). Function changes in spinal muscular atrophy II and III. *Neurology*, 47(4), 973–976. <https://doi.org/10.1212/WNL.47.4.973>
- Saudubray, J. M., Baumgartner, M. R., & Walter, J. (Eds.). (2016). *Inborn metabolic diseases: diagnosis and treatment* (6th edition). Springer.
- Schröder, R., & Schoser, B. (2009). Myofibrillar Myopathies: A Clinical and Myopathological Guide. *Brain Pathology*, 19(3), 483–492. <https://doi.org/10.1111/j.1750-3639.2009.00289.x>
- Shavlakadze, T., & Grounds, M. (2006). Of bears, frogs, meat, mice and men: complexity of factors affecting skeletal muscle mass and fat. *BioEssays*, 28(10), 994–1009. <https://doi.org/10.1002/bies.20479>
- Sveen, M.-L., Andersen, S. P., Ingelsrud, L. H., Blichter, S., Olsen, N. E., Jønck, S., Krag, T. O., & Vissing, J. (2013). Resistance training in patients with limb-girdle and becker muscular dystrophies. *Muscle & Nerve*, 47(2), 163–169. <https://doi.org/10.1002/mus.23491>
- Thomas Wieser. (2017). Carnitine Palmitoyltransferase II Deficiency. *Gene Reviews*. <https://www.ncbi.nlm.nih.gov/books/NBK1253/>
- Velloso, C. P. (2008). Regulation of muscle mass by growth hormone and IGF-I. *British Journal of Pharmacology*, 154(3), 557–568. <https://doi.org/10.1038/bjp.2008.153>
- Voet, N. B., van der Kooij, E. L., Riphagen, I. I., Lindeman, E., van Engelen, B. G., & Geurts, A. C. (2013). Strength training and aerobic exercise training for muscle disease. *Cochrane Database of Systematic Reviews*. <https://doi.org/10.1002/14651858.CD003907.pub4>
- Wang, C. H., Finkel, R. S., Bertini, E. S., Schroth, M., Simonds, A., Wong, B., Aloysius, A., Morrison, L., Main, M., Crawford, T. O., & Trela, A. (2007). Consensus Statement for Standard of Care in Spinal Muscular Atrophy. *Journal of Child Neurology*, 22(8), 1027–1049. <https://doi.org/10.1177/0883073807305788>
- Wattjes, M. P., & Fischer, D. (2013). *Neuromuscular imaging*. Springer.

http://ucl.alma.exlibrisgroup.com/view/action/uresolver.do?operation=resolveService&package_service_id=3320980190004761&institutionId=4761&customerId=4760

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

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

Zammit, P. S., Golding, J. P., Nagata, Y., Hudon, V., Partridge, T. A., & Beauchamp, J. R. (2004). Muscle satellite cells adopt divergent fates. *The Journal of Cell Biology*, 166(3), 347–357. <https://doi.org/10.1083/jcb.200312007>