

CLNE0020: Motoneurons, Neuromuscular Junctions and Associated Disease

View Online



Al-Chalabi A, van den Berg LH and Veldink J, 'Gene Discovery in Amyotrophic Lateral Sclerosis: Implications for Clinical Management' (2017) 13 Nature Reviews Neurology 96

Andreasson U, Blennow K and Zetterberg H, 'Update on Ultrasensitive Technologies to Facilitate Research on Blood Biomarkers for Central Nervous System Disorders' (2016) 3 Alzheimer's & Dementia: Diagnosis, Assessment & Disease Monitoring 98

Badders NM and others, 'Selective Modulation of the Androgen Receptor AF2 Domain Rescues Degeneration in Spinal Bulbar Muscular Atrophy' (2018) 24 Nature Medicine 427
Beitel LK and others, 'Mechanisms Mediating Spinal and Bulbar Muscular Atrophy: Investigations into Polyglutamine-Expanded Androgen Receptor Function and Dysfunction' (2013) 4 Frontiers in Neurology

Belaya K and others, 'Mutations in [\[redacted\]](#) Cause Congenital Myasthenic Syndrome and Bridge Myasthenic Disorders with Dystroglycanopathies' (2015) 138 Brain 2493

Benatar M and others, 'ALS Biomarkers for Therapy Development: State of the Field and Future Directions' (2016) 53 Muscle & Nerve 169

Berlowitz DJ and others, 'Identifying Who Will Benefit from Non-Invasive Ventilation in Amyotrophic Lateral Sclerosis/Motor Neurone Disease in a Clinical Cohort' (2016) 87 Journal of Neurology, Neurosurgery & Psychiatry 280

Birnkrant DJ and others, 'Diagnosis and Management of Duchenne Muscular Dystrophy, Part 1: Diagnosis, and Neuromuscular, Rehabilitation, Endocrine, and Gastrointestinal and Nutritional Management' (2018) 17 The Lancet Neurology 251

—, 'Diagnosis and Management of Duchenne Muscular Dystrophy, Part 2: Respiratory, Cardiac, Bone Health, and Orthopaedic Management' (2018) 17 The Lancet Neurology 347

—, 'Diagnosis and Management of Duchenne Muscular Dystrophy, Part 3: Primary Care, Emergency Management, Psychosocial Care, and Transitions of Care across the Lifespan' (2018) 17 The Lancet Neurology 445

Bonanomi D and Pfaff SL, 'Motor Axon Pathfinding' (2010) 2 Cold Spring Harbor Perspectives in Biology a001735

Brownstone RM and Bui TV, 'Spinal Interneurons Providing Input to the Final Common Path

during Locomotion', *Breathe, Walk and Chew: The Neural Challenge: Part I*, vol 187 (Elsevier 2010) <<https://linkinghub.elsevier.com/retrieve/pii/B978044453613600006X>>

Carrì MT, D'Ambrosi N and Cozzolino M, 'Pathways to Mitochondrial Dysfunction in ALS Pathogenesis' (2017) 483 *Biochemical and Biophysical Research Communications* 1187

Cortes CJ and others, 'Muscle Expression of Mutant Androgen Receptor Accounts for Systemic and Motor Neuron Disease Phenotypes in Spinal and Bulbar Muscular Atrophy' (2014) 82 *Neuron* 295

Couratier P and others, 'Epidemiology of Amyotrophic Lateral Sclerosis: A Review of Literature' (2016) 172 *Revue Neurologique* 37

Crisp SJ, Kullmann DM and Vincent A, 'Autoimmune Synaptopathies' (2016) 17 *Nature Reviews Neuroscience* 103

—, 'Autoimmune Synaptopathies' (2016) 17 *Nature Reviews Neuroscience* 103

Cruz PMR, Palace J and Beeson D, 'Congenital Myasthenic Syndromes and the Neuromuscular Junction' (2014) 27 *Current Opinion in Neurology* 566

