

Appendix 4. New publications from Centre since last review

*denotes PI (UCL/NCL) collaboration

Bold indicates UCL/NCL PI

Total number of publications: 810

Total number of publications with more than one PI as author: 311

Michael Hanna

1. Rose MR, et al. ENMC International Workshop: Inclusion Body Myositis, 2-4 December 2011, Naarden, The Netherlands. *Neuromuscular Disorders*. 2013 Dec; 23(12):1044:55. PMID: 24268584
2. Fratta P, **Hanna MG**, **Fisher EM**, Sidle K. An unusual presentation for SOD1-ALS: isolated facial diplegia. *Muscle Nerve*. 2013 Dec;48(6):994-5. PMID: 238735403
3. Cortese A, Plagnol V, Brady S, Simone R, Lashley T, Acevedo-Arozena A, de Silva R, Greensmith L, Holton J, **Hanna MG**, **Fisher EM**, Fratta P. Widespread RNA metabolism impairment in sporadic inclusion body myositis TDP43-proteinopathy. *Neurobiol Aging*. 2013 Dec 30. [Epub ahead of print] PMID: 24462217
4. *Graham CD, Weinman J, Sadjadi R, Chalder T, Petty R, **Hanna MG**, Turner C, Parton M, Maddison P, Radunovic A, Longman C, Robb Y, **Bushby K**, Hilton-Jones D, Rose MR. A multicentre postal survey investigating the contribution of illness perceptions, coping and optimism to quality of life and mood in adults with muscle disease. *Clin Rehabil*. 2013 Nov 15. [Epub ahead of print] PMID: 24240060
5. Machado P, Brady S, **Hanna MG**. Update in inclusion body myositis. *Curr Opin Rheumatol*. 2013 Nov;25(6):763-71. PMID: 24067381
6. Jaffer F, **Reilly MM**, Quinlivan R, **Muntoni F**, Turner C, Parton M, **Lunn M**, Hilton-Jones D, Korkodilos M, **Hanna MG**. Emergency neuromuscular admissions are avoidable: a regional audit of unplanned hospital admissions of neuromuscular patients 2009-2011: final results and recommendations. *J Neurol Neurosurg Psychiatry*. 2013 Nov;84(11):e2. PMID: 24109054
7. Pitceathly RD, Taanman JW, Rahman S, Meunier B, Sadowski M, Cirak S, Hargreaves I, Land JM, Nanji T, Polke JM, Woodward CE, Sweeney MG, Solanki S, Foley AR, Hurles ME, Stalker J, Blake J, Holton JL, Phadke R, **Muntoni F**, **Reilly MM**, **Hanna MG**. COX10 Mutations Resulting in Complex Multisystem Mitochondrial Disease That Remains Stable Into Adulthood. *JAMA Neurol*. 2013 Oct 7. [Epub ahead of print] PMID: 24100867
8. Fischmann A, Morrow JM, Sinclair CD, **Reilly MM**, **Hanna MG**, **Yousry T**, **Thornton JS**. Improved anatomical reproducibility in quantitative lower-limb muscle MRI. *J Magn Reson Imaging*. 2013 Oct 7. [Epub ahead of print] PMID: 24123788
9. Tan SV, Z'graggen WJ, Boërio D, Rayan DR, Norwood F, Ruddy D, Howard R, **Hanna MG**, Bostock H. Chloride channels in myotonia congenita assessed by velocity recovery cycles. *Muscle Nerve*. 2013 Sep 4. [Epub ahead of print] PMID: 24037712.
10. *Fratta P, Collins T, Pemble S, Nethisinghe S, Devoy A, Giunti P, Sweeney MG, **Hanna MG**, **Fisher EM**. Sequencing analysis of the spinal bulbar muscular atrophy CAG expansion reveals absence of repeat interruptions. *Neurobiol Aging*. 2013 Sep 13 [Epub ahead of print] PMID: 24041967

