MSc Neuromuscular Disease

View Online

1

D. Blottner and M. Salanova, The neuromuscular system: from earth to space life science : neuromuscular cell signalling in disuse and exercise, Springer, Cham, 2015, vol. Springer briefs in space life sciences.

2

M. K. D. Benson, Children's neuromuscular disorders, Springer, London, 2011.

3

A. Shaibani, A Video Atlas of Neuromuscular Disorders, Oxford University Press, 2014.

4

D. Hilton-Jones and M. R. Turner, Eds, Oxford textbook of neuromuscular disorders, Oxford University Press, [Oxford], 2014, vol. Oxford textbooks in clinical neurology.

5

D. Kernell, The Motoneurone and its Muscle Fibres, Oxford University Press, 2006.

6

A. A. Amato and J. A. Russell, Neuromuscular disorders, McGraw-Hill, New York, 2008.

7

K. K. Jain, Ed., Applied neurogenomics, Humana Press, New York, NY, 2015, vol. Neuromethods.

8

A. M. Davies, in Patterning and Cell Type Specification in the Developing CNS and PNS, Elsevier, 2013, pp. 303–311.

9

T. Gordon and O. A. Sulaiman, in Neuroglia, ed. H. Kettenmann, Oxford University Press, 2012, pp. 701–714.

10

M. B. Welch and C. M. Brummett, in Neuroscientific Foundations of Anesthesiology, eds G. A. Mashour and R. Lydic, Oxford University Press, 2011, pp. 133–140.

11

A. M. Rossor, P. J. Tomaselli and M. M. Reilly, Current Opinion in Neurology, DOI:10.1097/WCO.0000000000000373.

12

A. M. Rossor, M. R. B. Evans and M. M. Reilly, Practical Neurology, 2015, **15**, 187–198.

13

AM Rossor, Current opinion in neurology, DOI:10.1097/WCO.0000000000000373.

14

M. Reilly and V. Fridman, Seminars in Neurology, 2015, **35**, 407–423.

15

A. M. Rossor, M. R. B. Evans and M. M. Reilly, Practical Neurology, 2015, **15**, 187–198.

16

R. J. Pasterkamp, Nature Reviews Neuroscience, 2012, **13**, 605–618.

17

R Klein, The FASEB Journal, 1994, **8**, 738–744.

18

G. Schlosser, Developmental Biology, 2006, **294**, 303–351.

19

N. K. Thiede-Stan and M. E. Schwab, Journal of Cell Science, 2015, **128**, 2403–2414.

20

Irina Dudanova, Journal of Neuroscience, 2012, **32**, 5209–5215.

21

L. Wang, R. Klein, B. Zheng and T. Marquardt, Neuron, 2011, **71**, 263–277.

22

E. Y. Van Battum, S. Brignani and R. J. Pasterkamp, The Lancet Neurology, 2015, **14**, 532–546.

23

T.-J. Kao, C. Law and A. Kania, Seminars in Cell & Developmental Biology, 2012, **23**, 83–91.

24

Byung-Yong Park, .

25

M. Taniguchi, S. Yuasa, H. Fujisawa, I. Naruse, S. Saga, M. Mishina and T. Yagi, Neuron, 1997, **19**, 519–530.

26

A. Ebens, K. Brose, E. D. Leonardo, M. G. H. Jr, F. Bladt, C. Birchmeier, B. A. Barres and M. Tessier-Lavigne, Neuron, 1996, **17**, 1157–1172.

27

A. Caton, Development, **127**, 1751–1766.

28

J. C. Conover, J. T. Erickson, D. M. Katz, L. M. Bianchi, W. T. Poueymirou, J. McClain, L. Pan, M. Helgren, N. Y. Ip, P. Boland, B. Friedman, S. Wiegand, R. Vejsada, A. C. Kato, T. M. DeChiara and G. D. Yancopoulos, Nature, 1995, **375**, 235–238.

29

K. R. Jessen and R. Mirsky, Nature Reviews Neuroscience, 2005, **6**, 671–682.

30

K. R. Monk, M. L. Feltri and C. Taveggia, Glia, 2015, **63**, 1376–1393.

31

K. R. Jessen, R. Mirsky and A. C. Lloyd, Cold Spring Harbor Perspectives in Biology, DOI:10.1101/cshperspect.a020487.

32

J. L. Salzer, Cold Spring Harbor Perspectives in Biology, DOI:10.1101/cshperspect.a020529.

33

P. J. Arthur-Farraj, M. Latouche, D. K. Wilton, S. Quintes, E. Chabrol, A. Banerjee, A. Woodhoo, B. Jenkins, M. Rahman, M. Turmaine, G. K. Wicher, R. Mitter, L. Greensmith, A. Behrens, G. Raivich, R. Mirsky and K. R. Jessen, Neuron, 2012, **75**, 633–647.

34

A. Brosius Lutz and B. A. Barres, Developmental Cell, 2014, **28**, 7–17.

35

K. R. Jessen and R. Mirsky, The Journal of Physiology, 2016, **594**, 3521–3531.

36

M. Auer-Grumbach, in Peripheral Nerve Disorders, Elsevier, 2013, vol. 115, pp. 893–906.

37

G. L. Davidson, S. M. Murphy, J. M. Polke, M. Laura, M. A. M. Salih, F. Muntoni, J. Blake, S. Brandner, N. Davies, R. Horvath, S. Price, M. Donaghy, M. Roberts, N. Foulds, G. Ramdharry, D. Soler, M. P. Lunn, H. Manji, M. B. Davis, H. Houlden and M. M. Reilly, Journal of Neurology, 2012, **259**, 1673–1685.

