

CLNE0022: Skeletal Muscle and Associated Diseases

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@article{Ahmed_Machado_Miller_Spicer_Herbelin_He_Noel_Wang_McVey_Pasnoor_et al._2016, title={Targeting protein homeostasis in sporadic inclusion body myositis}, volume={8}, DOI={10.1126/scitranslmed.aad4583}, number={331}, journal={Science Translational Medicine}, author={Ahmed, Mhoriam and Machado, Pedro M. and Miller, Adrian and Spicer, Charlotte and Herbelin, Laura and He, Jianghua and Noel, Janelle and Wang, Yunxia and McVey, April L. and Pasnoor, Mamatha and Gallagher, Philip and Statland, Jeffrey and Lu, Ching-Hua and Kalmar, Bernadett and Brady, Stefen and Sethi, Huma and Samandouras, George and Parton, Matt and Holton, Janice L. and Weston, Anne and Collinson, Lucy and Taylor, J. Paul and Schiavo, Giampietro and Hanna, Michael G. and Barohn, Richard J. and Dimachkie, Mazen M. and Greensmith, Linda}, year={2016}, month={Mar}, pages={331ra41-331ra41} }

@article{Amato_Greenberg_2013, title={Inflammatory Myopathies}, volume={19}, DOI={10.1212/01.CON.0000440662.26427.bd}, journal={CONTINUUM: Lifelong Learning in Neurology}, author={Amato, Anthony A. and Greenberg, Steven A.}, year={2013}, month={Dec}, pages={1615-1633} }

@article{Atherton_Smith_2012a, title={Muscle protein synthesis in response to nutrition and exercise}, volume={590}, DOI={10.1113/jphysiol.2011.225003}, number={5}, journal={The Journal of Physiology}, author={Atherton, P. J. and Smith, K.}, year={2012}, month={Mar}, pages={1049-1057} }

@article{Atherton_Smith_2012b, title={Muscle protein synthesis in response to nutrition and exercise}, volume={590}, DOI={10.1113/jphysiol.2011.225003}, number={5}, journal={The Journal of Physiology}, author={Atherton, P. J. and Smith, K.}, year={2012}, month={Mar}, pages={1049-1057} }

@article{Baar_2014a, title={Using Molecular Biology to Maximize Concurrent Training}, volume={44}, DOI={10.1007/s40279-014-0252-0}, number={S2}, journal={Sports Medicine}, author={Baar, Keith}, year={2014}, month={Nov}, pages={117-125} }

@article{Baar_2014b, title={Using Molecular Biology to Maximize Concurrent Training}, volume={44}, DOI={10.1007/s40279-014-0252-0}, number={S2}, journal={Sports Medicine}, author={Baar, Keith}, year={2014}, month={Nov}, pages={117-125} }

@article{Baioni_Ambiel_2010, title={Spinal muscular atrophy: diagnosis, treatment and future prospects}, volume={86}, DOI={10.2223/JPED.1988}, number={4}, journal={Jornal de Pediatria}, author={Baioni, Mariana T. C. and Ambiel, Celia R.}, year={2010}, month={Aug}, pages={261-270} }

@article{Barohn_Dimachkie_Jackson_2014, title={A Pattern Recognition Approach to Patients with a Suspected Myopathy}, volume={32}, DOI={10.1016/j.ncl.2014.04.008}, number={3}, journal={Neurologic Clinics}, author={Barohn, Richard J. and Dimachkie, Mazen M. and Jackson, Carlayne E.}, year={2014}, month={Aug}, pages={569-593} }

@article{Berthelsen_Husu_Christensen_Prahm_Vissing_Jensen_2014, title={Anti-gravity training improves walking capacity and postural balance in patients with muscular dystrophy}, volume={24}, DOI={10.1016/j.nmd.2014.03.001}, number={6}, journal={Neuromuscular Disorders}, author={Berthelsen, Martin Peter and Husu, Edith and Christensen, Sofie Bouschinger and Prahm, Kira Philipsen and Vissing, John and Jensen, Bente Rona}, year={2014}, month={Jun}, pages={492-498} }

@article{Boldrin_Morgan_2007, title={Activating muscle stem cells: therapeutic potential in muscle diseases}, volume={20}, DOI={10.1097/WCO.0b013e3282ef5919}, number={5}, journal={Current Opinion in Neurology}, author={Boldrin, Luisa and Morgan, Jennifer E}, year={2007}, month={Oct}, pages={577-582} }

@article{Boldrin_Zammit_Morgan_2015, title={Satellite cells from dystrophic muscle retain regenerative capacity}, volume={14}, DOI={10.1016/j.scr.2014.10.007}, number={1}, journal={Stem Cell Research}, author={Boldrin, Luisa and Zammit, Peter S. and Morgan, Jennifer E.}, year={2015}, month={Jan}, pages={20-29} }

@article{Briggs_Morgan_2013, title={Recent progress in satellite cell/myoblast engraftment - relevance for therapy}, volume={280}, DOI={10.1111/febs.12273}, number={17}, journal={FEBS Journal}, author={Briggs, Deborah and Morgan, Jennifer E.}, year={2013}, month={Sep}, pages={4281-4293} }

@article{Bushby_Finkel_Birnkrant_Clemens_Cripe_Kaul_Kinnett_McDonald_Pandya_et al._2010a, title={Diagnosis and management of Duchenne muscular dystrophy, part 1: diagnosis, and pharmacological and psychosocial management}, volume={9}, DOI={10.1016/S1474-4422(09)70271-6}, number={1}, journal={The Lancet Neurology}, author={Bushby, Katharine and Finkel, Richard and Birnkrant, David J and Case, Laura E and Clemens, Paula R and Cripe, Linda and Kaul, Ajay and Kinnett, Kathi and McDonald, Craig and Pandya, Shree and Poysky, James and Shapiro, Frederic and Tomezsko, Jean and Constantin, Carolyn}, year={2010}, month={Jan}, pages={77-93} }