11. *Turner C, Hilton-Jones D, **Lochmüller H, Hanna MG**. MRC Centre for Neuromuscular Diseases 1st (1st December 2010), and 2nd (2nd May 2012) myotonic dystrophy workshops, London, UK and the myotonic dystrophy standards of care and national registry meeting, Newcastle, UK July 2011. *Neuromuscul Disord*. 2013 Sep 18. [Epub ahead of print] PMID: 24054840
12. Spillane J, Fialho D, **Hanna MG**. Diagnosis of skeletal muscle channelopathies. *Expert Opin Med Diagn*. 2013 Sep 26. [Epub ahead of print] PMID: 24066928
13. *Willis TA, **Hollingsworth K**, Coombs A, Sveen M, Andersen S, Stojkovic T et al. Quantitative muscle MRI as an assessment tool for monitoring disease progression in LGMD2I: a multicentre longitudinal study, *PLoS One*, 2013 Aug 14;8(8). PMID: 23967145
14. *Pfeffer G, Horvath R, Klopstock T, Mootha VK, Suomalainen A, Koene S et al. New treatments for mitochondrial disease – no time to drop our standards. *Nature Reviews Neurology*, July 2013, PMID: 23817350.
15. Ke Q et al. Rare disease centers for periodic paralysis: China vs the US and UK. *Muscle Nerve*. 2013 July 28. PMID: 23893386
16. *Fratta P, **Hanna MG, Fisher EM**, Sidle K. An unusual presentation for SOD1-ALS: Isolated facial diplegia. *Muscle Nerve*. 2013 Jul 19. [Epub ahead of print] PMID: 23873540
17. *Pitceathly RD, Rahman S, Wedatilake Y, Polke JM, Cirak S, Foley AR, Sailer A, Hurles ME, Stalker J, Hargreaves I, Woodward CE, Sweeney MG, **Muntoni F, Houlden H**, Taanman JW, **Hanna MG**; UK10K Consortium. NDUFA4 mutations underlie dysfunction of a cytochrome c oxidase subunit linked to human neurological disease. *Cell Rep*. 2013 Jun 27;3(6):1795-805. Epub 2013 Jun 6. PMID: 23746447
18. Trivedi JR, Bundy B, Statland J, Salajegheh M, Rayan DR, Venance SL, Wang Y, Fialho D, Matthews E, Cleland J, Gorham N, Herbelin L, Cannon S, Amato A, Griggs RC, **Hanna MG**, Barohn RJ; CINCH Consortium. Non-dystrophic myotonia: prospective study of objective and patient reported outcomes. *Brain*. 2013 Jul;136(Pt 7):2189-200. Epub 2013 Jun 13. PMID: 23771340
19. *Morrow JM, Matthews E, Raja Rayan DL, Fischmann A, Sinclair CD, **Reilly MM, Thornton JS, Hanna MG, Yousry TA**. Muscle MRI reveals distinct abnormalities in genetically proven non-dystrophic myotonias. *Neuromuscul Disord*. 2013 Aug;23(8):637-46. Epub 2013 Jun 27. PMID: 23810313
20. *Morrow JM, **Reilly MM, Hanna MG**. Reliability and accuracy of skeletal muscle imaging in limb-girdle muscular dystrophies. *Neurology*. 2013 Jun 11;80(24):2276. PMID: 23905174
21. Wallace A, Dewar E, Skorupinska M, Laura M, Morrow JM, Sterr A, **Hanna MG** et al. Evaluating the benefits of community based aerobic training on the physical health and well-being of people with neuromuscular diseases: a pilot study. *Journal of the Peripheral Nervous System*. 18: 123-123. June 2013.
22. Burge JA, **Hanna MG**, Schorge S. Non-genomic actions of progesterone and 17 β -estradiol on the chloride conductance of skeletal muscle. *Muscle Nerve*. 2013 Apr 26. [Epub ahead of print] PMID: 23625574.
23. Thorne T, Fratta P, **Hanna MG**, Cortese A, Plagnol V, **Fisher EM**, Stumpf MP. Graphical modelling of molecular networks underlying sporadic inclusion body myositis. *Mol Biosyst*. 2013 Apr 17. [Epub ahead of print] PMID: 23595110.