38

A. M. Rossor, M. R. B. Evans and M. M. Reilly, Practical Neurology, 2015, **15**, 187–198.

39

A. Chhabra, Neuroimaging Clinics of North America, 2014, **24**, 79–89.

40

D. Purves, Neuroscience, National Library of Medicine, Bethesda, 2001.

41

Effects of axon diameter and myelination (video) | Khan Academy, https://www.khanacademy.org/science/health-and-medicine/nervous-system-and-sensory-infor/neuron-membrane-potentials-2014-03-27T17:58:17.207Z/v/effects-of-axon-diameter-and-myelination.

42

Nerve Signaling, https://www.nobelprize.org/educational/medicine/nerve_signaling/index.html.

43

J. S. Duncan, G. P. Winston, M. J. Koepp and S. Ourselin, The Lancet Neurology, 2016, **15**, 420–433.

44

M. Nowell, R. Sparks, G. Zombori, A. Miserocchi, R. Rodionov, B. Diehl, T. Wehner, M. White, S. Ourselin, A. McEvoy and J. Duncan, British Journal of Neurosurgery, 2017, **31**, 468–470.

45

V. N. Vakharia, R. Sparks, A. G. O'Keeffe, R. Rodionov, A. Miserocchi, A. McEvoy, S. Ourselin and J. Duncan, Epilepsia, 2017, **58**, 921–932.

46

A. Michell, Understanding EMG, Oxford University Press, 2013.

47

D. C. Preston and B. E. Shapiro, Electromyography and neuromuscular disorders: clinical-electrophysiologic correlations, Elsevier Saunders, London, 3rd ed., 2013.

48

Top tips for writing a lay summary | The Academy of Medical Sciences, https://acmedsci.ac.uk/more/news/10-tips-for-writing-a-lay-summary.

49

How to Write a Lay Summary | Digital Curation Centre, http://www.dcc.ac.uk/resources/how-guides/write-lay-summary.

50

Free guides, http://www.plainenglish.co.uk/free-guides.html.

51

Part two - The specifics - Access to Understanding, http://www.access2understanding.org/guidance/part-two-the-specifics/.

52

Readable | Free Readability Test Tool, https://www.webpagefx.com/tools/read-able/.

53

J. H. Rees, Journal of Neurology, Neurosurgery & Psychiatry, 2004, **75**, ii43–ii50.

54

J.-C. Antoine and J.-P. Camdessanché, La Presse Médicale, 2013, **42**, e235–e244.

55

How to Write a Lay Summary | DCC How-to Guides, http://www.dcc.ac.uk/resources/how-guides/.

56

A. M. Rossor, M. R. B. Evans and M. M. Reilly, Practical Neurology, 2015, **15**, 187–198.

57

A. M. Rossor, B. Kalmar, L. Greensmith and M. M. Reilly, Journal of Neurology, Neurosurgery & Psychiatry, 2012, **83**, 6–14.

58

A. M. Rossor, A. S. Carr, H. Devine, H. Chandrashekar, A. L. Pelayo-Negro, D. Pareyson, M. E. Shy, S. S. Scherer and M. M. Reilly, Journal of Neurology, Neurosurgery & Psychiatry, 2017, **88**, 846–863.

59

A. S. Carr, A. L. Pelayo-Negro, M. R. Evans, M. Laurà, J. Blake, C. Stancanelli, V. Iodice, A. D. Wechalekar, C. J. Whelan, J. D. Gillmore, P. N. Hawkins and M. M. Reilly, Journal of Neurology, Neurosurgery & Psychiatry, 2016, **87**, 620–627.

60

P. N. Hawkins, Y. Ando, A. Dispenzeri, A. Gonzalez-Duarte, D. Adams and O. B. Suhr, Annals of Medicine, 2015, **47**, 625–638.

61

V. Plante-Bordeneuve, A. Ferreira, T. Lalu, C. Zaros, C. Lacroix, D. Adams and G. Said,

Neurology, 2007, **69**, 693–698.

62

S. Dubrey, E. Ackermann and J. Gillmore, Postgraduate Medical Journal, 2015, **91**, 439–448.

63

M. M. Dimachkie and R. J. Barohn, Neurologic Clinics, 2013, **31**, 491–510.

64

H. J. Willison, B. C. Jacobs and P. A. van Doorn, The Lancet, 2016, **388**, 717–727.

65

M. P. Collins and R. D. Hadden, Nature Reviews Neurology, 2017, **13**, 302–316.

66

M. Yates, R. A. Watts, I. M. Bajema, M. C. Cid, B. Crestani, T. Hauser, B. Hellmich, J. U. Holle, M. Laudien, M. A. Little, R. A. Luqmani, A. Mahr, P. A. Merkel, J. Mills, J. Mooney, M. Segelmark, V. Tesar, K. Westman, A. Vaglio, N. Yalçındağ, D. R. Jayne and C. Mukhtyar, Annals of the Rheumatic Diseases, 2016, **75**, 1583–1594.

