

MRC Centre for Neuromuscular Diseases
PI Publications 2008 – Present

Total number of publications: 563

Total number of publications with more than one PI as author: 138

**paper with more than one PI as author

Fialho D, Schorge S, Pucovska U, Davies NP, Labrum R, Haworth A, Stanley E, Sud R, Wakeling W, Davis MB, **Kullmann DM, Hanna MG. Chloride channel myotonia: exon 8 hot-spot for dominant-negative interactions. *Brain*. 2007 Dec;130(Pt 12):3265-74. PubMed PMID: 17932099.

Taylor RW, **Chinnery PF, Turnbull DM. Investigation of metabolic myopathies. *Handb Clin Neurol*. 2007;86:193-204. PMID: 18809001.

Matthews E, Tan SV, Fialho D, Sweeney MG, Sud R, Haworth A, Stanley E, Cea G, Davis MB, **Hanna MG**. What causes paramyotonia in the United Kingdom? Common and new SCN4A mutations revealed. *Neurology*. 2008 Jan 1;70(1):50-3. PMID: 18166706.

Cree LM, Samuels DC, Chuva de Sousa Lopes S, Rajasimha HK, Wonnapijit P, Mann JR, Dahl H-H.M. **Chinnery PF**. A reduction in the number of mitochondrial DNA molecules during embryogenesis explains the rapid segregation of genotypes. *Nature Genetics* 2008; 40(2):249-54. PMID: 18223651

Kirkman M,A, Yu-Wai-Man P, **Chinnery PF**. The clinical spectrum of mitochondrial disorders. *Clinical Medicine* 2008;8:601-6: PMID: 19149282

McNeill A, Birchall D, Hayflick SJ, Gregory A, Schenck JF, Zimmerman EA, Shang H, Miyajima H, **Chinnery PF**. T2* and FSE MRI distinguishes four subtypes of Neurodegeneration with Brain Iron Accumulation. *Neurology* 2008;70(18):1614-9. PMID: 18443312

Krishnan KJ, Reeve AK, Samuels DC, **Chinnery PF**, Blackwood JK, Taylor RW, Wanrooij S, Spelbrink JN, Lightowlers RN, Turnbull DM. What causes mitochondrial DNA deletions in human cells? *Nature Genetics* 2008;40(3):275-9. PMID: 18305478

Hudson G, Amati-Bonneau P, Blakely EL, Stewart JD, He L, Schaefer AM, Griffiths PG, Ahlqvist K, Suomalainen A, Reynier P, McFarland R, **Turnbull DM, Chinnery PF, Taylor RW. Mutation of OPA1 causes dominant optic atrophy with external ophthalmoplegia, ataxia, deafness and multiple mitochondrial DNA deletions: a novel disorder of mtDNA maintenance. *Brain* 2008;131(Pt 2):329-37. PMID 18065439.

Hudson G, Yu-Wai-Man P, **Chinnery PF**. Leber hereditary optic neuropathy. *Expert Opinion in Medical Diagnostics* 2008;2:789-800. PMID: 17573650

Nemes A, De Coo IMF, Spruijt L, Smeets HJT, **Chinnery PF**, Soliman OII, Geleijnse ML ten Cate FJ. Is there alteration in aortic stiffness in Leber's hereditary optic neuropathy? *European Journal of Ophthalmology* 2008;18(2):309-12. PMID:18320530

Hudson G, Mowbray C, Elson J, Jacob A, Boggild M, Torroni A, **Chinnery PF**. Does mitochondrial DNA predispose to neuromyelitis optica (Devic's disease)? *Brain* 2008;131(Pt 4):e93. PMID: 17967805

Abramov, AY and **Duchen, MR**. (2008) Mechanisms underlying the loss of mitochondrial membrane potential in glutamate excitotoxicity, *BBA bioenergetics*, 1777(7-8):953-64.

Szabadkai, G and **Duchen MR**, (2008) Mitochondria: The Hub of Cellular Ca²⁺ Signaling, *Physiology*, 23(2):84-94.

Duchen MR, Verkhratsky A, Muallem S. (2008) Mitochondria and calcium in health and disease. *Cell Calcium*. 44(1):1-5.

Wood-Kaczmar A, Gandhi S, Yao Z, Abramov AS, Miljan EA, Keen G, Stanyer L, Hargreaves I, Klupsch K, Deas E, Downward J, Mansfield L, Jat P, Taylor J, Heales S, **Duchen MR**, Latchman D, Tabrizi SJ,

Wood NW. (2008) PINK1 Is Necessary for Long Term Survival and Mitochondrial Function in Human Dopaminergic Neurons. PLoS ONE., 3(6):e2455

Hall AM, Unwin, **Hanna MG and **Duchen MR**. (2008) New insights into mitochondrial cytopathies and their relevance to the kidney. Quarterly Journal of Medicine, 177(7-8):953-64.

Campanella, M, Casswell, E, Chong, S, Farah, Z, Wieckowski, MR, Abramov, AY, Tinker, A and **Duchen, MR**. (2008) The cell biology of IF₁: a major regulator of mitochondrial structure and function. Cell Metabolism, 8(1):13-25.

Milton R.H., Abeti R., Averaimo S., DeBiasi S., Vitellaro L., Jiang L., Curmi P.M.G., Breit S.N., **Duchen M.R.** and Mazzanti M. (2008) CLIC1 function is required for beta-amyloid induced generation of reactive oxygen species by microglia, J Neurosci. 28(45):11488-99

L.G. Bilstrand, N. Nirmalanathan, J. Yip, **L. Greensmith & M.R. Duchen (2008) Expression of mutant SOD1G93A in astrocytes induces functional deficits in motoneuron mitochondria. J Neurochem. 107(5):1271-83

Isman, O., Roberts, M. L., **Morgan, JE.**, Graham, I. R., Goldring, K., Lawrence-Watt, D. J., Lu, Q. L., Dunckley, M. G., Porter, A. C., Partridge, T. A., Dickson, G. (2008). Adenovirus-based targeting in myoblasts is hampered by nonhomologous vector integration. Hum. Gene Ther. 19(10), 1000-1008. PMID: 21935475

Jessen KR, Mirsky R. Negative regulation of myelination: relevance for development, injury, and demyelinating disease. Glia. 2008, 56:1552-65. PMID: 18803323

Parkinson DB, Bhaskaran A, Arthur-Farraj P, Noon LA, Woodhoo A, Lloyd AC, Feltri ML, Wrabetz L, Behrens A, Mirsky R, **Jessen KR**. c-Jun is a negative regulator of myelination. J Cell Biol. 2008 181:625-37. PMID:18490512

Mirsky R, Woodhoo A, Parkinson DB, Arthur-Farraj P, Bhaskaran A, **Jessen KR**. Novel signals controlling embryonic Schwann cell development, myelination and dedifferentiation. J Peripher Nerv Syst. 2008 13:122-35. PMID: 8601657

Agudo M, Woodhoo A, Webber D, Mirsky R, **Jessen KR**, McMahon SB. Schwann cell precursors transplanted into the injured spinal cord multiply, integrate and are permissive for axon growth. Glia. 2008, 56:1263-70. PMID: 18484102

Clement EM, Godfrey C, Tan J, Brockington M, Torelli S, Feng L, **Brown SC, Jimenez-Mallebrera C, Sewry CA, Longman C, Mein R, Abbs S, Vajsaar J, Schachter H, **Muntoni F**. Mild POMGnT1 mutations underlie a novel limb-girdle muscular dystrophy variant. Arch Neurol. 2008 Jan;65(1):137-41. PMID:18195152

Houlden H, Laura M, Wavrant-DeVrieze F, Blake J, **Reilly M M**. Mutations in the Heat Shock Protein 27 cause autosomal dominant, recessive and sporadic distal hereditary motor neuropathy / Charcot-Marie-Tooth disease type 2. Neurology 2008; 71; 1660-1668. PMID 18832141

Bennett DL, Groves M, Blake J, Holton JL, King RH, Orrell RW, Ginsberg L, **Reilly MM**. The use of nerve and muscle biopsies in the diagnosis of vasculitis: a 5 year retrospective study. JNNP 2008; 79; 1376-1381. PMID 18819942

Dick KJ, McEntagart M, Alwan W, **Reilly M**, Crosby AH. Refinement of the locus for distal Hereditary Motor neuropathy VII (dHMN-VII) and exclusion of candidate genes. Genome 2008; 51; 959-962.

Bosnell R, Wegner C, Kincses ZT, Korteweg T, Agosta F, Ciccarelli O, De Stefano N, Gass A, Hirsch J, Johansen-Berg H, Barkhof F, Mancini L, Manfredonia F, Marino S, Miller DH, Montalban X, Palace J, Rocca M, Enzinger C, Ropele S, Rovira A, Smith S, **Thompson A, Thornton J, **Yousry T**, Whitcher B, Filippi M, Matthews PM. Reproducibility of fMRI in the clinical setting: implications for trial designs. NeuroImage 2008;42:603-610. PMID 18579411

Cano SJ, Warner TT, **Thompson AJ**, Bhatia KP, Fitzpatrick R, Hobart JC. The Cervical Dystonia Impact Profile: Can a Rasch developed patient reported outcome measure satisfy traditional psychometric criteria? *Health and Quality of Life Outcomes* 2008;6:58. PMID 18684327

Kerrison S, Laws S, Cane M, **Thompson A**. The patient's experience of being a human subject. *J Roy Soc Med* 2008; 101:416-422. PMID 18687865

Schessl J, Zou Y, McGrath M, Cowling B, Maiti B, Chin S, Sewry CA, Battini R, Hu Y, Cottle D, Ganguly A, Roseblatt M, Spruce L, Kirschner J, Judkins A, Golden J, Goebel H, **Muntoni F**, Flanigan K, Mitchell C, Bonnemann C. Proteomic identification of the LIM domain protein FHL1 as the gene-product mutated in sporadic and familial reducing body myopathy. *Journal Clinical Investigation*, 2008; 3;118(3):904-912. PMID: 18274675

Windpassinger C, Schoser B, **Straub V, Hochmeister S, Noor A, Lohberger B, Farra N, Petek E, Schwarzbraun T, Ofner L, Löscher WN, Wagner K, **Lochmüller H**, Vincent JB, Quasthoff S. An X-linked myopathy with postural muscle atrophy and generalized hypertrophy, termed XMPMA, is caused by mutations in FHL1. *Am J Hum Genet.* 2008 Jan;82(1):88-99. PMID: 18179888

Andäng M, Hjerling-Leffler J, Moliner A, Lundgren TK, Castelo-Branco G, Nanou E, Pozas E, Bryja V, Halliez S, Nishimaru H, Wilbertz J, Arenas E, **Koltzenburg M**, Charnay P, El Manira A, Ibañez CF, Ernfors P. Histone H2AX-dependent GABA(A) receptor regulation of stem cell proliferation. *Nature.* 2008 Jan 24;451(7177):460-4. Epub 2008 Jan 9. PMID: 18185516.

Reeve AK, Krishnan KJ, Elson JL, Morris CM, Bender A, Lightowers RN, **Turnbull DM**. Nature of mitochondrial DNA deletions in substantia nigra neurons. *Am J Hum Genet.* 2008 Jan;82(1):228-35. PMID: 18179904

DD Ateh, IK Hussain, AH Mustafa, KM Price, R Gulati, CD Nickols, MM. Bird, **L Greensmith, M Hafezparast, **EMC Fisher**, CS. Baker & JE Martin (2008) Dynein–dynactin complex subunits are differentially localized in brain and spinal cord, with selective involvement in pathological features of neurodegenerative disease. *Neuropathology and Applied Neurobiology* 34, 88–94 PMID18086204

LG Bilsland and **L Greensmith** (2008) The Endocannabinoid System in Amyotrophic Lateral Sclerosis in The Endocannabinoid System in Neuroinflammatory Diseases. *Current Pharmaceutical Design.* 14, 2306-2316. PMID 18781981

B Kalmar, S Novoselov, A Gray, ME Cheetham, B Margulis and **L Greensmith** (2008) Late stage treatment with Arimoclomol delays disease progression and prevents protein aggregation in the SOD1 mouse model of ALS. *Journal of Neurochemistry.* 107, 339-50. PMID 18673445

LG Bilsland, N Nirmalanathan, J Yip, **L Greensmith & **MR Duchen** (2008) Expression of mutant SOD1^{G93A} in astrocytes *Journal of Neurochemistry.* 107, 1271-1283 induces functional deficits in motoneuron mitochondria. PMID 18808448

Rajasimha HK, **Chinnery PF**, Samuels DC. Selection against pathogenic mtDNA mutations in a stem cell population leads to the loss of the 3243A>G mutation in blood. *American Journal of Human Genetics* 2008; Feb;82(2):333-43. PMID: 18252214

McFarland R, Hudson G, Taylor RW, Green SH, Hodges S, McKiernan PJ, **Chinnery PF**, Ramesh V. Reversible Valproate hepatotoxicity due to mutations in mitochondrial DNA polymerase γ (POLG1). in *Archives of Disease in Childhood* 2008 Feb;93(2):151-3. PMID:18208989

Chinnery PF, Zeviani M. 155th ENMC Workshop: Polymerase gamma and disorders of mitochondrial DNA synthesis. *Neuromuscular Disorders* 2008 Mar;18(3):259-67. PMID: 18160290

Sylantsev S, Savtchenko LP, Niu YP, Ivanov AI, Jensen TP, **Kullmann DM**, Xiao MY, Rusakov DA. Electric fields due to synaptic currents sharpen excitatory transmission. *Science.* 2008 Mar 28;319(5871):1845-9. PubMed PMID: 18369150.

Kullmann DM, Lamsa K. Roles of distinct glutamate receptors in induction of anti-Hebbian long-term potentiation. *J Physiol*. 2008 Mar 15;586(6):1481-6. Epub 2008 Jan 10. Review. PubMed PMID: 18187472.

Graves TD, **Hanna MG**. Channelling into the epilepsies. *Epilepsy Curr*. 2008 Mar-Apr;8(2):37-8. PMID: 18330464.

****Muntoni F, Bushby K**, vanOmmen GJ. 149th ENMC International Workshop and 1st TREAT-NMD Workshop on: "Planning Phase I/II Clinical trials using Systemically Delivered Antisense Oligonucleotides in Duchenne Muscular Dystrophy". *Neuromuscul Disord*. 2008 Mar;18(3):268-75. PMID: 18207401

****Spuler S, Carl M, Zabojszcza J, Straub V, Bushby K**, Moore SA, Bähring S, Wenzel K, Vinkemeier U, Rocken C. Dysferlin-deficient muscular dystrophy features amyloidosis. *Ann Neurol*. 2008 Mar;63(3):323-8. PMID: 18306167.

Mihaylova V, Müller JS, Vilchez JJ, Salih MA, Kabiraj MM, D'Amico A, Bertini E, Wölfle J, Schreiner F, Kurlemann G, Rasic VM, Siskova D, Colomer J, Herczegfalvi A, Fabriciova K, Weschke B, Scola R, Hoellen F, Schara U, Abicht A, **Lochmüller H**. Clinical and molecular genetic findings in COLQ-mutant congenital myasthenic syndromes. *Brain*. 2008 Mar;131(Pt 3):747-59. Epub 2008 Jan 7. PMID: 18180250.

****Healy DG, Goadsby PJ, Kitchen ND, Yousry T, Hanna MG**. Neurological picture. Spontaneous intracranial hypotension, hygromata and haematomata. *J Neurol Neurosurg Psychiatry*. 2008 Apr;79(4):442. PMID: 18344394.

****Hicks D, Lampe AK, Barresi R, Charlton R, Fiorillo C, Bonnemann CG, Hudson J, Sutton R, Lochmüller H, Straub V, Bushby K**. A refined diagnostic algorithm for Bethlem myopathy. *Neurology*. 2008 Apr 1;70(14):1192-9. PMID: 18378883.

****Klinge L, Dean AF, Kress W, Dixon P, Charlton R, Müller JS, Anderson LV, Straub V, Barresi R, Lochmüller H, Bushby K**. Late onset in dysferlinopathy widens the clinical spectrum. *Neuromuscul Disord*. 2008 Apr;18(4):288-90. PMID: 18396043.

Betts J, Barron MJ, Needham SJ, Schaefer AM, Taylor RW, **Turnbull DM**. Gastrointestinal tract involvement associated with the 3243A>G mitochondrial DNA mutation. *Neurology*. 2008 Apr 8;70(15):1290-2. PMID: 18391161.

Murphy R, **Turnbull DM**, Walker M, Hattersley AT. Clinical features, diagnosis and management of maternally inherited diabetes and deafness (MIDD) associated with the 3243A>G mitochondrial point mutation. *Diabet Med*. 2008 Apr;25(4):383-99. Epub 2008 Feb 18. Review. PMID: 18294221.

Moriarty KT, McFarland R, Whittaker R, Burch J, Turnbull HE, Taylor RW, **Turnbull DM**. Pre-eclampsia and magnesium toxicity with therapeutic plasma level in a woman with .3243A>G melas mutation. *J Obstet Gynaecol*. 2008 Apr;28(3):349. PMID: 18569490.

****Wagner KR, Fleckenstein JL, Amato AA, Barohn RJ, Bushby K, Escolar DM, Flanigan KM, Pestronk A, Tawil R, Wolfe GI, Eagle M, Florence JM, King WM, Pandya S, Straub V, Juneau P, Meyers K, Csimma C, Araujo T, Allen R, Parsons SA, Wozney JM, Lavallie ER, Mendell JR**. A phase I/II trial of MYO-029 in adult subjects with muscular dystrophy. *Ann Neurol*. 2008 May;63(5):561-71. PMID: 18335515.

Hjermind LE, Vissing J, Asmus F, Krag T, **Lochmüller H**, Walter MC, Erdal J, Blake DJ, Nielsen JE. No muscle involvement in myoclonus-dystonia caused by epsilon-sarcoglycan gene mutations. *Eur J Neurol*. 2008 May;15(5):525-9. Epub 2008 Mar 18. PMID: 18355305.

Rorbach J, Yusoff AA, Tuppen H, Abg-Kamaludin DP, Chrzanowska-Lightowlers ZM, Taylor RW, **Turnbull DM**, McFarland R, Lightowlers RN. Overexpression of human mitochondrial valyl tRNA synthetase can partially restore levels of cognate mt-tRNA^{Val} carrying the pathogenic C25U mutation. *Nucleic Acids Res*. 2008 May;36(9):3065-74. Epub 2008 Apr 8. PMID: 18400783

Yu-Wai-Man P, Bateman D, Hudson G, Griffiths PG, **Chinnery PF**. Leber hereditary optic neuropathy in a 75 year old man. *Journal of Neuroophthalmology* 2008 Jun;28(2):155. PMID: 18562849

Lampe AK, Zou Y, Sudano D, O'Brien KK, Hicks D, Laval SH, Charlton R, Jimenez-Mallebrera C, Zhang RZ, Finkel RS, Tennekoon G, Schreiber G, van der Knaap MS, Marks H, **Straub V, Flanigan KM, Chu ML, **Muntoni F**, **Bushby KM**, Bönnemann CG. Exon skipping mutations in collagen VI are common and are predictive for severity and inheritance. *Hum Mutat*. 2008 Jun;29(6):809-22. PMID: 18366090

Treves S, Jungbluth H, **Muntoni F**, Zorzato F. Congenital muscle disorders with cores: the ryanodine receptor calcium channel paradigm. *Curr Opin Pharmacol* 2008 Jun;8(3):319-326. PMID: 18313359

Patel P, Harris R, Geddes SM, Strehle EM, Watson JD, Bashir R, **Bushby K**, Driscoll PC, Keep NH. Solution structure of the inner DysF domain of myoferlin and implications for limb girdle muscular dystrophy type 2b. *J Mol Biol*. 2008 Jun 20;379(5):981-90. Epub 2008 Apr 26. PMID: 18495154.

Huang Y, de Morrée A, van Remoortere A, **Bushby K**, Frants RR, Dunnen JT, van der Maarel SM. Calpain 3 is a modulator of the dysferlin protein complex in skeletal muscle. *Hum Mol Genet*. 2008 Jun 15;17(12):1855-66. Epub 2008 Mar 11. PMID: 18334579

Thornhill P, Bassett D, **Lochmüller H, **Bushby K**, **Straub V**. Developmental defects in a zebrafish model for muscular dystrophies associated with the loss of fukutin-related protein (FKRP). *Brain*. 2008 Jun;131(Pt 6):1551-61. Epub 2008 May 13. PMID: 18477595.

