

CLNE0022: Skeletal Muscle and Associated Diseases

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[1]

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 [Online]. Available: <https://webarchive.nationalarchives.gov.uk/20150410163038/http://archive.audit-commission.gov.uk/auditcommission/aboutus/publications/pages/national-reports-and-studies-archive.aspx.html>

[2]

'An information leaflet for surgical patients.', *Annals of The Royal College of Surgeons of England*, vol. 65, no. 4, 1983 [Online]. Available: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2494353/>

[3]

Department of Health, 'Toolkit for Producing Patient Information, Version 2'. Crown copyright, London, 2003 [Online]. Available: <https://www.uea.ac.uk/documents/246046/0/Toolkit+for+producing+patient+information.pdf>

[4]

M. Garner, Z. Ning, and J. Francis, 'A framework for the evaluation of patient information leaflets', *Health Expectations*, vol. 15, no. 3, pp. 283–294, Sep. 2012, doi: 10.1111/j.1369-7625.2011.00665.x.

[5]

'Prescription information leaflets: a pilot study in general practice.', *British Medical Journal* (Clinical research ed.), vol. 287, no. 6400, 1983 [Online]. Available: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1549423/>

[6]

M. P. Berthelsen, E. Husu, S. B. Christensen, K. P. Prahm, J. Vissing, and B. R. Jensen, 'Anti-gravity training improves walking capacity and postural balance in patients with muscular dystrophy', *Neuromuscular Disorders*, vol. 24, no. 6, pp. 492–498, Jun. 2014, doi: 10.1016/j.nmd.2014.03.001.

[7]

E. H. Cup et al., 'Exercise Therapy and Other Types of Physical Therapy for Patients With Neuromuscular Diseases: A Systematic Review', *Archives of Physical Medicine and Rehabilitation*, vol. 88, no. 11, pp. 1452–1464, Nov. 2007, doi: 10.1016/j.apmr.2007.07.024.

[8]

N. B. Voet, E. L. van der Kooi, I. I. Riphagen, E. Lindeman, B. G. van Engelen, and A. C. Geurts, 'Strength training and aerobic exercise training for muscle disease', *Cochrane Database of Systematic Reviews*, Jul. 2013, doi: 10.1002/14651858.CD003907.pub4.

[9]

M.-L. Sveen et al., 'Resistance training in patients with limb-girdle and becker muscular dystrophies', *Muscle & Nerve*, vol. 47, no. 2, pp. 163–169, Feb. 2013, doi: 10.1002/mus.23491.

[10]

T. D. Jeppesen et al., 'Aerobic training is safe and improves exercise capacity in patients with mitochondrial myopathy', *Brain*, vol. 129, no. 12, pp. 3402–3412, Jun. 2006, doi: 10.1093/brain/awl149.

[11]

'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'. [Online]. Available: <https://www.minervamedica.it/en/journals/europa-medicophysica/article.php?cod=R33Y2013N02A0169>

[12]

Institute of Neurology, Queen Square and National Hospital for Neurology and Neurosurgery (London, England), Neurology: a Queen Square textbook, Second edition. Chichester, West Sussex, UK: John Wiley & Sons, Inc, 2016 [Online]. Available: <https://onlinelibrary.wiley.com/doi/book/10.1002/9781118486160>

[13]

'Neuromuscular Disease Center'. [Online]. Available: <https://neuromuscular.wustl.edu/>

[14]

R. J. Barohn, M. M. Dimachkie, and C. E. Jackson, 'A Pattern Recognition Approach to Patients with a Suspected Myopathy', *Neurologic Clinics*, vol. 32, no. 3, pp. 569-593, Aug. 2014, doi: 10.1016/j.ncl.2014.04.008.