67

M. P. Collins, P. J. B. Dyck, G. S. Gronseth, L. Guillevin, R. D. M. Hadden, D. Heuss, J.-M. Léger, N. C. Notermans, J. D. Pollard, G. Said, G. Sobue, A. F. J. E. Vrancken and J. T. Kissel, Journal of the Peripheral Nervous System, 2010, **15**, 176–184.

68

M. P. Berthelsen, E. Husu, S. B. Christensen, K. P. Prahm, J. Vissing and B. R. Jensen, Neuromuscular Disorders, 2014, **24**, 492–498.

69

E. H. Cup, A. J. Pieterse, J. M. ten Broek-Pastoor, M. Munneke, B. G. van Engelen, H. T. Hendricks, G. J. van der Wilt and R. A. Oostendorp, Archives of Physical Medicine and Rehabilitation, 2007, **88**, 1452–1464.

70

71

M.-L. Sveen, S. P. Andersen, L. H. Ingelsrud, S. Blichter, N. E. Olsen, S. Jønck, T. O. Krag and J. Vissing, Muscle & Nerve, 2013, **47**, 163–169.

72

T. D. Jeppesen, M. Schwartz, D. B. Olsen, F. Wibrand, T. Krag, M. Duno, S. Hauerslev and J. Vissing, Brain, 2006, **129**, 3402–3412.

73

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, https://www.minervamedica.it/en/journals/europa-medicophysica/article.php?cod=R33Y2013N02A0169.

74

Michael P. Wiggs, Frontiers in Physiology, DOI:10.3389/fphys.2015.00063.

75

Keith Baar, Sports Medicine (Auckland, N.z.), DOI:10.1007/s40279-014-0252-0.

76

D. M. Craig, S. P. Ashcroft, M. Y. Belew, B. Stocks, K. Currell, K. Baar and A. Philp, Frontiers in Physiology, DOI:10.3389/fphys.2015.00296.

77

B. Hoier and Y. Hellsten, Microcirculation, 2014, **21**, 301–314.

78

D. G. Hardie, Physiology, 2006, **21**, 48–60.

79

J. A. Hawley, M. Hargreaves, M. J. Joyner and J. R. Zierath, Cell, 2014, **159**, 738–749.

80

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, Churchill Livingstone, Edinburgh, 2004.

81

A. M. Gordon, Physiological Reviews, 2000, **80**, 853–924.

82

R Bottinelli, The Journal of Physiology.

83

S. D. R. Harridge, R. Bottinelli, M. Canepari, M. A. Pellegrino, C. Reggiani, M. Esbjörnsson and B. Saltin, Pflügers Archiv - European Journal of Physiology, 1996, **432**, 913–920.

84

By:Hunter, S (Hunter, S); White, M (White, M); Thompson, M (Thompson, M), JOURNALS OF GERONTOLOGY SERIES A-BIOLOGICAL SCIENCES AND MEDICAL SCIENCES JOURNALS OF GERONTOLOGY SERIES A-BIOLOGICAL SCIENCES AND MEDICAL SCIENCES.

85

T. D. O'Brien, N. D. Reeves, V. Baltzopoulos, D. A. Jones and C. N. Maganaris, Experimental Physiology, 2010, **95**, 202–210.

86

C. Clarke, R. Howard, M. Rossor, S. D. Shorvon, National Hospital for Neurology and Neurosurgery (London, England), and Institute of Neurology, Queen Square, Neurology: a Queen Square textbook, Wiley-Blackwell, Chichester, 2009.

87

Neuromuscular Disease Centre, http://neuromuscular.wustl.edu/.

88

Richard J. Barohn, Neurologic clinics, DOI:10.1016/j.ncl.2014.04.008.

89

M. P. Wattjes, R. A. Kley and D. Fischer, European Radiology, 2010, **20**, 2447–2460.

90

J. M. Morrow, C. D. J. Sinclair, A. Fischmann, P. M. Machado, M. M. Reilly, T. A. Yousry, J. S. Thornton and M. G. Hanna, The Lancet Neurology, 2016, **15**, 65–77.

91

D. Briggs and J. E. Morgan, FEBS Journal, 2013, **280**, 4281–4293.

92

Peter S. Zammit, The Journal of Cell Biology, DOI:10.1083/jcb.200312007.

93

L. Boldrin and J. E. Morgan, Current Opinion in Neurology, 2007, **20**, 577–582.

94

L. Boldrin, P. S. Zammit and J. E. Morgan, Stem Cell Research, 2015, **14**, 20–29.

95

J. Ross, A. Benn, J. Jonuschies, L. Boldrin, F. Muntoni, J. E. Hewitt, S. C. Brown and J. E. Morgan, STEM CELLS, 2012, **30**, 2330–2341.

96

G. Ravenscroft, M. R. Davis, P. Lamont, A. Forrest and N. G. Laing, Seminars in Cell & Developmental Biology, 2017, **64**, 160–170.

97

G. Ravenscroft, N. G. Laing and C. G. Bönnemann, Brain, 2015, **138**, 246–268.

98

K. N. North, C. H. Wang, N. Clarke, H. Jungbluth, M. Vainzof, J. J. Dowling, K. Amburgey, S. Quijano-Roy, A. H. Beggs, C. Sewry, N. G. Laing and C. G. Bönnemann, Neuromuscular Disorders, 2014, **24**, 97–116.

99

S. Quijano-Roy, R. Y. Carlier and D. Fischer, Seminars in Pediatric Neurology, 2011, **18**, 221–229.

100

Kimberly Amburgey, Neurology, DOI:10.1212/WNL.0000000000004415.