Strach K, Sommer T, Grohé C, Meyer C, Fischer D, Walter MC, Vorgerd M, Reilich P, Bär H, Reimann J, Reuner U, Germing A, Goebel HH, **Lochmüller H**, Wintersperger B, Schröder R. Clinical, genetic, and cardiac magnetic resonance imaging findings in primary desminopathies. *Neuromuscul Disord*. 2008 Jun;18(6):475-82. Epub 2008 May 27. PMID: 18504128.

Argyriou AA, **Koltzenburg M**, Polychronopoulos P, Papapetropoulos S, Kalofonos HP. Peripheral nerve damage associated with administration of taxanes in patients with cancer. *Crit Rev Oncol Hematol*. 2008 Jun;66(3):218-28. Epub 2008 Mar 7. Review. PMID: 18329278.

Reeve AK, Krishnan KJ, **Turnbull DM**. Age related mitochondrial degenerative disorders in humans. *Biotechnol J*. 2008 Jun;3(6):750-6. Review. PMID: 18512864.

Craig K, Takiyama Y, Soong B-W, Jardim LB, Saraiva-Pereira ML, Lythgow K, Morino H, Maruyama H, Kawakami H, **Chinnery PF**. Pathogenic expansions of the SCA6 locus are associated with a common CACNA1A haplotype across the globe: founder effect or predisposing chromosome? *European Journal of Human Genetics* 2008 Jul;16(7):841-7. PMID: 18285829

Scott R, Ruiz A, Henneberger C, **Kullmann DM**, Rusakov DA. Analog modulation of mossy fiber transmission is uncoupled from changes in presynaptic Ca²⁺. *J Neurosci*. 2008 Jul 30;28(31):7765-73. PubMed PMID: 18667608.

Graves TD, **Hanna MG**. Episodic ataxia: SLC1A3 and CACNB4 do not explain the apparent genetic heterogeneity. *J Neurol*. 2008 Jul;255(7):1097-9. Epub 2008 May 2. PMID: 18446307.

Pane M, Staccioli S, Messina S, D'Amico A, Pelliccioni M, Mazzone ES, Cuttini M, Alfieri P, Battini R, Main M, **Muntoni F**, Bertini E, Villanova M, Mercuri E. Daily salbutamol in young patients with SMA type II. *Neuromuscul Disord*. 2008 Jul;18(7):536-40. PMID: 18579379

Aboumoussa A, Hoogendijk J, Charlton R, Barresi R, Herrmann R, Voit T, Hudson J, Roberts M, Hilton-Jones D, Eagle M, **Bushby K, **Straub V**. Caveolinopathy—new mutations and additional symptoms. *Neuromuscul Disord*. 2008 Jul;18(7):572-8. Epub 2008 Jun 25. PMID: 18583131.

Müller OJ, **Lochmüller H**. Sarcoglycans take center stage in gene transfer therapy. *Neurology*. 2008 Jul 22;71(4):234-5. Epub 2008 Jun 4. PMID: 18525030.

Bender A, Schwarzkopf RM, McMillan A, Krishnan KJ, Rieder G, Neumann M, Elstner M, **Turnbull DM**,

Klopstock T. Dopaminergic midbrain neurons are the prime target for mitochondrial DNA deletions. *J Neurol*. 2008 Aug;255(8):1231-5. Epub 2008 Jul 11. PMID: 18604467.

Blakely E, He L, Gardner JL, Hudson G, Walter J, Hughes I, **Turnbull DM**, Taylor RW. Novel mutations in the TK2 gene associated with fatal mitochondrial DNA depletion myopathy. *Neuromuscul Disord*. 2008 Jul;18(7):557-60. Epub 2008 May 27. PMID: 18508266.

Elliott HR, Samuels DC, Eden JA, Relton CL, **Chinnery PF**. Pathogenic mitochondrial DNA mutations are common in the general population. *American Journal of Human Genetics* 2008 Aug;83(2):254-60. PMID: 18674747

Ban M, Elson J, Walton A, **Turnbull D, Compston A, **Chinnery P**, Sawcer S. Investigation of the role of mitochondrial DNA in multiple sclerosis susceptibility. *PLoS ONE*. 2008 Aug 6;3(8):e2891. PMID: 18682780

Cree LM, Patel S, Pyle A, Lynn S, **Turnbull DM, **Chinnery PF**, Walker M. Age-related decline in mitochondrial DNA copy number in isolated human pancreatic islets. *Diabetologia* 2008 Aug;51(8):1440-3. PMID: 18528676

Verma V, Dhar A, **Chinnery PF**. Occult GI cause of spastic paresis of the legs. *Gut* 2008 Aug;57(8):1064. PMID: 18628374

Quijano-Roy S, Mbieleu B, Bönnemann CG, Jeannet PY, Colomer J, Clarke NF, Cuisset JM, Roper H, De Meirleir L, D'Amico A, Ben Yaou R, Nascimento A, Barois A, Demay L, Bertini E, Ferreiro A, Sewry CA, Romero NB, Ryan M, **Muntoni F**, Guicheney P, Richard P, Bonne G, Estournet B. De novo Imna mutations cause a new form of congenital muscular dystrophy. *Annals of Neurology*, 2008 Aug;64(2):177-86. PMID: 18551513

McSweeney N, Cowan F, Manzur A, Robb S, **Muntoni F**. Perinatal dyskinesia as a presenting feature of Prader Willi syndrome. *Eur J Paediatr Neurol*. 2008 Aug 20. PMID: 18722147

M Kinali, D Beeson, MC Pitt, H Jungbluth, AK Simonds, A Aloysius, H Cockerill, T Davis, J Palace, AY Manzur, C Jimenez-Mallebrera, C Sewry, **F Muntoni**, SA Robb. Congenital Myasthenic Syndromes in childhood: diagnostic and management challenges. *J. Neuroimmunology*, 2008 15; 201-202:6-12. PMID: 18707767

Schwab N, Waschbisch A, Wrobel B, **Lochmüller H**, Sommer C, Wiendl H. Human myoblasts modulate the function of antigen-presenting cells. *J Neuroimmunol*. 2008 Aug 30;200(1-2):62-70. Epub 2008 Jul 21. PMID: 18644633.

Yu-Wai-Man P, Elliott C, Griffiths PG, Johnson IJ, **Chinnery PF**. Investigation of auditory dysfunction in Leber Hereditary Optic Neuropathy. *Acta Ophthalmologica Scandinavica* 2008 Sep;86(6):630-3. PMID:18070226

Stewart JD, Gavin Hudson G, Patrick Yu-Wai-Man P, Blakeley EL, He L-P, **Horvath R, Maddison P, Wright A, Griffiths PG, **Turnbull DM**, Taylor RW, **Chinnery PF**. OPA1 in multiple mitochondrial DNA deletion disorders. *Neurology* 2008; 25;71(22):1829-31. PMID: 19029523

Zsurka G, Baron M, Stewart JD, Kornblum C, Bös M, Sassen R, Taylor RW, Elger C, **Chinnery PF**, Kunz WS. Clonally expanded mitochondrial DNA mutations in patients with Alpers-Huttenlocher syndrome. *Journal of Neuropathology and Experimental Neurology* 2008; Sep;67(9):857-66. PMID: 18716558

Klein A, Clement E, Mercuri E, **Muntoni F**. Differential diagnosis of congenital muscular dystrophies. *European Journal of Paediatric Neurology*, 2008 Sep;12(5):371-7. PMID: 18588847

Otto A, Corina Schmidt, Graham Luke, Steve Allen, Petr Valasek, **Muntoni F**, Diana Lawrence-Watt, and Ketan Patel. Canonical Wnt signalling induces satellite cell proliferation and is active during adult skeletal muscle regeneration. *Journal of Cell Science*, 2008 Sep 1;121:2939-50. PMID: 18697834

Budde BS, Namavar Y, Barth PG, Poll-The BT, Nürnberg G, Becker C, van Ruissen F, Weterman MA, Fluiter K, T Te Beek E, Aronica E, van der Knaap MS, Höhne W, Toliaat MR, Crow YJ, Steinlin M, Voit T,

Roelens F, Brussel W, Brockmann K, Kyllerman M, Boltshauser E, Hammersen G, Willemsen M, Basel-Vanagaite L, Krägeloh-Mann I, de Vries LS, Sztriha L, **Muntoni F**, Ferrie CD, Battini R, Hennekam RC, Grillo E, Beemer FA, Stoets LM, Wollnik B, Nürnberg P, Baas F.; tRNA splicing endonuclease mutations cause pontocerebellar hypoplasia. *Nature Genetics*, 2008 Sep;40(9):1113-8. PMID: 18711368

Bauer R, Macgowan GA, Blain A, **Bushby K, **Straub V**. Steroid treatment causes deterioration of myocardial function in the δ -sarcoglycan-deficient mouse model for dilated cardiomyopathy. *Cardiovasc Res*. 2008 Sep 1;79(4):652-61. Epub 2008 May 20. PMID: 18495669.

Guglieri M, **Bushby K**. How to go about diagnosing and managing the limb-girdle muscular dystrophies. *Neurol India*. 2008 Jul-Sep;56(3):271-80. Review. PMID: 18974553.

Heuss D, Klascinski J, Schubert SW, Moriabadi T, **Lochmüller H**, Hashemolhosseini S. Examination of transcript amounts and activity of protein kinase CK2 in muscle lysates of different types of human muscle pathologies. *Mol Cell Biochem*. 2008 Sep;316(1-2):135-40. Epub 2008 Jun 14. PMID: 18553059.

Devos D, Tchofo PJ, Vuillaume I, Destée A, Batey S, Burn S, **Chinnery PF**. Clinical features and natural history of neuroferritinopathy caused by the 458dupA FTL mutation. *Brain* 2008;Oct 14th. PMID 18854324

Gorman G, Fairgrieve S, Birchall D, **Chinnery PF**. Fragile X premutation presenting as essential tremor. *Journal of Neurology Neurosurgery and Psychiatry* 2008 Oct;79(10):1195-6. PMID: 18796598

Graves TD, Imbrici P, Kors EE, Terwindt GM, Eunson LH, Frants RR, Haan J, Ferrari MD, Goadsby PJ, **Hanna MG, van den Maagdenberg AM, **Kullmann DM**. Premature stop codons in a facilitating EF-hand splice variant of CaV2.1 cause episodic ataxia type 2. *Neurobiol Dis*. 2008 Oct;32(1):10-5. PubMed PMID: 18606230.

Hall AM, Unwin RJ, **Hanna MG, **Duchen MR**. Renal function and mitochondrial cytopathy (MC): more questions than answers? *QJM*. 2008 Oct;101(10):755-66. Epub 2008 May 16. Review. PMID: 18487272

Sewry CA, Jemenez-Mallebrera C, **Muntoni F**. Congenital myopathies. *Curr Opin Neurol*. 2008 Oct;21(5):569-575. PMID: 18769251

Jungbluth H, **Muntoni F**, Ferreira A. 150th ENMC International Workshop: Core Myopathies, 9-11th March 2007, Naarden, The Netherlands. *Neuromuscul Disord*. 2008 Oct 21. PMID: 18948004

Muntoni F, Torelli S, Brockington M. Muscular dystrophies due to glycosylation defects. *Neurotherapeutics*. 2008 Oct;5(4):627-632. PMID: 21825985

Guglieri M, **Straub V, **Bushby K**, **Lochmüller H**. Limb-girdle muscular dystrophies. *Curr Opin Neurol*. 2008 Oct;21(5):576-84. Review. PMID:18769252.

****Straub V**, **Bushby K**. Therapeutic possibilities in the autosomal recessive limb-girdle muscular dystrophies. *Neurotherapeutics*. 2008 Oct;5(4):619-26. Review. PMID: 19019315.

Schessl J, Walter MC, Schreiber G, Schara U, Müller CR, **Lochmüller H**, Bönnemann CG, Korinthenberg R, Kirschner J. Phenotypic variability in siblings with calpainopathy (LGMD2A). *Acta Myol*. 2008 Oct;27:54-8. PMID: 19364062

Schara U, **Lochmüller H**. Therapeutic strategies in congenital myasthenic syndromes. *Neurotherapeutics*. 2008 Oct;5(4):542-7. Review. PMID: 19019305.

Lochmüller H, Griggs RC. New treatments for neuromuscular disease: optimism and obstacles. *Neurotherapeutics*. 2008 Oct;5(4):497-8. PMID: 19019299.

Swalwell H, Blakely EL, Sutton R, Tonska K, Elstner M, He L, Taivassalo T, Burns DK, **Turnbull DM**, Haller RG, Davidson MM, Taylor RW. A homoplasmic mtDNA variant can influence the phenotype of the pathogenic m.7472Cins MTTTS1 mutation: are two mutations better than one? *Eur J Hum Genet*. 2008 Oct;16(10):1265-74. Epub 2008 Apr 9. PMID: 18398437.

Wonnapijit P, **Chinnery PF**, Samuels DC. The Distribution of Mitochondrial DNA Heteroplasmy due to Random Genetic Drift. *American Journal of Human Genetics* 2008 Nov;83(5):582-93. PMID: 18976726

Kullmann DM. Benign neonatal convulsions and spontaneous network activity in the developing brain: is there a link? *J Physiol*. 2008 Nov 15;586(Pt 22):5281. PubMed PMID: 19011128

Fialho D, **Kullmann DM, **Hanna MG**, Schorge S. Non-genomic effects of sex hormones on CLC-1 may contribute to gender differences in myotonia congenita. *Neuromuscul Disord*. 2008 Nov;18(11):869-72. PubMed PMID: 18815035

Matthews E, Guet A, Mayer M, Vicart S, Pemble S, Sternberg D, Fontaine B, **Hanna MG**. Neonatal hypotonia can be a sodium channelopathy: recognition of a new phenotype. *Neurology*. 2008 Nov 18;71(21):1740-2. PMID: 19015492

Manzur AY, Kinali M, **Muntoni F**. Update on the management of Duchenne muscular dystrophy. *Arch Dis Child*. 2008 Nov;93(11):986-90. PMID: 18667451

Mercuri E, Mahyew A, **Muntoni F, Messina S, **Straub V**, van Ommen GJ, Voit T, Bertini E, **Bushby K**, On Behalf of the TREAT-NMD Neuromuscular Network. Towards harmonisation of outcome measures for DMD and SMA within TREAT-NMD; Report of three expert workshops: TREAT-NMD/ENMC Workshop on outcome measures, 12th-13th May 2007, Naarden, The Netherlands; TREAT-NMD Workshop on outcome measures in experimental trials for DMD, 30th June-1st July 2007, Naarden, The Netherlands; TREAT-NMD Meeting on physical activity monitoring in neuromuscular disorders, 11th July 2007, Paris, France. *Neuromuscul Disord*. 2008 Nov;18(11):894-903. PMID: 18818076

Bovolenta M, Neri M, Fini S, Fabris M, TrabANELLI C, Venturoli A, Martoni E, Bassi E, Spitali P, Brioschi S, Falzarano MS, Rimessi P, Ciccone R, Ashton E, McCauley J, Yau S, Abbs S, **Muntoni F**, Merlini L, Gualandi F, Ferlini A. A novel custom high density-comparative genomic hybridisation array detects common rearrangements as well as deep intronic mutations in dystrophinopathies. *BMC Genomics*. 2008 Nov 28;9(1):572. PMID: 19040728

Clement E, Mercuri E, Godfrey C, Smith J, Robb S, Kinali M, **Straub V, **Bushby V**, Manzur A, Talim B, Cowan F, Quinlivan R, Klein A, Topaloglu H, Mein R, Abbs S, North K, Barkovich J, Rutherford M, **Muntoni F**. Brain involvement in dystroglycanopathies. 2008. *Annals of Neurology*, Nov;64(5):573-82. PMID: 19067344

Klinge L, Dekomien G, Aboumoussa A, Charlton R, Epplen JT, Barresi R, **Bushby K, **Straub V**. Sarcoglycanopathies: can muscle immunoanalysis predict the genotype? *Neuromuscul Disord*. 2008 Dec;18(12):934-41. Epub 2008 Nov 7. PMID: 18996010.

Waschbisch A, Wintterle S, **Lochmüller H**, Walter MC, Wischhusen J, Kieseier BC, Wiendl H. Human muscle cells express the costimulatory molecule B7-H3, which modulates muscle-immune interactions. *Arthritis Rheum*. 2008 Nov;58(11):3600-8. PMID: 18975328.

Suominen T, Schoser B, Raheem O, Auvinen S, Walter M, Krahe R, **Lochmüller H**, Kress W, Udd B. High frequency of co-segregating CLCN1 mutations among Myotonic dystrophy type 2 patients from Finland and Germany. *J Neurol*. 2008 Nov;255(11):1731-6. Epub 2008 Sep 24. PMID: 18807109.

Schmalhofer WA, Calhoun J, Burrows R, Bailey T, Kohler MG, Weinglass AB, Kaczorowski GJ, Garcia ML, **Koltzenburg M**, Priest BT. ProTx-II, a selective inhibitor of NaV1.7 sodium channels, blocks action potential propagation in nociceptors. *Mol Pharmacol*. 2008 Nov;74(5):1476-84. Epub 2008 Aug 26. PMID: 18728100.

Murphy JL, Blakely EL, Schaefer AM, He L, Wyrick P, Haller RG, Taylor RW, **Turnbull DM**, Taivassalo T. Resistance training in patients with single, large-scale deletions of mitochondrial DNA. *Brain*. 2008 Nov;131(Pt 11):2832-40. PMID: 18984605.

Fernandez-Fuente M, Ames EG, Wagner ML, Zhou H, Strom M, Zammit PS, Mickelson JR, **Muntoni F, **Brown SC**, Piercy RJ. Assessment of the transformation of equine skin-derived fibroblasts to

multinucleated skeletal myotubes following lentiviral-induced expression of equine myogenic differentiation 1. *Am J Vet Res.* 2008 Dec;69(12):1637-45. PMID:19046012

Scott R, Lalic T, **Kullmann DM**, Capogna M, Rusakov DA. Target-cell specificity of kainate autoreceptor and Ca²⁺-store-dependent short-term plasticity at hippocampal mossy fiber synapses. *J Neurosci.* 2008 Dec 3;28(49):13139-49. PubMed PMID: 19052205.

****Brown SC**, Piercy RJ, **Muntoni F**, Sewry CA. Investigating the pathology of Emery Dreifuss muscular dystrophy. *Biochem Soc Trans.* 2008 Dec 1;36(6):1335-1338. PMID: 19021551

****Sárközy A**, **Bushby K**, Béroud C, **Lochmüller H**. 157th ENMC International Workshop: patient registries for rare, inherited muscular disorders 25-27 January 2008 Naarden, The Netherlands. *Neuromuscul Disord.* 2008 Dec;18(12):997-1001. Epub 2008 Oct 22. PMID: 18948006.

Richard P, Gaudon K, Haddad H, Ammar AB, Genin E, Bauché S, Paturneau-Jouas M, Müller JS, **Lochmüller H**, Grid D, Hamri A, Nouioua S, Tazir M, Mayer M, Desnuelle C, Barois A, Chabrol B, Pouget J, Koenig J, Gouider-Khouja N, Hentati F, Eymard B, Hantaï D. The CHRNE 1293insG founder mutation is a frequent cause of congenital myasthenia in North Africa. *Neurology.* 2008 Dec 9;71(24):1967-72. PMID: 19064877.

Collie A, Landsverk M, Ruzzo E, Mefford H, Buysse K, Adkins J, Knutzen D, Barnett K, Brown R Jr, Parry G, Yum S, Simpson D, Olney R, **Chinnery P**, Eichler E, Chance P, Hannibal M. Non-recurrent SEPT9 duplications cause Hereditary Neuralgic Amyotrophy. *Journal of Medical Genetics* 2009 PMID: 19939853

Hudson G, Yu-Wai-Man P, Zeviani M, **Chinnery PF**. Genetic variation in the methylenetetrahydrofolate reductase gene MTHFR does not alter the risk of visual failure in Leber's hereditary optic neuropathy. *Molecular Vision* 2009;15:870-5. PMID 1942144

****R.G. Whittaker RW**, J.K. Blackwood JK, Alston CL, Blakely EL, Elson JL, McFarland R, **Chinnery PF**, **Turnbull DM**, Taylor RW. Urine heteroplasmy is the best predictor of clinical outcome in the m.3243A>G mtDNA mutation. *Neurology* 2009;72:568-9. PMID: 19204268

****Stewart JD**, Tennant S, Powell H, Pyle A, Blakeley EL, He L, Hudson G, Roberts M, du Plessis D, Gow D, Mewasingh LD, **Hanna MG**, Omer S, Morris AA, Roxburgh R, Livingston JH, McFarland R, **Turnbull DM**, **Chinnery PF**, Taylor RW. Novel POLG1 mutations associated with neuromuscular and liver phenotypes in adults and children. *Journal of Medical Genetics* 2009;46:209–214. PMID 19251978

Yu-Wai-Man P, Gorman G, Bateman DE, Leigh RJ, **Chinnery PF**. Vertigo and Vestibular Abnormalities in Spinocerebellar Ataxia Type 6. *Journal of Neurology* 2009 Jan;256(1):78-82. PMID 19224313

Hall A, Unwin RJ, **Duchen MR**. (2009) Imaging mitochondrial function in intact rat kidney slices by multi-photon microscopy. *Journal of the American Society for Nephrology.* 20(6):1293-302

Cantley, J., Selman, C., Shukla, D., Abramov, AY, Forstreuter, F., Estban, M., Claret, M., lingard, SJ., Clements, M., Harten., SK., Asare-Anane, H., Batterham, RL., Herrera, PL., Persaud, SJ., **Duchen MR.**, Maxwell., PH Withers, DJ. Deletion of the von-Hippel-Lindau gene in pancreatic β cells impairs glucose homeostasis in mice. *J Clin Invest.*, 2009 Jan;119(1):125-35

Campanella, M, Seraphim, A, Casswell, E, Echave P, Abeti R and **Duchen MR** (2009) IF1, the F₁F₀-ATP synthase endogenous regulator, defines mitochondrial volume fraction in HeLa cells by regulating autophagy *BBA (bioenergetics)* 1787(5):393-401.