Darabid H, Perez-Gonzalez AP and Robitaille R, 'Neuromuscular Synaptogenesis: Coordinating Partners with Multiple Functions' (2014) 15 *Nature Reviews Neuroscience* 703

Dasen JS and Jessell TM, 'Chapter Six Hox Networks and the Origins of Motor Neuron Diversity', *Hox Genes*, vol 88 (Elsevier 2009)
<<https://linkinghub.elsevier.com/retrieve/pii/S007021530988006X>>

Drory VE and others, 'The Value of Muscle Exercise in Patients with Amyotrophic Lateral Sclerosis' (2001) 191 *Journal of the Neurological Sciences* 133

Engel AG and others, 'Congenital Myasthenic Syndromes: Pathogenesis, Diagnosis, and Treatment' (2015) 14 *The Lancet Neurology* 420

Finkel RS and others, 'Diagnosis and Management of Spinal Muscular Atrophy: Part 2: Pulmonary and Acute Care; Medications, Supplements and Immunizations; Other Organ Systems; and Ethics' (2018) 28 *Neuromuscular Disorders* 197

Fratta P and others, 'Correlation of Clinical and Molecular Features in Spinal Bulbar Muscular Atrophy' (2014) 82 *Neurology* 2077

Fuller G and Manford M, *Neurology: An Illustrated Colour Text* (3rd ed, Churchill Livingstone Elsevier 2010)
<http://ucl.alma.exlibrisgroup.com/view/action/uresolver.do?operation=resolveService&package_service_id=3669595080004761&institutionId=4761&customerId=4760>

Gendron TF and others, 'Poly(GP) Proteins Are a Useful Pharmacodynamic Marker for -Associated Amyotrophic Lateral Sclerosis' (2017) 9 *Science Translational Medicine*

- Gibson S and Haringer V, 'Amyotrophic Lateral Sclerosis: Clinical Perspectives' [2015] Orphan Drugs: Research and Reviews
- Gilhus NE, 'Myasthenia Gravis' (2016) 375 New England Journal of Medicine 2570
- Gordon E, Rohrer JD and Fox NC, 'Advances in Neuroimaging in Frontotemporal Dementia' (2016) 138 Journal of Neurochemistry 193
- Harland R, 'Neural Induction' (2000) 10 Current Opinion in Genetics & Development 357
- Harwood CA, McDermott CJ and Shaw PJ, 'Clinical Aspects of Motor Neurone Disease' (2012) 40 Medicine 540
- Hughes J and others, 'Principles of Early Drug Discovery' (2011) 162 British Journal of Pharmacology 1239
- Jacobson L and others, 'Plasma from Human Mothers of Fetuses with Severe Arthrogryposis Multiplex Congenita Causes Deformities in Mice' (1999) 103 Journal of Clinical Investigation 1031
- Jessell TM, 'Neuronal Specification in the Spinal Cord: Inductive Signals and Transcriptional Codes' (2000) 1 Nature Reviews Genetics 20
- Jones RA and others, 'Cellular and Molecular Anatomy of the Human Neuromuscular Junction' (2017) 21 Cell Reports 2348
- Kanning KC, Kaplan A and Henderson CE, 'Motor Neuron Diversity in Development and Disease' (2010) 33 Annual Review of Neuroscience 409
- Koneczny I and others, 'MuSK Myasthenia Gravis IgG4 Disrupts the Interaction of LRP4 with MuSK but Both IgG4 and IgG1-3 Can Disperse Preformed Agrin-Independent AChR Clusters' (2013) 8 PLoS ONE
- Koneczny I, Cossins J and Vincent A, 'The Role of Muscle-Specific Tyrosine Kinase (MuSK) and Mystery of MuSK Myasthenia Gravis' (2014) 224 Journal of Anatomy 29
- Kusner LL and Kaminski HJ, 'Myasthenia Gravis', Neurobiology of Brain Disorders (Elsevier 2015) <<https://linkinghub.elsevier.com/retrieve/pii/B9780123982704000100>>
- Ladle DR, Pecho-Vrieseling E and Arber S, 'Assembly of Motor Circuits in the Spinal Cord: Driven to Function by Genetic and Experience-Dependent Mechanisms' (2007) 56 Neuron 270
- Laurá M and others, 'Prevalence and Orthopedic Management of Foot and Ankle Deformities in Charcot-Marie-Tooth Disease' (2018) 57 Muscle & Nerve 255
- Leung DG, Other Proven and Putative Autoimmune Disorders of the Peripheral Nervous System, vol 1 (Oxford University Press 2017) <<http://oxfordmedicine.com/view/10.1093/med/9780199937837.001.0001/med-9780199937837-chapter-98>>