101

Irene Colombo, Neurology, DOI:10.1212/WNL.0000000000001110.

102

Guidance for Paediatric Physiotherapists - Managing Neuromuscular Disorders, http://apcp.csp.org.uk/publications/guidance-paediatric-physiotherapists-managing-neuromuscular-disorders.

103

APCP, http://apcp.csp.org.uk/.

104

Neuromuscular | Department of Neurology, https://neuro.wustl.edu/education/fellowships/neuromuscular/.

105

Muscular Dystrophy UK, http://www.musculardystrophyuk.org/.

106

C. E. M. Hollak and R. Lachmann, Eds, Inherited metabolic disease in adults: a clinical guide, Oxford University Press, [New York], 2016.

107

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

108

G. F. Hoffmann, J. Zschocke and W. L. Nyhan, Inherited metabolic diseases: a clinical

approach, Springer, Heidelberg, 2009.

109

P. Machado, S. Brady and M. G. Hanna, Current Opinion in Rheumatology, 2013, **25**, 763–771.

110

P. M. Machado, M. M. Dimachkie and R. J. Barohn, Current Opinion in Neurology, 2014, **27**, 591–598.

111

P. M. Machado, M. Ahmed, S. Brady, Q. Gang, E. Healy, J. M. Morrow, A. C. Wallace, L. Dewar, G. Ramdharry, M. Parton, J. L. Holton, H. Houlden, L. Greensmith and M. G. Hanna, Current Rheumatology Reports, DOI:10.1007/s11926-014-0477-9.

112

Mhoriam Ahmed, Science translational medicine, DOI:10.1126/scitranslmed.aad4583.

113

M. Needham and F. L. Mastaglia, Clinical Neurophysiology, 2016, **127**, 1764–1773.

114

R. Schröder and B. Schoser, Brain Pathology, 2009, **19**, 483–492.

115

A. A. Amato and S. A. Greenberg, CONTINUUM: Lifelong Learning in Neurology, 2013, **19**, 1615–1633.

116

M. Olivé, R. A. Kley and L. G. Goldfarb, Current Opinion in Neurology, 2013, **26**, 527–535.

117

P.-O. Carstens and J. Schmidt, Clinical & Experimental Immunology, 2014, **175**, 349–358.

118

M. C. Dalakas, New England Journal of Medicine, 2015, **372**, 1734–1747.

119

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.

120

T. D. Bunker, Annals of The Royal College of Surgeons of England.

121

122

M. Garner, Z. Ning and J. Francis, Health Expectations, 2012, **15**, 283–294.

123

C F George, British Medical Journal (Clinical research ed.).

124

K. G. Hollingsworth, P. L. de Sousa, V. Straub and P. G. Carlier, Neuromuscular Disorders, 2012, **22**, S54–S67.

125

M. P. Wattjes and D. Fischer, Neuromuscular imaging, Springer, New York, 2013.

126

S. C. Forbes, R. J. Willcocks, W. T. Triplett, W. D. Rooney, D. J. Lott, D.-J. Wang, J. Pollaro, C. R. Senesac, M. J. Daniels, R. S. Finkel, B. S. Russman, B. J. Byrne, E. L. Finanger, G. I. Tennekoon, G. A. Walter, H. L. Sweeney and K. Vandenborne, PLoS ONE, DOI:10.1371/journal.pone.0106435.

127

G. H. Glover and E. Schneider, Magnetic Resonance in Medicine, 1991, **18**, 371–383.

128

R. J. Willcocks, W. D. Rooney, W. T. Triplett, S. C. Forbes, D. J. Lott, C. R. Senesac, M. J. Daniels, D.-J. Wang, A. T. Harrington, G. I. Tennekoon, B. S. Russman, E. L. Finanger, B. J. Byrne, R. S. Finkel, G. A. Walter, H. L. Sweeney and K. Vandenborne, Annals of Neurology, 2016, **79**, 535–547.

129

Jean-Yves Hogrel, Neurology, DOI:10.1212/WNL.0000000000002464.

130

131

C. E. M. Hollak and R. Lachmann, Eds, Inherited metabolic disease in adults: a clinical guide, Oxford University Press, [New York], 2016.

132

Nancy D Leslie, .

133

Thomas Wieser, .

134

Miguel A Martín, .

135

Nancy Leslie, .

136

M. C. Ørngreen and J. Vissing, Current Treatment Options in Neurology, DOI:10.1007/s11940-017-0473-2.

137

S. E. Olpin, E. Murphy, R. J. Kirk, R. W. Taylor and R. Quinlivan, Journal of Clinical Pathology, 2015, **68**, 410–417.

138

F. Muntoni, S. Torelli and A. Ferlini, The Lancet Neurology, 2003, **2**, 731–740.

139

K. Bushby, R. Finkel, D. J. Birnkrant, L. E. Case, P. R. Clemens, L. Cripe, A. Kaul, K. Kinnett, C. McDonald, S. Pandya, J. Poysky, F. Shapiro, J. Tomezsko and C. Constantin, The Lancet Neurology, 2010, **9**, 77–93.

140

K. Bushby, R. Finkel, D. J. Birnkrant, L. E. Case, P. R. Clemens, L. Cripe, A. Kaul, K. Kinnett, C. McDonald, S. Pandya, J. Poysky, F. Shapiro, J. Tomezsko and C. Constantin, The Lancet

Neurology, 2010, **9**, 177–189.