[15]

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

[16]

By:Gordon, AM (Gordon, AM); Homsher, E (Homsher, E); Regnier, M (Regnier, M), 'Regulation of contraction in striated muscle', *PHYSIOLOGICAL REVIEWS* *PHYSIOLOGICAL REVIEWS*, vol. 80, no. 2, pp. 853-924, 2000 [Online]. Available: http://apps.webofknowledge.com/full_record.do?product=WOS&search_mode=GeneralSearch&qid=13&SID=C49BOGnSGP9s3PqA3ow&page=1&doc=1

[17]

'Force-velocity properties of human skeletal muscle fibres: myosin heavy chain isoform and temperature dependence.', *The Journal of Physiology*, vol. 495, no. Pt 2, 1996 [Online]. Available: <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1160815/>

[18]

S. D. R. Harridge et al., 'Whole-muscle and single-fibre contractile properties and myosin heavy chain isoforms in humans', *Pflügers Archiv - European Journal of Physiology*, vol. 432, no. 5, pp. 913–920, Sep. 1996, doi: 10.1007/s004240050215.

[19]

S. Hunter, M. White, and M. Thompson, 'Techniques to Evaluate Elderly Human Muscle Function: A Physiological Basis', *The Journals of Gerontology Series A: Biological Sciences and Medical Sciences*, vol. 53A, no. 3, pp. B204–B216, May 1998, doi: 10.1093/gerona/53A.3.B204.

[20]

T. D. O'Brien, N. D. Reeves, V. Baltzopoulos, D. A. Jones, and C. N. Maganaris, 'In vivo measurements of muscle specific tension in adults and children', *Experimental Physiology*, vol. 95, no. 1, pp. 202–210, Jan. 2010, doi: 10.1113/expphysiol.2009.048967.

[21]

P. J. Atherton and K. Smith, 'Muscle protein synthesis in response to nutrition and exercise', *The Journal of Physiology*, vol. 590, no. 5, pp. 1049–1057, Mar. 2012, doi: 10.1113/jphysiol.2011.225003.

[22]

K. Baar, 'Using Molecular Biology to Maximize Concurrent Training', *Sports Medicine*, vol. 44, no. S2, pp. 117–125, Nov. 2014, doi: 10.1007/s40279-014-0252-0.

[23]

M. J. Rennie, H. Wackerhage, E. E. Spangenburg, and F. W. Booth, 'Control of the Size of the Human Muscle Mass', *Annual Review of Physiology*, vol. 66, no. 1, pp. 799–828, Mar. 2004, doi: 10.1146/annurev.physiol.66.052102.134444.

[24]

F. Kadi, 'Cellular and molecular mechanisms responsible for the action of testosterone on human skeletal muscle. A basis for illegal performance enhancement', *British Journal of Pharmacology*, vol. 154, no. 3, pp. 522–528, Jun. 2008, doi: 10.1038/bjp.2008.118.

[25]

C. P. Velloso, 'Regulation of muscle mass by growth hormone and IGF-I', *British Journal of Pharmacology*, vol. 154, no. 3, pp. 557–568, Jun. 2008, doi: 10.1038/bjp.2008.153.

[26]

G. R. Marcotte, D. W. D. West, and K. Baar, 'The Molecular Basis for Load-Induced Skeletal Muscle Hypertrophy', *Calcified Tissue International*, vol. 96, no. 3, pp. 196–210, Mar. 2015, doi: 10.1007/s00223-014-9925-9.

[27]

I. M. Egner, J. C. Bruusgaard, E. Eftestøl, and K. Gundersen, 'A cellular memory mechanism aids overload hypertrophy in muscle long after an episodic exposure to anabolic steroids', *The Journal of Physiology*, vol. 591, no. 24, pp. 6221–6230, Dec. 2013, doi: 10.1113/jphysiol.2013.264457.

[28]

P. J. Atherton and K. Smith, 'Muscle protein synthesis in response to nutrition and exercise', *The Journal of Physiology*, vol. 590, no. 5, pp. 1049–1057, Mar. 2012, doi: 10.1113/jphysiol.2011.225003.

[29]

R. H. Fitts et al., 'Prolonged space flight-induced alterations in the structure and function of human skeletal muscle fibres', *The Journal of Physiology*, vol. 588, no. 18, pp. 3567–3592, Sep. 2010, doi: 10.1113/jphysiol.2010.188508.