@article{Bushby_Finkel_Birnkrant_Clemens_Cripe_Kaul_Kinnett_McDonald_Pandya_et al._2010b, title={Diagnosis and management of Duchenne muscular dystrophy, part 2: implementation of multidisciplinary care}, volume={9}, DOI={10.1016/S1474-4422(09)70272-8}, number={2}, journal={The Lancet Neurology}, author={Bushby, Katharine and Finkel, Richard and Birnkrant, David J and Case, Laura E and Clemens, Paula R and Cripe, Linda and Kaul, Ajay and Kinnett, Kathi and McDonald, Craig and Pandya, Shree and Poysky, James and Shapiro, Frederic and Tomezsko, Jean and Constantin, Carolyn}, year={2010}, month={Feb}, pages={177-189} }

@article{Bushby_Finkel_Wong_Barohn_Campbell_Comi_Connolly_Day_Flanigan_Goemans_et al._2014, title={Ataluren treatment of patients with nonsense mutation dystrophinopathy}, volume={50}, DOI={10.1002/mus.24332}, number={4},

journal={Muscle & Nerve}, author={Bushby, Katharine and Finkel, Richard and Wong, Brenda and Barohn, Richard and Campbell, Craig and Comi, Giacomo P. and Connolly, Anne M. and Day, John W. and Flanigan, Kevin M. and Goemans, Nathalie and Jones, Kristi J. and Mercuri, Eugenio and Quinlivan, Ros and Renfroe, James B. and Russman, Barry and Ryan, Monique M. and Tulinius, Mar and Voit, Thomas and Moore, Steven A. and Lee Sweeney, H. and Abresch, Richard T. and Coleman, Kim L. and Eagle, Michelle and Florence, Julaine and Gappmaier, Eduard and Glanzman, Allan M. and Henricson, Erik and Barth, Jay and Elfring, Gary L. and Reha, Allen and Spiegel, Robert J. and O'donnell, Michael W. and Peltz, Stuart W. and Mcdonald, Craig M.}, year={2014}, month={Oct}, pages={477-487} }

@article{By:Gordon, AM (Gordon, AM); Homsher, E (Homsher, E); Regnier, M (Regnier, M)_2000, title={Regulation of contraction in striated muscle}, volume={80}, url={http://apps.webofknowledge.com/full_record.do?product=WOS&search_mode=GeneralSearch&qid=13&SID=C49BOGnSGP9s3PqA3ow&page=1&doc=1}, number={2}, journal={PHYSIOLOGICAL REVIEWS PHYSIOLOGICAL REVIEWS}, author={By:Gordon, AM (Gordon, AM); Homsher, E (Homsher, E); Regnier, M (Regnier, M)}, year={2000}, pages={853-924} }

@article{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, title={Disuse atrophy and exercise rehabilitation in humans profoundly affects the expression of genes associated with the regulation of skeletal muscle mass}, volume={18}, url={http://apps.webofknowledge.com/full_record.do?product=WOS&search_mode=GeneralSearch&qid=6&SID=C49BOGnSGP9s3PqA3ow&page=1&doc=1}, DOI={10.1096/fj.03-1228fje}, number={6}, journal={FASEB JOURNAL FASEB JOURNAL}, author={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)}, year={2004} }

@article{Carstens_Schmidt_2014, title={Diagnosis, pathogenesis and treatment of myositis: recent advances}, volume={175}, DOI={10.1111/cei.12194}, number={3}, journal={Clinical & Experimental Immunology}, author={Carstens, P. -O. and Schmidt, J.}, year={2014}, month={Mar}, pages={349-358} }

@article{Cirak_Arechavala-Gomez_Guglieri_Feng_Torelli_Anthony_Abbs_Garralda_Bourke_Wells_et al._2011, title={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}, volume={378}, DOI={10.1016/S0140-6736(11)60756-3}, number={9791}, journal={The Lancet}, author={Cirak, Sebahattin and Arechavala-Gomez, Virginia and Guglieri, Michela and Feng, Lucy and Torelli, Silvia and Anthony, Karen and Abbs, Stephen and Garralda, Maria Elena and Bourke, John and Wells, Dominic J and Dickson, George and Wood, Matthew JA and Wilton, Steve D and Straub, Volker and Kole, Ryszard and Shrewsbury, Stephen B and Sewry, Caroline and Morgan, Jennifer E and Bushby, Kate and Muntoni, Francesco}, year={2011}, month={Aug}, pages={595-605} }

@article{Craig_Ashcroft_Belew_Stocks_Currell_Baar_Philp_2015, title={Utilizing small nutrient compounds as enhancers of exercise-induced mitochondrial biogenesis}, volume={6}, DOI={10.3389/fphys.2015.00296}, journal={Frontiers in Physiology},

author={Craig, Daniel M. and Ashcroft, Stephen P. and Belew, Micah Y. and Stocks, Ben and Currell, Kevin and Baar, Keith and Philp, Andrew}, year={2015}, month={Oct} }

@article{Cup_Pieterse_ten Broek-Pastoor_Munneke_van Engelen_Hendricks_van der Wilt_Oostendorp_2007, title={Exercise Therapy and Other Types of Physical Therapy for Patients With Neuromuscular Diseases: A Systematic Review}, volume={88}, DOI={10.1016/j.apmr.2007.07.024}, number={11}, journal={Archives of Physical Medicine and Rehabilitation}, author={Cup, Edith H. and Pieterse, Allan J. and ten Broek-Pastoor, Jessica M. and Munneke, Marten and van Engelen, Baziel G. and Hendricks, Henk T. and van der Wilt, Gert J. and Oostendorp, Rob A.}, year={2007}, month={Nov}, pages={1452-1464} }