141

V. Ricotti, D. A. Ridout, E. Scott, R. Quinlivan, S. A. Robb, A. Y. Manzur, F. Muntoni, A. Manzur, F. Muntoni, S. Robb, R. Quinlivan, V. Ricotti, M. Main, K. Bushby, V. Straub, A. Sarkozy, M. Guglieri, E. Strehle, M. Eagle, A. Mayhew, H. Roper, H. McMurchie, A. Childs, K. Pysden, L. Pallant, S. Spinty, G. Peachey, A. Shillington, E. Wraige, H. Jungbluth, J. Sheehan, R. Spahr, I. Hughes, E. Bateman, C. Cammiss, T. Willis, L. Groves, N. Emery, P. Baxter, M. Senior, L. Hartley, B. Parsons, A. Majumdar, L. Jenkins, K. Naismith, A. Keddie, I. Horrocks, M. Di Marco, G. Chow and A. Miah, Journal of Neurology, Neurosurgery & Psychiatry, 2013, **84**, 698–705.

142

N. M. Goemans, M. Tulinius, J. T. van den Akker, B. E. Burm, P. F. Ekhart, N. Heuvelmans, T. Holling, A. A. Janson, G. J. Platenburg, J. A. Sipkens, J. M. A. Sitsen, A. Aartsma-Rus, G.-J. B. van Ommen, G. Buyse, N. Darin, J. J. Verschuuren, G. V. Campion, S. J. de Kimpe and J. C. van Deutekom, New England Journal of Medicine, 2011, **364**, 1513–1522.

143

J. R. Mendell, L. R. Rodino-Klapac, Z. Sahenk, K. Roush, L. Bird, L. P. Lowes, L. Alfano, A. M. Gomez, S. Lewis, J. Kota, V. Malik, K. Shontz, C. M. Walker, K. M. Flanigan, M. Corridore, J. R. Kean, H. D. Allen, C. Shilling, K. R. Melia, P. Sazani, J. B. Saoud and E. M. Kaye, Annals of Neurology, 2013, **74**, 637–647.

144

J. R. Mendell, N. Goemans, L. P. Lowes, L. N. Alfano, K. Berry, J. Shao, E. M. Kaye and E. Mercuri, Annals of Neurology, 2016, **79**, 257–271.

145

M. Kinali, V. Arechavala-Gomeza, L. Feng, S. Cirak, D. Hunt, C. Adkin, M. Guglieri, E. Ashton, S. Abbs, P. Nihoyannopoulos, M. E. Garralda, M. Rutherford, C. Mcculley, L. Popplewell, I. R. Graham, G. Dickson, M. J. Wood, D. J. Wells, S. D. Wilton, R. Kole, V. Straub, K. Bushby, C. Sewry, J. E. Morgan and F. Muntoni, The Lancet Neurology, 2009, **8**, 918–928.

146

S. Cirak, V. Arechavala-Gomeza, M. Guglieri, L. Feng, S. Torelli, K. Anthony, S. Abbs, M. E. Garralda, J. Bourke, D. J. Wells, G. Dickson, M. J. Wood, S. D. Wilton, V. Straub, R. Kole, S. B. Shrewsbury, C. Sewry, J. E. Morgan, K. Bushby and F. Muntoni, The Lancet, 2011, **378**, 595–605.

147

K. Bushby, R. Finkel, B. Wong, R. Barohn, C. Campbell, G. P. Comi, A. M. Connolly, J. W. Day, K. M. Flanigan, N. Goemans, K. J. Jones, E. Mercuri, R. Quinlivan, J. B. Renfroe, B. Russman, M. M. Ryan, M. Tulinius, T. Voit, S. A. Moore, H. Lee Sweeney, R. T. Abresch, K. L. Coleman, M. Eagle, J. Florence, E. Gappmaier, A. M. Glanzman, E. Henricson, J. Barth, G. L. Elfring, A. Reha, R. J. Spiegel, M. W. O'donnell, S. W. Peltz and C. M. Mcdonald, Muscle & Nerve, 2014, **50**, 477–487.

148

Victorian Department of Health / University of Melbourne, https://www2.health.vic.gov.au/.

149

150

E. Matthews, D. Fialho, S. V. Tan, S. L. Venance, S. C. Cannon, D. Sternberg, B. Fontaine, A. A. Amato, R. J. Barohn, R. C. Griggs and M. G. Hanna, Brain, 2010, **133**, 9–22.

151

S. L. Venance, S. C. Cannon, D. Fialho, B. Fontaine, M. G. Hanna, L. J. Ptacek, M. Tristani-Firouzi, R. Tawil and R. C. Griggs, Brain, 2006, **129**, 8–17.

152

S. C. Cannon, in Comprehensive Physiology, ed. R. Terjung, John Wiley & Sons, Inc., Hoboken, NJ, USA, 2011, pp. 761–790.

153

S. V. Tan, E. Matthews, M. Barber, J. A. Burge, S. Rajakulendran, D. Fialho, R. Sud, A. Haworth, M. Koltzenburg and M. G. Hanna, Annals of Neurology, 2011, **69**, 328–340.