[30]

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), '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, vol. 18, no. 6, 2004, doi: 10.1096/fj.03-1228fje. [Online]. Available: http://apps.webofknowledge.com/full_record.do?product=WOS&search_mode=GeneralSearch&qid=6&SID=C49BOGnSGP9s3PqA3ow&page=1&doc=1

[31]

M. J. Rennie, H. Wackerhage, E. E. Spangenburg, and F. W. Booth, 'Control of the Size of the Human Muscle Mass', *Annual Review of Physiology*, vol. 66, no. 1, pp. 799–828, Mar. 2004, doi: 10.1146/annurev.physiol.66.052102.134444.

[32]

T. Shavlakadze and M. Grounds, 'Of bears, frogs, meat, mice and men: complexity of factors affecting skeletal muscle mass and fat', *BioEssays*, vol. 28, no. 10, pp. 994–1009, Oct. 2006, doi: 10.1002/bies.20479.

[33]

Z. A. Puthuchery et al., 'Acute Skeletal Muscle Wasting in Critical Illness', *JAMA*, vol. 310, no. 15, Oct. 2013, doi: 10.1001/jama.2013.278481.

[34]

M. P. Wiggs, 'Can endurance exercise preconditioning prevention disuse muscle atrophy?', *Frontiers in Physiology*, vol. 6, Mar. 2015, doi: 10.3389/fphys.2015.00063.

[35]

K. Baar, 'Using Molecular Biology to Maximize Concurrent Training', *Sports Medicine*, vol. 44, no. S2, pp. 117–125, Nov. 2014, doi: 10.1007/s40279-014-0252-0.

[36]

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

[37]

B. Hoier and Y. Hellsten, 'Exercise-Induced Capillary Growth in Human Skeletal Muscle and the Dynamics of VEGF', *Microcirculation*, vol. 21, no. 4, pp. 301–314, May 2014, doi: 10.1111/micc.12117.

[38]

D. G. Hardie and K. Sakamoto, 'AMPK: A Key Sensor of Fuel and Energy Status in Skeletal Muscle', *Physiology*, vol. 21, no. 1, pp. 48–60, Feb. 2006, doi: 10.1152/physiol.00044.2005.

[39]

J. A. Hawley, M. Hargreaves, M. J. Joyner, and J. R. Zierath, 'Integrative Biology of Exercise', *Cell*, vol. 159, no. 4, pp. 738–749, Nov. 2014, doi: 10.1016/j.cell.2014.10.029.

[40]

D. Briggs and J. E. Morgan, 'Recent progress in satellite cell/myoblast engraftment - relevance for therapy', *FEBS Journal*, vol. 280, no. 17, pp. 4281–4293, Sep. 2013, doi: 10.1111/febs.12273.

[41]

P. S. Zammit, J. P. Golding, Y. Nagata, V. Hudon, T. A. Partridge, and J. R. Beauchamp, 'Muscle satellite cells adopt divergent fates', *The Journal of Cell Biology*, vol. 166, no. 3, pp. 347–357, Aug. 2004, doi: 10.1083/jcb.200312007.

[42]

L. Boldrin and J. E. Morgan, 'Activating muscle stem cells: therapeutic potential in muscle diseases', *Current Opinion in Neurology*, vol. 20, no. 5, pp. 577–582, Oct. 2007, doi: 10.1097/WCO.0b013e3282ef5919.

[43]

L. Boldrin, P. S. Zammit, and J. E. Morgan, 'Satellite cells from dystrophic muscle retain regenerative capacity', *Stem Cell Research*, vol. 14, no. 1, pp. 20–29, Jan. 2015, doi:

10.1016/j.scr.2014.10.007.

[44]

J. Ross et al., 'Defects in Glycosylation Impair Satellite Stem Cell Function and Niche Composition in the Muscles of the Dystrophic Large Mouse', *STEM CELLS*, vol. 30, no. 10, pp. 2330–2341, Oct. 2012, doi: 10.1002/stem.1197.

[45]

'Neuropathology and Applied Neurobiology', vol. Volume 43, Issue 1 [Online]. Available: <https://onlinelibrary.wiley.com/toc/13652990/2017/43/1>

[46]

V. Dubowitz, C. A. Sewry, and A. Oldfors, *Muscle biopsy: a practical approach*, Fourth edition. [Philadelphia, Pa.]: Saunders Elsevier, 2013.