@article{Dalakas_2015, title={Inflammatory Muscle Diseases}, volume={372}, DOI={10.1056/NEJMra1402225}, number={18}, journal={New England Journal of Medicine}, author={Dalakas, Marinos C.}, year={2015}, month={Apr}, pages={1734-1747} }

@book{Darras_De Vivo_Jones_2003, address={Philadelphia, Penn. ; London}, title={Neuromuscular disorders of infancy, childhood, and adolescence: a clinician's approach}, publisher={Butterworth-Heinemann}, author={Darras, Basil T. and De Vivo, Darryl C. and Jones, H. Royden}, year={2003} }

@misc{2003, address={London}, title={Toolkit for Producing Patient Information, Version 2}, url={https://www.uea.ac.uk/documents/246046/0/Toolkit+for+producing+patient+information.pdf}, publisher={Crown copyright}, author={Department of Health}, year={2003} }

@book{Dubowitz_Sewry_Oldfors_2013, address={Philadelphia, Pa.}, edition={Fourth edition}, title={Muscle biopsy: a practical approach}, publisher={Saunders Elsevier}, author={Dubowitz, Victor and Sewry, Caroline A. and Oldfors, Anders}, year={2013} }

@article{Egner_Bruusgaard_Eftestøl_Gundersen_2013, title={A cellular memory mechanism aids overload hypertrophy in muscle long after an episodic exposure to anabolic steroids}, volume={591}, DOI={10.1113/jphysiol.2013.264457}, number={24}, journal={The Journal of Physiology}, author={Egner, Ingrid M. and Bruusgaard, Jo C. and Eftestøl, Einar and Gundersen, Kristian}, year={2013}, month={Dec}, pages={6221-6230} }

@article{Feingold_Mahle_Auerbach_Clemens_Domenighetti_Jefferies_Judge_Lal_Markham_Parks_et al._2017, title={Management of Cardiac Involvement Associated With Neuromuscular Diseases: A Scientific Statement From the American Heart Association}, volume={136}, DOI={10.1161/CIR.0000000000000526}, number={13}, journal={Circulation}, author={Feingold, Brian and Mahle, William T. and Auerbach, Scott and Clemens, Paula and Domenighetti, Andrea A. and Jefferies, John L. and Judge, Daniel P. and Lal, Ashwin K. and Markham, Larry W. and Parks, W. James and Tsuda, Takeshi and Wang, Paul J. and Yoo, Shi-Joon}, year={2017}, month={Sep} }

@article{Fitts_Trappe_Costill_Gallagher_Creer_Colloton_Peters_Romatowski_Bain_Riley_20

10, title={Prolonged space flight-induced alterations in the structure and function of human skeletal muscle fibres}, volume={588}, DOI={10.1113/jphysiol.2010.188508}, number={18}, journal={The Journal of Physiology}, author={Fitts, R. H. and Trappe, S. W. and Costill, D. L. and Gallagher, P. M. and Creer, A. C. and Colloton, P. A. and Peters, J. R. and Romatowski, J. G. and Bain, J. L. and Riley, D. A.}, year={2010}, month={Sep}, pages={3567-3592} }

@article{Forbes_Willcocks_Triplett_Rooney_Lott_Wang_Pollaro_Senesac_Daniels_Finkel_et al._2014, title={Magnetic Resonance Imaging and Spectroscopy Assessment of Lower Extremity Skeletal Muscles in Boys with Duchenne Muscular Dystrophy: A Multicenter Cross Sectional Study}, volume={9}, DOI={10.1371/journal.pone.0106435}, number={9}, journal={PLoS ONE}, author={Forbes, Sean C. and Willcocks, Rebecca J. and Triplett, William T. and Rooney, William D. and Lott, Donovan J. and Wang, Dah-Jyuu and Pollaro, Jim and Senesac, Claudia R. and Daniels, Michael J. and Finkel, Richard S. and Russman, Barry S. and Byrne, Barry J. and Finanger, Erika L. and Tennekoon, Gihan I. and Walter, Glenn A. and Sweeney, H. Lee and Vandenborne, Krista}, year={2014}, month={Sep} }

@article{Fuglsang-Frederiksen_2006, title={The role of different EMG methods in evaluating myopathy}, volume={117}, DOI={10.1016/j.clinph.2005.12.018}, number={6}, journal={Clinical Neurophysiology}, author={Fuglsang-Frederiksen, Anders}, year={2006}, month={Jun}, pages={1173-1189} }

@article{Garner_Ning_Francis_2012, title={A framework for the evaluation of patient information leaflets}, volume={15}, DOI={10.1111/j.1369-7625.2011.00665.x}, number={3}, journal={Health Expectations}, author={Garner, Mark and Ning, Zhenye and Francis, Jill}, year={2012}, month={Sep}, pages={283-294} }

@article{Glover_Schneider_1991, title={Three-point dixon technique for true water/fat decomposition with B0 inhomogeneity correction}, volume={18}, DOI={10.1002/mrm.1910180211}, number={2}, journal={Magnetic Resonance in Medicine}, author={Glover, G. H. and Schneider, E.}, year={1991}, month={Apr}, pages={371-383} }