154

L. Sharp and J. R. Trivedi, Current Treatment Options in Neurology, DOI:10.1007/s11940-014-0313-6.

155

S. Paganoni and A. Amato, Physical Medicine and Rehabilitation Clinics of North America, 2013, **24**, 193–207.

156

A. Fuglsang-Frederiksen, Clinical Neurophysiology, 2006, **117**, 1173–1189.

157

A. Ferlini, C. Scotton and G. Novelli, Public Health Genomics, 2013, **16**, 313–321.

158

Boers, M (Boers, M); Brooks, P (Brooks, P); Strand, CV (Strand, CV); Tugwell, P (Tugwell, P), JOURNAL OF RHEUMATOLOGY JOURNAL OF RHEUMATOLOGY, 1998, **25**, 198–199.

159

R. A. Conwit, M. J. Bhanushali, J. D. Porter, P. Kaufmann and L. Gutmann, Muscle & Nerve, 2011, **44**, 695–702.

160

161

M. Ahmed, P. M. Machado, A. Miller, C. Spicer, L. Herbelin, J. He, J. Noel, Y. Wang, A. L. McVey, M. Pasnoor, P. Gallagher, J. Statland, C.-H. Lu, B. Kalmar, S. Brady, H. Sethi, G. Samandouras, M. Parton, J. L. Holton, A. Weston, L. Collinson, J. P. Taylor, G. Schiavo, M. G. Hanna, R. J. Barohn, M. M. Dimachkie and L. Greensmith, Science Translational Medicine, 2016, **8**, 331ra41-331ra41.

162

A Cruz-Martínez, Electromyography and clinical neurophysiology.

163

J. Hull, R. Aniapravan, E. Chan, M. Chatwin, J. Forton, J. Gallagher, N. Gibson, J. Gordon, I. Hughes, R. McCulloch, R. R. Russell and A. Simonds, Thorax, 2012, **67**, i1–i40.

164

165

S. Ward, Thorax, 2005, **60**, 1019–1024.

166

The Lancet Neurology, 2015, **14**, 883–892.

167

I. O. C. Woollacott and J. D. Rohrer, Journal of Neurochemistry, 2016, **138**, 6–31.

168

E. Gordon, J. D. Rohrer and N. C. Fox, Journal of Neurochemistry, 2016, **138**, 193–210.

169

L. Li, W.-C. Xiong and L. Mei, Annual Review of Physiology,

DOI:10.1146/annurev-physiol-022516-034255.

170

N. Singhal and P. T. Martin, Developmental Neurobiology, 2011, **71**, 982–1005.

171

H. Nishimune, G. Valdez, G. Jarad, C. L. Moulson, U. Müller, J. H. Miner and J. R. Sanes, The Journal of Cell Biology, 2008, **182**, 1201–1215.

172

H. Nishimune, Annals of the New York Academy of Sciences, 2012, **1274**, 24–32.

173

R. Rudolf, M. M. Khan, S. Labeit and M. R. Deschenes, Frontiers in Aging Neuroscience, DOI:10.3389/fnagi.2014.00099.

174

M. Otto, R. Bowser, M. Turner, J. Berry, J. Brettschneider, J. Connor, J. Costa, M. Cudkowicz, J. Glass, O. Jahn, S. Lehnert, A. Malaspina, L. Parnetti, A. Petzold, P. Shaw, A. Sherman, P. Steinacker, S. Süßmuth, C. Teunissen, H. Tumani, A. Wuolikainen and A. Ludolph, Amyotrophic Lateral Sclerosis, 2012, **13**, 1–10.

175

N. G. Simon, M. R. Turner, S. Vucic, A. Al-Chalabi, J. Shefner, C. Lomen-Hoerth and M. C. Kiernan, Annals of Neurology, 2014, **76**, 643–657.

176

C.-H. Lu, C. Macdonald-Wallis, E. Gray, N. Pearce, A. Petzold, N. Norgren, G. Giovannoni, P. Fratta, K. Sidle, M. Fish, R. Orrell, R. Howard, K. Talbot, L. Greensmith, J. Kuhle, M. R. Turner and A. Malaspina, Neurology, 2015, **84**, 2247–2257.

177

D. Caballero-Hernandez, M. G. Toscano, M. Cejudo-Guillen, M. L. Garcia-Martin, S. Lopez, J. M. Franco, F. J. Quintana, C. Roodveldt and D. Pozo, Trends in Molecular Medicine, 2016, **22**, 53–67.

178

Michael Benatar, Muscle & nerve, DOI:10.1002/mus.24979.

179

Ulf Andreasson, Alzheimer's & Dementia : Diagnosis, Assessment & Disease Monitoring, DOI:10.1016/j.dadm.2016.05.005.

180

J.-H. Kang, M. Korecka, M. J. Figurski, J. B. Toledo, K. Blennow, H. Zetterberg, T. Waligorska, M. Brylska, L. Fields, N. Shah, H. Soares, R. A. Dean, H. Vanderstichele, R. C. Petersen, P. S. Aisen, A. J. Saykin, M. W. Weiner, J. Q. Trojanowski and L. M. Shaw, Alzheimer's & Dementia, 2015, **11**, 772–791.

181

A. Al-Chalabi, L. H. van den Berg and J. Veldink, Nature Reviews Neurology, 2016, **13**, 96–104.

182

M. T. Carrì, N. D'Ambrosi and M. Cozzolino, Biochemical and Biophysical Research Communications, 2017, **483**, 1187–1193.