24. *Sarkozy A, Hicks D, Hudson J, Laval SH, Barresi R, Hilton-Jones D, et al. ANO5 Gene Analysis in a Large Cohort of Patients with Anoctaminopathy: Confirmation of Male Prevalence and High Occurrence of the Common Exon 5 Gene Mutation. *Hum Mutat.* 2013 Apr 18. PubMed PMID:23606453.
25. Horga A, Raja Rayan DL, Matthews E, Sud R, Fialho D, Durran SC, Burge JA, Portaro S, Davis MB, Haworth A, **Hanna MG**. Prevalence study of genetically defined skeletal muscle channelopathies in England. *Neurology.* 2013 Apr 16;80(16):1472-5. Epub 2013 Mar 20. PMID: 23516313.
26. Cortese A, Machado P, Morrow J, Dewar L, Hiscock A, Miller A, Brady S, Hilton-Jones D, Parton M, **Hanna MG**. Longitudinal observational study of sporadic inclusion body myositis: Implications for clinical trials. *Neuromuscul Disord.* 2013 May;23(5):404-12. Epub 2013 Mar 11. PMID: 23489664.
27. Rajakulendran S, Roberts J, Koltzenburg M, **Hanna MG**, Stewart H. Deletion of chromosome 12q21 affecting KCNC2 and ATXN7L3B in a family with neurodevelopmental delay and ataxia. *J Neurol Neurosurg Psychiatry.* 2013 Mar 9. [Epub ahead of print] PMID: 23475819.
28. Rajakulendran S, Roberts J, **Koltzenburg M, Hanna MG**, Stewart H. Deletion of chromosome 12q21 affecting KCNC2 and ATXN7L3B in a family with neurodevelopmental delay and ataxia. *J Neurol Neurosurg Psychiatry.* 2013 Nov;84(11):1255-7. Epub 2013 Mar 9. PMID: 23475819
29. Malik B, Nirmalanathan N, Gray AL, La Spada AR, **Hanna MG, Greensmith L**. 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 Mar;136(Pt 3):926-43. Epub 2013 Feb 7. PMID: 23393146
30. Smith MD, Seth JH, **Hanna MG**, Panicker JN. Detrusor overactivity in Becker muscular dystrophy. *Muscle Nerve.* 2013 Mar;47(3):464-5. Epub 2013 Feb 4. PMID: 23382079.
31. Tomlinson SE, Rajakulendran S, Tan SV, Graves TD, Bamiou DE, Labrum RW, Burke D, Sue CM, Giunti P, Schorge S, **Kullmann DM, Hanna MG**. Clinical, genetic, neurophysiological and functional study of new mutations in episodic ataxia type 1. *J Neurol Neurosurg Psychiatry.* 2013 Jan 24. [Epub ahead of print] PMID: 23349320.
32. *Nesbitt V, Pitceathly RD, **Turnbull DM, Taylor RW**, Sweeney MG, Mudanohwo EE, Rahman S, **Hanna MG, McFarland R**. The UK MRC Mitochondrial Disease Patient Cohort Study: clinical phenotypes associated with the m.3243A>G mutation—implications for diagnosis and management. *J Neurol Neurosurg Psychiatry.* 2013 Jan 25. [Epub ahead of print] PMID: 23355809
33. Portaro S, Musumeci O, Rizzo V, Rodolico C, Buccasfusca M, Toscano A, Sweeney MG, **Hanna MG**. Stiffness as a presenting symptom of an odd clinical condition caused by multiple sclerosis and myotonia congenita. *Neuromuscular Disorders* 23(1):52-55 Jan 2013. PMID: 22921319
34. Morrow JM, Matthews E, Raja Rayan D, Fischmann A, Sinclair CDJ, **Reilly MM**, Thornton JS, **Hanna MG, Yousry T**. Muscle MRI reveals distinct abnormalities genetically proven non-dystrophic myotonias. *Neuromuscular Disorders.* 2013. PMID: 23810313
35. Pitceathly RDS, Tomlinson SE, Holton JL, Morrow JM, Rahman S, **Hanna MG** et al. Distal myopathy with cachexia: an unrecognized phenotype caused by dominantly-inherited mitochondrial polymerase γ mutations. *JNNP* 84(1):107-110. Jan 2013. PMID: 22933815