[47]

K. G. Hollingsworth, P. L. de Sousa, V. Straub, and P. G. Carlier, '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*, vol. 22, pp. S54–S67, Oct. 2012, doi: 10.1016/j.nmd.2012.06.005.

[48]

M. P. Wattjes and D. Fischer, *Neuromuscular imaging*. New York: Springer, 2013 [Online]. Available: http://ucl.alma.exlibrisgroup.com/view/action/uresolver.do?operation=resolveService&package_service_id=3320980190004761&institutionId=4761&customerId=4760

[49]

S. C. Forbes et al., 'Magnetic Resonance Imaging and Spectroscopy Assessment of Lower Extremity Skeletal Muscles in Boys with Duchenne Muscular Dystrophy: A Multicenter Cross Sectional Study', *PLoS ONE*, vol. 9, no. 9, Sep. 2014, doi:

10.1371/journal.pone.0106435.

[50]

G. H. Glover and E. Schneider, 'Three-point dixon technique for true water/fat decomposition with B0 inhomogeneity correction', *Magnetic Resonance in Medicine*, vol. 18, no. 2, pp. 371–383, Apr. 1991, doi: 10.1002/mrm.1910180211.

[51]

R. J. Willcocks et al., 'Multicenter prospective longitudinal study of magnetic resonance biomarkers in a large duchenne muscular dystrophy cohort', *Annals of Neurology*, vol. 79, no. 4, pp. 535–547, Apr. 2016, doi: 10.1002/ana.24599.

[52]

J.-Y. Hogrel et al., 'Longitudinal functional and NMR assessment of upper limbs in Duchenne muscular dystrophy', *Neurology*, vol. 86, no. 11, pp. 1022–1030, Mar. 2016, doi: 10.1212/WNL.0000000000002464.

[53]

H. K. Kim et al., '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*, vol. 205, no. 2, pp. W216–W223, Aug. 2015, doi: 10.2214/AJR.14.13755.

[54]

F. Muntoni, S. Torelli, and A. Ferlini, 'Dystrophin and mutations: one gene, several proteins, multiple phenotypes', *The Lancet Neurology*, vol. 2, no. 12, pp. 731–740, Dec. 2003, doi: 10.1016/S1474-4422(03)00585-4.

[55]

K. Bushby et al., 'Diagnosis and management of Duchenne muscular dystrophy, part 1: diagnosis, and pharmacological and psychosocial management', *The Lancet Neurology*, vol. 9, no. 1, pp. 77–93, Jan. 2010, doi: 10.1016/S1474-4422(09)70271-6.

[56]

K. Bushby et al., 'Diagnosis and management of Duchenne muscular dystrophy, part 2: implementation of multidisciplinary care', *The Lancet Neurology*, vol. 9, no. 2, pp. 177–189, Feb. 2010, doi: 10.1016/S1474-4422(09)70272-8.

[57]

V. Ricotti et al., 'Long-term benefits and adverse effects of intermittent versus daily glucocorticoids in boys with Duchenne muscular dystrophy', *Journal of Neurology, Neurosurgery & Psychiatry*, vol. 84, no. 6, pp. 698–705, Jun. 2013, doi: 10.1136/jnnp-2012-303902.

[58]

N. M. Goemans et al., 'Systemic Administration of PRO051 in Duchenne's Muscular Dystrophy', *New England Journal of Medicine*, vol. 364, no. 16, pp. 1513–1522, Apr. 2011, doi: 10.1056/NEJMoa1011367.

[59]

J. R. Mendell et al., 'Eteplirsen for the treatment of Duchenne muscular dystrophy', *Annals of Neurology*, vol. 74, no. 5, pp. 637–647, Nov. 2013, doi: 10.1002/ana.23982.

[60]

J. R. Mendell et al., 'Longitudinal effect of eteplirsen versus historical control on ambulation in Duchenne muscular dystrophy', *Annals of Neurology*, vol. 79, no. 2, pp. 257–271, Feb. 2016, doi: 10.1002/ana.24555.

[61]

M. Kinali et al., '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*, vol. 8, no. 10, pp. 918–928, Oct. 2009, doi: 10.1016/S1474-4422(09)70211-X.