183

G. Lin, D. Mao and H. J. Bellen, in Fly Models of Human Diseases, Elsevier, 2017, vol. 121, pp. 111–171.

184

Z. Monahan, F. Shewmaker and U. B. Pandey, Brain Research, 2016, **1649**, 189–200.

185

C. Ruegsegger and S. Saxena, Brain Research, 2016, **1648**, 571–579.

186

A. E. Renton, A. Chiò and B. J. Traynor, Nature Neuroscience, 2014, **17**, 17–23.

187

T. M. Jessell, Nature Reviews Genetics, 2000, **1**, 20–29.

188

R. Harland, Current Opinion in Genetics & Development, 2000, **10**, 357–362.

189

J. S. Dasen and T. M. Jessell, in Hox Genes, Elsevier, 2009, vol. 88, pp. 169–200.

190

D. Bonanomi and S. L. Pfaff, Cold Spring Harbor Perspectives in Biology, 2010, **2**, a001735–a001735.

191

H. Darabid, A. P. Perez-Gonzalez and R. Robitaille, Nature Reviews Neuroscience, 2014, **15**, 703–718.

192

K. C. Kanning, A. Kaplan and C. E. Henderson, Annual Review of Neuroscience, 2010, **33**, 409–440.

193

D. R. Ladle, E. Pecho-Vrieseling and S. Arber, Neuron, 2007, **56**, 270–283.

194

Robert M. Brownstone, Progress in brain research, DOI:10.1016/B978-0-444-53613-6.00006-X.

195

E. O'Connor, A. Töpf, R. Zahedi, S. Spendiff, D. Cox, A. Roos and H. Lochmüller, Annals of the New York Academy of Sciences, DOI:10.1111/nyas.13520.

196

Andrew G. Engel, The Lancet. Neurology, DOI:10.1016/S1474-4422(14)70201-7.

197

P. M. R. Cruz, J. Palace and D. Beeson, Current Opinion in Neurology, 2014, **27**, 566–575.

198

P. M. Rodríguez Cruz, J. Palace and D. Beeson, Journal of Neurology, 2014, **261**, 2234–2243.

199

K. Belaya, P. M. Rodríguez Cruz, W. W. Liu, S. Maxwell, S. McGowan, M. E. Farrugia, R. Petty, T. J. Walls, M. Sedghi, K. Basiri, W. W. Yue, A. Sarkozy, M. Bertoli, M. Pitt, R. Kennett, A. Schaefer, K. Bushby, M. Parton, H. Lochmüller, J. Palace, F. Muntoni and D. Beeson, Brain , 2015, **138**, 2493–2504.

200

P. M. Rodríguez Cruz, C. Sewry, D. Beeson, S. Jayawant, W. Squier, R. McWilliam and J. Palace, Neuromuscular Disorders, 2014, **24**, 1103–1110.

201

S. J. Crisp, D. M. Kullmann and A. Vincent, Nature Reviews Neuroscience, 2016, **17**, 103–117.

202

N. E. Gilhus, New England Journal of Medicine, 2016, **375**, 2570–2581.

203

Matthew N Meriggioli, Lancet neurology, DOI:10.1016/S1474-4422(09)70063-8.

204

J. Spillane, D. J. Beeson and D. M. Kullmann, Journal of Neurology, Neurosurgery & Psychiatry, 2010, **81**, 850–857.

205

Orrell, Richard WBarclay, Chris, Practitioner, **260**, 17–21.

206

S. Morgan and R. W. Orrell, British Medical Bulletin, 2016, **119**, 87–98.

207

G. Fuller and M. Manford, Neurology: an illustrated colour text, Churchill Livingstone, Edinburgh, 3rd ed., 2010.

208

P. Couratier, P. Corcia, G. Lautrette, M. Nicol, P.-M. Preux and B. Marin, Revue Neurologique , 2016, **172**, 37–45.

209

210

A. Vincent, Nature Reviews Immunology, 2002, **2**, 797–804.

211

Leslie Jacobson, Journal of Clinical Investigation, DOI:10.1172/JCI5943.

212

W. Hoch, J. McConville, S. Helms, J. Newsom-Davis, A. Melms and A. Vincent, Nature Medicine, 2001, **7**, 365–368.

213

I. Koneczny, J. Cossins and A. Vincent, Journal of Anatomy, 2014, **224**, 29–35.

214

S. Viegas, L. Jacobson, P. Waters, J. Cossins, S. Jacob, M. I. Leite, R. Webster and A. Vincent, Experimental Neurology, 2012, **234**, 506–512.

215

I. Koneczny, J. Cossins, P. Waters, D. Beeson and A. Vincent, PLoS ONE, DOI:10.1371/journal.pone.0080695.

216

S. J. Crisp, D. M. Kullmann and A. Vincent, Nature Reviews Neuroscience, 2016, **17**, 103–117.

217

Beryl B. Cummings, Science translational medicine, DOI:10.1126/scitranslmed.aal5209.

218

D. Schofield, K. Alam, L. Douglas, R. Shrestha, D. G. MacArthur, M. Davis, N. G. Laing, N. F. Clarke, J. Burns, S. T. Cooper, K. N. North, S. A. Sandaradura and G. L. O'Grady, npj Genomic Medicine, DOI:10.1038/s41525-017-0006-7.