@article{Goemans_Tulinius_van den Akker_Burm_Ekhart_Heuvelmans_Holling_Janson_Platenburg_Sipkens_et al._2011, title={Systemic Administration of PRO051 in Duchenne's Muscular Dystrophy}, volume={364}, DOI={10.1056/NEJMoa1011367}, number={16}, journal={New England Journal of Medicine}, author={Goemans, Nathalie M. and Tulinius, Mar and van den Akker, Johanna T. and Burm, Brigitte E. and Ekhart, Peter F. and Heuvelmans, Niki and Holling, Tjadine and Janson, Anneke A. and Platenburg, Gerard J. and Sipkens, Jessica A. and Sitsen, J.M. Ad and Aartsma-Rus, Annemieke and van Ommen, Gert-Jan B. and Buyse, Gunnar and Darin, Niklas and Verschuuren, Jan J. and Campion, Giles V. and de Kimpe, Sjef J. and van Deutekom, Judith C.}, year={2011}, month={Apr}, pages={1513-1522} }

@book{Great Britain. Audit Commission for Local Authorities and the National Health Service in England and Wales., title={What Seems to Be the Matter Communication (National Health Service Report)}, url={https://webarchive.nationalarchives.gov.uk/20150410163038/http://archive.audit-commission.gov.uk/auditcommission/aboutus/publications/pages/national-reports-and-studies-archive.aspx.html}, publisher={Stationery Office}, author={Great Britain. Audit

Commission for Local Authorities and the National Health Service in England and Wales.} }

@article{Hardie_Sakamoto_2006, title={AMPK: A Key Sensor of Fuel and Energy Status in Skeletal Muscle}, volume={21}, DOI={10.1152/physiol.00044.2005}, number={1}, journal={Physiology}, author={Hardie, D. Grahame and Sakamoto, Kei}, year={2006}, month={Feb}, pages={48-60} }

@article{Harridge_Bottinelli_Canepari_Pellegrino_Reggiani_Esbjörnsson_Saltin_1996, title={Whole-muscle and single-fibre contractile properties and myosin heavy chain isoforms in humans}, volume={432}, DOI={10.1007/s004240050215}, number={5}, journal={Pflügers Archiv - European Journal of Physiology}, author={Harridge, S. D. R. and Bottinelli, R. and Canepari, M. and Pellegrino, M. A. and Reggiani, C. and Esbjörnsson, M. and Saltin, B.}, year={1996}, month={Sep}, pages={913-920} }

@article{Hawley_Hargreaves_Joyner_Zierath_2014, title={Integrative Biology of Exercise}, volume={159}, DOI={10.1016/j.cell.2014.10.029}, number={4}, journal={Cell}, author={Hawley, John A. and Hargreaves, Mark and Joyner, Michael J. and Zierath, Juleen R.}, year={2014}, month={Nov}, pages={738-749} }

@article{Hogrel_Wary_Morauz_Azzabou_Decostre_Ollivier_Canal_Lilien_Ledoux_Annoussamy_et al._2016, title={Longitudinal functional and NMR assessment of upper limbs in Duchenne muscular dystrophy}, volume={86}, DOI={10.1212/WNL.0000000000002464}, number={11}, journal={Neurology}, author={Hogrel, Jean-Yves and Wary, Claire and Morauz, Amélie and Azzabou, Noura and Decostre, Valérie and Ollivier, Gwenn and Canal, Aurélie and Lilien, Charlotte and Ledoux, Isabelle and Annoussamy, Mélanie and Reguiba, Nacera and Gidaro, Teresa and Le Moing, Anne Gaelle and Cardas, Ruxandra and Voit, Thomas and Carlier, Pierre G. and Servais, Laurent}, year={2016}, month={Mar}, pages={1022-1030} }

@article{Hoier_Hellsten_2014, title={Exercise-Induced Capillary Growth in Human Skeletal Muscle and the Dynamics of VEGF}, volume={21}, DOI={10.1111/micc.12117}, number={4}, journal={Microcirculation}, author={Hoier, Birgitte and Hellsten, Ylva}, year={2014}, month={May}, pages={301-314} }

@article{Hollingsworth_de Sousa_Straub_Carlier_2012, title={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}, volume={22}, DOI={10.1016/j.nmd.2012.06.005}, journal={Neuromuscular Disorders}, author={Hollingsworth, Kieren G. and de Sousa, Paulo L. and Straub, Volker and Carlier, Pierre G.}, year={2012}, month={Oct}, pages={S54-S67} }

@article{Hunter_White_Thompson_1998, title={Techniques to Evaluate Elderly Human Muscle Function: A Physiological Basis}, volume={53A}, DOI={10.1093/gerona/53A.3.B204}, number={3}, journal={The Journals of Gerontology Series A: Biological Sciences and Medical Sciences}, author={Hunter, S. and White, M. and Thompson, M.}, year={1998}, month={May}, pages={B204-B216} }

@book{Institute of Neurology, Queen Square_National Hospital for Neurology and

Neurosurgery (London, England)_2016, address={Chichester, West Sussex, UK},
edition={Second edition}, title={Neurology: a Queen Square textbook},
url={https://onlinelibrary.wiley.com/doi/book/10.1002/9781118486160}, publisher={John
Wiley & Sons, Inc}, author={Institute of Neurology, Queen Square and National Hospital
for Neurology and Neurosurgery (London, England)}, editor={Clarke, Charles and Howard,
Robin and Rossor, M. and Shorvon, S. D.}, year={2016} }

@article{Jeppesen_Schwartz_Olsen_Wibrand_Krag_Duno_Hauerslev_Vissing_2006,
title={Aerobic training is safe and improves exercise capacity in patients with
mitochondrial myopathy}, volume={129}, DOI={10.1093/brain/awl149}, number={12},
journal={Brain}, author={Jeppesen, T. D. and Schwartz, M. and Olsen, D. B. and Wibrand,
F. and Krag, T. and Duno, M. and Hauerslev, S. and Vissing, J.}, year={2006},
month={Jun}, pages={3402-3412} }

