CLNE0020: Motoneurons, Neuromuscular Junctions and Associated Disease

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1

Morren JA, Galvez-Jimenez N. Current and prospective disease-modifying therapies for amyotrophic lateral sclerosis. Expert Opinion on Investigational Drugs 2012;**21**:297–320. doi:10.1517/13543784.2012.657303

2

Mitsumoto H, Brooks BR, Silani V. Clinical trials in amyotrophic lateral sclerosis: why so many negative trials and how can trials be improved? The Lancet Neurology 2014;**13**:1127–38. doi:10.1016/S1474-4422(14)70129-2

3

Maragakis NJ. What can we learn from the edaravone development program for ALS? Amyotrophic Lateral Sclerosis and Frontotemporal Degeneration 2017;**18**:98–103. doi:10.1080/21678421.2017.1361446

4

Hughes J, Rees S, Kalindjian S, et al. Principles of early drug discovery. British Journal of Pharmacology 2011;**162**:1239–49. doi:10.1111/j.1476-5381.2010.01127.x

5

Birnkrant DJ, Bushby K, Bann CM, et al. Diagnosis and management of Duchenne muscular dystrophy, part 1: diagnosis, and neuromuscular, rehabilitation, endocrine, and gastrointestinal and nutritional management. The Lancet Neurology 2018;**17**:251–67. doi:10.1016/S1474-4422(18)30024-3

6

Birnkrant DJ, Bushby K, Bann CM, et al. Diagnosis and management of Duchenne muscular dystrophy, part 2: respiratory, cardiac, bone health, and orthopaedic management. The Lancet Neurology 2018;**17**:347–61. doi:10.1016/S1474-4422(18)30025-5

7

Birnkrant DJ, Bushby K, Bann CM, et al. Diagnosis and management of Duchenne muscular dystrophy, part 3: primary care, emergency management, psychosocial care, and transitions of care across the lifespan. The Lancet Neurology 2018;**17**:445–55. doi:10.1016/S1474-4422(18)30026-7

8

Mercuri E, Finkel RS, Muntoni F, et al. Diagnosis and management of spinal muscular atrophy: Part 1: Recommendations for diagnosis, rehabilitation, orthopedic and nutritional care. Neuromuscular Disorders 2018;**28**:103–15. doi:10.1016/j.nmd.2017.11.005

9

Finkel RS, Mercuri E, Meyer OH, et al. Diagnosis and management of spinal muscular atrophy: Part 2: Pulmonary and acute care; medications, supplements and immunizations; other organ systems; and ethics. Neuromuscular Disorders 2018;**28**:197–207. doi:10.1016/j.nmd.2017.11.004

10

Scoto M, Finkel RS, Mercuri E, et al. Therapeutic approaches for spinal muscular atrophy (SMA). Gene Therapy 2017;**24**:514–9. doi:10.1038/gt.2017.45

11

Ramsey D, Scoto M, Mayhew A, et al. Revised Hammersmith Scale for spinal muscular atrophy: A SMA specific clinical outcome assessment tool. PLOS ONE 2017;**12**. doi:10.1371/journal.pone.0172346

12

Mazzone ES, Mayhew A, Montes J, et al. Revised upper limb module for spinal muscular atrophy: Development of a new module. Muscle & Nerve 2017;**55**:869–74. doi:10.1002/mus.25430

13

Westerberg E, Molin CJ, Spörndly Nees S, et al. The impact of physical exercise on neuromuscular function in Myasthenia gravis patients. Medicine 2018;**97**. doi:10.1097/MD.0000000000011510

14

Peragallo JH. Pediatric Myasthenia Gravis. Seminars in Pediatric Neurology 2017;**24**:116–21. doi:10.1016/j.spen.2017.04.003

15

Laurá M, Singh D, Ramdharry G, et al. Prevalence and orthopedic management of foot and ankle deformities in Charcot-Marie-Tooth disease. Muscle & Nerve 2018;**57**:255–9. doi:10.1002/mus.25724