219

G. L. O'Grady, M. Lek, S. R. Lamande, L. Waddell, E. C. Oates, J. Punetha, R. Ghaoui, S. A. Sandaradura, H. Best, S. Kaur, M. Davis, N. G. Laing, F. Muntoni, E. Hoffman, D. G. MacArthur, N. F. Clarke, S. Cooper and K. North, Annals of Neurology, 2016, **80**, 101–111.

220

C. G. Bönnemann, C. H. Wang, S. Quijano-Roy, N. Deconinck, E. Bertini, A. Ferreiro, F. Muntoni, C. Sewry, C. Béroud, K. D. Mathews, S. A. Moore, J. Bellini, A. Rutkowski and K. N. North, Neuromuscular Disorders, 2014, **24**, 289–311.

221

K. N. North, C. H. Wang, N. Clarke, H. Jungbluth, M. Vainzof, J. J. Dowling, K. Amburgey, S. Quijano-Roy, A. H. Beggs, C. Sewry, N. G. Laing and C. G. Bönnemann, Neuromuscular Disorders, 2014, **24**, 97–116.

222

M. P. Menezes and K. N. North, Journal of Paediatrics and Child Health, 2012, **48**, 458–465.

223

Milestones timeline : Nature Milestones in DNA, https://www.nature.com/milestones/miledna/timeline.html.

224

M. L. Metzker, Nature Reviews Genetics, 2010, **11**, 31–46.

225

Y. Yang, D. M. Muzny, J. G. Reid, M. N. Bainbridge, A. Willis, P. A. Ward, A. Braxton, J. Beuten, F. Xia, Z. Niu, M. Hardison, R. Person, M. R. Bekheirnia, M. S. Leduc, A. Kirby, P. Pham, J. Scull, M. Wang, Y. Ding, S. E. Plon, J. R. Lupski, A. L. Beaudet, R. A. Gibbs and C. M. Eng, New England Journal of Medicine, 2013, **369**, 1502–1511.

226

Y. Sun, C. A. L. Ruivenkamp, M. J. V. Hoffer, T. Vrijenhoek, M. Kriek, C. J. van Asperen, J. T. den Dunnen and G. W. E. Santen, Human Mutation, 2015, **36**, 648–655.

227

R. Ghaoui, S. T. Cooper, M. Lek, K. Jones, A. Corbett, S. W. Reddel, M. Needham, C. Liang, L. B. Waddell, G. Nicholson, G. O'Grady, S. Kaur, R. Ong, M. Davis, C. M. Sue, N. G. Laing, K. N. North, D. G. MacArthur and N. F. Clarke, JAMA Neurology, DOI:10.1001/jamaneurol.2015.2274.

228

S. Richards, N. Aziz, S. Bale, D. Bick, S. Das, J. Gastier-Foster, W. W. Grody, M. Hegde, E. Lyon, E. Spector, K. Voelkerding and H. L. Rehm, Genetics in Medicine, 2015, **17**, 405–423.

229

Gabrielle Natalie Samuel, New Genetics and Society, DOI:10.1080/14636778.2017.1370671.

230

G. N. Samuel and B. Farsides, Medicine, Health Care and Philosophy, DOI:10.1007/s11019-017-9810-1.

231

A. G. Vaithinathan and V. Asokan, Journal of Evidence-Based Medicine, 2017, **10**, 76–80.

232

B. H. Griffin, L. S. Chitty and M. Bitner-Glindzicz, Archives of disease in childhood - Education & practice edition, 2017, **102**, 105–107.

233

M. M. Evers, L. J. A. Toonen and W. M. C. van Roon-Mom, Advanced Drug Delivery Reviews, 2015, **87**, 90–103.

234

Karin E. Lundin, Human Gene Therapy, DOI:10.1089/hum.2015.070.

235

O. Khorkova and C. Wahlestedt, Nature Biotechnology, 2017, **35**, 249–263.

236

A. Aartsma-Rus, in Exon Skipping, ed. A. Aartsma-Rus, Humana Press, Totowa, NJ, 2012, vol. 867, pp. 117–129.

237

A. M. Rossor, M. M. Reilly and J. N. Sleigh, Practical Neurology, DOI:10.1136/practneurol-2017-001764.

238

N. Wood, Neurogenetics, Cambridge University Press, Cambridge, 2012.

239

C. Clarke, R. Howard, M. Rossor and S. Shorvon, Eds, Neurology, John Wiley &;#38; Sons, Ltd, Chichester, UK, 2016.

240

OMIM - Online Mendelian Inheritance in Man, https://www.omim.org/.

241

L. Teboul, Y. Hérault, C. Smith and B. Whitelaw, Mammalian Genome, 2017, **28**, 235–236.

242

A. Fernández, S. Josa and L. Montoliu, Mammalian Genome, 2017, **28**, 237–246.

243

Marie-Christine Birling, Mammalian Genome, DOI:10.1007/s00335-017-9703-x.

244

A. Greenfield, Mammalian Genome, 2017, **28**, 388–393.

245

Addgene: CRISPR Guide, https://www.addgene.org/crispr/guide/.

246

MGI-Mouse Genome Informatics -The international database resource for the laboratory mouse, http://www.informatics.jax.org/.

247

A. M. Rossor, P. J. Tomaselli and M. M. Reilly, Current Opinion in Neurology, DOI:10.1097/WCO.0000000000000373.

248

A. M. Rossor, M. R. B. Evans and M. M. Reilly, Practical Neurology, 2015, **15**, 187–198.