@book{Jones_Haan_Round_2004, address={Edinburgh}, title={Skeletal muscle from
molecules to movement: a textbook of muscle physiology for sport, exercise,
physiotherapy and medicine}, publisher={Churchill Livingstone}, author={Jones, D. A.
and Haan, Arnold de and Round, Joan M.}, year={2004} }

@article{Kadi_2008, title={Cellular and molecular mechanisms responsible for the action
of testosterone on human skeletal muscle. A basis for illegal performance enhancement},
volume={154}, DOI={10.1038/bjp.2008.118}, number={3}, journal={British Journal of
Pharmacology}, author={Kadi, F}, year={2008}, month={Jun}, pages={522-528} }

@article{Kim_Serai_Lindquist_Merrow_Horn_Kim_Wong_2015, title={Quantitative Skeletal
Muscle MRI: Part 2, MR Spectroscopy and T2 Relaxation Time Mapping—Comparison
Between Boys With Duchenne Muscular Dystrophy and Healthy Boys}, volume={205},
DOI={10.2214/AJR.14.13755}, number={2}, journal={American Journal of
Roentgenology}, author={Kim, Hee Kyung and Serai, Suraj and Lindquist, Diana and
Merrow, Arnold C. and Horn, Paul S. and Kim, Dong Hoon and Wong, Brenda L.},
year={2015}, month={Aug}, pages={W216-W223} }

@article{Kinali_Arechavala-Gomez_Feng_Cirak_Hunt_Adkin_Guglieri_Ashton_Abbs_Nihoya
nnopoulos_et al._2009, title={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}, volume={8},
DOI={10.1016/S1474-4422(09)70211-X}, number={10}, journal={The Lancet
Neurology}, author={Kinali, Maria and Arechavala-Gomez, Virginia and Feng, Lucy and
Cirak, Sebahattin and Hunt, David and Adkin, Carl and Guglieri, Michela and Ashton, Emma
and Abbs, Stephen and Nihoyannopoulos, Petros and Garralda, Maria Elena and
Rutherford, Mary and Mcculley, Caroline and Popplewell, Linda and Graham, Ian R and
Dickson, George and Wood, Matthew JA and Wells, Dominic J and Wilton, Steve D and Kole,
Ryszard and Straub, Volker and Bushby, Kate and Sewry, Caroline and Morgan, Jennifer E
and Muntoni, Francesco}, year={2009}, month={Oct}, pages={918-928} }

@article{Machado_Brady_Hanna_2013, title={Update in inclusion body myositis},
volume={25}, DOI={10.1097/01.bor.0000434671.77891.9a}, number={6},
journal={Current Opinion in Rheumatology}, author={Machado, Pedro and Brady, Stefen
and Hanna, Michael G.}, year={2013}, month={Nov}, pages={763-771} }

@article{Machado_Ahmed_Brady_Gang_Healy_Morrow_Wallace_Dewar_Ramdharry_Parton_et al._2014, title={Ongoing Developments in Sporadic Inclusion Body Myositis}, volume={16}, DOI={10.1007/s11926-014-0477-9}, number={12}, journal={Current Rheumatology Reports}, author={Machado, Pedro M. and Ahmed, Mhoriam and Brady, Stefen and Gang, Qiang and Healy, Estelle and Morrow, Jasper M. and Wallace, Amanda C. and Dewar, Liz and Ramdharry, Gita and Parton, Matthew and Holton, Janice L. and Houlden, Henry and Greensmith, Linda and Hanna, Michael G.}, year={2014}, month={Dec} }

@article{Machado_Dimachkie_Barohn_2014, title={Sporadic inclusion body myositis}, volume={27}, DOI={10.1097/WCO.000000000000129}, number={5}, journal={Current Opinion in Neurology}, author={Machado, Pedro M. and Dimachkie, Mazen M. and Barohn, Richard J.}, year={2014}, month={Oct}, pages={591-598} }

@article{Marcotte_West_Baar_2015, title={The Molecular Basis for Load-Induced Skeletal Muscle Hypertrophy}, volume={96}, DOI={10.1007/s00223-014-9925-9}, number={3}, journal={Calcified Tissue International}, author={Marcotte, George R. and West, Daniel W. D. and Baar, Keith}, year={2015}, month={Mar}, pages={196-210} }

@inbook{Matthews_Hanna_2014, title={Skeletal muscle channelopathies}, url={http://oxfordmedicine.com/view/10.1093/med/9780199698073.001.0001/med-9780199698073-chapter-31}, DOI={10.1093/med/9780199698073.003.0031}, booktitle={Oxford Textbook of Neuromuscular Disorders}, publisher={Oxford University Press}, author={Matthews, Emma and Hanna, Michael G.}, editor={Hilton-Jones, David and Turner, Martin R.}, year={2014}, month={May}, pages={316-325} }

@article{Mendell_Goemans_Lowes_Alfano_Berry_Shao_Kaye_Mercuri_2016, title={Longitudinal effect of eteplirsen versus historical control on ambulation in Duchenne muscular dystrophy}, volume={79}, DOI={10.1002/ana.24555}, number={2}, journal={Annals of Neurology}, author={Mendell, Jerry R. and Goemans, Nathalie and Lowes, Linda P. and Alfano, Lindsay N. and Berry, Katherine and Shao, James and Kaye, Edward M. and Mercuri, Eugenio}, year={2016}, month={Feb}, pages={257-271} }