Li L, Xiong W-C and Mei L, 'Neuromuscular Junction Formation, Aging, and Disorders' (2018) 80 Annual Review of Physiology 159

Lieberman AP and others, 'Peripheral Androgen Receptor Gene Suppression Rescues Disease in Mouse Models of Spinal and Bulbar Muscular Atrophy' (2014) 7 Cell Reports 774

Lin G, Mao D and Bellen HJ, '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, vol 121 (Elsevier 2017) <<https://linkinghub.elsevier.com/retrieve/pii/S0070215316301454>>

Lu C-H and others, 'Neurofilament Light Chain: A Prognostic Biomarker in Amyotrophic Lateral Sclerosis' (2015) 84 Neurology 2247

Malik B and others, 'Absence of Disturbed Axonal Transport in Spinal and Bulbar Muscular Atrophy' (2011) 20 Human Molecular Genetics 1776

Malik B and others, 'Co-Induction of the Heat Shock Response Ameliorates Disease Progression in a Mouse Model of Human Spinal and Bulbar Muscular Atrophy: Implications for Therapy' (2013) 136 Brain 926

Manzano R and others, 'Beyond Motor Neurons: Expanding the Clinical Spectrum in Kennedy's Disease' (2018) 89 Journal of Neurology, Neurosurgery & Psychiatry 808

Maragakis NJ, 'What Can We Learn from the Edaravone Development Program for ALS?' (2017) 18 Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration 98

Mazzone ES and others, 'Revised Upper Limb Module for Spinal Muscular Atrophy: Development of a New Module' (2017) 55 Muscle & Nerve 869

Mercuri E and others, 'Diagnosis and Management of Spinal Muscular Atrophy: Part 1: Recommendations for Diagnosis, Rehabilitation, Orthopedic and Nutritional Care' (2018) 28 Neuromuscular Disorders 103

Meriggioli MN and Sanders DB, 'Autoimmune Myasthenia Gravis: Emerging Clinical and Biological Heterogeneity' (2009) 8 The Lancet Neurology 475

Milioto C and others, 'Beta-Agonist Stimulation Ameliorates the Phenotype of Spinal and Bulbar Muscular Atrophy Mice and Patient-Derived Myotubes' (2017) 7 Scientific Reports

Mitsumoto H, Brooks BR and Silani V, 'Clinical Trials in Amyotrophic Lateral Sclerosis: Why so Many Negative Trials and How Can Trials Be Improved?' (2014) 13 The Lancet Neurology 1127

Monahan Z, Shewmaker F and Pandey UB, 'Stress Granules at the Intersection of Autophagy and ALS' (2016) 1649 Brain Research 189

Morgan S and Orrell RW, 'Pathogenesis of Amyotrophic Lateral Sclerosis' (2016) 119 British Medical Bulletin 87