16

Reilly MM, Pareyson D, Burns J, et al. 221st ENMC International Workshop: Neuromuscular Disorders 2017;**27**:1138–42. doi:10.1016/j.nmd.2017.09.005

17

Ramdharry GM, Pollard A, Anderson C, et al. A pilot study of proximal strength training in Charcot-Marie-Tooth disease. Journal of the Peripheral Nervous System 2014;**19**:328–32. doi:10.1111/jns.12100

18

Gibson S, Haringer V. Amyotrophic lateral sclerosis: clinical perspectives. Orphan Drugs: Research and Reviews Published Online First: April 2015. doi:10.2147/ODRR.S63585

19

Berlowitz DJ, Howard ME, Fiore JF, et al. Identifying who will benefit from non-invasive ventilation in amyotrophic lateral sclerosis/motor neurone disease in a clinical cohort. Journal of Neurology, Neurosurgery & Psychiatry 2016;**87**:280–6. doi:10.1136/jnnp-2014-310055

20

Harwood CA, McDermott CJ, Shaw PJ. Clinical aspects of motor neurone disease. Medicine 2012;**40**:540–5. doi:10.1016/j.mpmed.2012.07.003

21

Drory VE, Goltsman E, Goldman Reznik J, et al. The value of muscle exercise in patients with amyotrophic lateral sclerosis. Journal of the Neurological Sciences 2001;**191**:133–7. doi:10.1016/S0022-510X(01)00610-4

22

Al-Chalabi A, van den Berg LH, Veldink J. Gene discovery in amyotrophic lateral sclerosis: implications for clinical management. Nature Reviews Neurology 2017;**13**:96–104. doi:10.1038/nrneurol.2016.182

23

Carrì MT, D'Ambrosi N, Cozzolino M. Pathways to mitochondrial dysfunction in ALS pathogenesis. Biochemical and Biophysical Research Communications 2017;**483**:1187–93. doi:10.1016/j.bbrc.2016.07.055

24

Lin G, Mao D, Bellen HJ. Amyotrophic Lateral Sclerosis Pathogenesis Converges on Defects in Protein Homeostasis Associated with TDP-43 Mislocalization and Proteasome-Mediated Degradation Overload. In: Fly Models of Human Diseases. Elsevier 2017. 111–71. doi:10.1016/bs.ctdb.2016.07.004

25

Monahan Z, Shewmaker F, Pandey UB. Stress granules at the intersection of autophagy and ALS. Brain Research 2016;**1649**:189–200. doi:10.1016/j.brainres.2016.05.022

26

Ruegsegger C, Saxena S. Proteostasis impairment in ALS. Brain Research 2016;**1648**:571–9. doi:10.1016/j.brainres.2016.03.032

27

Renton AE, Chiò A, Traynor BJ. State of play in amyotrophic lateral sclerosis genetics. Nature Neuroscience 2014;**17**:17–23. doi:10.1038/nn.3584

28

Jessell TM. Neuronal specification in the spinal cord: inductive signals and transcriptional codes. Nature Reviews Genetics 2000;**1**:20–9. doi:10.1038/35049541

29

Harland R. Neural induction. Current Opinion in Genetics & Development 2000;**10**:357–62. doi:10.1016/S0959-437X(00)00096-4

30

Dasen JS, Jessell TM. Chapter Six Hox Networks and the Origins of Motor Neuron Diversity. In: Hox Genes. Elsevier 2009. 169–200. doi:10.1016/S0070-2153(09)88006-X

31

Bonanomi D, Pfaff SL. Motor Axon Pathfinding. Cold Spring Harbor Perspectives in Biology 2010;**2**:a001735–a001735. doi:10.1101/cshperspect.a001735

32

Darabid H, Perez-Gonzalez AP, Robitaille R. Neuromuscular synaptogenesis: coordinating partners with multiple functions. Nature Reviews Neuroscience 2014;**15**:703–18. doi:10.1038/nrn3821