Szabadkai, G and **Duchen, MR** (2009) Mitochondria mediated cell death in Diabetes. *Apoptosis.* 14(12):1405-23

Abramov and **Duchen** (2009) Impaired mitochondrial bioenergetics determines glutamate-induced delayed calcium deregulation in neurons. *BBA*, 1800(3):297-304.

Mann, ZF, **Duchen MR** & Gale JE (2009) Mitochondria modulate the spatio-temporal properties of intra and intercellular Ca²⁺ signals in cochlear supporting cells. *Cell Calcium*, 46(2):136-46.

Galkin A, Abramov AY, Frakich N, **Duchen MR**, Moncada S. Lack of oxygen deactivates mitochondrial complex I: implications for ischemic injury? *J Biol Chem*. 2009 284(52):36055-61.

Echave, P, Machado-da-Silva, G, Arkell, RS, **Duchen MR**, Jacobson, J, Mitter, R and Lloyd AC (2009) Extracellular growth factors and mitogens cooperate to drive mitochondrial biogenesis. *J Cell Sci*, 122(Pt 24):4516-25.

Achilli, F., Bros-Facer, V., Williams, H., Banks, G., AlQatari, M., Chia, R., Tucci, V., Groves, M., Nickols, C., Seburn, K., Kendall, R., Cader, M., Talbot, K., van Minnen, J., Burgess, R., **Brandner, S., Martin, J., **Koltzenburg, M.**, **Greensmith, L.**, Nolan, P., **Fisher, E.M.C.** (2009) An ENU-induced mutation in a mouse glycyl-tRNA synthetase (GARS) causes peripheral sensory and motor phenotypes creating a model of Charcot-Marie-Tooth type 2D peripheral neuropathy. *Dis Model Mech* 2: 359-373 PMID: 19470612

Banks, G.T., Bros-Facer, V., Williams, H.P., Chia, R., Achilli, F., Bryson, J.B., **Greensmith, L., **Fisher, E.M.C.** (2009) Mutant glycyl-tRNA synthetase ameliorates SOD1G93A motor neuron degeneration phenotype but has little effect on Loa dynein heavy chain mutant mice. *PLoS ONE* 4: e6218. PMID: 19593442

Arechavala-Gomez, V., Kinali, M., Feng, L., **Brown, S. C., Sewry, C., **Morgan, J.E.**, **Muntoni, F.** (2009). Immunohistological intensity measurements as a tool to assess. *Neuropathol Appl Neurobiol* 36(4):265-74. PMID:2002311

Meng, J., Adkin, C. F., Arechavala-Gomez, V., Boldrin, L., **Muntoni, F., **Morgan, J.E.** (2009). The contribution of human synovial stem cells to skeletal muscle regeneration. *Neuromuscul Disord* 20(1):6-15. PMID: 20034794

Kinali, M., Arechavala-Gomez, V., Feng, L., Cirak, S., Hunt, D., Adkin, C., Guglieri, M., Ashton, E., Abbs, S., Nihoyannopoulos, P., Garralda, M. E., Rutherford, M., McCulley, C., Popplewell, L., Graham, I. R., Dickson, G., Wood, M. J., Wells, D. J., Wilton, S. D., Kole, R., Straub, V., Bushby, K., Sewry, C., **Morgan, J. E., **Muntoni, F.** (2009). 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. *Lancet Neurol*. 8(10), 918-928. PMID: 19713152

Collins, C. A., Gnocchi, V. F., White, R. B., Boldrin, L., Perez-Ruiz, A., Relaix, F., **Morgan, J.E.**, Zammit, P. S. (2009). Integrated functions of Pax3 and Pax7 in the regulation of proliferation, cell size and myogenic differentiation. *PLoS ONE* 4(2), e4475. PMID: 19221588

Boldrin, L., Zammit, P. S., **Muntoni, F., **Morgan, J.E.** (2009). Mature Adult Dystrophic Mouse Muscle Environment Does Not Impede Efficient Engrafted Satellite Cell Regeneration and Self-Renewal. *Stem Cells* 27(10), 2478-2487. PMID: 19575422

Woodhoo A, Alonso MB, Droggiti A, Turmaine M, D'Antonio M, Parkinson DB, Wilton DK, Al-Shawi R, Simons P, Shen J, Guillemot F, Radtke F, Meijer D, Feltri ML, Wrabetz L, Mirsky R, **Jessen KR**. Notch controls embryonic Schwann cell differentiation, postnatal myelination and adult plasticity. *Nat Neurosci*. 2009, 12:839-47. PMID:19525946

Ackroyd MR, Skordis L, Kaluarachchi M, Godwin J, Prior S, Fidanboyly M, Piercy RJ, **Muntoni F, **Brown SC**. Reduced expression of fukutin related protein in mice results in a model for fukutin related protein associated muscular dystrophies. *Brain*. 2009 Feb;132(Pt 2):439-51. Epub 2009 Jan 20. PMID:19155270

Oren I, Nissen W, **Kullmann DM**, Somogyi P, Lamsa KP. Role of ionotropic glutamate receptors in long-term potentiation in rat hippocampal CA1 oriens-lacunosum moleculare interneurons. *J Neurosci*. 2009 Jan 28;29(4):939-50. PubMed PMID: 19176803

Reilly MM, Shy M. Diagnosis and new treatments in genetic neuropathies. *JNNP* 2009; 80: 1304-1314. PMID 19917815

Reilly MM. Classification and diagnosis of the inherited neuropathies. *Ann Indian Acad Neur* 2009; 12: 80-88. PMID 20142852

Petzold A, Brettschneider J, Jin K, Murray N, Hirsch N, Itayena Y, **Reilly MM**, Takeda A, Taneda H. CSF protein biomarkers for proximal axonal damage improves prognostic accuracy in the acute phase of Guillain-Barre- Syndrome. *Muscle Nerve* 2009; 40; 42-49. PMID 19533642

Ramdharry G, Day B, **Reilly MM**, Marsden J. Hip flexor fatigue limits walking in Charcot Marie Tooth disease. *Muscle Nerve* 2009; 40; 103-111. PMID 19405092

Mitchell A, Laura M, Blake J, Lunn M, Cox A, Gibbons V, Davies M, Wood N, Manji H, Houlden H, Murray N, **Reilly MM.** GJB1 gene mutations in inflammatory demyelinating neuropathies not responding to treatment. *Letter JNNP* 2009; 80; 699-700. PMID 19448103

H. Houlden, M. Laura, L. Ginsberg, H. Jungbluth, S. Robb, J. Blake, S. Robinson, R.H.M. King and **MM Reilly.** The phenotype of SH3TC2 mutations and possible predisposition to an inflammatory neuropathy. *Neuromuscular Disorders* 2009; 19; 264-269. PMID 19272779

Houlden H, **Reilly MM**, Smith S. Pupil abnormalities in 131 cases of genetically defined inherited peripheral neuropathy. *Eye* 2009; 23: 966-974. PMID 18636082

Houlden H, Hammans S, Katifi H, **Reilly MM.** A novel Frabin (FGD4) nonsense mutation p.R275X associated with phenotypic variability in CMT4H. *Neurology* 2009; 72; 617-620. PMID 19221294

RMC trial group. Randomised controlled trial of methotrexate for chronic inflammatory demyelinating polyradiculopathy (RMC trial): a pilot multicentre study. *Lancet Neurology* 2009; 8: 158-164. PMID 19136303

Hicks D, Lampe AK, Laval SH, Allamand V, Jimenez-Mallebrera C, Walter MC, **Muntoni F, Quijano-Roy S, Richard P, **Straub V, Lochmüller H, Bushby KM.** Cyclosporine A treatment for Ullrich congenital muscular dystrophy: a cellular study of mitochondrial dysfunction and its rescue. *Brain.* 2009 Jan;132(Pt 1):147-55. PMID: 19015158

Selcen D, **Muntoni F**, Burton B, Pegoraro E, Sewry C, Bite A, Engel A. Mutation in BAG3 Causes Severe Dominant Childhood Muscular Dystrophy. *Annals of Neurology.* 2009, 65(1):83-9. PMID: 19085932

Schessl J, Taratuto AL, Sewry C, Battini R, Chin SS, Maiti B, Dubrovsky AL, Erro MG, Espada G, Robertella M, Saccoliti M, Olmos P, Bridges LR, Standring P, Hu Y, Zou Y, Swoboda KJ, Scavina M, Goebel HH, Mitchell CA, Flanigan KM, **Muntoni F**, Bönnemann CG. Clinical, morphological and genetic findings in reducing body myopathy caused by mutations in FHL1. *Brain*, 2009; 132(Pt 2):452-64. PMID: 19181672

Jungbluth H, Lillis S, Zhou H, Abbs S, Sewry C, Swash M, **Muntoni F.** Late Onset axial myopathy with cores due to a novel heterozygous dominant mutation in the skeletal muscle ryanodine receptor (RYR1) gene. *Neuromuscular Disorders*;19(5):344-7. 2009. PMID: 19303294

Arbogast S, Beuvin M, Fraysse B, Zhou H, **Muntoni F** and Ferreira A. Oxidative stress in SEPNI-related myopathy: from pathophysiology to treatment. *Annals of Neurology*, 2009;65(6):677-686. PMID: 19557870

Whittaker RG, **Turnbull DM.** A diagnostic tattoo. *Clin Genet.* 2009 Jan;75(1):37-8. PMID: 19128396.

J Riddoch-Contreras, S-Y Yang, JRT Dick, G Goldspink, RW Orrell & **L Greensmith** (2009) Mechano-Growth Factor, an IGF-I splice variant, rescues motoneurons and improves muscle function in SOD1^{G93A} mice. *Experimental Neurology.* 215, 281–289 PMID 19038252

B Kalmar and **L Greensmith** (2009) Activation of the heat shock response in a cellular model of ALS-evidence for neuroprotective and neurotoxic effects *Cellular & Molecular Biology Letters* 14(2):319-35. PMID 19183864

D Boërio, **L Greensmith** and H Bostock (2009) Excitability properties of motor axons in the maturing mouse *Journal of the Peripheral Nervous System* 14(1):45-53 PMID 19335539

B Kalmar and **L Greensmith** (2009) Induction of heat shock proteins for protection against oxidative stress in "Controlling Oxidative Stress- therapeutic and delivery strategies *Advanced Drug Delivery Reviews* 61, 310–318 PMID 19248813

F Mackenzie, R Romero, F Wong, D Williams, T Gillingwater, H Hilton, J Dick, J Riddoch-Contreras, L Ireson, N Powles-Glover, P Underhill, T Hough, R Arkell, **L Greensmith**, R Ribchester & G Blanco (2009) The TRPP protein PKD1L2 is a regulator of neuromuscular homeostasis *Human Molecular Genetics* 18(19):3553-66. PMID 19578180

Fuhrmann N, Alavi MV, Bitoun P, Woernle S, Auburger G, Leo-Kottler B, Yu-Wai-Man P, **Chinnery P**, Wissinger B. Genomic rearrangements in OPA1 are frequent in patients with autosomal dominant optic atrophy. *Journal of Medical Genetics* 2009 Feb;46(2):136-44. PMID: 19181907

Healy DG, Goadsby PJ, Kitchen ND, Yousry T, **Hanna MG. Spontaneous intracranial hypotension, hygromata and haematomata. *BMJ Case Rep.*2009;. Epub 2009 Feb 16. PMID: 21687310

Merrison AF, **Hanna MG**. The bare essentials: muscle disease. *Pract Neurol.* 2009 Feb;9(1):54-65. PMID: 19151243

Ghassemi F, Vukcevic M, Xu L, Zhou H, Meissner G, **Muntoni F**, Jungbluth H, Zorzato F, Treves S. A recessive ryanodine receptor 1 mutation in a CCD patient increases channel activity. *Cell Calcium.* 2009 Feb;45(2):192-7. PMID: 19027160

Baumeister SK, Todorovic S, Milić-Rasić V, Dekomien G, **Lochmüller H**, Walter MC. Eosinophilic myositis as presenting symptom in gamma-sarcoglycanopathy. *Neuromuscul Disord.* 2009 Feb;19(2):167-71. Epub 2009 Jan 23. PMID: 19167890.

McFarland R, **Turnbull DM**. Batteries not included: diagnosis and management of mitochondrial disease. *J Intern Med.* 2009 Feb;265(2):210-28. Review. PMID: 19192037.

Fellous TG, McDonald SA, Burkert J, Humphries A, Islam S, De-Alwis NM, Gutierrez-Gonzalez L, Tadrous PJ, Elia G, Kocher HM, Bhattacharya S, Mears L, El-Bahrawy M, **Turnbull DM, Taylor RW, Greaves LC, **Chinnery PF**, Day CP, Wright NA, Alison MR. A Methodological Approach to Tracing Cell Lineage in Human Epithelial Tissues. *Stem Cells* 2009 Mar 19;27(6):1410-1420. PMID: 19489031

Yu-Wai-Man P, Griffiths PG, Hudson G, **Chinnery PF**. Inherited mitochondrial optic neuropathies. *Journal of Medical Genetics* 2009 Mar;46(3):145-58. PMID 19001017

Wolny S, McFarland R, **Chinnery P**, Cheetham T. Abnormal growth in mitochondrial disease. *Acta Paediatrica* 2009 Mar;98(3):553-4. PMID 19120037

Owe SG, Jensen V, Evergren E, Ruiz A, Shupliakov O, **Kullmann DM**, Storm-Mathisen J, Walaas SI, Hvalby Ø, Bergersen LH. Synapsin- and actin-dependent frequency enhancement in mouse hippocampal mossy fiber synapses. *Cereb Cortex.* 2009 Mar;19(3):511-23. PubMed PMID: 18550596

Van Der Linden FAH, Kragt JJ, Hobart JC, Klein M, **Thompson AJ**, Van Der Ploeg HM, Polman CH, Uitdehaag BMJ. The size of the treatment effect: do patients and proxies agree? *BMC Neurology*, 2009; 9:12 published online 25th March. PMID 19317921

Muntoni F, Guicheney P, Voit T. 158th ENMC international workshop on congenital muscular dystrophy (Xth international CMD workshop) 8th-10th February 2008 Naarden, The Netherlands. *Neuromuscul Disord.* 2009 Mar;19(3):229-34. PMID: 19097897

Bauer R, Hudson J, Müller HD, Sommer C, Dekomien G, Bourke J, Routledge D, **Bushby K, Klepper J, **Straub V**. Does delta-sarcoglycan-associated autosomal-dominant cardiomyopathy exist? *Eur J Hum Genet.* 2009 Sep;17(9):1148-53. Epub 2009 Mar 4. PMID: 19259135

Gutierrez-Gonzalez L, Deheragoda M, Elia G, Leedham SJ, Shankar A, Imber C, Jankowski JA, **Turnbull DM**, Novelli M, Wright NA, McDonald SA. Analysis of the clonal architecture of the human small intestinal epithelium establishes a common stem cell for all lineages and reveals a mechanism for the fixation and spread of mutations. *J Pathol.* 2009 Mar;217(4):489-96. PMID: 19156773.

Machado P, Miller A, Holton J, **Hanna M**. Sporadic inclusion body myositis: an unsolved mystery. *Acta Reumatol Port*. 2009 Apr-Jun;34(2A):161-82. Review. PMID: 19474772

Meola G, **Hanna MG**, Fontaine B. Diagnosis and new treatment in muscle channelopathies. *J Neurol Neurosurg Psychiatry*. 2009 Apr;80(4):360-5. Review. PMID: 19289476

Waschbisch A, Meuth SG, Herrmann AM, Wrobel B, Schwab N, **Lochmüller H**, Wiendl H. Intercellular exchanges of membrane fragments (trocytosis) between human muscle cells and immune cells: a potential mechanism for the modulation of muscular immune responses. *J Neuroimmunol*. 2009 Apr 30;209(1-2):131-8. Epub 2009 Mar 9. PMID: 19269695.

Senderek J, Garvey SM, Krieger M, Guergueltcheva V, Urtizberea A, Roos A, Elbracht M, Stendel C, Tournev I, Mihailova V, Feit H, Tramonte J, Hedera P, Crooks K, Bergmann C, Rudnik-Schöneborn S, Zerres K, **Lochmüller H**, Seboun E, Weis J, Beckmann JS, Hauser MA, Jackson CE. Autosomal-dominant distal myopathy associated with a recurrent missense mutation in the gene encoding the nuclear matrix protein, matrin 3. *Am J Hum Genet*. 2009 Apr;84(4):511-8. Epub 2009 Apr 2. PMID: 19344878

Brabec P, Vondráček P, Klimes D, Baumeister S, **Lochmüller H**, Pavlík T, Gregor J. Characterization of the DMD/BMD patient population in Czech Republic and Slovakia using an innovative registry approach. *Neuromuscul Disord*. 2009 Apr;19(4):250-4. Epub 2009 Mar 9. PMID: 19269824.

Bulst S, Abicht A, Holinski-Feder E, Müller-Ziermann, Koehler U, Thirion C, Walter MC, Stewart J, **Chinnery PF, Lochmüller H, Horvath R. In vitro supplementation with dAMP/dGMP leads to partial restoration of mtDNA levels in mitochondrial depletion syndromes. *Human Molecular Genetics* 2009 May 1;18(9):1590-9. PMID: 19221117

Shatunov A, Olivé M, Odgerel Z, Stadelmann-Nessler C, Irlbacher K, van Landeghem F, Bayarsaikhan M, Lee H-S, Goudeau B, **Chinnery PF, Straub V, Hilton-Jones D, Damian MS, Kaminska A, Vicart P, **Bushby K**, Dalakas MC, Sambuughin M, Ferrer I, Goebel HH, Goldfarb LG. In-frame deletion in the seventh immunoglobulin-like repeat of filamin C in a family with myofibrillar myopathy. *European Journal of Human Genetics* 2009 May;17(5):656-63. PMID 19050726

Matthews E, Labrum R, Sweeney MG, Sud R, Haworth A, **Chinnery PF, Meola G, Schorge S, **Kullmann DM**, Davis MB, **Hanna MG**. Voltage sensor charge loss accounts for most cases of hypokalemic periodic paralysis. *Neurology*. 2009 May 5;72(18):1544-7. PMID 19118277

Dumollard R, Carroll J, **Duchen MR**, Campbell K, Swann K Mitochondrial function and redox state in mammalian embryos. *Semin Cell Dev Biol*. 2009 May;20(3):346-53.

Bauer R, **Straub V, Blain A, **Bushby K**, MacGowan GA. Contrasting effects of steroids and angiotensin-converting-enzyme inhibitors in a mouse model of dystrophin-deficient cardiomyopathy. *Eur J Heart Fail*. 2009 May;11(5):463-71. Epub 2009 Feb 20. PMID: 19233868.

Sejerson T, **Bushby K**; TREAT-NMD EU Network of Excellence. Standards of care for Duchenne muscular dystrophy: brief TREAT-NMD recommendations. *Adv Exp Med Biol*. 2009;652:13-21. PMID: 20225016.

Horváth R, Bender A, Abicht A, Holinski-Feder E, Czermin B, Trips T, Schneiderat P, **Lochmüller H**, Klopstock T. Heteroplasmic mutation in the anticodon-stem of mitochondrial tRNA(Val) causing MNGIE-like gastrointestinal dysmotility and cachexia. *J Neurol*. 2009 May;256(5):810-5. Epub 2009 Mar 1. PMID: 19252805.