- Morren JA and Galvez-Jimenez N, 'Current and Prospective Disease-Modifying Therapies for Amyotrophic Lateral Sclerosis' (2012) 21 Expert Opinion on Investigational Drugs 297
- 'Motor Neurone Disease: Assessment and Management | Guidance and Guidelines | NICE' <<https://www.nice.org.uk/guidance/ng42>>
- Nishimune H and others, 'Laminins Promote Postsynaptic Maturation by an Autocrine Mechanism at the Neuromuscular Junction' (2008) 182 The Journal of Cell Biology 1201
- O'Connor E and others, 'Clinical and Research Strategies for Limb-Girdle Congenital Myasthenic Syndromes' (2018) 1412 Annals of the New York Academy of Sciences 102
- Orrell, Richard WBarclay, Chris, 'Diagnosis and Management of Motor Neurone Disease' 260 Practitioner 17
<<https://search.proquest.com/docview/1844334383/64C39DCAF3D346C0PQ/1?accountid=14511>>
- Otto M and others, 'Roadmap and Standard Operating Procedures for Biobanking and Discovery of Neurochemical Markers in ALS' (2012) 13 Amyotrophic Lateral Sclerosis 1
- Peragallo JH, 'Pediatric Myasthenia Gravis' (2017) 24 Seminars in Pediatric Neurology 116
- Ramdharry GM and others, 'A Pilot Study of Proximal Strength Training in Charcot-Marie-Tooth Disease' (2014) 19 Journal of the Peripheral Nervous System 328
- Ramsey D and others, 'Revised Hammersmith Scale for Spinal Muscular Atrophy: A SMA Specific Clinical Outcome Assessment Tool' (2017) 12 PLOS ONE
- Reilly MM and others, '221st ENMC International Workshop': (2017) 27 Neuromuscular Disorders 1138
- Renton AE, Chiò A and Traynor BJ, 'State of Play in Amyotrophic Lateral Sclerosis Genetics' (2014) 17 Nature Neuroscience 17
- Rodríguez Cruz PM and others, 'Congenital Myopathies with Secondary Neuromuscular Transmission Defects; A Case Report and Review of the Literature' (2014) 24 Neuromuscular Disorders 1103
- Rodríguez Cruz PM, Palace J and Beeson D, 'Inherited Disorders of the Neuromuscular Junction: An Update' (2014) 261 Journal of Neurology 2234
- Rudolf R and others, 'Degeneration of Neuromuscular Junction in Age and Dystrophy' (2014) 6 Frontiers in Aging Neuroscience
- Ruegsegger C and Saxena S, 'Proteostasis Impairment in ALS' (2016) 1648 Brain Research 571
- Scoto M and others, 'Therapeutic Approaches for Spinal Muscular Atrophy (SMA)' (2017) 24 Gene Therapy 514

Singhal N and Martin PT, 'Role of Extracellular Matrix Proteins and Their Receptors in the Development of the Vertebrate Neuromuscular Junction' (2011) 71 *Developmental Neurobiology* 982

Spillane J and others, 'Lambert-Eaton Syndrome IgG Inhibits Transmitter Release via P/Q Ca²⁺ Channels' (2015) 84 *Neurology* 575

Spillane J, Beeson DJ and Kullmann DM, 'Myasthenia and Related Disorders of the Neuromuscular Junction' (2010) 81 *Journal of Neurology, Neurosurgery & Psychiatry* 850

Viegas S and others, 'Passive and Active Immunization Models of MuSK-Ab Positive Myasthenia: Electrophysiological Evidence for Pre and Postsynaptic Defects' (2012) 234 *Experimental Neurology* 506

Vincent A, 'Unravelling the Pathogenesis of Myasthenia Gravis' (2002) 2 *Nature Reviews Immunology* 797

'Volume 58, Issue 3, March 2016' Volume 58, Issue 3, March 2016
<<https://link.springer.com/journal/12031/58/3>>

Westerberg E and others, 'The Impact of Physical Exercise on Neuromuscular Function in Myasthenia Gravis Patients' (2018) 97 *Medicine*

Wolfe GI and others, 'Randomized Trial of Thymectomy in Myasthenia Gravis' (2016) 375 *New England Journal of Medicine* 511

Woollacott IOC and Rohrer JD, 'The Clinical Spectrum of Sporadic and Familial Forms of Frontotemporal Dementia' (2016) 138 *Journal of Neurochemistry* 6