@article{Mendell_Rodino-Klapac_Sahenk_Roush_Bird_Lowes_Alfano_Gomez_Lewis_Kota_et al._2013, title={Eteplirsen for the treatment of Duchenne muscular dystrophy}, volume={74}, DOI={10.1002/ana.23982}, number={5}, journal={Annals of Neurology}, author={Mendell, Jerry R. and Rodino-Klapac, Louise R. and Sahenk, Zarife and Roush, Kandice and Bird, Loren and Lowes, Linda P. and Alfano, Lindsay and Gomez, Ann Maria and Lewis, Sarah and Kota, Janaiah and Malik, Vinod and Shontz, Kim and Walker, Christopher M. and Flanigan, Kevin M. and Corridore, Marco and Kean, John R. and Allen, Hugh D. and Shilling, Chris and Melia, Kathleen R. and Sazani, Peter and Saoud, Jay B. and Kaye, Edward M.}, year={2013}, month={Nov}, pages={637-647} }

@article{Mercuri_Bertini_Iannaccone_2012, title={Childhood spinal muscular atrophy: controversies and challenges}, volume={11}, DOI={10.1016/S1474-4422(12)70061-3}, number={5}, journal={The Lancet Neurology}, author={Mercuri, Eugenio and Bertini, Enrico and Iannaccone, Susan T}, year={2012}, month={May}, pages={443-452} }

@article{Miguel A Martín_2014, title={Glycogen Storage Disease Type V}, url={https://www.ncbi.nlm.nih.gov/books/NBK1344/}, journal={Gene Reviews},

publisher={University of Washington, Seattle}, author={Miguel A Martín}, year={2014} }

@article{Muntoni_Torelli_Ferlini_2003, title={Dystrophin and mutations: one gene, several proteins, multiple phenotypes}, volume={2}, DOI={10.1016/S1474-4422(03)00585-4}, number={12}, journal={The Lancet Neurology}, author={Muntoni, Francesco and Torelli, Silvia and Ferlini, Alessandra}, year={2003}, month={Dec}, pages={731-740} }

@article{Nancy D Leslie_2018, title={Very Long-Chain Acyl-Coenzyme A Dehydrogenase Deficiency}, url={https://www.ncbi.nlm.nih.gov/books/NBK6816/}, journal={Gene Reviews}, publisher={University of Washington, Seattle}, author={Nancy D Leslie}, year={2018} }

@article{Nancy Leslie_2017, title={Pompe Disease}, url={https://www.ncbi.nlm.nih.gov/books/NBK1261/}, journal={Gene Reviews}, publisher={University of Washington, Seattle}, author={Nancy Leslie}, year={2017} }

@article{Needham_Mastaglia_2016, title={Sporadic inclusion body myositis: A review of recent clinical advances and current approaches to diagnosis and treatment}, volume={127}, DOI={10.1016/j.clinph.2015.12.011}, number={3}, journal={Clinical Neurophysiology}, author={Needham, Merrilee and Mastaglia, Frank L.}, year={2016}, month={Mar}, pages={1764-1773} }

@article{O'Brien_Reeves_Baltzopoulos_Jones_Maganaris_2010, title={In vivo measurements of muscle specific tension in adults and children}, volume={95}, DOI={10.1113/expphysiol.2009.048967}, number={1}, journal={Experimental Physiology}, author={O'Brien, Thomas D. and Reeves, Neil D. and Baltzopoulos, Vasilios and Jones, David A. and Maganaris, Constantinos N.}, year={2010}, month={Jan}, pages={202-210} }

@article{Olivé_Kley_Goldfarb_2013, title={Myofibrillar myopathies}, volume={26}, DOI={10.1097/WCO.0b013e328364d6b1}, number={5}, journal={Current Opinion in Neurology}, author={Olivé, Montse and Kley, Rudolf A. and Goldfarb, Lev G.}, year={2013}, month={Oct}, pages={527-535} }

@article{Olpin_Murphy_Kirk_Taylor_Quinlivan_2015, title={The investigation and management of metabolic myopathies}, volume={68}, DOI={10.1136/jclinpath-2014-202808}, number={6}, journal={Journal of Clinical Pathology}, author={Olpin, Simon Edward and Murphy, Elaine and Kirk, Richard James and Taylor, Robert William and Quinlivan, Rosaline}, year={2015}, month={Jun}, pages={410-417} }

@article{Ørngreen_Vissing_2017, title={Treatment Opportunities in Patients With Metabolic Myopathies}, volume={19}, DOI={10.1007/s11940-017-0473-2}, number={11}, journal={Current Treatment Options in Neurology}, author={Ørngreen, Mette Cathrine and Vissing, John}, year={2017}, month={Nov} }

@article{Paganoni_Amato_2013, title={Electrodiagnostic Evaluation of Myopathies}, volume={24}, DOI={10.1016/j.pmr.2012.08.017}, number={1}, journal={Physical Medicine and Rehabilitation Clinics of North America}, author={Paganoni, Sabrina and

Amato, Anthony}, year={2013}, month={Feb}, pages={193-207} }

@article{Puthucheary_Rawal_McPhail_Connolly_Ratnayake_Chan_Hopkinson_Padhke_Dew_Sidhu_et al._2013, title={Acute Skeletal Muscle Wasting in Critical Illness}, volume={310}, DOI={10.1001/jama.2013.278481}, number={15}, journal={JAMA}, author={Puthucheary, Zudin A. and Rawal, Jaikity and McPhail, Mark and Connolly, Bronwen and Ratnayake, Gamunu and Chan, Pearl and Hopkinson, Nicholas S. and Padhke, Rahul and Dew, Tracy and Sidhu, Paul S. and Velloso, Cristiana and Seymour, John and Agle, Chibez C. and Selby, Anna and Limb, Marie and Edwards, Lindsay M. and Smith, Kenneth and Rowlerson, Anthea and Rennie, Michael John and Moxham, John and Harridge, Stephen D. R. and Hart, Nicholas and Montgomery, Hugh E.}, year={2013}, month={Oct} }