Miller TD, Jackson AP, Barresi R, Smart CM, Eugenicos M, Summers D, Clegg S, **Straub V**, Stone J. Inclusion body myopathy with Paget disease and frontotemporal dementia (IBMPFD): clinical features including sphincter disturbance in a large pedigree. *J Neurol Neurosurg Psychiatry*. 2009 May;80(5):583-4. PMID: 19372299.

Greaves LC, **Turnbull DM**. Mitochondrial DNA mutations and ageing. *Biochim Biophys Acta*. 2009 Oct;1790(10):1015-20. Epub 2009 May 4. Review. PMID: 19409965.

Aitken H, Gorman G, McFarland R, Roberts M, Taylor RW, **Turnbull DM**. Clinical reasoning: Blurred vision and dancing feet: restless legs syndrome presenting in mitochondrial disease. *Neurology*. 2009 May 5;72(18):e86-90. PMID: 19414717.

Wright AF, Murphy MP, **Turnbull DM**. Do organellar genomes function as long-term redox damage sensors? *Trends Genet*. 2009 Jun;25(6):253-61. Epub 2009 May 27. PMID: 19481287.

Goizet C, Boukhris A, Durr A, Beetz C, Truchetto J, Tesson C, Tsaousidou M, Forlani S, Guyant-Marechal L, Fontaine B, Guimaraes J, Isidor B, Chazouilleres O, Wendum D, Grid D, Chevy F, **Chinnery PF**, Coutinho P, Azulay J-P, Feki I, Mochel F, Wolf C, Mhiri C, Crosby A, Brice A, Stevanin G. CYP7B1 mutations in pure and complex forms of hereditary spastic paraplegia type 5. *Brain* 2009 Jun;132(Pt 6):1589-600. PMID: 19439420

Mercuri E, Manzur A, Main M, Alsopp J, **Muntoni F**. Is there post-natal muscle growth in amyoplasia? A sequential MRI study. *Neuromuscul Disord*. 2009 Jun;19(6):444-5. PMID: 19477646

Chiu YH, Hornsey MA, Klinge L, Jørgensen LH, Laval SH, Charlton R, Barresi R, **Straub V, Lochmüller H, Bushby K. Attenuated muscle regeneration is a key factor in dysferlin-deficient muscular dystrophy. *Hum Mol Genet*. 2009 Jun 1;18(11):1976-89. Epub 2009 Mar 13. PMID: 19286669

Jørgensen LH, Larochelle N, Orlopp K, Dunant P, Dudley RW, Stucka R, Thirion C, Walter MC, Laval SH, **Lochmüller H**. Efficient and fast functional screening of microdystrophin constructs in vivo and in vitro for therapy of duchenne muscular dystrophy. *Hum Gene Ther*. 2009 Jun;20(6):641-50. PMID: 19239382.

Ghezzi D, Goffrini P, Uziel G, Horvath R, Klopstock T, **Lochmüller H**, D'Adamo P, Gasparini P, Strom TM, Prokisch H, Invernizzi F, Ferrero I, Zeviani M. SDHAF1, encoding a LYR complex-II specific assembly factor, is mutated in SDH-defective infantile leukoencephalopathy. *Nat Genet*. 2009 Jun;41(6):654-6. Epub 2009 May 24. PMID: 19465911.

Goehring C, Rutschow D, Bauer R, Schinkel S, Weichenhan D, Bekeredjian R, **Straub V**, Kleinschmidt JA, Katus HA, Müller OJ. Prevention of cardiomyopathy in delta-sarcoglycan knockout mice after systemic transfer of targeted adeno-associated viral vectors. *Cardiovasc Res*. 2009 Jun 1;82(3):404-10. Epub 2009 Feb 13. PMID: 19218289.

Greaves LC, Mathers JC, Taylor RW, **Turnbull DM**. Modelling mitochondrial DNA mutations in bacterial cytochrome c oxidase: link to colon cancer? *Proc Natl Acad Sci U S A*. 2009 Jun 2;106(22):E57. Epub 2009 May 15. PMID: 19447926

White KE, Davies VJ, Hogan VE, Piechota MJ, Nichols PP, **Turnbull DM**, Votruba M. OPA1 deficiency associated with increased autophagy in retinal ganglion cells in a murine model of dominant optic atrophy. *Invest Ophthalmol Vis Sci*. 2009 Jun;50(6):2567-71. Epub 2009 Feb 21. PMID: 19234344.

De Alwis N, Hudson G, Burt AD, Day CP, **Chinnery PF**. Human liver stem cells originate from the canals of Hering *Hepatology* 2009; Hepatology. 2009 Jul 7;50(3):992-993. PMID: 19714715

McNeill A, Birchall D, **Straub V, Goldfarb L, Reilich P, Walter MC, Schramm N, **Lochmüller H, Chinnery PF**. Lower Limb Radiology of Distal Myopathy due to the S60F Myotilin Mutation. *European Neurology* 2009 Jul 3;62(3):161-166. PMID: 19590214

Kirkman MA, Korsten A, Leonhardt M, Dimitriadis K, De Coo IF, Klopstock T, Griffiths PG, Hudson G, **Chinnery PF**, Yu-Wai-Man P. Quality of Life in Patients with Leber Hereditary Optic Neuropathy. *Investigative Ophthalmology and Visual Science* 2009 Jul;50(7):3112-5. PMID: 19255150

Campanella, M., Parker, N., Tan, C-H., Hall AM., **Duchen MR** (2009) IF1: setting the pace of the F₁F_o-ATP synthase, *Trends in Biochemical Sciences*, Jul;34(7):343-50. PMID: 19559621

Schorge S, **Kullmann DM**. Sodium channelopathy of peripheral nerve: tightening the genotype-phenotype relationship. *Brain*. 2009 Jul;132(Pt 7):1690-2. Epub 2009 May 8. PubMed PMID: 19429904.

Nadeau A, Kinali M, Main M, Jimenez Mallebrera C, Aloysius A, Clement E, North B, Manzur A, Robb S, Mercuri E, **Muntoni F**. Natural history of Ullrich congenital muscular dystrophy. *Neurology*, 2009 Jul 7;73(1):25-31. PMID: 19564581

Manzur AY, **Muntoni F**. Diagnosis and new treatments in muscular dystrophies. *J Neurol Neurosurg Psychiatry*. 2009 Jul;80(7):706-14. PMID: 19531685

Manzur AY, **Muntoni F**. Diagnosis and new treatments in muscular dystrophies. *Postgraduate Med J* 2009; 85(1009): 622-30. PMID: 19892898

****Bushby K**, Lynn S, **Straub V**; TREAT-NMD Network. Collaborating to bring new therapies to the patient-the TREAT-NMD model. *Acta Myol*. 2009 Jul;28(1):12-5. PMID: 19772190

Charlton R, Henderson M, Richards J, Hudson J, **Straub V, **Bushby K**, Barresi R. Immunohistochemical analysis of calpain 3: advantages and limitations in diagnosing LGMD2A. *Neuromuscul Disord*. 2009 Jul;19(7):449-57. Epub 2009 Jun 24. PMID: 19556129.

Giacomotto J, Pertl C, Borrel C, Walter MC, Bulst S, Johnsen B, Baillie DL, **Lochmüller H**, Thirion C, Ségalat L. Evaluation of the therapeutic potential of carbonic anhydrase inhibitors in two animal models of dystrophin deficient muscular dystrophy. *Hum Mol Genet*. 2009 Nov 1;18(21):4089-101. Epub 2009 Jul 31. PMID: 19648295.

Weraarpachai W, Antonicka H, Sasarman F, Seeger J, Schrank B, Kolesar JE, **Lochmüller H**, Chevrette M, Kaufman BA, Horvath R, Shoubridge EA. Mutation in TACO1, encoding a translational activator of COX I, results in cytochrome c oxidase deficiency and late-onset Leigh syndrome. *Nat Genet*. 2009 Jul;41(7):833-7. Epub 2009 Jun 7. PMID: 19503089.

Garcia-Angarita N, Kirschner J, Heiliger M, Thirion C, Walter MC, Schnitfeld-Acarlioglu S, Albrecht M, Müller K, Wieczorek D, **Lochmüller H**, Krause S. Severe nemaline myopathy associated with consecutive mutations E74D and H75Y on a single ACTA1 allele. *Neuromuscul Disord*. 2009 Jul;19(7):481-4. Epub 2009 Jun 23. PMID: 19553116.

Trender-Gerhard I, Sweeney MG, Schwingenschuh P, Mir P, Edwards MJ, Gerhard A, Polke JM, **Hanna MG**, Davis MB, Wood NW, Bhatia KP. Autosomal-dominant GTPCH1-deficient DRD: clinical characteristics and long-term outcome of 34 patients. *J Neurol Neurosurg Psychiatry*. 2009 Aug;80(8):839-45. Epub 2009 Mar 29 PMID: 19332422.

Mitropant C, Adams AM, Meloni PL, **Muntoni F**, Fletcher S, Wilton SD. Rational Design of Antisense Oligomers to Induce Dystrophin Exon Skipping. *Mol Ther*. 2009 Aug;17(8):1418-26. PMID: 19293776

Onopiuk M, Brutkowski W, Wierzbicka K, Wojciechowska S, Szczepanowska J, Fronk J, **Lochmüller H**, Górecki DC, Zabłocki K. Mutation in dystrophin-encoding gene affects energy metabolism in mouse myoblasts. *Biochem Biophys Res Commun*. 2009 Aug 28;386(3):463-6. Epub 2009 Jun 13. PMID: 19527684.

Reeve AK, Krishnan KJ, Taylor G, Elson JL, Bender A, Taylor RW, Morris CM, **Turnbull DM**. The low abundance of clonally expanded mitochondrial DNA point mutations in aged substantia nigra neurons. *Aging Cell*. 2009 Aug;8(4):496-8. Epub 2009 May 31. PMID: 19489744

Kraytsberg Y, Simon DK, **Turnbull DM**, Khrapko K. Do mtDNA deletions drive premature aging in mtDNA mutator mice? *Aging Cell*. 2009 Aug;8(4):502-6. Epub 2009 Apr 22. PMID: 19416127

Kirby DM, Rennie KJ, Smulders-Srinivasan TK, Acin-Perez R, Whittington M, Enriquez JA, Trevelyan AJ, **Turnbull DM**, Lightowlers RN. Transmitochondrial embryonic stem cells containing pathogenic mtDNA mutations are compromised in neuronal differentiation. *Cell Prolif*. 2009 Aug;42(4):413-24. Epub 2009 Jun 23. PMID: 19552636

Kirkman MA, Yu-Wai-Man P, Korsten A, Leonhardt M, Dimitriadis K, De Coo IF, Klopstock T, Chinnery

PF. Gene-Environment Interactions in Leber Hereditary Optic Neuropathy Brain 2009;Sep;132(Pt 9):2317-26 PMID: 19525327

Pavlov I, Walker MC, **Kullmann DM**. Computational sophistication at a single GABAergic connection. Neuron. 2009 Sep 24;63(6):716-8. PubMed PMID: 19778499.

Rajakulendran S, Tan SV, Matthews E, Tomlinson SE, Labrum R, Sud R, **Kullmann DM, Schorge S, **Hanna MG**. A patient with episodic ataxia and paramyotonia congenita due to mutations in KCNA1 and SCN4A. Neurology. 2009 Sep 22;73(12):993-5. PubMed PMID: 19770477

Tomlinson SE, **Hanna MG, **Kullmann DM**, Tan SV, Burke D. Clinical neurophysiology of the episodic ataxias: insights into ion channel dysfunction in vivo. Clin Neurophysiol. 2009 Oct;120(10):1768-76. Review. PubMed PMID: 19734086.

Rahman S, **Hanna MG**. Diagnosis and therapy in neuromuscular disorders: diagnosis and new treatments in mitochondrial diseases. J Neurol Neurosurg Psychiatry. 2009 Sep;80(9):943-53. Review. PMID: 19684231

Laing NG, Dye DE, Wallgren-Pettersson C, Richard G, Monnier N, Lillis S, Winder TL, **Lochmüller H**, Graziano C, Mitrani-Rosenbaum S, Twomey D, Sparrow JC, Beggs AH, Nowak KJ. Mutations and polymorphisms of the skeletal muscle alpha-actin gene (ACTA1). Hum Mutat. 2009 Sep;30(9):1267-77. PMID: 19562689

Borg K, Stucka R, Locke M, Melin E, Ahlberg G, Klutzny U, Hagen M, Huebner A, **Lochmüller H**, Wrogemann K, Thornell LE, Blake DJ, Schoser B. Intragenic deletion of TRIM32 in compound heterozygotes with sarcofibrillar myopathy/LGMD2H. Hum Mutat. 2009 Sep;30(9):E831-44. PMID: 19492423.

Bauer R, Hudson J, Müller HD, Sommer C, Dekomien G, Bourke J, Routledge D, Bushby K, Klepper J, **Straub V**. Does delta-sarcoglycan-associated autosomal-dominant cardiomyopathy exist? Eur J Hum Genet. 2009 Sep;17(9):1148-53. Epub 2009 Mar 4. PMID: 19259135

Ogundare O, Jumma O, **Turnbull DM**, Woywodt A. Searching for the needle in the Haystacks. Lancet. 2009 Sep 5;374(9692):850. PMID: 19733782.

Greaves LC, Beadle NE, Taylor GA, Commane D, Mathers JC, Khrapko K, **Turnbull DM**. Quantification of mitochondrial DNA mutation load. Aging Cell. 2009 Sep;8(5):566-72. Epub 2009 Jul 18. PMID: 19624578

Yu-Wai-Man P, Davies VJ, Piechota MJ, Cree LM, Votruba M, Chinnery PF. Secondary mtDNA defects do not cause optic nerve dysfunction in a mouse model of dominant optic atrophy. Investigative Ophthalmology & Visual Science 2009;Oct;50(10):4561-6. PMID: 19443720

Jimenez-Mallebrera C, Torelli S, Feng L, Kim J, Godfrey C, Clement E, Mein R, Abbs S, **Brown SC, Campbell KP, Kröger S, Talim B, Topaloglu H, Quinlivan R, Roper H, Childs AM, Kinali M, Sewry CA, **Muntoni F**. A comparative study of alpha-dystroglycan glycosylation in dystroglycanopathies suggests that the hypoglycosylation of alpha-dystroglycan does not consistently correlate with clinical severity. Brain Pathol. 2009 Oct;19(4):596-611. Epub 2008 Aug 7. PMID:18691338

Astrea G, Schessl J, Clement E, Tosetti M, Mercuri E, Rutherford M, Cioni G, Bönnemann CG, **Muntoni F**, Battini R. Muscle MRI in FHL1-linked reducing body myopathy. Neuromuscul Disord. 2009 Oct;19(10):689-91. PMID: 19616434

Read J, Kinali M, **Muntoni F**, Garralda ME. Psychosocial adjustment in siblings of young people with Duchenne muscular dystrophy. Eur J Paediatr Neurol. 2009 Oct 20.

O'Rourke K, Buddles MR, Farrell M, Howley R, Sukuraman S, Connolly S, **Turnbull DM**, Hutchinson M, Taylor RW. Phenotypic diversity associated with the mitochondrial m.8313G>A point mutation. Muscle Nerve. 2009 Oct;40(4):648-51. PMID: 19618438.

Samuels DC, Burn DJ, **Chinnery PF**. Detecting new neurodegenerative disease genes: does phenotype accuracy limit the horizon? *Trends in Genetics* 2009 Nov;25(11):486-8. PMID: 19819581

****Horvath R**, Kemp JP, Tuppen H, Hudson G, Oldfors A, Marie SKN, Moslemi A-R, Servidei S, Holme E, Shanske S, Kollberg G, Jayakar P, Pyle A, Marks AM, Holinski-Feder E, Scavina M, Walter MC, Çoku J, Günther-Scholz A, Smith PM, McFarland R, Chrzanowska-Lightowlers ZMA, Lightowlers RN, Hirano M, **Lochmüller H**, Taylor RW, **Chinnery PF**, Tulinius M, DiMauro S. Molecular basis of infantile reversible COX deficiency myopathy. *Brain* 2009 Nov;132(Pt 11):3165-74. PMID: 19720722

****Norwood FLM**, Harling C, **Chinnery PF**, Eagle M, **Bushby K**, **Straub V**. Prevalence of genetic muscle disease in Northern England: in-depth analysis of a muscle clinic population. *Brain* 2009; Nov;132(Pt 11):3175-86. PMID: 19767415

Pyle A, Ibbett IM, Gordon C, Keers SM, Walker M, **Chinnery PF**, Baudouin SC. A common UCP2 polymorphism predisposes to stress hyperglycaemia in severe sepsis. *Journal of Medical Genetics* 2009 Nov;46(11):773-5. PMID: 19556617

****Labrum RW**, Rajakulendran S, Graves TD, Eunson LH, Bevan R, Sweeney MG, Hammans SR, Tubridy N, Britton T, Carr LJ, Ostergaard JR, Kennedy CR, Al-Memmar A, **Kullmann DM**, Schorge S, Temple K, Davis MB, **Hanna MG**. Large scale calcium channel gene rearrangements in episodic ataxia and hemiplegic migraine: implications for diagnostic testing. *J Med Genet.* 2009 Nov;46(11):786-91. PubMed PMID: 19586927.

****Bushby K**, **Lochmüller H**, Lynn S, **Straub V**. Interventions for muscular dystrophy: molecular medicines entering the clinic. *Lancet.* 2009 Nov 28;374(9704):1849-56. Review. PMID: 19944865.

****Garrod P**, Hollingsworth KG, Eagle M, Aribisala BS, Birchall D, **Bushby K**, **Straub V**. MR imaging in Duchenne muscular dystrophy: quantification of T1-weighted signal, contrast uptake, and the effects of exercise. *J Magn Reson Imaging.* 2009 Nov;30(5):1130-8. PMID: 19856446.

Mahad DJ, Ziabreva I, Campbell G, Laulund F, Murphy JL, Reeve AK, Greaves L, Smith KJ, **Turnbull DM**. Detection of cytochrome c oxidase activity and mitochondrial proteins in single cells. *J Neurosci Methods.* 2009 Nov 15;184(2):310-9. Epub 2009 Aug 31. PMID: 19723540.

Elson JL, Swalwell H, Blakely EL, McFarland R, Taylor RW, **Turnbull DM**. Pathogenic mitochondrial tRNA mutations--which mutations are inherited and why? *Hum Mutat.* 2009 Nov;30(11):E984-92. PMID: 19718780.

Alavi MV, Fuhrmann N, Nguyen H P, Yu-Wai-Man P, Heiduschka P, **Chinnery PF**, Wissinger B. Subtle neurological and metabolic abnormalities in an Opa1 mouse model of autosomal dominant optic atrophy. *Experimental Neurology* 2009 Dec;220(2):404-9. PMID: 19815013

Cree LM, Samuels DC, P.F. **Chinnery PF**. The inheritance of pathogenic mitochondrial DNA mutations. *Biochimica et Biophysica Acta (Molecular basis of disease)* 2009 Dec;1792(12):1097-102. PMID: 19303927

Pavlov I, Savtchenko LP, **Kullmann DM**, Semyanov A, Walker MC. Outwardly rectifying tonically active GABAA receptors in pyramidal cells modulate neuronal offset, not gain. *J Neurosci.* 2009 Dec 2;29(48):15341-50. PubMed PMID: 19955387.

****Heeroma JH**, Henneberger C, Rajakulendran S, **Hanna MG**, Schorge S, **Kullmann DM**. Episodic ataxia type 1 mutations differentially affect neuronal excitability and transmitter release. *Dis Model Mech.* 2009 Nov-Dec;2(11-12):612-9. PubMed PMID: 19779067.

Bushby K. Diagnosis and management of the limb girdle muscular dystrophies. *Pract Neurol.* 2009 Dec;9(6):314-23. Review. PMID: 19923111.

Mihaylova V, Salih MA, Mukhtar MM, Abuzeid HA, El-Sadig SM, von der Hagen M, Huebner A, Nürnberg G, Abicht A, Müller JS, **Lochmüller H**, Guergueltcheva V. Refinement of the clinical

phenotype in musk-related congenital myasthenic syndromes. *Neurology*. 2009 Dec 1;73(22):1926-8. PMID: 19949040.

Thompson R, Schoser B, Monckton DG, Blonsky K, **Lochmüller H**. Patient Registries and Trial Readiness in Myotonic Dystrophy--TREAT-NMD/Marigold International Workshop Report. *Neuromuscul Disord*. 2009 Dec;19(12):860-6. Epub 2009 Oct 20. PMID: 19846307.