36. *Willis TA, **Hollingsworth KG**, Coombs A, Sveen ML, Andersen S, Stojkovic T, Eagle M, Mayhew A, de Sousa PL, Dewar L, Morrow JM, Sinclair CD, **Thornton JS**, **Bushby K**, **Lochmüller H**, **Hanna MG**, Hogrel JY, Carlier PG, Vissing J, **Straub V**. Quantitative muscle MRI as an assessment tool for monitoring disease progression in LGMD2I: a multicentre longitudinal study. *PLoS One*. 2013;8(8):e70993. eCollection 2013. PMID: 23967145
37. Machado PM, Ahmed M, Brady S, Gang Q, Healy E, Morrow JM, Wallace AC, Dewar L, Ramdharry G, Parton M, Holton JL, **Houlden H**, **Greensmith L**, **Hanna MG**. Ongoing developments in sporadic inclusion body myositis. *Curr Rheumatol Rep*. 2014 Dec;16(12):477. PMID: 25399751
38. Corrochano S, Männikkö R, Joyce PI, McGoldrick P, Wettstein J, Lassi G, Raja Rayan DL, Blanco G, Quinn C, Liavas A, Lionikas A, Amior N, Dick J, Healy EG, Stewart M, Carter S, Hutchinson M, Bentley L, Fratta P, Cortese A, Cox R, Brown SD, Tucci V, Wackerhage H, Amato AA, **Greensmith L**, **Koltzenburg M**, **Hanna MG**, Acevedo-Arozena A. Novel mutations in human and mouse SCN4A implicate AMPK in myotonia and periodic paralysis. *Brain*. 2014 Dec;137(Pt 12):3171-85. PMID: 25348630
39. *Horga A, Pitceathly RD, Blake JC, Woodward CE, Zapater P, Fratter C, Mudanohwo EE, Plant GT, **Houlden H**, Sweeney MG, **Hanna MG**, **Reilly MM**. Peripheral neuropathy predicts nuclear gene defect in patients with mitochondrial ophthalmoplegia. *Brain*. 2014 Dec;137(Pt 12):3200-12. PMID: 25281868
40. *Cottenie E, Kochanski A, Jordanova A, Bansagi B, Zimon M, Horga A, Jaunmuktane Z, Saveri P, Rasic VM, Baets J, Bartsakoulia M, Ploski R, Teterycz P, Nikolic M, Quinlivan R, Laura M, Sweeney MG, Taroni F, **Lunn MP**, Moroni I, Gonzalez M, **Hanna MG**, Bettencourt C, Chabrol E, Franke A, von Au K, Schilhabel M, Kabzińska D, Hausmanowa-Petrusewicz I, Brandner S, Lim SC, Song H, Choi BO, **Horvath R**, Chung KW, Zuchner S, Pareyson D, Harms M, **Reilly MM**, **Houlden H**. Truncating and missense mutations in IGHMBP2 cause Charcot-Marie Tooth disease type 2. *Am J Hum Genet*. 2014 Nov 6;95(5):590-601. PMID: 25439726
41. Singh RR, Tan SV, **Hanna MG**, Robb SA, Clarke A, Jungbluth H. Mutations in SCN4A: a rare but treatable cause of recurrent life-threatening laryngospasm. *Pediatrics*. 2014 Nov;134(5):e1447-50. PMID: 25311598.
42. Matthews E, **Hanna MG**. Repurposing of sodium channel antagonists as potential new anti-myotonic drugs. *Exp Neurol*. 2014 Nov;261:812-5. PMID: 25218042.
43. Suetterlin K, Männikkö R, **Hanna MG**. Muscle channelopathies: recent advances in genetics, pathophysiology and therapy. *Curr Opin Neurol*. 2014 Oct;27(5):583-90. PMID: 25188014.
44. Quinlivan R, Matthews E, **Hanna MG**. Innovative care model for patients with complex muscle diseases. *Curr Opin Neurol*. 2014 Oct;27(5):607-13. PMID: 25188015.
45. Gang Q, Bettencourt C, Machado P, **Hanna MG**, **Houlden H**. Sporadic inclusion body myositis: the genetic contributions to the pathogenesis. *Orphanet J Rare Dis*. 2014 Jun 19;9:88. Review. PubMed PMID: 24948216
46. Montague K, Malik B, Gray AL, La Spada AR, **Hanna MG**, Szabadkai G, **Greensmith L**. Endoplasmic reticulum stress in spinal and bulbar muscular atrophy: a potential target for therapy. *Brain*. 2014 Jul;137(Pt 7):1894-906. Epub 2014 Jun 4. PMID: 24898351
47. Fratta P, Nirmalanathan N, Masset L, Skorupinska I, Collins T, Cortese A, Pemble S, Malaspina A, **Fisher EM**, **Greensmith L**, **Hanna MG**. Correlation of clinical and molecular