@article{Rennie_Wackerhage_Spangenburg_Booth_2004a, title={Control of the Size of the Human Muscle Mass}, volume={66}, DOI={10.1146/annurev.physiol.66.052102.134444}, number={1}, journal={Annual Review of Physiology}, author={Rennie, Michael J. and Wackerhage, Henning and Spangenburg, Espen E. and Booth, Frank W.}, year={2004}, month={Mar}, pages={799-828} }

@article{Rennie_Wackerhage_Spangenburg_Booth_2004b, title={Control of the Size of the Human Muscle Mass}, volume={66}, DOI={10.1146/annurev.physiol.66.052102.134444}, number={1}, journal={Annual Review of Physiology}, author={Rennie, Michael J. and Wackerhage, Henning and Spangenburg, Espen E. and Booth, Frank W.}, year={2004}, month={Mar}, pages={799-828} }

@article{Ricotti_Ridout_Scott_Quinlivan_Robb_Manzur_Muntoni_Manzur_Muntoni_Robb_et al._2013, title={Long-term benefits and adverse effects of intermittent versus daily glucocorticoids in boys with Duchenne muscular dystrophy}, volume={84}, DOI={10.1136/jnnp-2012-303902}, number={6}, journal={Journal of Neurology, Neurosurgery & Psychiatry}, author={Ricotti, V. and Ridout, D. A. and Scott, E. and Quinlivan, R. and Robb, S. A. and Manzur, A. Y. and Muntoni, F. and Manzur, A. and Muntoni, F. and Robb, S. and Quinlivan, R. and Ricotti, V. and Main, M. and Bushby, K. and Straub, V. and Sarkozy, A. and Guglieri, M. and Strehle, E. and Eagle, M. and Mayhew, A. and Roper, H. and McMurchie, H. and Childs, A. and Pysden, K. and Pallant, L. and Spinty, S. and Peachey, G. and Shillington, A. and Wraige, E. and Jungbluth, H. and Sheehan, J. and Spahr, R. and Hughes, I. and Bateman, E. and Cammiss, C. and Willis, T. and Groves, L. and Emery, N. and Baxter, P. and Senior, M. and Hartley, L. and Parsons, B. and Majumdar, A. and Jenkins, L. and Naismith, K. and Keddie, A. and Horrocks, I. and Di Marco, M. and Chow, G. and Miah, A.}, year={2013}, month={Jun}, pages={698-705} }

@article{Ross_Benn_Jonuschies_Boldrin_Muntoni_Hewitt_Brown_Morgan_2012, title={Defects in Glycosylation Impair Satellite Stem Cell Function and Niche Composition in the Muscles of the Dystrophic Large Mouse}, volume={30}, DOI={10.1002/stem.1197}, number={10}, journal={STEM CELLS}, author={Ross, Jacob and Benn, Abigail and Jonuschies, Jacqueline and Boldrin, Luisa and Muntoni, Francesco and Hewitt, Jane E. and Brown, Susan C. and Morgan, Jennifer E.}, year={2012}, month={Oct}, pages={2330-2341} }

@article{Rudnik-Schöneborn_Hausmanowa-Petrusewicz_Borkowska_Zerres_2001, title={The Predictive Value of Achieved Motor Milestones Assessed in 441 Patients with Infantile Spinal Muscular Atrophy Types II and III}, volume={45}, DOI={10.1159/000052118}, number={3}, journal={European Neurology}, author={Rudnik-Schöneborn, Sabine and Hausmanowa-Petrusewicz, Irena and Borkowska, Janina and Zerres, Klaus}, year={2001}, pages={174-181} }

@article{Russman_Buncher_White_Samaha_Iannaccone_1996, title={Function changes in spinal muscular atrophy II and III}, volume={47}, DOI={10.1212/WNL.47.4.973}, number={4}, journal={Neurology}, author={Russman, B. S. and Buncher, C. R. and White, M. and Samaha, F. J. and Iannaccone, S. T.}, year={1996}, month={Oct}, pages={973-976} }

@article{Schröder_Schoser_2009, title={Myofibrillar Myopathies: A Clinical and Myopathological Guide}, volume={19}, DOI={10.1111/j.1750-3639.2009.00289.x}, number={3}, journal={Brain Pathology}, author={Schröder, Rolf and Schoser, Benedikt}, year={2009}, month={Jul}, pages={483-492} }

@article{Shavlakadze_Grounds_2006, title={Of bears, frogs, meat, mice and men: complexity of factors affecting skeletal muscle mass and fat}, volume={28}, DOI={10.1002/bies.20479}, number={10}, journal={BioEssays}, author={Shavlakadze, Thea and Grounds, Miranda}, year={2006}, month={Oct}, pages={994-1009} }

@article{Sveen_Andersen_Ingelsrud_Blichter_Olsen_Jønck_Krag_Vissing_2013, title={Resistance training in patients with limb-girdle and becker muscular dystrophies}, volume={47}, DOI={10.1002/mus.23491}, number={2}, journal={Muscle & Nerve}, author={Sveen, Marie-Louise and Andersen, Søren P. and Ingelsrud, Lina H. and Blichter, Sarah and Olsen, Niels E. and Jønck, Simon and Krag, Thomas O. and Vissing, John}, year={2013}, month={Feb}, pages={163-169} }

@article{Thomas Wieser_2017, title={Carnitine Palmitoyltransferase II Deficiency}, url={https://www.ncbi.nlm.nih.gov/books/NBK1253/}, journal={Gene Reviews}, publisher={University of Washington, Seattle}, author={Thomas Wieser}, year={2017} }