Schara U, Barisic N, Deschauer M, Lindberg C, **Straub V, Strigl-Pill N, Wendt M, Abicht A, Müller JS, **Lochmüller H**. Ephedrine therapy in eight patients with congenital myasthenic syndrome due to DOK7 mutations. *Neuromuscul Disord*. 2009 Dec;19(12):828-32. Epub 2009 Oct 17. PMID: 19837590.

Al-Dosary M, Whittaker RG, Haughton J, McFarland R, Goodship J, **Turnbull DM**, Taylor RW. Neuromuscular disease presentation with three genetic defects involving two genomes. *Neuromuscul Disord*. 2009 Dec;19(12):841-4. Epub 2009 Oct 22. PMID: 19853445.

Hardy J, Trabzuni D, Ryten M. Whole genome expression as a quantitative trait. *Biochem Soc Trans*. 2009 Dec;37(Pt 6):1276-7. PMID 19909261

Matthews E, Fialho D, Tan SV, Venance SL, Cannon SC, Sternberg D, Fontaine B, Amato AA, Barohn RJ, Griggs RC, **Hanna MG**; CINCH Investigators. The non-dystrophic myotonias: molecular pathogenesis, diagnosis and treatment. *Brain*. 2010 Jan;133(Pt 1):9-22. Epub 2009 Nov 16. Review. PMID: 19917643

Hilton-Jones D, Miller A, Parton M, Holton J, Sewry C, **Hanna MG**. Inclusion body myositis: MRC Centre for Neuromuscular Diseases, IBM workshop, London, 13 June 2008. *Neuromuscul Disord*. 2010 Feb;20(2):142-7. Epub 2010 Jan 13. PMID: 20074951

Cooper FE, Grube M, Elsegood KJ, Welch JL, Kelly TP, **Chinnery PF**, Griffiths TD. The contribution of the cerebellum to cognition in Spinocerebellar Ataxia Type 6. *Behavioural Neurology* 2010;23(1-2):3-15. PMID: 20714057

Batey S, Vuillaume I, Devos D, Destée A, Curtis AJ, Lombes A, Curtis A, Burn J, **Chinnery PF**. A novel FTL insertion causing neuroferritinopathy. *Journal of Medical Genetics* 2010 Jan;47(1):71-2. PMID: 20065344

Hassani A, Horvath R, **Chinnery PF**. Mitochondrial Myopathies: Developments in treatment. *Current Opinion in Neurology* 2010. PMID: 20651591

Yu-Wai-Man P, Bailie M, Atawan A, **Chinnery PF**, Griffiths PG. Pattern of Retinal Ganglion Cell Loss in Dominant Optic Atrophy due to OPA1 Mutations. *Eye* 2010: PMID:21478995

Andrey Y. Abramov⁵, Tora K. Smulders-Srinivasan², Denise M. Kirby^{2,3}, Rebeca Acin-Perez⁴, José Antonio Enriquez⁴, Robert N. Lightowlers², **Michael R. Duchen*¹, **Doug M. Turnbull***² (2010) Mechanism of neurodegeneration of neurons with mitochondrial DNA mutations, *Brain*, 133(Pt 3):797-807

Duchen MR, Szabadkai G. Roles of mitochondria in human disease. *Essays Biochem*. 2010;47:115-37

Hall AM, Campanella M, Loesch A, Duchen MR, Unwin RJ. Albumin uptake in OK cells exposed to rotenone: a model for studying the effects of mitochondrial dysfunction on endocytosis in the proximal tubule? *Nephron Physiol*. 2010;115(2):p9-p19.

Ono, Y., Boldrin, L., Knopp, P., **Morgan, JE**, Zammit, P. S. (2010). Muscle satellite cells are a functionally heterogeneous population in both somite-derived and branchiomeric muscles. *Dev.Biol*. 337(1):29-41. PMID: 19835858

Poplewell, L. J., Adkin, C., Arechavala-Gomez, V., Aartsma-Rus, A., De Winter, C. L., Wilton, S. D., **Morgan, JE, **Muntoni, F.**, Graham, I. R., Dickson, G. (2010). Comparative analysis of antisense oligonucleotide sequences targeting exon 53 of the human DMD gene: Implications for future clinical trials. *Neuromuscul Disord* 20(2), 102-110. PMID: 20079639

- **Meng, J., Adkin, C. F., Arechavala-Gomez, V., Boldrin, L., Muntoni, F., Morgan, JE (2010).** The contribution of human synovial stem cells to skeletal muscle regeneration. *Neuromuscul Disord* 20(1), 6-15. PMID: 20034794
- **Arechavala-Gomez, V., Kinali, M., Feng, L., Guglieri, M., Edge, G., Main, M., Hunt, D., Lehovsky, J., Straub, V., Bushby, K., Sewry, C. A., Morgan, JE, Muntoni, F. (2010).** Revertant fibres and dystrophin traces in Duchenne muscular dystrophy: implication for clinical trials. *Neuromuscul Disord* 20(5), 295-301. PMID: 20395141
- **Arechavala-Gomez, V., Kinali, M., Feng, L., Brown, S. C., Sewry, C., Morgan, JE, Muntoni, F. (2010).** Immunohistological intensity measurements as a tool to assess sarcolemma-associated protein expression. *Neuropathol Appl Neurobiol* 36(4), 265-27. PMID: 20002311
- Morgan J, Rouche A., Bausero P., Houssaïni A., Gross J., Fiszman M.Y., Alameddine H.S (2010).** MMP-9 overexpression improves myogenic cell migration and engraftment. *Muscle Nerve* 42(4):584-95. PMID: 20734311
- Mirsky R. and **Jessen KR.** *F1000 Biol Rep.* 2010 2: 19. PMID: 20948814
- **Rajakulendran S, Schorge S, Kullmann DM, Hanna MG.** Dysfunction of the Ca(V)2.1 calcium channel in cerebellar ataxias. *F1000 Biol Rep.* 2010 Jan 18;2. pii: 4. PubMed PMID: 20948794
- Kullmann DM.** Neurological channelopathies. *Annu Rev Neurosci.* 2010;33:151-72. Review. PubMed PMID: 20331364.
- **Saifee TA, Elliott KJ, Lunn MP, Blake J, Rabin N, Yong KL, D'Sa S, Brandner S, Reilly MM.** Bortezomib-induced inflammatory neuropathy. 2010: 15; 366-368. PMID 21199108
- **Reilly MM, Shy M, Muntoni F, Pareyson D.** 168th ENMC International workshop: Outcome measures and clinical trials in Charcot Marie Tooth disease (CMT). 168th ENMC International Workshop: Outcome measures and clinical trials in Charcot-Marie-Tooth disease (CMT). *Neuromuscul Disord* 2010 20(12):839-46. PMID 20850975
- Zimon M, Baets J, Auer-Grumbach M, Berciano J, Garcia A, Lopez-Laso E, Merlini L, Hilton-Jones D, McEntagart M, Crosby AH, Barisic N, Boltshauser E, Shaw CE, Landouze G, Ludlow CL, Gaudet R, Houlden H, **Reilly MM**, Fischbeck CJ, Timmermann V, Jordonova A, Jonghe PD. Dominant mutations in the cation channel gene transient receptor potential vanilloid 4 causes an unusual spectrum of neuropathies. *Brain* 2010: 133; 1789-809. PMID 20460441
- Khaleeli Z, Healy DG, Bridson A, Lunn M, **Reilly MM**, Land J, Giovannoni G. Copper deficiency as a treatable cause of poor balance. *BMJ* 2010: 340; c508. PMID 20385722
- Penno A, **Reilly MM**, Houlden H, Laura M, Rentsch K, Niederkofler V, Stoeckli ET, Nicholson G, Eichler F, Brown RH, Von Eckardstein A, Hornemann T. Hereditary sensory neuropathy type 1 is caused by the accumulation of two neurotoxic sphingolipids. *JBC* 2010: 285; 11178-87. PMID 20097765
- Marsh EA, Hirst CL, Llewellyn JG, **Reilly MM**, Krishnan A, Doran M, Ryan AM, Coles AJ, Jones JL, Robertson NP. Alemtuzumab in the treatment of IVIg-dependent chronic inflammatory demyelinating polyneuropathy. *J of Neurol* 2010: 257; 913-919. PMID 20049473
- Roberts RC, Peden AA, Buss F, Bright NA, Latouche M, **Reilly MM**, Kendrick-Jones J, Luzio JP. Mistargeting of SH3TC2 away from the recycling endosome causes Charcot-Marie-Tooth disease type 4C. *Hum Mol Genet* 2010: 19; 1009-1018. PMID 20028792
- Reilly MM.** NEFL-related Charcot-Marie-Tooth disease (editorial). *Ann Neurol* 2010: 66: 714-716. PMID 20033987
- Hobart J, Cano S, **Thompson A.** Effect sizes can be misleading: is it time to change the way we measure change? *J Neurol Neurosurg Psychiatr* 2010;81(9):1044-1048. PMID 20627964

Jungbluth H, Cullup T, Lillis S, Zhou H, Abbs S, Sewry CA, **Muntoni F**. Centronuclear myopathy with cataracts due to a novel dynamin 2 (DNM2) mutation. *Neuromuscular Disorders*, 2010 Jan;20(1):49-52. PMID: 19932620

Wamsley GL, Arechavala-Gomez V, Fernandez-Fuente M, Burke MM, Nagel N, Holder A, Stanley R, Chandler K, Marks SL, **Muntoni F**, Shelton GD, Piercy RY. A duchenne muscular dystrophy gene hot spot mutation in dystrophin-deficient cavalier king charles spaniels is amenable to exon 51 skipping. *PLoS One*. 2010 Jan 13;5(1):e8647. PMID: 20072625

Bushby K. Lifeline. An interview of Kate Bushby. *Lancet Neurol*. 2010 Jan;9(1):37. PMID: 20083037.

Bushby K, Finkel R, Birnkrant DJ, Case LE, Clemens PR, Cripe L, Kaul A, Kinnett K, McDonald C, Pandya S, Poysky J, Shapiro F, Tomezsko J, Constantin C; DMD Care Considerations Working Group. Diagnosis and management of Duchenne muscular dystrophy, part 1: diagnosis, and pharmacological and psychosocial management. *Lancet Neurol*. 2010 Jan;9(1):77-93. Epub 2009 Nov 27. Review. PMID: 19945913.

Bauer R, Blain A, Greally E, Lochmüller H, **Bushby K, MacGowan GA, **Straub V**. Attenuation of adverse cardiac effects in prednisolone-treated delta-sarcoglycan-deficient mice by mineralocorticoid-receptor-antagonism. *Neuromuscul Disord*. 2010 Jan;20(1):21-8. Epub 2009 Nov 12. PMID: 19913415.

Greaves LC, Barron MJ, Plusa S, Kirkwood TB, Mathers JC, Taylor RW, **Turnbull DM**. Defects in multiple complexes of the respiratory chain are present in ageing human colonic crypts. *Exp Gerontol*. 2010 Aug;45(7-8):573-9. Epub 2010 Jan 22. PMID: 20096767

D Boërio, B Kalmar, **L Greensmith** and H. Bostock (2010) Changes in excitability of mouse motor axons in the mutant SOD1^{G93A} model of amyotrophic lateral sclerosis. *Muscle and Nerve* 41(6):774-84 PMID 20095022

JC Stevens, R Chia, WT Hendriks, V Bros-Facer, J van Minnen, JE. Martin, GS Jackson, **L Greensmith, G Schiavo, and **EMC Fisher** (2010) Modification of superoxide dismutase 1 (SOD1) properties by a GFP tag – implications for research into amyotrophic lateral sclerosis (ALS) *PLoS One* 2010 Mar 8;5(3):e9541 PMID 20221404

AC Ludolph, C Bendotti, E Blaugrund, A Chio, **L Greensmith**, JP Loeffler, R Mead, HG Niessen, S Petri, PF Pradat PF, W Robberecht, M Ruegg, B Schwalenstöcker, D Stiller, L van den Berg, F Vieira and S von Horsten (2010) Guidelines for preclinical animal research in ALS/MND: A consensus meeting. *Amyotrophic Lateral Sclerosis* 11(1-2):38-45 PMID 20184514

AM El-Kadi, V Bros-Facer, W Deng, A Philpott, E Stoddart, G Banks, GS Jackson, **EMC Fisher, MR Duchon, **L Greensmith**, Al Moore and M Hafezparast (2010) The legs at odd angles (*loa*) mutation in cytoplasmic dynein ameliorates mitochondrial function in SOD1G93A mouse model for Motor Neuron Disease *Journal of Biological Chemistry* 285 (24):18627-39 PMID 20382740

LG Bilsland, E. Sahai, G Kelly, M Golding, **L. Greensmith** and G. Schiavo (2010) Deficits in axonal transport precede ALS symptoms in vivo *Proc.Natl.Acad.Sci USA*.107(47):20523-8 PMID 21059924

Yu-Wai-Man P, Stewart JD, Hudson G, Andrews RM, Griffiths PG, Birch MK, **Chinnery PF** OPA1 increases the risk of normal but not high tension glaucoma. *Journal of Medical Genetics* 2010 Feb;47(2):120-5. PMID: 19581274

Walker MC, Pavlov I, **Kullmann DM**. A 'sustain pedal' in the hippocampus? *Nat Neurosci*. 2010 Feb;13(2):146-8. PubMed PMID: 20104205.

Davies G, Irani SR, Coltart C, Ingle G, Amin Y, Taylor C, Radcliffe J, Hirsch NP, Howard RS, Vincent A, **Kullmann DM**. Anti-N-methyl-D-aspartate receptor antibodies: a potentially treatable cause of encephalitis in the intensive care unit. *Crit Care Med*. 2010 Feb;38(2):679-82. PubMed PMID: 20016378

Tomlinson S, Burke D, **Hanna M, **Koltzenburg M**, Bostock H. In vivo assessment of HCN channel current (I_h) in human motor axons. *Muscle Nerve*. 2010 Feb;41(2):247-56. PMID: 19813191

Limongelli G, Tome-Esteban M, Dejthevaporn C, Rahman S, **Hanna MG**, Elliott PM. Prevalence and natural history of heart disease in adults with primary mitochondrial respiratory chain disease. *Eur J Heart Fail*. 2010 Feb;12(2):114-21. PMID: 20083621

Mercuri E, Clements E, Offiah A, Pichiecchio A, Vasco G, Bianco F, Berardinelli A, Manzur A, Pane M, Messina S, Gualandi F, Ricci E, Rutherford M, **Muntoni F**. Muscle MRI involvement in muscular dystrophies with rigidity of the spine. *Annals of Neurology*. 2010 Feb;67(2):201-8. PMID: 20225280

Klinge L, Harris J, Sewry C, Charlton R, Anderson L, Laval S, Chiu YH, Hornsey M, **Straub V, Barresi R, **Lochmüller H**, **Bushby K**. Dysferlin associates with the developing T-tubule system in rodent and human skeletal muscle. *Muscle Nerve*. 2010 Feb;41(2):166-73. PMID: 20082313.

Lu PJ, Zillmer A, Wu X, **Lochmuller H**, Vachris J, Blake D, Chan YM, Lu QL. Mutations alter secretion of fukutin-related protein. *Biochim Biophys Acta*. 2010 Feb;1802(2):253-8. Epub 2009 Nov 10. PMID: 19900540.

Auer-Grumbach M, Olschewski A, Papić L, Kremer H, McEntagart ME, Uhrig S, Fischer C, Fröhlich E, Bálint Z, Tang B, Strohmaier H, **Lochmüller H**, Schlotter-Weigel B, Senderek J, Krebs A, Dick KJ, Petty R, Longman C, Anderson NE, Padberg GW, Schelhaas HJ, van Ravenswaaij-Arts CM, Pieber TR, Crosby AH, Guelly C. Alterations in the ankyrin domain of TRPV4 cause congenital distal SMA, scapuloperoneal SMA and HMSN2C. *Nat Genet*. 2010 Feb;42(2):160-4. Epub 2009 Dec 27. PMID: 20037588.

Lochmüller H, Schneiderat P. Biobanking in rare disorders. *Adv Exp Med Biol*. 2010;686:105-13. Review. PMID: 20824442.

Nooteboom M, Johnson R, Taylor RW, Wright NA, Lightowlers RN, Kirkwood TB, Mathers JC, **Turnbull DM**, Greaves LC. Age-associated mitochondrial DNA mutations lead to small but significant changes in cell proliferation and apoptosis in human colonic crypts. *Aging Cell*. 2010 Feb;9(1):96-9. Epub 2009 Oct 30. PMID: 19878146

Tuppen HA, Blakely EL, **Turnbull DM**, Taylor RW. Mitochondrial DNA mutations and human disease. *Biochim Biophys Acta*. 2010 Feb;1797(2):113-28. Epub 2009 Sep 15. Review. PMID: 19761752.

Krishnan KJ, **Turnbull DM**. Mitochondrial DNA and genetic disease. *Essays Biochem*. 2010;47:139-51. Review. PMID: 20533905.

Krishnan KJ, Blackwood JK, Reeve AK, **Turnbull DM**, Taylor RW. Detection of mitochondrial DNA variation in human cells. *Methods Mol Biol*. 2010;628:227-57. PMID: 20238085.

Van Deerlin VM, Sleiman PM, Martinez-Lage M, Chen-Plotkin A, Wang LS, Graff-Radford NR, Dickson DW, Rademakers R, Boeve BF, Grossman M, Arnold SE, Mann DM, Pickering-Brown SM, Seelaar H, Heutink P, van Swieten JC, Murrell JR, Ghetti B, Spina S, Grafman J, Hodges J, Spillantini MG, Gilman S, Lieberman AP, Kaye JA, Woltjer RL, Bigio EH, Mesulam M, Al-Sarraj S, Troakes C, Rosenberg RN, White CL 3rd, Ferrer I, Lladó A, Neumann M, Kretschmar HA, Hulette CM, Welsh-Bohmer KA, Miller BL, Alzualde A, de Munain AL, McKee AC, Gearing M, Levey AI, Lah JJ, **Hardy J**, Rohrer JD, Lashley T, Mackenzie IR, Feldman HH, Hamilton RL, Dekosky ST, van der Zee J, Kumar-Singh S, Van Broeckhoven C, Mayeux R, Vonsattel JP, Troncoso JC, Kril JJ, Kwok JB, Halliday GM, Bird TD, Ince PG, Shaw PJ, Cairns NJ, Morris JC, McLean CA, Decarli C, Ellis WG, Freeman SH, Frosch MP, Growdon JH, Perl DP, Sano M, Bennett DA, Schneider JA, Beach TG, Reiman EM, Woodruff BK, Cummings J, Vinters HV, Miller CA, Chui HC, Alafuzoff I, Hartikainen P, Seilhean D, Galasko D, Masliah E, Cotman CW, Tuñón MT, Martínez MC, Muñoz DG, Carroll SL, Marson D, Riederer PF, Bogdanovic N, Schellenberg GD, Hakonarson H, Trojanowski JQ, Lee VM. Common variants at 7p21 are associated with frontotemporal lobar degeneration with TDP-43 inclusions. *Nat Genet*. 2010 Feb 14. PMID 20154673

Guerreiro RJ, Washecka N, **Hardy J**, Singleton A. A thorough assessment of benign genetic variability in GRN and MAPT. *Hum Mutat*. 2010 Feb;31(2):E1126-40. PMID 20020531

****Zhou H, Lillis S, Loy RE, Ghassemi F, Rose MR, Norwood F, Mills K, Al-Sarraj S, Lane RJ, Feng L, Matthews E, Sewry CA, Abbs S, Buk S, Hanna M, Treves S, Dirksen RT, Meissner G, Muntoni F, Jungbluth H.** Multi-minicore disease and atypical periodic paralysis associated with novel mutations in the skeletal muscle ryanodine receptor (RYR1) gene. *Neuromuscul Disord.* 2010 Mar;20(3):166-73. Epub 2010 Jan 18. PMID: 20080402

Yu Wai Man P, **Chinnery PF.** Heterozygous OPA1 mutations and Behr Syndrome. *Brain* 2010; Apr;134(Pt 4):e169; Chinnery PF. Defining neurogenetic phenotypes (or how to compare needles in haystacks). *Brain* 2010 Mar;133(Pt 3):649-51. PMID: 20157007

****Yu-Wai-Man P, Griffiths PG, Gorman GS, Lourenco CM, Wright AF, Auer-Grumbach M, Toscano A, Musumeci O, Valentino ML, Caporali L, Lamperti C, Tallaksen CM, Duffey P, Miller J, Whittaker RG, Baker MR, Jackson MJ, Clarke MP, Dhillon B, Czermin B, Stewart JD, Hudson G, Reynier P, Bonneau P, Marques Jr W, Lenaers G, McFarland R, Taylor RW, Turnbull DM, Votruba M, Zeviani M, Carelli V, Bindoff LA, Horvath R, Amati-Bonneau P, Chinnery PF.** Multi-system neurological disease is common in patients with OPA1 mutations. *Brain* 2010 Mar;133(Pt 3):771-86. PMID: 20157015

Zhou H, Lillis S, Loy RE, Ghassemi F, Rose MR, Norwood F, Mills K, Al-Sarraj S, Lane RJ, Feng L, Matthews E, Sewry CA, Abbs S, Buk S, Hanna M, Treves S, Dirksen RT, Meissner G, **Muntoni F, Jungbluth H.** Multi-minicore disease and atypical periodic paralysis associated with novel mutations in the skeletal muscle ryanodine receptor (RYR1) gene. *Neuromuscul Disord.* 2010 Mar;20(3):166-73. PMID: 20080402

Bushby K, Finkel R, Birnkrant DJ, Case LE, Clemens PR, Cripe L, Kaul A, Kinnett K, McDonald C, Pandya S, Poysky J, Shapiro F, Tomezsko J, Constantin C; DMD Care Considerations Working Group. Diagnosis and management of Duchenne muscular dystrophy, part 2: implementation of multidisciplinary care. *Lancet Neurol.* 2010 Feb;9(2):177-89. Epub 2009 Nov 27. Review. Erratum in: *Lancet Neurol.* 2010 Mar;9(3):237. PMID: 19945914.