33

Kanning KC, Kaplan A, Henderson CE. Motor Neuron Diversity in Development and Disease. Annual Review of Neuroscience 2010;**33**:409–40. doi:10.1146/annurev.neuro.051508.135722

34

Ladle DR, Pecho-Vrieseling E, Arber S. Assembly of Motor Circuits in the Spinal Cord: Driven to Function by Genetic and Experience-Dependent Mechanisms. Neuron 2007;**56**:270–83. doi:10.1016/j.neuron.2007.09.026

35

Brownstone RM, Bui TV. Spinal interneurons providing input to the final common path during locomotion. In: Breathe, Walk and Chew: The Neural Challenge: Part I. Elsevier 2010. 81–95. doi:10.1016/B978-0-444-53613-6.00006-X

36

Li L, Xiong W-C, Mei L. Neuromuscular Junction Formation, Aging, and Disorders. Annual Review of Physiology 2018;**80**:159–88. doi:10.1146/annurev-physiol-022516-034255

37

Singhal N, Martin PT. Role of extracellular matrix proteins and their receptors in the development of the vertebrate neuromuscular junction. Developmental Neurobiology 2011;**71**:982–1005. doi:10.1002/dneu.20953

38

Nishimune H, Valdez G, Jarad G, et al. Laminins promote postsynaptic maturation by an autocrine mechanism at the neuromuscular junction. The Journal of Cell Biology 2008;**182**:1201–15. doi:10.1083/jcb.200805095

39

Rudolf R, Khan MM, Labeit S, et al. Degeneration of Neuromuscular Junction in Age and Dystrophy. Frontiers in Aging Neuroscience 2014;**6**. doi:10.3389/fnagi.2014.00099

40

Jones RA, Harrison C, Eaton SL, et al. Cellular and Molecular Anatomy of the Human Neuromuscular Junction. Cell Reports 2017;**21**:2348–56. doi:10.1016/j.celrep.2017.11.008

41

O'Connor E, Töpf A, Zahedi RP, et al. Clinical and research strategies for limb-girdle congenital myasthenic syndromes. Annals of the New York Academy of Sciences 2018; **1412**:102–12. doi:10.1111/nyas.13520

42

Engel AG, Shen X-M, Selcen D, et al. Congenital myasthenic syndromes: pathogenesis, diagnosis, and treatment. The Lancet Neurology 2015;**14**:420–34. doi:10.1016/S1474-4422(14)70201-7

43

Cruz PMR, Palace J, Beeson D. Congenital myasthenic syndromes and the neuromuscular junction. Current Opinion in Neurology 2014;**27**:566–75. doi:10.1097/WCO.0000000000000134

44

Rodríguez Cruz PM, Palace J, Beeson D. Inherited disorders of the neuromuscular junction: an update. Journal of Neurology 2014;**261**:2234–43. doi:10.1007/s00415-014-7520-7

45

Belaya K, Rodríguez Cruz PM, Liu WW, et al. Mutations in cause

congenital myasthenic syndrome and bridge myasthenic disorders with dystroglycanopathies. Brain 2015;**138**:2493–504. doi:10.1093/brain/awv185

46

Rodríguez Cruz PM, Sewry C, Beeson D, et al. Congenital myopathies with secondary neuromuscular transmission defects; A case report and review of the literature. Neuromuscular Disorders 2014;**24**:1103–10. doi:10.1016/j.nmd.2014.07.005

47

Crisp SJ, Kullmann DM, Vincent A. Autoimmune synaptopathies. Nature Reviews Neuroscience 2016;**17**:103–17. doi:10.1038/nrn.2015.27

48

Gilhus NE. Myasthenia Gravis. New England Journal of Medicine 2016;**375**:2570–81. doi:10.1056/NEJMra1602678

49

Kusner LL, Kaminski HJ. Myasthenia Gravis. In: Neurobiology of Brain Disorders. Elsevier 2015. 135–50. doi:10.1016/B978-0-12-398270-4.00010-0