features in spinal bulbar muscular atrophy. *Neurology*. 2014 May 9. [Epub ahead of print] PMID: 24814851

Birbeck GL, **Hanna MG**, Griggs RC. Global opportunities and challenges for clinical neuroscience. *JAMA*. 2014 Apr 23-30;311(16):1609-10. PMID: 24756506

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50. Graves TD, Cha YH, Hahn AF, Barohn R, Salajegheh MK, Griggs RC, Bundy BN, Jen JC, Baloh RW, **Hanna MG**; CINCH Investigators. Episodic ataxia type 1: clinical characterization, quality of life and genotype-phenotype correlation. *Brain*. 2014 Apr;137(Pt 4):1009-18. Epub 2014 Feb 26. PMID: 24578548

51. *Willis TA, Hollingsworth KG, Coombs A, Sveen ML, Andersen S, Stojkovic T, Eagle M, Mayhew A, de Sousa PL, Dewar L, Morrow JM, Sinclair CD, **Thornton JS**, **Bushby K**, **Lochmuller H**, **Hanna MG**, Hogrel JY, Carlier PG, Vissing J, **Straub V**. Quantitative magnetic resonance imaging in limb-girdle muscular dystrophy 2I: a multinational cross-sectional study. *PLoS One*. 2014 Feb 28;9(2) PMID: 24587344

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53. Suetterlin KJ, Bugiardini E, Kaski JP, **Morrow JM**, Matthews E, **Hanna MG**, Fialho D. Long-term Safety and Efficacy of Mexiletine for Patients With Skeletal Muscle Channelopathies. *JAMA Neurol*. 2015 Dec;72(12):1531-3. PubMed PMID: 26658970.

54. Poole OV, **Hanna MG**, Pitceathly RD. Mitochondrial disorders: disease mechanisms and therapeutic approaches. *Discov Med*. 2015 Nov;20(111):325-31. PubMed PMID: 26645904.

55. Gang Q, Bettencourt C, **Houlden H**, **Hanna MG**, Machado PM. Genetic advances in sporadic inclusion body myositis. *Curr Opin Rheumatol*. 2015 Nov;27(6):586-94. Review. PubMed PMID: 26335925.

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Myositis Genetics Consortium. Dense genotyping of immune-related loci in idiopathic inflammatory myopathies confirms HLA alleles as the strongest genetic risk factor and suggests different genetic background for major clinical subgroups. *Ann Rheum Dis*. 2016 Aug;75(8):1558-66. Epub 2015 Sep 11. PubMed PMID: 26362759.

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