Rajab A, **Straub V, McCann LJ, Seelow D, Varon R, Barresi R, Schulze A, Lucke B, Lützkendorf S, Karbasiyan M, Bachmann S, Spuler S, Schuelke M.** Fatal cardiac arrhythmia and long-QT syndrome in a new form of congenital generalized lipodystrophy with muscle rippling (CGL4) due to PTRF-CAVIN mutations. *PLoS Genet.* 2010 Mar 12;6(3):e1000874. PMID: 20300641

Goecke TW, Ekici AB, Niesler B, Loehberg CR, Hammer C, Rappold G, Schanze D, **Straub V, Altmann HH, Strissel P, Strick R, Beckmann MW, Fasching PA.** Two naturally occurring variants of the serotonin receptor gene HTR3C are associated with nausea in pregnancy. *Acta Obstet Gynecol Scand.* 2010;89(1):7-14. PMID: 20021265.

Blackwood JK, Whittaker RG, Blakely EL, Alston CL, **Turnbull DM, Taylor RW.** The investigation and diagnosis of pathogenic mitochondrial DNA mutations in human urothelial cells. *Biochem Biophys Res Commun.* 2010 Mar 19;393(4):740-5. Epub 2010 Feb 18. PMID: 20171163.

Trevelyan AJ, Kirby DM, Smulders-Srinivasan TK, Nooteboom M, Acin-Perez R, Enriquez JA, Whittington MA, Lightowlers RN, **Turnbull DM.** Mitochondrial DNA mutations affect calcium handling in differentiated neurons. *Brain.* 2010 Mar;133(Pt 3):787-96. Epub 2010 Mar 5. PMID: 20207702

Houlden H, Schneider SA, Paudel R, Melchers A, Schwingenschuh P, Edwards M, **Hardy J, Bhatia KP.** THAP1 mutations (DYT6) are an additional cause of early-onset dystonia. *Neurology.* 2010 Mar 9;74(10):846-50. PMID 20211909

Rajakulendran S, Kuntzer T, Dunand M, Yau SC, Ashton EJ, Storey H, McCauley J, Abbs S, Thonney F, Leturcq F, Lobrinus JA, Yousry T, Farmer S, Holton JL, **Hanna MG.** Marked hemiatrophy in carriers of Duchenne muscular dystrophy. *Arch Neurol.* 2010 Apr;67(4):497-500. PMID: 20385919.

****Walter MC, Czermin B, Muller-Ziermann S, Bulst S, Stewart JD, Hudson G, Schneiderat P, Abicht A, Holinski-Feder E, Lochmüller H, Chinnery PF, Klopstock T, Horvath R.** Late-onset ptosis and myopathy in a patient with a heterozygous insertion in POLG2. *Journal of Neurology* 2010 Apr 20. PMID: 20405137

Wonnapijit P, **Chinnery PF**, Samuels DC. Previous estimates of mitochondrial DNA mutation level variance did not account for sampling error: Comparing the mtDNA genetic bottleneck in mice and humans. *American Journal of Human Genetics* 2010 Apr 9;86(4):540-50. PMID: 20362273

Yu-Wai-Man P, Griffiths PG, Burke A, Sellar PW, Clarke MP, Gnanaraj L, Ah-Kine D, Hudson G, Czermin B, Taylor RW, Horvath R, **Chinnery PF**. The Prevalence and Natural History of Dominant Optic Atrophy due to OPA1 Mutations. *Ophthalmology* Aug;117(8):1538-46, 1546.e1. Epub 2010 Apr 24. PMID: 20417570

Igosheva N, Abramov AY, Poston L, Eckert JJ, Fleming TP, **Duchen MR**, McConnell J. Maternal diet-induced obesity alters mitochondrial activity and redox status in mouse oocytes and zygotes. *PLoS One*. 2010 Apr 9;5(4):e10074.

Ng KE, Schwarzer S, **Duchen MR**, Tinker A. The intracellular localization and function of the ATP-sensitive K⁺ channel subunit Kir6.1. *J Membr Biol*. 2010 Apr;234(2):137-47.

Ruiz A, Campanac E, Scott RS, Rusakov DA, **Kullmann DM**. Presynaptic GABAA receptors enhance transmission and LTP induction at hippocampal mossy fiber synapses. *Nat Neurosci*. 2010 Apr;13(4):431-8. Epub 2010 Mar 21. PubMed PMID: 20305647

Geranmayeh F, Clement E, Feng LH, Sewry C, Pagan J, Diprcpath RM, Abbs S, Brueton L, Childs AM, Jungbluth H, Goede CG, Lynch B, Lin JP, Chow G, Sousa CD, O'Mahony O, Majumdar A, Bushby K, **Muntoni F**. Genotype-Phenotype correlation in a large population of muscular dystrophy patients with LAMA2 mutations. *Neuromuscul Disord*. 2010 Apr;20(4):241-50. PMID: 20207543

Levin J, Bulst S, Thirion C, Schmidt F, Bötzel K, Krause S, Pertl C, Kretzschmar H, Walter MC, Giese A, **Lochmüller H**. Divergent molecular effects of desmin mutations on protein assembly in myofibrillar myopathy. *J Neuropathol Exp Neurol*. 2010 Apr;69(4):415-24. PMID: 20448486.

Reilich P, Schoser B, Schramm N, Krause S, Schessl J, Kress W, Müller-Höcker J, Walter MC, **Lochmüller H**. The p.G154S mutation of the alpha-B crystallin gene (CRYAB) causes late-onset distal myopathy. *Neuromuscul Disord*. 2010 Apr;20(4):255-9. Epub 2010 Feb 19. PMID: 20171888.

van de Leemput J, Wavrant-De Vrièze F, Rafferty I, Bras JM, Giunti P, **Fisher EM, Hardy JA, Singleton AB, Houlden H Sequencing analysis of the ITPR1 gene in a pure autosomal dominant spinocerebellar ataxia series. *Mov Disord*. 2010 Apr 30;25(6):763-5. PMID 20437544

Hardy J. Neurological diagnoses identify molecular processes. *Arch Neurol*. 2010 Apr;67(4):400-1 PMID 20385904

Yu-Wai-Man P, Sitarz KS, Samuels DC, Griffiths PG, Reeve AK, Bindoff LA, Horvath R, **Chinnery PF**. OPA1 mutations cause cytochrome c oxidase deficiency due to loss of wild-type mtDNA molecules. *Human Molecular Genetics* 2010 Aug 1;19(15):3043-52. Epub 2010 May 18. PMID: 20484224

Craven L, Tuppen HA, Greggains GD, Harbottle SJ, Murphy JL, Cree LM, Murdoch AP, **Chinnery PF, Taylor RW, Lightowlers RN, Herbert M, **Turnbull DM**. Pronuclear transfer in human embryos to prevent transmission of mitochondrial DNA disease. *Nature* 2010 May 6;465(7294):82-5. PMID: 20393463

Chinnery PF, Elliott HR, Syed A, Rothwell P Mitochondrial DNA sub-haplogroup K reduces the risk of transient ischaemic attack and ischaemic stroke. *Lancet Neurology* 2010 May;9(5):498-503. PMID: 20362514. Accompanying commentaries in *Lancet Neurology*, *New Scientist*, and *Nature Reviews Neurology*.

Fratton C, Gorman GS, Stewart JD, Buddles M, Smith C, Evans J, Sellar A, Poulton J, Roberts M, **Hanna MG, Rahman S, Omer SE, Klopstock T, Schoser B, Kornblum K, Czermin B, Lecky B, Blakely EL, Craig K, **Chinnery PF, Turnbull DM, Horvath R**, Taylor RW. The clinical, histochemical and molecular spectrum of PEO1 (Twinkle)-linked adPEO. *Neurology* 2010 May 18;74(20):1619-26. PMID: 20479361

Michot C, Hubert L, Brivet M, De Meirleir L, Valayannopoulos V, Müller-Felber W, Venkateswaran R, Ogier H, Desguerre I, Altuzarra C, Thompson E, Smitka M, Huebner A, Husson M, Horvath R, **Chinnery P**, Vaz FM, Munnich A, Elpeleg O, Delahodde A, de Keyser Y, de Lonlay P. LPIN1 gene mutations: a

major cause of severe rhabdomyolysis in early childhood. *Human Mutation* 2010 May 17;31(7):E1564-E1573. PMID: 20583302

Averaimo S, Milton RH, **Duchen MR**, Mazzanti M. Chloride intracellular channel 1 (CLIC1): Sensor and effector during oxidative stress. *FEBS Lett.* 2010 May 17;584(10):2076-84.

Clarke NF, Waddell LB, Cooper ST, Perry M, Smith RL, Kornberg AJ, **Muntoni F, Lillis S, **Straub V**, **Bushby K**, Guglieri M, King MD, Farrell MA, Marty I, Lunardi J, Monnier N, North KN. Recessive mutations in RYR1 are a common cause of congenital fibre type disproportion. *Hum Mutat.* 2010 May 11;31(7):E1544-E1550. PMID: 20583297

Muntoni F. The development of antisense oligonucleotide therapies for Duchenne muscular dystrophy: Report on a TREAT-NMD workshop hosted by the European Medicines Agency (EMA), on September 25th 2009. *Neuromuscul Disord.* 2010 May;20(5):355-62. PMID: 20347306

Müller JS, Jepson CD, Laval SH, **Bushby K, **Straub V**, **Lochmüller H**. Dok-7 promotes slow muscle integrity as well as neuromuscular junction formation in a zebrafish model of congenital myasthenic syndromes. *Hum Mol Genet.* 2010 May 1;19(9):1726-40. Epub 2010 Feb 10. PMID: 20147321.

Allamand V, Merlini L, **Bushby K**; Consortium for Collagen VI-Related Myopathies. 166th ENMC International Workshop on Collagen type VI-related Myopathies, 22-24 May 2009, Naarden, The Netherlands. *Neuromuscul Disord.* 2010 May;20(5):346-54. Epub 2010 Mar 7. PMID: 20211562.

Stober A, Aleo A, Kuhl V, Bornemann A, Walter MC, **Lochmüller H**, Lindner A, Krause S. Novel missense mutation p.A310P in the GNE gene in autosomal-recessive hereditary inclusion-body myopathy/distal myopathy with rimmed vacuoles in an Italian family. *Neuromuscul Disord.* 2010 May;20(5):335-6. Epub 2010 Mar 25. PMID: 20346669.

Escher C, **Lochmüller H**, Fischer D, Frank S, Reimann J, Walter MC, Ehrat M, Ruegg MA, Gygyax D. Reverse protein arrays as novel approach for protein quantification in muscular dystrophies. *Neuromuscul Disord.* 2010 May;20(5):302-9. Epub 2010 Mar 20. PMID: 20304647.

Craven L, Tuppen HA, Greggains GD, Harbottle SJ, Murphy JL, Cree LM, Murdoch AP, **Chinnery PF, Taylor RW, Lightowlers RN, Herbert M, **Turnbull DM**. Pronuclear transfer in human embryos to prevent transmission of mitochondrial DNA disease. *Nature.* 2010 May 6;465(7294):82-5. Epub 2010 Apr 14. PMID: 20393463

Pohjoismäki JL, Goffart S, Taylor RW, **Turnbull DM**, Suomalainen A, Jacobs HT, Karhunen PJ. Developmental and pathological changes in the human cardiac muscle mitochondrial DNA organization, replication and copy number. *PLoS One.* 2010 May 3;5(5):e10426. PMID: 20454654

Schorge S, van de Leemput J, Singleton A, Houlden H, **Hardy J**. Human ataxias: a genetic dissection of inositol triphosphate receptor (ITPR1)-dependent signaling. *Trends Neurosci.* 2010 May;33(5):211-9. PMID 20226542

Matthews E, **Hanna MG**. Muscle channelopathies: does the predicted channel gating pore offer new treatment insights for hypokalaemic periodic paralysis? *J Physiol.* 2010 Jun 1;588(Pt 11):1879-86. Epub 2010 Feb 1. Review. PMID:20123788

Grube M, Cooper FE, **Chinnery PF**, Griffiths TD. Dissociation of duration-based and beat-based auditory timing in cerebellar degeneration. *Proceedings of the National Academy of Sciences USA* 2010 Jun 22;107(25):11597-601. PMID: 20534501

Isaacs JD, **Chinnery PF**. Translating cutting edge science into novel and effective therapies. *Current Opinion in Pharmacology* 2010 Jun;10(3):300-1. PMID: 20627681

Ritchie AE, Griffiths PG, **Chinnery PF**, Davidson AW. Eye movement recordings to investigate a supranuclear component in chronic progressive external ophthalmoplegia: a cross-sectional study. *British Journal of Ophthalmology* 2010 Jun 24. PMID: 20576783

Pyle A, Burn DJ, Gordon C, Swan C, **Chinnery PF**, Baudouin SV. Fall in circulating mononuclear cell mitochondrial DNA content in human sepsis. *Intensive Care Medicine* 2010 Jun;36(6):956-62. PMID: 20224905

Ritchie AE, Griffiths PG, **Chinnery PF**, Davidson AW. Eye movement recordings to investigate a supranuclear component in CPEO: a cross sectional study. *British Journal of Ophthalmology* 2010; 2010 Sep;94(9):1165-8. Epub 2010 Jun 24; PMID: 20576783

El-Kadi AM, Bros-Facer V, Deng W, Philpott A, Stoddart E, Banks G, Jackson GS, **Fisher EM, Duchen MR, Greensmith L, Moore AL, Hafezparast M. The legs at odd angles (Loa) mutation in cytoplasmic dynein ameliorates mitochondrial function in SOD1G93A mouse model for motor neuron disease. *J Biol Chem*. 2010 Jun 11;285(24):18627-39.

Mankad K, **Kullmann DM**, Davagnanam I. Neurological manifestation of vitamin B12 deficiency. *Am J Med*. 2010 Jun;123(6):e1-2. PubMed PMID: 20569736.

Tai YF, **Kullmann DM**, Howard RS, Scott GM, Hirsch NP, Revesz T, Leary SM. Central nervous system histoplasmosis in an immunocompetent patient. *J Neurol*. 2010 Nov;257(11):1931-3. Epub 2010 Jun 22. PubMed PMID: 20567842.

Spillane J, Beeson DJ, **Kullmann DM**. Myasthenia and related disorders of the neuromuscular junction. *J Neurol Neurosurg Psychiatry*. 2010 Aug;81(8):850-7. Epub 2010 Jun 14. PubMed PMID: 20547629.

Irani SR, Bera K, Waters P, Zuliani L, Maxwell S, Zandi MS, Friese MA, Galea I, **Kullmann DM**, Beeson D, Lang B, Bien CG, Vincent A. N-methyl-D-aspartate antibody encephalitis: temporal progression of clinical and paraclinical observations in a predominantly non-paraneoplastic disorder of both sexes. *Brain*. 2010 Jun;133(Pt 6):1655-67. PubMed PMID: 20511282.

Kullmann DM, Waxman SG. Neurological channelopathies: new insights into disease mechanisms and ion channel function. *J Physiol*. 2010 Jun 1;588(Pt 11):1823-7. Epub 2010 Apr 7. Review. PubMed PMID: 20375141.

Rajakulendran S, Graves TD, Labrum RW, Kotzadimitriou D, Eunson L, Davis MB, Davies R, Wood NW, **Kullmann DM, Hanna MG, Schorge S. Genetic and functional characterisation of the P/Q calcium channel in episodic ataxia with epilepsy. *J Physiol*. 2010 Jun 1;588(Pt 11):1905-13. Epub 2010 Feb 15. PubMed PMID: 20156848.

Pavlov I, **Kullmann DM**. How much inhibition in an epileptiform burst? *J Physiol*. 2010 Jan 1;588(Pt 1):17-8. PubMed PMID: 20045901; PubMed Central PMCID: PMC2821539. 26: Lamsa KP, Kullmann DM, Woodin MA. Spike-timing dependent plasticity in inhibitory circuits. *Front Synaptic Neurosci*. 2010 Jun 21;2:8. PubMed PMID: 21423494.

Guglieri M, **Bushby K**. Molecular treatments in Duchenne muscular dystrophy. *Curr Opin Pharmacol*. 2010 Jun;10(3):331-7. Epub 2010 Apr 29. Review. PMID: 20434401.

Burke G, Hillier C, Cole J, Sampson M, Bridges L, **Bushby K**, Barresi R, Hammans SR. Calpainopathy presenting as foot drop in a 41 year old. *Neuromuscul Disord*. 2010 Jun;20(6):407-10. PMID: 20580976.

Spengos K, Walter MC, Dekomien G, Papadopoulos K, **Lochmüller H**, Manta P. C283Y mutation in the gamma-sarcoglycan gene in Greek Gypsies with severe limb girdle muscular dystrophy. *Eur J Neurol*. 2010 Jun 1;17(6):e41-2. Epub 2010 Mar 22. PMID: 20345928.

Ferrari R, Kapogiannis D, Huey ED, Grafman J, **Hardy J**, Momeni P. Novel Missense Mutation in Charged Multivesicular Body Protein 2B in a Patient With Frontotemporal Dementia. *Alzheimer Dis Assoc Disord*. 2010 Jun 29 PMID 20592581

Christopher D. J. Sinclair, Rebecca S. Samson, David L. Thomas, Nikolaus Weiskopf, Antoine Lutti, John S. Thornton and **Xavier Golay**; Quantitative magnetization transfer in in vivo healthy human skeletal muscle at 3T Magnetic Resonance in Medicine, *Magn Reson Med*. 2010 Dec;64(6):1739-48. Epub 2010 Jul 27. PMID: 20665899

Chinnery PF. The age of single-gene neurological disorders is not dead. *Brain* 2010 Jul;133(Pt 7):1865-8. PMID: 20584944

Akam T, **Kullmann DM.** Oscillations and filtering networks support flexible routing of information. *Neuron*. 2010 Jul 29;67(2):308-20. PubMed PMID: 20670837.

Graves TD, Rajakulendran S, Zuberi SM, Morris HR, Schorge S, **Hanna MG, Kullmann DM. Nongenetic factors influence severity of episodic ataxia type 1 in monozygotic twins. *Neurology*. 2010 Jul 27;75(4):367-72. PubMed PMID: 20660867.

****Bushby K, Straub V.** One gene, one or many diseases? Simplifying dysferlinopathy. *Neurology*. 2010 Jul 27;75(4):298-9. Epub 2010 Jun 23. PMID: 20574035.

Clarke NF, Waddell LB, Cooper ST, Perry M, Smith RL, Kornberg AJ, Muntoni F, Lillis S, **Straub V, Bushby K, Guglieri M, King MD, Farrell MA, Marty I, Lunardi J, Monnier N, North KN. Recessive mutations in RYR1 are a common cause of congenital fiber type disproportion. *Hum Mutat*. 2010 Jul;31(7):E1544-50. PMID: 20583297.

Odgerel Z, Sarkozy A, Lee HS, McKenna C, Rankin J, **Straub V, Lochmüller H, Paola F, D'Amico A, Bertini E, **Bushby K,** Goldfarb LG. Inheritance patterns and phenotypic features of myofibrillar myopathy associated with a BAG3 mutation. *Neuromuscul Disord*. 2010 Jul;20(7):438-42. Epub 2010 Jun 3. PMID: 20605452

Schara U, Christen HJ, Durmus H, Hietala M, Krabetz K, Rodolico C, Schreiber G, Topaloglu H, Talim B, Voss W, Pihko H, Abicht A, Müller JS, **Lochmüller H.** Long-term follow-up in patients with congenital myasthenic syndrome due to CHAT mutations. *Eur J Paediatr Neurol*. 2010 Jul;14(4):326-33. Epub 2009 Nov 8. PMID: 19900826.