[62]

S. Cirak et al., '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*, vol. 378, no. 9791, pp. 595–605, Aug. 2011, doi: 10.1016/S0140-6736(11)60756-3.

[63]

K. Bushby et al., 'Ataluren treatment of patients with nonsense mutation dystrophinopathy', *Muscle & Nerve*, vol. 50, no. 4, pp. 477–487, Oct. 2014, doi: 10.1002/mus.24332.

[64]

S. Paganoni and A. Amato, 'Electrodiagnostic Evaluation of Myopathies', *Physical Medicine and Rehabilitation Clinics of North America*, vol. 24, no. 1, pp. 193–207, Feb. 2013, doi: 10.1016/j.pmr.2012.08.017.

[65]

A. Fuglsang-Frederiksen, 'The role of different EMG methods in evaluating myopathy', *Clinical Neurophysiology*, vol. 117, no. 6, pp. 1173–1189, Jun. 2006, doi: 10.1016/j.clinph.2005.12.018.

[66]

P. Machado, S. Brady, and M. G. Hanna, 'Update in inclusion body myositis', *Current Opinion in Rheumatology*, vol. 25, no. 6, pp. 763–771, Nov. 2013, doi: 10.1097/01.bor.0000434671.77891.9a.

[67]

P. M. Machado, M. M. Dimachkie, and R. J. Barohn, 'Sporadic inclusion body myositis', *Current Opinion in Neurology*, vol. 27, no. 5, pp. 591–598, Oct. 2014, doi: 10.1097/WCO.000000000000129.

[68]

P. M. Machado et al., 'Ongoing Developments in Sporadic Inclusion Body Myositis', *Current Rheumatology Reports*, vol. 16, no. 12, Dec. 2014, doi: 10.1007/s11926-014-0477-9.

[69]

M. Ahmed et al., 'Targeting protein homeostasis in sporadic inclusion body myositis', *Science Translational Medicine*, vol. 8, no. 331, pp. 331ra41-331ra41, Mar. 2016, doi: 10.1126/scitranslmed.aad4583.

[70]

M. Needham and F. L. Mastaglia, 'Sporadic inclusion body myositis: A review of recent clinical advances and current approaches to diagnosis and treatment', *Clinical Neurophysiology*, vol. 127, no. 3, pp. 1764-1773, Mar. 2016, doi: 10.1016/j.clinph.2015.12.011.

[71]

R. Schröder and B. Schoser, 'Myofibrillar Myopathies: A Clinical and Myopathological Guide', *Brain Pathology*, vol. 19, no. 3, pp. 483-492, Jul. 2009, doi: 10.1111/j.1750-3639.2009.00289.x.

[72]

A. A. Amato and S. A. Greenberg, 'Inflammatory Myopathies', *CONTINUUM: Lifelong Learning in Neurology*, vol. 19, pp. 1615-1633, Dec. 2013, doi: 10.1212/01.CON.0000440662.26427.bd.

[73]

M. Olivé, R. A. Kley, and L. G. Goldfarb, 'Myofibrillar myopathies', *Current Opinion in Neurology*, vol. 26, no. 5, pp. 527-535, Oct. 2013, doi: 10.1097/WCO.0b013e328364d6b1.

[74]

P.-O. Carstens and J. Schmidt, 'Diagnosis, pathogenesis and treatment of myositis: recent advances', *Clinical & Experimental Immunology*, vol. 175, no. 3, pp. 349-358, Mar. 2014, doi: 10.1111/cei.12194.

[75]

M. C. Dalakas, 'Inflammatory Muscle Diseases', *New England Journal of Medicine*, vol. 372, no. 18, pp. 1734–1747, Apr. 2015, doi: 10.1056/NEJMra1402225.

[76]

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

[77]

M. T. C. Baioni and C. R. Ambiel, 'Spinal muscular atrophy: diagnosis, treatment and future prospects', *Jornal de Pediatria*, vol. 86, no. 4, pp. 261–270, Aug. 2010, doi: 10.2223/JPED.1988.