50

Leung DG. Other Proven and Putative Autoimmune Disorders of the Peripheral Nervous System. Oxford University Press 2017. doi:10.1093/med/9780199937837.003.0098

51

Spillane J, Beeson DJ, Kullmann DM. Myasthenia and related disorders of the neuromuscular junction. Journal of Neurology, Neurosurgery & Psychiatry 2010;**81**:850–7. doi:10.1136/jnnp.2008.169367

52

Meriggioli MN, Sanders DB. Autoimmune myasthenia gravis: emerging clinical and biological heterogeneity. The Lancet Neurology 2009;**8**:475–90. doi:10.1016/S1474-4422(09)70063-8

53

Spillane J, Ermolyuk Y, Cano-Jaimez M, et al. Lambert-Eaton syndrome IgG inhibits transmitter release via P/Q Ca2+ channels. Neurology 2015;**84**:575–9. doi:10.1212/WNL.0000000000001225

54

Wolfe GI, Kaminski HJ, Aban IB, et al. Randomized Trial of Thymectomy in Myasthenia Gravis. New England Journal of Medicine 2016;**375**:511–22. doi:10.1056/NEJMoa1602489

55

Orrell, Richard WBarclay, Chris. Diagnosis and management of motor neurone disease. Practitioner;**260**:17–21.https://search.proquest.com/docview/1844334383/64C39DCAF3D346C0PQ/1?accountid=14511

56

Morgan S, Orrell RW. Pathogenesis of amyotrophic lateral sclerosis. British Medical Bulletin 2016;**119**:87–98. doi:10.1093/bmb/ldw026

57

Fuller G, Manford M. Neurology: an illustrated colour text. 3rd ed. Edinburgh: : Churchill Livingstone Elsevier 2010. http://ucl.alma.exlibrisgroup.com/view/action/uresolver.do?operation=resolveService&package_service_id=3669595080004761&institutionId=4761&customerId=4760

58

Motor neurone disease: assessment and management | Guidance and guidelines | NICE. https://www.nice.org.uk/guidance/ng42

59

Couratier P, Corcia P, Lautrette G, et al. Epidemiology of amyotrophic lateral sclerosis: A review of literature. Revue Neurologique 2016;**172**:37–45. doi:10.1016/j.neurol.2015.11.002

60

Otto M, Bowser R, Turner M, et al. Roadmap and standard operating procedures for biobanking and discovery of neurochemical markers in ALS. Amyotrophic Lateral Sclerosis 2012;**13**:1–10. doi:10.3109/17482968.2011.627589

61

Lu C-H, Macdonald-Wallis C, Gray E, et al. Neurofilament light chain: A prognostic biomarker in amyotrophic lateral sclerosis. Neurology 2015;**84**:2247–57. doi:10.1212/WNL.0000000000001642

62

Benatar M, Boylan K, Jeromin A, et al. ALS biomarkers for therapy development: State of the field and future directions. Muscle & Nerve 2016;**53**:169–82. doi:10.1002/mus.24979

63

Andreasson U, Blennow K, Zetterberg H. Update on ultrasensitive technologies to facilitate research on blood biomarkers for central nervous system disorders. Alzheimer's & Dementia: Diagnosis, Assessment & Disease Monitoring 2016;**3**:98–102. doi:10.1016/j.dadm.2016.05.005

64

Gendron TF, Chew J, Stankowski JN, et al. Poly(GP) proteins are a useful pharmacodynamic marker for -associated amyotrophic lateral sclerosis. Science Translational Medicine 2017;**9**. doi:10.1126/scitranslmed.aai7866

65

Vincent A. Unravelling the pathogenesis of myasthenia gravis. Nature Reviews Immunology 2002;**2**:797–804. doi:10.1038/nri916

66

Jacobson L, Polizzi A, Morriss-Kay G, et al. Plasma from human mothers of fetuses with severe arthrogryposis multiplex congenita causes deformities in mice. Journal of Clinical Investigation 1999;**103**:1031–8. doi:10.1172/JCI5943