@article{Velloso_2008, title={Regulation of muscle mass by growth hormone and IGF-I}, volume={154}, DOI={10.1038/bjp.2008.153}, number={3}, journal={British Journal of Pharmacology}, author={Velloso, C P}, year={2008}, month={Jun}, pages={557-568} }

@article{Voet_van der Kooi_Riphagen_Lindeman_van Engelen_Geurts_2013, title={Strength training and aerobic exercise training for muscle disease}, DOI={10.1002/14651858.CD003907.pub4}, journal={Cochrane Database of Systematic Reviews}, author={Voet, Nicoline BM and van der Kooi, Elly L and Riphagen, Ingrid I and Lindeman, Eline and van Engelen, Baziël GM and Geurts, Alexander CH}, year={2013}, month={Jul} }

@article{Wang_Finkel_Bertini_Schroth_Simonds_Wong_Aloysius_Morrison_Main_Crawford_e t al._2007, title={Consensus Statement for Standard of Care in Spinal Muscular Atrophy}, volume={22}, DOI={10.1177/0883073807305788}, number={8}, journal={Journal of

Child Neurology}, author={Wang, Ching H. and Finkel, Richard S. and Bertini, Enrico S. and Schroth, Mary and Simonds, Anita and Wong, Brenda and Aloysius, Annie and Morrison, Leslie and Main, Marion and Crawford, Thomas O. and Trela, Anthony}, year={2007}, month={Aug}, pages={1027-1049} }

@book{Wattjes_Fischer_2013, address={New York}, title={Neuromuscular imaging}, url={http://ucl.alma.exlibrisgroup.com/view/action/uresolver.do?operation=resolveService&package_service_id=3320980190004761&institutionId=4761&customerId=4760}, publisher={Springer}, author={Wattjes, Mike P. and Fischer, Dirk}, year={2013} }

@article{Wiggs_2015, title={Can endurance exercise preconditioning prevention disuse muscle atrophy?}, volume={6}, DOI={10.3389/fphys.2015.00063}, journal={Frontiers in Physiology}, author={Wiggs, Michael P.}, year={2015}, month={Mar} }

@article{Willcocks_Rooney_Triplett_Forbes_Lott_Senesac_Daniels_Wang_Harrington_Tennekoon_et_al._2016, title={Multicenter prospective longitudinal study of magnetic resonance biomarkers in a large duchenne muscular dystrophy cohort}, volume={79}, DOI={10.1002/ana.24599}, number={4}, journal={Annals of Neurology}, author={Willcocks, Rebecca J. and Rooney, William D. and Triplett, William T. and Forbes, Sean C. and Lott, Donovan J. and Senesac, Claudia R. and Daniels, Michael J. and Wang, Dah-Jyuu and Harrington, Ann T. and Tennekoon, Gihan I. and Russman, Barry S. and Finanger, Erika L. and Byrne, Barry J. and Finkel, Richard S. and Walter, Glenn A. and Sweeney, H. Lee and Vandenborne, Krista}, year={2016}, month={Apr}, pages={535-547} }

@article{Zammit_Golding_Nagata_Hudon_Partridge_Beauchamp_2004, title={Muscle satellite cells adopt divergent fates}, volume={166}, DOI={10.1083/jcb.200312007}, number={3}, journal={The Journal of Cell Biology}, author={Zammit, Peter S. and Golding, Jon P. and Nagata, Yosuke and Hudon, Valérie and Partridge, Terence A. and Beauchamp, Jonathan R.}, year={2004}, month={Aug}, pages={347-357} }

@article{An information leaflet for surgical patients._1983, volume={65}, url={https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2494353/}, number={4}, journal={Annals of The Royal College of Surgeons of England}, publisher={Royal College of Surgeons of England}, year={1983} }

@article{Prescription information leaflets: a pilot study in general practice._1983, volume={287}, url={https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1549423/}, number={6400}, journal={British Medical Journal (Clinical research ed.)}, publisher={BMJ Publishing Group}, year={1983} }

@article{Force-velocity properties of human skeletal muscle fibres: myosin heavy chain isoform and temperature dependence._1996, volume={495}, url={https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1160815/}, number={Pt 2}, journal={The Journal of Physiology}, publisher={Wiley-Blackwell}, year={1996} }

@book{Saudubray_Baumgartner_Walter_2016, address={Berlin}, edition={6th edition}, title={Inborn metabolic diseases: diagnosis and treatment}, publisher={Springer}, year={2016} }

```
@book{Hollak_Lachmann_2016a, title={Inherited Metabolic Disease in Adults},  
volume={1},  
url={http://www.oxfordmedicine.com/view/10.1093/med/9780199972135.001.0001/med-9  
780199972135}, DOI={10.1093/med/9780199972135.001.0001}, publisher={Oxford  
University Press}, year={2016}, month={Sep} }
```

```
@book{Hollak_Lachmann_2016b, title={Inherited Metabolic Disease in Adults},  
volume={1},  
url={http://www.oxfordmedicine.com/view/10.1093/med/9780199972135.001.0001/med-9  
780199972135}, DOI={10.1093/med/9780199972135.001.0001}, publisher={Oxford  
University Press}, year={2016}, month={Sep} }
```

```
@book{Hoffmann_Zschocke_Nyhan_2017, address={Berlin}, edition={Second edition},  
title={Inherited metabolic diseases: a clinical approach}, publisher={Springer},  
year={2017} }
```

```
@misc{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,  
url={https://www.minervamedica.it/en/journals/europa-medicophysica/article.php?cod=R3  
3Y2013N02A0169} }
```

```
@misc{Neuromuscular Disease Center, url={https://neuromuscular.wustl.edu/} }
```

```
@article{Neuropathology and Applied Neurobiology, volume={Volume 43, Issue 1},  
url={https://onlinelibrary.wiley.com/toc/13652990/2017/43/1} }
```