[78]

S. Rudnik-Schöneborn, I. Hausmanowa-Petrusewicz, J. Borkowska, and K. Zerres, 'The Predictive Value of Achieved Motor Milestones Assessed in 441 Patients with Infantile Spinal Muscular Atrophy Types II and III', *European Neurology*, vol. 45, no. 3, pp. 174–181, 2001, doi: 10.1159/000052118.

[79]

B. S. Russman, C. R. Buncher, M. White, F. J. Samaha, and S. T. Iannaccone, 'Function changes in spinal muscular atrophy II and III', *Neurology*, vol. 47, no. 4, pp. 973–976, Oct. 1996, doi: 10.1212/WNL.47.4.973.

[80]

C. H. Wang et al., 'Consensus Statement for Standard of Care in Spinal Muscular Atrophy', *Journal of Child Neurology*, vol. 22, no. 8, pp. 1027–1049, Aug. 2007, doi: 10.1177/0883073807305788.

[81]

E. Mercuri, E. Bertini, and S. T. Iannaccone, 'Childhood spinal muscular atrophy: controversies and challenges', *The Lancet Neurology*, vol. 11, no. 5, pp. 443–452, May 2012, doi: 10.1016/S1474-4422(12)70061-3.

[82]

C. E. M. Hollak and R. Lachmann, Eds., *Inherited Metabolic Disease in Adults*, vol. 1. Oxford University Press, 2016 [Online]. Available: <http://www.oxfordmedicine.com/view/10.1093/med/9780199972135.001.0001/med-9780199972135>

[83]

Nancy D Leslie, 'Very Long-Chain Acyl-Coenzyme A Dehydrogenase Deficiency', *Gene Reviews*, 2018 [Online]. Available: <https://www.ncbi.nlm.nih.gov/books/NBK6816/>

[84]

Thomas Wieser, 'Carnitine Palmitoyltransferase II Deficiency', *Gene Reviews*, 2017 [Online]. Available: <https://www.ncbi.nlm.nih.gov/books/NBK1253/>

[85]

Miguel A Martín, 'Glycogen Storage Disease Type V', *Gene Reviews*, 2014 [Online]. Available: <https://www.ncbi.nlm.nih.gov/books/NBK1344/>

[86]

Nancy Leslie, 'Pompe Disease', *Gene Reviews*, 2017 [Online]. Available: <https://www.ncbi.nlm.nih.gov/books/NBK1261/>

[87]

M. C. Ørngreen and J. Vissing, 'Treatment Opportunities in Patients With Metabolic Myopathies', *Current Treatment Options in Neurology*, vol. 19, no. 11, Nov. 2017, doi: 10.1007/s11940-017-0473-2.

[88]

S. E. Olpin, E. Murphy, R. J. Kirk, R. W. Taylor, and R. Quinlivan, 'The investigation and management of metabolic myopathies', *Journal of Clinical Pathology*, vol. 68, no. 6, pp. 410-417, Jun. 2015, doi: 10.1136/jclinpath-2014-202808.

[89]

B. Feingold et al., 'Management of Cardiac Involvement Associated With Neuromuscular Diseases: A Scientific Statement From the American Heart Association', *Circulation*, vol. 136, no. 13, Sep. 2017, doi: 10.1161/CIR.0000000000000526.

[90]

E. Matthews and M. G. Hanna, 'Skeletal muscle channelopathies', in *Oxford Textbook of Neuromuscular Disorders*, D. Hilton-Jones and M. R. Turner, Eds. Oxford University Press, 2014, pp. 316-325 [Online]. Available: <http://oxfordmedicine.com/view/10.1093/med/9780199698073.001.0001/med-9780199698073-chapter-31>

[91]

C. E. M. Hollak and R. Lachmann, Eds., *Inherited Metabolic Disease in Adults*, vol. 1. Oxford University Press, 2016 [Online]. Available: <http://www.oxfordmedicine.com/view/10.1093/med/9780199972135.001.0001/med-9780199972135>

[92]

J. M. Saudubray, M. R. Baumgartner, and J. Walter, Eds., *Inborn metabolic diseases: diagnosis and treatment*, 6th edition. Berlin: Springer, 2016.

[93]

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