67

Viegas S, Jacobson L, Waters P, et al. Passive and active immunization models of MuSK-Ab positive myasthenia: Electrophysiological evidence for pre and postsynaptic defects. Experimental Neurology 2012;**234**:506–12. doi:10.1016/j.expneurol.2012.01.025

68

Koneczny I, Cossins J, Vincent A. The role of muscle-specific tyrosine kinase (MuSK) and mystery of MuSK myasthenia gravis. Journal of Anatomy 2014;**224**:29–35. doi:10.1111/joa.12034

69

Koneczny I, Cossins J, Waters P, et al. MuSK Myasthenia Gravis IgG4 Disrupts the Interaction of LRP4 with MuSK but Both IgG4 and IgG1-3 Can Disperse Preformed Agrin-Independent AChR Clusters. PLoS ONE 2013;**8**. doi:10.1371/journal.pone.0080695

70

Crisp SJ, Kullmann DM, Vincent A. Autoimmune synaptopathies. Nature Reviews Neuroscience 2016;**17**:103–17. doi:10.1038/nrn.2015.27

71

Woollacott IOC, Rohrer JD. The clinical spectrum of sporadic and familial forms of frontotemporal dementia. Journal of Neurochemistry 2016;**138**:6–31.

doi:10.1111/jnc.13654

72

Gordon E, Rohrer JD, Fox NC. Advances in neuroimaging in frontotemporal dementia. Journal of Neurochemistry 2016;**138**:193–210. doi:10.1111/jnc.13656

73

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

74

Badders NM, Korff A, Miranda HC, et al. Selective modulation of the androgen receptor AF2 domain rescues degeneration in spinal bulbar muscular atrophy. Nature Medicine 2018;**24**:427–37. doi:10.1038/nm.4500

75

Beitel LK, Alvarado C, Mokhtar S, et al. Mechanisms Mediating Spinal and Bulbar Muscular Atrophy: Investigations into Polyglutamine-Expanded Androgen Receptor Function and Dysfunction. Frontiers in Neurology 2013;**4**. doi:10.3389/fneur.2013.00053

76

Cortes CJ, Ling S-C, Guo LT, et al. Muscle Expression of Mutant Androgen Receptor Accounts for Systemic and Motor Neuron Disease Phenotypes in Spinal and Bulbar Muscular Atrophy. Neuron 2014;**82**:295–307. doi:10.1016/j.neuron.2014.03.001

77

Fratta P, Nirmalananthan N, Masset L, et al. Correlation of clinical and molecular features in spinal bulbar muscular atrophy. Neurology 2014;**82**:2077–84. doi:10.1212/WNL.0000000000000507

78

Lieberman AP, Yu Z, Murray S, et al. Peripheral Androgen Receptor Gene Suppression Rescues Disease in Mouse Models of Spinal and Bulbar Muscular Atrophy. Cell Reports 2014;**7**:774–84. doi:10.1016/j.celrep.2014.02.008

79

Malik B, Nirmalananthan N, Bilsland LG, et al. Absence of disturbed axonal transport in spinal and bulbar muscular atrophy. Human Molecular Genetics 2011;**20**:1776–86. doi:10.1093/hmg/ddr061

80

Malik B, Nirmalananthan N, Gray AL, et al. Co-induction of the heat shock response ameliorates disease progression in a mouse model of human spinal and bulbar muscular atrophy: implications for therapy. Brain 2013;**136**:926–43. doi:10.1093/brain/aws343

81

Manzano R, Sorarú G, Grunseich C, et al. Beyond motor neurons: expanding the clinical spectrum in Kennedy's disease. Journal of Neurology, Neurosurgery & Psychiatry 2018;**89**:808–12. doi:10.1136/jnnp-2017-316961

82

Milioto C, Malena A, Maino E, et al. Beta-agonist stimulation ameliorates the phenotype of spinal and bulbar muscular atrophy mice and patient-derived myotubes. Scientific Reports 2017;**7**. doi:10.1038/srep41046