Reilich P, Schramm N, Schoser B, Schneiderat P, Strigl-Pill N, Müller-Höcker J, Kress W, Ferbert A, Rudnik-Schöneborn S, Noth J, **Lochmüller H,** Weis J, Walter MC. Facioscapulohumeral muscular dystrophy presenting with unusual phenotypes and atypical morphological features of vacuolar myopathy. *J Neurol*. 2010 Jul;257(7):1108-18. Epub 2010 Feb 10. PMID: 20146070.

Srouf M, Bolduc V, Guergueltcheva V, **Lochmüller H,** Gendron D, Shevell MI, Poulin C, Mathieu J, Bouchard JP, Brais B. DOK7 mutations presenting as a proximal myopathy in French Canadians. *Neuromuscul Disord*. 2010 Jul;20(7):453-7. Epub 2010 Jun 17. PMID: 20610155.

Greaves LC, Yu-Wai-Man P, Blakely EL, Krishnan KJ, Beadle NE, Kerin J, Barron MJ, Griffiths PG, Dickinson AJ, Turnbull DM, **Taylor RW.** Mitochondrial DNA defects and selective extraocular muscle involvement in CPEO. *Invest Ophthalmol Vis Sci*. 2010 Jul;51(7):3340-6. Epub 2010 Feb 17. PMID: 20164463

Yu-Wai-Man P, Lai-Cheong J, Borthwick GM, He L, Taylor GA, Greaves LC, Taylor RW, Griffiths PG, **Turnbull DM.** Somatic mitochondrial DNA deletions accumulate to high levels in aging human extraocular muscles. *Invest Ophthalmol Vis Sci*. 2010 Jul;51(7):3347-53. Epub 2010 Feb 17. PMID: 20164450

Rajakulendran S, Tan SV, **Hanna MG.** Muscle weakness, palpitations and a small chin: the Andersen-Tawil syndrome. *Pract Neurol*. 2010 Aug;10(4):227-31. Review. PMID: 20647529

Birnkrant DJ, **Bushby KM,** Amin RS, Bach JR, Benditt JO, Eagle M, FINDER JD, Kalra MS, Kissel JT, Koumbourlis AC, Kravitz RM. The respiratory management of patients with duchenne muscular dystrophy: a DMD care considerations working group specialty article. *Pediatr Pulmonol*. 2010 Aug;45(8):739-48. PMID: 20597083.

Tuppen HA, Fehmi J, Czermin B, Goffrini P, Meloni F, Ferrero I, He L, Blakely EL, McFarland R, Horvath R, **Turnbull DM,** Taylor RW. Long-term survival of neonatal mitochondrial complex III deficiency associated with a novel BCS1L gene mutation. *Mol Genet Metab*. 2010 Aug;100(4):345-8. Epub 2010 Apr 24. PMID: 20472482.

Guo X, Popadin KY, Markuzon N, Orlov YL, Kravtsov Y, Krishnan KJ, Zsurka G, **Turnbull DM**, Kunz WS, Khrapko K. Repeats, longevity and the sources of mtDNA deletions: evidence from 'deletional spectra'. *Trends Genet.* 2010 Aug;26(8):340-3. Epub 2010 Jun 28. PMID: 20591530

Singleton AB, **Hardy J**, Traynor BJ, Houlden H. Towards a complete resolution of the genetic architecture of disease. *Trends Genet.* 2010 PMID 20813421

Shatunov A, Mok K, Newhouse S, Weale ME, Smith B, Vance C, Johnson L, Veldink JH, van Es MA, van den Berg LH, Robberecht W, Van Damme P, Hardiman O, Farmer AE, Lewis CM, Butler AW, Abel O, Andersen PM, Fogh I, Silani V, Chiò A, Traynor BJ, Melki J, Meininger V, Landers JE, McGuffin P, Glass JD, Pall H, Leigh PN, **Hardy J**, Brown RH Jr, Powell JF, Orrell RW, Morrison KE, Shaw PJ, Shaw CE, Al-Chalabi A. Chromosome 9p21 in sporadic amyotrophic lateral sclerosis in the UK and seven other countries: a genome-wide association study. *Lancet Neurol.* 2010 Aug 27. PMID 20801717

Jephson CG, Mills NA, Pitt MC, Beeson D, Aloysius A, **Muntoni F**, Robb SA, Bailey CM. Congenital stridor with feeding difficulty as a presenting symptom of Dok7 congenital myasthenic syndrome. *Int J Pediatr Otorhinolaryngol.* 2010 Sep;74(9):991-4. PMID: 20554332

Klinge L, Aboumoussa A, Eagle M, Hudson J, Sarkozy A, Vita G, Charlton R, Roberts M, **Straub V, Barresi R, **Lochmüller H**, **Bushby K**. New aspects on patients affected by dysferlin deficient muscular dystrophy. *J Neurol Neurosurg Psychiatry.* 2010 Sep;81(9):946-53. Epub 2009 Jun 14. PMID: 19528035.

Mihaylova V, Scola RH, Gervini B, Lorenzoni PJ, Kay CK, Werneck LC, Stucka R, Guergueltcheva V, von der Hagen M, Huebner A, Abicht A, Müller JS, **Lochmüller H**. Molecular characterisation of congenital myasthenic syndromes in Southern Brazil. *J Neurol Neurosurg Psychiatry.* 2010 Sep;81(9):973-7. Epub 2010 Jun 20. PMID: 20562457

McFarland R, Taylor RW, **Turnbull DM**. A neurological perspective on mitochondrial disease. *Lancet Neurol.* 2010 Aug;9(8):829-40. Review. PMID: 20650404.

Sinclair CD, Miranda MA, Cowley P, Morrow JM, Davagnanam I, Mehta H, **Hanna MG, **Koltzenburg M**, **Reilly MM**, **Yousry TA**, Thornton JS. MRI shows increased sciatic nerve cross sectional area in inherited and inflammatory neuropathies. *J Neurol Neurosurg Psychiatry.* 2010 Oct 22. PMID: 20971754

Raja Rayan DL, **Hanna MG**. Skeletal muscle channelopathies: nondystrophic myotonias and periodic paralysis. *Curr Opin Neurol.* 2010 Oct;23(5):466-76. Review. PMID: 20634695

Elachouri G, Vidoni S, Zanna C, Pattyn A, Boukhaddaoui H, Gaget K, Yu-Wai-Man P, Gasparre G, Sarzi E, Delettre C, Olichon A, Loiseau D, Reynier P, **Chinnery PF**, Rotig A, Carelli V, Hamel CP, Rugolo M, Lenaers G. OPA1 links human mitochondrial genome maintenance to mtDNA replication and distribution. *Genome Research* 2010 Oct 25. PMID: 20974897

Ondo WG, Adam OR, Jankovic J, **Chinnery PF**. Dramatic response of facial stereotype/tic to tetrabenazine in the first reported cases of neuroferritinopathy in the United States. *Movement Disorders* 2010 Oct 30;25(14):2470-2. PMID: 20818611

Yu-Wai-Man P, Trenell MI, Hollingsworth KG, Griffiths PG, **Chinnery PF**, OPA1 mutations impair mitochondrial function in both pure and complicated dominant optic atrophy. *Brain* 2011 Apr;134(Pt 4):e164. Epub 2010 Oct 14 PMID: 20952381

Yu-Wai-Man P, Griffiths PG, **Chinnery PF**. Mitochondrial Optic Neuropathies - Disease Mechanisms and Therapeutic Strategies. *Progress in Retinal and Eye Research* 2010 Nov 25. PMID: 21112411

Stewart JD, Horvath R, Baruffini E, Ferrero I, Bulst S, Watkins PB, Fontana RJ, Day CP, **Chinnery PF**. POLG determines the risk of sodium valproate induced liver toxicity. *Hepatology* 2010 Nov;52(5):1791-6. PMID: 21038416

Schorge S, **Kullmann DM**. Sodium channel mutations and epilepsy: association and causation. *Exp Neurol.* 2010 Nov;226(1):8-10. Epub 2010 Aug 13. PubMed PMID: 20709059.

- **Murphy SM, Davidson G, **Brandner S**, Houlden H, **Reilly MM**. Mutation in FAM134B causing severe hereditary sensory neuropathy. *JNNP* 2010 Nov 28 (epub ahead of press) PMID 21115472
- **Boldrin L, **Muntoni F**, **Morgan JE**. Are Human and Mouse Satellite Cells Really the Same? *J Histochem Cytochem*. 2010 Nov;58(11):941-55. PMID: 20644208
- **Wilmshurst JM, Lillis S, Zhou H, Pillay K, Henderson H, Kress W, Müller CR, Ndong A, Cloke V, Cullup T, Bertini E, Boennemann C, **Straub V**, Quinlivan R, Dowling JJ, Al-Sarraj S, Treves S, Abbs S, Manzur AY, Sewry CA, **Muntoni F**, Jungbluth H. RYR1 mutations are a common cause of congenital myopathies with central nuclei. *Ann Neurol* 2010 Nov;68(5):717-26. PMID: 20839240
- **Bauer R, Blain A, Greally E, **Bushby K**, **Lochmüller H**, Laval S, **Straub V**, MacGowan GA. Intolerance to β -blockade in a mouse model of δ -sarcoglycan-deficient muscular dystrophy cardiomyopathy. *Eur J Heart Fail*. 2010 Nov;12(11):1163-70. Epub 2010 Jul 30. PMID: 20675662.
- Quinlivan R, Shaw N, **Bushby K**. 170th ENMC International Workshop: bone protection for corticosteroid treated Duchenne muscular dystrophy. 27-29 November 2009, Naarden, The Netherlands. *Neuromuscul Disord*. 2010 Nov;20(11):761-9. Epub 2010 Aug 17. PMID: 20724157.
- Seeger J, Schrank B, Pyle A, Stucka R, Lörcher U, Müller-Ziermann S, Abicht A, Czermin B, Holinski-Feder E, **Lochmüller H**, Horvath R. Clinical and neuropathological findings in patients with TACO1 mutations. *Neuromuscul Disord*. 2010 Nov;20(11):720-4. Epub 2010 Aug 19. PMID: 20727754.
- Alston CL, Lowe J, **Turnbull DM**, Maddison P, Taylor RW. A novel mitochondrial tRNAGlu (MTTE) gene mutation causing chronic progressive external ophthalmoplegia at low levels of heteroplasmy in muscle. *J Neurol Sci*. 2010 Nov 15;298(1-2):140-4. PMID: 20810132.
- Stewart JD, Schoeler S, Sitarz K, Horvath R, Hallmann K, Pyle A, Yu-Wai-Man P, Taylor RW, Samuels DC, Kunz WS, **Chinnery PF**. POLG mutations cause decreased mitochondrial DNA repopulation rates following induced depletion in human fibroblasts. *Biochimica et Biophysica Acta - Molecular Basis of Disease* 2011 Mar;1812(3):321-5. Epub 2010 Dec 5. PMID:21138766
- Hudson G, Yu-Wai-Man P, Griffiths PG, Carelli V, Zeviani V, **Chinnery PF**. Variation in OPA1 does not explain the incomplete penetrance of Leber hereditary optic neuropathy. *Molecular Vision* 2010 Dec 15;16:2760-4. PMID: 21203403
- Bathgate B, Yu-Wai-Man P, Webb B, Taylor RW, Fowler B, **Chinnery PF**. Recessive spastic paraparesis associated with complex I deficiency due to MTHFR mutations. *Journal of Neurology Neurosurgery and Psychiatry* 2010 Dec 2. [Epub ahead of print]. PMID: 21131308
- Lythgow KT, Hudson G, Andras P, **Chinnery PF**. A critical analysis of the combined usage of protein localization prediction methods: increasing the number of independent data sets can reduce the accuracy of predicted mitochondrial localization. *Mitochondrion* 2011 May;11(3):444-9. Epub 2010 Dec 31. PMID:21195798
- Matsakas A, Otto A, Elashry MI, **Brown SC**, Patel K. Rejuvenation Res. Altered primary and secondary myogenesis in the myostatin-null mouse. 2010 Dec;13(6):717-27. Epub 2011 Jan 4. PMID:21204650
- **Brockington M, Torelli S, Sharp PS, Liu K, Cirak S, **Brown SC**, Wells DJ, **Muntoni F**. Transgenic overexpression of LARGE induces α -dystroglycan hyperglycosylation in skeletal and cardiac muscle. *PLoS One*. 2010 Dec 28;5(12):e14434. PMID:21203384
- **Tomlinson SE, Tan SV, **Kullmann DM**, Griggs RC, Burke D, **Hanna MG**, Bostock H. Nerve excitability studies characterize Kv1.1 fast potassium channel dysfunction in patients with episodic ataxia type 1. *Brain*. 2010 Dec;133(Pt 12):3530-40. PMID: 21106501
- Robb SA, **Muntoni F**, Simonds AK. Respiratory management of congenital myasthenic syndromes in childhood: Workshop 8th December 2009, UCL Institute of Neurology, London, UK. *Neuromuscul Disord*. 2010 Dec;20(12):833-8. PMID: 20850318

Wang CH, Bonnemann CG, Rutkowski A, Sejersen T, Bellini J, Battista V, Florence JM, Schara U, Schuler PM, Wahbi K, Aloysius A, Bash RO, Bérout C, Bertini E, **Bushby K, Cohn RD, Connolly AM, Deconinck N, Desguerre I, Eagle M, Estournet-Mathiaud B, Ferreira A, Fajak A, Goemans N, Iannaccone ST, Jouinot P, Main M, Melacini P, Mueller-Felber W, **Muntoni F**, Nelson LL, Rahbek J, Quijano-Roy S, Sewry C, Storhaug K, Simonds A, Tseng B, Vajsar J, Vianello A, Zeller R. Consensus Statement on Standard of Care for Congenital Muscular Dystrophies. *J Child Neurol*. 2010 Dec;25(12):1559-81. PMID: 21078917

Read J, Simonds A, Kinali M, **Muntoni F**, Garralda ME. Sleep and well being in young men with neuromuscular disorders receiving non-invasive ventilation and their carers. *Neuromuscul Disord*. 2010 20(7):458-63. PMID: 20558064

Munot P, Lashley D, Jungbluth H, Feng L, Pitt M, Robb SA, Palace J, Jayawant S, Kennet R, Beeson D, Cullup T, Abbs S, Laing N, Sewry C, **Muntoni F**. Congenital fibre type disproportion associated with mutations in the tropomyosin 3 (TPM3) gene mimicking congenital myasthenia. *Neuromuscul Disord*. 2010 Dec;20(12):796-800. PMID: 20951040

Forrest S, Meloni PL, **Muntoni F**, Kim J, Fletcher S, Wilton SD. Personalized exon skipping strategies to address clustered non-deletion dystrophin mutations. *Neuromuscul Disord*. 2010 Dec;20(12):810-6. PMID: 20817455

Schoenmakers E, Agostini M, Mitchell C, Schoenmakers N, Papp L, Rajanayagam O, Padidela R, Ceron-Gutierrez L, Doffinger R, Prevosto C, Luan J, Montano S, Lu J, Castanet M, Clemons N, Groeneveld M, Castets P, Karbaschi M, Aitken S, Dixon A, Williams J, Campi I, Blount M, Burton H, **Muntoni F**, O'Donovan D, Dean A, Warren A, Brierley C, Baguley D, Guicheney P, Fitzgerald R, Coles A, Gaston H, Todd P, Holmgren A, Khanna KK, Cooke M, Semple R, Halsall D, Wareham N, Schwabe J, Grasso L, Beck-Peccoz P, Ogunko A, Dattani M, Gurnell M, Chatterjee K. Mutations in the selenocysteine insertion sequence-binding protein 2 gene lead to a multisystem selenoprotein deficiency disorder in humans. *J Clin Invest* 2010; 120(12):4220-4235. PMID: 21084748

Krahn M, Goicoechea M, Hanisch F, Groen E, Bartoli M, Pécheux C, Garcia-Bragado F, Leturcq F, Jeannet PY, Lohrbus JA, Jacquemont S, Strober J, Urtizbera JA, Saenz A, **Bushby K**, Lévy N, Lopez de Munain A. Eosinophilic infiltration related to CAPN3 mutations: a pathophysiological component of primary calpainopathy? *Clin Genet*. 2010 Dec 27.[Epub ahead of print] PMID: 21204801.

Vermeer S, Hoischen A, Meijer RP, Gilissen C, Neveling K, Wieskamp N, de Brouwer A, Koenig M, Anheim M, Assoum M, Drouot N, Todorovic S, Milic-Rasic V, **Lochmüller H**, Stevanin G, Goizet C, David A, Durr A, Brice A, Kremer B, van de Warrenburg BP, Schijvenaars MM, Heister A, Kwint M, Arts P, van der Wijst J, Veltman J, Kamsteeg EJ, Scheffer H, Knoers N. Targeted next-generation sequencing of a 12.5 Mb homozygous region reveals ANO10 mutations in patients with autosomal-recessive cerebellar ataxia. *Am J Hum Genet*. 2010 Dec 10;87(6):813-9. Epub 2010 Nov 18. PMID: 21092923

Sarkozy A, **Lochmüller H**. Neuromuscular disorders and 2010: recent advances. *J Neurol*. 2010 Dec;257(12):2117-21. Epub 2010 Sep 18. PMID: 20852879.

Forrest KM, Al-Sarraj S, Sewry C, Buk S, Tan SV, Pitt M, Durward A, McDougall M, Irving M, **Hanna MG, Matthews E, Sarkozy A, Hudson J, Barresi R, **Bushby K**, Jungbluth H, Wraige E. Infantile onset myofibrillar myopathy due to recessive CRYAB mutations. *Neuromuscul Disord*. 2011 Jan;21(1):37-40. PMID: 21130652

Matthews E, Manzur AY, Sud R, **Muntoni F, **Hanna MG**. Stridor as a neonatal presentation of skeletal muscle sodium channelopathy. *Arch Neurol*. 2011 Jan;68(1):127-9. PMID: 21220685

Dick D, Horvath R, **Chinnery PF**. AMACR mutations cause late-onset autosomal recessive cerebellar ataxia. *Neurology* 2011: PMID 21576695

Hicks D, Sarkozy A, Muelas N, Koehler K, Huebner A, Hudson G, **Chinnery PF, **Barresi R**, Eagle M, Polvikoski T, Bailey G, Miller J, Radunovic A, Hughes PJ, Roberts R, Krause S, Walter MC, Laval SH, **Straub V**, **Lochmüller H**, **Bushby K**. A founder mutation in Anoctamin 5 is a major cause of limb-girdle muscular dystrophy. *Brain* 2011 Jan;134(Pt 1):171-82. PMID: 21186264

Kemp JP, Smith PM, Pyle A, Neeve VC, Tuppen HA, Schara U, Talim B, Topaloglu H, Holinski-Feder E, Abicht A, Czermin B, **Lochmüller H, McFarland R, **Chinnery PF**, Chrzanowska-Lightowlers ZM, Lightowlers RN, Taylor RW, **Horvath R**. Nuclear factors involved in mitochondrial translation cause a subgroup of combined respiratory chain deficiency. *Brain* 2011 Jan;134(Pt 1):183-95. PMID: 21169334

Abramov AY, Ionov, M, Pavlov E, **Duchen, MR** (2011) Membrane cholesterol content plays a key role in the neurotoxicity of β -amyloid: implications for Alzheimer's Disease. *Ageing Cell*, 10(4):595-603.

Abeti R., Abramov AY and **Duchen MR** (2011) β -amyloid activates *Poly (ADP-ribose) polymerase* (PARP) causing astrocytic metabolic failure and neuronal death. *Brain*, 134(Pt 6):1658-72

Banks, G.T., Haas, M.A., Line, S., Shepherd, H.L., AlQatari, M., Stewart, S., Rishal, I., Philpott, A., Kalmar, B., Kuta, A., Groves, M., Parkinson, N., Acevedo-Arozena, A., **Brandner, S., Bannerman, B., **Greensmith, L.**, Hafezparast, M., **Koltzenburg, M.**, Deacon, R., Fainzilber, M., **Fisher, E.M.C.** (2011) Behavioural and other phenotypes in a cytoplasmic dynein light intermediate chain 1 mutant mouse. *J. Neurosci.* 31: 5483-5494. PMID 21471385

Acevedo-Arozena, A., Kalmar, B., Essa, S., Ricketts, T., Joyce, P., Kent, R., Rowe, C., Parker, A., Gray, A., Hafezparast, M., Thorpe, J.R., **Greensmith, L., **Fisher, E.M.C.** (2011) A comprehensive assessment of the SOD1G93A low-copy transgenic mouse, which models human amyotrophic lateral sclerosis. *Dis Model Mech* 4: 686-700. PMID 21540242

Kirschner J, **Lochmüller H**. Sarcoglycanopathies. *Handb Clin Neurol.* 2011;101:41-6. Review. PMID: 21496623.

**Kinali M, Arechavala-Gomez V, Cirak S, Glover A, Guglieri M, Feng L, Hollingsworth KG, Hunt D, Jungbluth H, Roper HP, Quinlivan RM, Gosalakal JA, Jayawant S, Nadeau A, Hughes-Carre L, Manzur AY, Mercuri E, Morgan JE, Straub V, Bushby K, Sewry C, Rutherford M, Muntoni F (2011). Muscle histology vs MRI in Duchenne muscular dystrophy. *Neurology* 76(4):346-353 25. PMID: 21263136

Ono Y, Calhabeu F, **Morgan JE**, Katagiri T, Amthor H, Zammit PS (2011). BMP signalling permits population expansion by preventing premature myogenic differentiation in muscle satellite cells. *Cell Death Differ.* 18(2):222-234. PMID: 20689554

Meng J, Adkin CF, Xu SW, **Muntoni F, **Morgan JE** (2011). Contribution of human muscle-derived cells to skeletal muscle regeneration in dystrophic host mice. *PLoS One* 6(3):e17454. PMID: 21408080
Lin YY, White R, Torelli S, Cirak S, **Muntoni F** Stemple DL. Zebrafish Fukutin family proteins link the unfolded protein response with dystroglycanopathies. *Human Molecular genetics*, 2011 ;20(9):1763-75. PMID: 21317159

Arthur-Farraj P, Wanek K, Hantke J, Davis CM, Jayakar A, Parkinson DB, Mirsky R, **Jessen KR**. Mouse schwann cells need both NRG1 and cyclic AMP to myelinate. *Glia.* 2011 59: 720-33. PMID: 21322058

Sinéad M Murphy, James Polke, Hadi Manji, Julian Blake, Lilla Reiniger , Mary Sweeney, Henry Houlden, **Sebastian Brandner, **Mary M Reilly**. A novel mutation in the nerve-specific 5'UTR of the GJB1 gene causes X-linked Charcot-Marie-Tooth Disease. *JPNS* 2011: 11: 65-70. PMID 21504505

Reilly MM, Murphy SM, Laura M. Charcot-Marie-Tooth disease. *JPNS* 2011: 16: 1-14. PMID 21504497

Feely SM, Laura M , Siskind CE, Sottile S, Davis M, Gibbons VS, **Reilly MM**, Shy ME. MFN2 mutations cause severe phenotypes in most patients with CMT2A. *Neurology* 2011: 76: 1690-1696. PMID 21508331

Saporta M, Katona I, Zhang X, Roper H, Carr L, Macdonald F, Brueton L, Blake J, Suter U, **Reilly MM**, Shy ME, Li J. Neuropathy in a Human without the PMP22 gene. *Arch Neurol* 2011: 68: 814-821. PMID 21670407

Siskind CE, Murphy SM, Ovens R, Polke J, **Reilly MM**, Shy ME. Phenotype expression in women with CMT1X. *J Peripher Nerve Sys* 2011: 16: 102-107. 21692908

Polke JM, Laurá M, Pareyson D, Taroni F, Milani M, Bergamin G, Gibbons VS, Houlden H, Chamley SC, Blake J, DeVile C, Sandford R, Sweeney MG, Davis MB, **Reilly MM**. Recessive axonal Charcot Marie Tooth disease due to compound heterozygous mitofusin 2 mutations. *Neurology* 2011; 77: 168-173. PMID 21715711

Pareyson D, **Reilly MM**, Schenone A, Fabrizi GM, Cavallaro T, Santoro L, Vita G, Quattrone A, Padua L, Gemignani F, Visioli F, Laurà M, Radice D, Calabrese D, Hughes RAC, Solari A, for the CMT-TRIAAL & CMT-TRAUK Group. Ascorbic acid in Charcot-Marie-Tooth disease type 1A (CMT-TRIAAL and CMT-TRAUK): a double blind randomised trial. *Lancet Neurology* 2011; 10: 320-328. 21393063

Ashbury AK, King RHM, **Reilly MM**, Dyck PJ, Said G, Compston A. Professor P. K. Thomas: clinician, editor and leader-a retrospective appreciation. *Brain* 2011; 134: 618-626. PMID 21278409

Guelly C, Zhu PP, Leonardis L, Papić L, Zidar J, Schabhüttl M, Strohmaier H, Weis J, Strom TM, Baets J, Willems J, De Jonghe P, **Reilly MM**, Fröhlich E, Hatz M, Trajanoski S, Pieber TR, Janecke AR, Blackstone C, Auer-Grumbach M. Targeted High-Throughput Sequencing Identifies Mutations in atlastin-1 as a Cause of Hereditary Sensory Neuropathy Type I. *Am J Hum Genet* 2011; 88: 99-105. PMID 21194697

Murphy SM, Laura M, Blake J, Polke J, Bremner F, **Reilly MM**. Conduction block and tonic pupils in Charcot-Marie-Tooth disease caused by a Myelin Protein Zero Ile112Thr mutation. *Neuromuscul Disord* 2011; 21: 223-226. PMID 21256749

Amato A, **Reilly MM**. The death of the CMT panel. *Ann Neurol* 2011; 69: 1-4. PMID 21280068

Russo M, Laurá M, Polke JM, Davis MB, Blake J, **Brandner S, Hughes RAC, Houlden H, Bennett DLH, Lunn MPT, **Reilly MM**. Variable phenotypes are associated with PMP22 missense mutations. *Neuromuscul Disord* 2011; 21: 106-114. PMID 21194947

Carvalho OP, Thornton GK, Hertecant J, Houlden H, Nicholas AK, Cox JJ, **Reilly M**, Al-Gazali L, Woods CG. A novel NGF mutation clarifies the molecular mechanism and extends the phenotypic spectrum of the HSAN5 neuropathy. *J Med Genet*. 2011; 48: 131-135. PMID 20978020

Sadnicka A, **Reilly MM, Mummery C, **Brandner S**, Hirsch N, Lunn M. Rituximab in the treatment of three coexistent neurological autoimmune diseases: Chronic inflammatory demyelinating polyradiculoneuropathy, Morvans syndrome and myasthenia gravis. *JNNP* 2011; 82: 230-232. PMID 20462915

Saifee TA, **Reilly MM, Ako E, Rugg-Gunn F, **Brandner S**, Lunn MP, Leary SM. Sarcoidosis presenting as acute inflammatory demyelinating polyradiculoneuropathy. *Muscle Nerve* 2011; 43: 296-298. PMID 21254101

Smith L-JE, Murphy SM, Holmes P, **Reilly MM**, Reiniger L, Thom M, Lunn PP. A painful right leg. *BMJ* 2011;16: 342. PMID 21411806

Yis U, Uyanik G, Heck PB, Smitka M, Nobel H, Ebinger F, Dirik E, Feng L, Kurul SH, Brocke K, Unalp A, Ozer E, Cakmakci H, Sewry C, Cirak S, **Muntoni F**, Hehr U, Morris-Rosendahl DJ. Fukutin mutations in non-Japanese patients with congenital muscular dystrophy: Less severe mutations predominate in patients with a non-Walker-Warburg phenotype. *Neuromuscular Disorders* 2011 Jan;21(1):20-30. PMID: 20961758

Lu QL, Yokota T, Takeda S, Garcia L, **Muntoni F**, Partridge T. The Status of Exon Skipping as a Therapeutic Approach to Duchenne Muscular Dystrophy. *Mol Ther*. 2011 Jan;19(1):9-15. PMID: 20978473

Namavar Y, Barth PG, Kasher PR, van Ruissen F, Brockmann K, Bernert G, Writzl K, Ventura K, Cheng EY, Ferriero DM, Basel-Vanagaite L, Eggens VR, Krägeloh-Mann I, De Meirleir L, King M, Graham JM Jr, von Moers A, Knoers N, Sztriha L, Korinthenberg R; PCH Consortium (**Muntoni F**), Dobyns WB, Baas F, Poll-The BT. Clinical, neuroradiological and genetic findings in pontocerebellar hypoplasia. *Brain*. 2011 Jan;134(Pt 1):143-56. PMID: 20952379

Ricotti V, Roberts RG, **Muntoni F**. Dystrophin and the brain. *Dev Med Child Neurol*. 2011 Jan;53(1):12. PMID: 21171237

Meng J, **Muntoni F, **Morgan JE**. Stem cells to treat muscular dystrophies - Where are we? *Neuromuscul Disord*. 2011 Jan;21(1):4-12. PMID: 21055938

Aartsma-Rus A; Cosigned by: Furlong P, Vroom E; AFM (France) LUMC Duchenne team, van Ommen GJ, Niks E, Straathof C, Verschuuren J; TREAT-NMD Project ethics council Members: Aartsma-Rus A, Ferlini A, Hagger L, Heslop E, Karcagi V, Kirschner J, McCormack P, Moeschel P, **Muntoni F**, Ouillade MC, Rahbeck J, Rehmann-Sutter C, Rouault F, Sejersen T, Vroom E, Woods S; The risks of therapeutic misconception and N 1 "trials" in rare diseases such as Duchenne dystrophy. *Neuromuscul Disord*. 2011 Jan;21(1):13-5. PMID: 21051233

Durmus H, Laval SH, Deymeer F, Parman Y, Kiyan E, Gokyigiti M, Ertekin C, Ercan I, Solakoglu S, Karcagi V, **Straub V, **Bushby K**, **Lochmüller H**, Serdaroglu-Ofazer P. Oculopharyngodistal myopathy is a distinct entity: clinical and genetic features of 47 patients. *Neurology*. 2011 Jan 18;76(3):227-35. PMID: 21242490.

Jørgensen LH, Blain A, Grealley E, Laval SH, Blamire AM, Davison BJ, Brinkmeier H, MacGowan GA, Schröder HD, **Bushby K, **Straub V**, **Lochmüller H**. Long-term blocking of calcium channels in mdx mice results in differential effects on heart and skeletal muscle. *Am J Pathol*. 2011 Jan;178(1):273-83. Epub 2010 Dec 23. PMID: 21224064

Bushby K. Neuromuscular diseases: milestones in development of treatments. *Lancet Neurol*. 2011 Jan;10(1):11-3. PMID: 21163438.

Vlachantoni D, Bramall AN, Murphy MP, Taylor RW, Shu X, Tulloch B, Van Veen T, **Turnbull DM**, McInnes RR, Wright AF. Evidence of severe mitochondrial oxidative stress and a protective effect of low oxygen in mouse models of inherited photoreceptor degeneration. *Hum Mol Genet*. 2011 Jan 15;20(2):322-35. Epub 2010 Nov 3. PMID: 21051333.

C-H Lu, B Kalmar, A Malaspina, **L Greensmith** & A Petzold (2011) A method to solubilise protein aggregates for immunoassay quantification which overcomes the neurofilament "hook" effect. *Journal of Neuroscience Methods* 195, 143-150 PMID 21134399

GT Banks, MA Haas, S Line, HL Shepherd, M AlQatari, S Stewart, I Rishal, A Philpott, B Kalmar, A Kuta, M Groves, N Parkinson, A Acevedo-Arozena, **S Brandner, **L Greensmith**, M Hafezparast, **M Koltzenburg**, R Deacon, M Fainzilber & **EMC Fisher** (2011) Behavioural and other phenotypes in a cytoplasmic dynein light intermediate chain 1 mutant mouse. *Journal of Neuroscience* 31(14):5483-5494 PMID 21471385

B Malik, N Nirmalanathan, L Bilsland, AR La Spada, **MG Hanna, G Schiavo, JM Gallo & **L Greensmith** (2011) Absence of disturbed axonal transport in Spinal and Bulbar Muscular Atrophy *Human Molecular Genetics* 20 (9):1776-1786 PMID 21317158

Tan SV, Matthews E, Barber M, Burge JA, Rajakulendran S, Fialho D, Sud R, Haworth A, **Koltzenburg M, **Hanna MG**. Refined exercise testing can aid DNA-based diagnosis in muscle channelopathies. *Ann Neurol*. 2011 Feb;69(2):328-40. PMID: 21387378

Cunningham MO, **Chinnery PF**. Mitochondria and cortical gamma oscillations: food for thought? *Brain* 2011 Feb; 134(Pt 2):330-2. PMID: 21278404

Gnocchi VF, Scharner J, Huang Z, Brady K, Lee JS, White RB, **Morgan JE**, Sun YB, Ellis JA, Zammit PS (2011). Uncoordinated transcription and compromised muscle function in the *Imna*-null mouse model of emery-dreifuss muscular dystrophy. *PLoS One*. 2011 Feb 22;6(2):e16651. PMID: 21364987

Pavlov I, Scimemi A, Savtchenko L, **Kullmann DM**, Walker MC. I(h)-mediated depolarization enhances the temporal precision of neuronal integration. *Nat Commun*. 2011 Feb 15;2:199. doi: 10.1038/ncomms1202. PubMed PMID: 21326231.

Kullmann DM, Lamsa KP. Interneurons go plastic. *Neuropharmacology*. 2011 Apr;60(5):711. Epub 2011 Feb 4. PubMed PMID: 21296094.

Gilbert GJ, Graves TD, **Kullmann DM**. Nongenetic factors influence severity of episodic ataxia type 1 in monozygotic twins. *Neurology*. 2011 Feb 1;76(5):490; author reply 490. PubMed PMID: 21282599.

Treves S, Vukcevic M, Jeannet PY, Levano S, Girard T, Urwyler A, Fischer D, Voit T, Jungbluth H, Lillis S, **Muntoni F, Quinlivan R, Sarkozy A, **Bushby K**, Zorzato F. Enhanced excitation coupled Ca²⁺ entry induces nuclear translocation of NFAT and contributes to IL-6 release from myotubes from patients with Central core disease. *Hum Mol Genet*. 2011 Feb 1;20(3):589-600. PMID: 21088110

Malerba A, Sharp PS, Graham IR, Arechavala-Gomez V, Foster K, **Muntoni F**, Wells DJ, Dickson G. Chronic Systemic Therapy With Low-dose Morpholino Oligomers Ameliorates the Pathology and Normalizes Locomotor Behavior in mdx Mice. *Mol Ther*. 2011 Feb;19(2):345-54. PMID: 21102560

Cacciottolo M, Belcastro V, Laval S, **Bushby K**, di Bernardo D, Nigro V. Reverse engineering gene network identifies new dysferlin-interacting proteins. *J Biol Chem*. 2011 Feb 18;286(7):5404-13. Epub 2010 Nov 30. PMID: 21119217

Senderek J, Müller JS, Dusl M, Strom TM, Guergueltcheva V, Diepolder I, Laval SH, Maxwell S, Cossins J, Krause S, Muelas N, Vilchez JJ, Colomer J, Mallebrera CJ, Nascimento A, Nafissi S, Kariminejad A, Nilipour Y, Bozorgmehr B, Najmabadi H, Rodolico C, Sieb JP, Steinlein OK, Schlotter B, Schoser B, Kirschner J, Herrmann R, Voit T, Oldfors A, Lindbergh C, Urtizberea A, von der Hagen M, Hübner A, Palace J, **Bushby K, **Straub V**, Beeson D, Abicht A, **Lochmüller H**. Hexosamine biosynthetic pathway mutations cause neuromuscular transmission defect. *Am J Hum Genet*. 2011 Feb 11;88(2):162-72. PMID: 21310273

Schara U, von Kleist-Retzow JC, Lainka E, Gerner P, Pyle A, Smith PM, **Lochmüller H**, Czermin B, Abicht A, Holinski-Feder E, Horvath R. Acute liver failure with subsequent cirrhosis as the primary manifestation of TRMU mutations. *J Inher Metab Dis*. 2011 Feb;34(1):197-201. Epub 2010 Dec 10. PMID: 21153446.

Colombo E, Romaggi S, Medico E, Menon R, Mora M, Falcone C, **Lochmüller H**, Confalonieri P, Mantegazza R, Morandi L, Farina C. Human neurotrophin receptor p75NTR defines differentiation-oriented skeletal muscle precursor cells: implications for muscle regeneration. *J Neuropathol Exp Neurol*. 2011 Feb;70(2):133-42. PMID: 21343882.

Larochelle N, Stucka R, Rieger N, Schermelleh L, Schiedner G, Kochanek S, Wolf E, **Lochmüller H**. Genomic integration of adenoviral gene transfer vectors following transduction of fertilized mouse oocytes. *Transgenic Res*. 2011 Feb;20(1):123-35. Epub 2010 May 13. PMID: 20464633.

Pitceathly RD, Fassone E, Taanman JW, Sadowski M, Fratter C, Mudanohwo EE, Woodward CE, Sweeney MG, Holton JL, **Hanna MG**, Rahman S. Kearns-Sayre syndrome caused by defective R1/p53R2 assembly. *J Med Genet*. 2011 Mar 22. [Epub ahead of print] PMID: 21378381

Hara Y, Balci-Hayta B, Yoshida-Moriguchi T, Kanagawa M, Beltrán-Valero de Bernabé D, Gündeşli H, Willer T, Satz JS, Crawford RW, Burden SJ, Kunz S, Oldstone MB, Accardi A, Talim B, **Muntoni F**, Topaloğlu H, Dinçer P, Campbell KP. A Dystroglycan Mutation Associated with Limb-Girdle Muscular Dystrophy. *New England Journal of Medicine*, 2011 Mar 10;364(10):939-46. PMID: 21388311

Dick E, Matsa E, Bispham J, Reza M, Guglieri M, Staniforth A, Watson S, Kumari R, **Lochmüller H**, Young L, Darling D, Denning C. Two new protocols to enhance the production and isolation of human induced pluripotent stem cell lines. *Stem Cell Res*. 2011 Mar;6(2):158-67. Epub 2010 Nov 20. PMID: 21095172.

Blain AM, **Straub VW**. δ -Sarcoglycan-deficient muscular dystrophy: from discovery to therapeutic approaches. *Skelet Muscle*. 2011 Mar 17;1(1):13. PMID: 21798091

Lo HP, Bertini E, Mirabella M, Domazetovska A, Dale RC, Petrini S, D'Amico A, Valente EM, Barresi R, Roberts M, Tozzi G, Tasca G, Cooper ST, **Straub V**, North KN. Mosaic caveolin-3 expression in acquired rippling muscle disease without evidence of myasthenia gravis or acetylcholine receptor autoantibodies. *Neuromuscul Disord*. 2011 Mar;21(3):194-203. Epub 2011 Feb 4. PMID: 21295981.

Lax NZ, **Turnbull DM**, Reeve AK. Mitochondrial Mutations: Newly Discovered Players in Neuronal Degeneration. *Neuroscientist*. 2011 Mar 30. [Epub ahead of print] PMID: 21454322.

Keogh MJ, Khan A, Gorman G, McNeill A, Horvath R, Burn J, **Chinnery PF**. An unusual gait following the discovery of a new disease. *Practical Neurology* 2011 Apr;11(2):81-4. PMID:21385964

Le Duigou C, **Kullmann DM**. Group I mGluR agonist-evoked long-term potentiation in hippocampal oriens interneurons. *J Neurosci*. 2011 Apr 13;31(15):5777-81. PubMed PMID: 21490219;

Kullmann DM, Lamsa KP. LTP and LTD in cortical GABAergic interneurons: emerging rules and roles. *Neuropharmacology*. 2011 Apr;60(5):712-9. Epub 2010 Dec 23. Review. PubMed PMID: 21185319.

Le Duigou C, Holden T, **Kullmann DM**. Short- and long-term depression at glutamatergic synapses on hippocampal interneurons by group I mGluR activation. *Neuropharmacology*. 2011 Apr;60(5):748-56. Epub 2010 Dec 23. PubMed PMID: 21185314.

Bianchi ML, Biggar D, **Bushby K**, Rogol AD, Rutter MM, Tseng B. Endocrine aspects of Duchenne muscular dystrophy. *Neuromuscul Disord*. 2011 Apr;21(4):298-303. Epub 2011 Feb 25. PMID: 21353552.

Rollinson S, Mead S, Snowden J, Richardson A, Rohrer J, Halliwell N, Usher S, Neary D, Mann D, **Hardy J**, Pickering-Brown S. Frontotemporal lobar degeneration genome wide association study replication confirms a risk locus shared with amyotrophic lateral sclerosis. *NEUROBIOL AGING* 32(4): Article number 758.e1 Apr 2011. PMID 21257233

Peuralinna T, Tanskanen M, Makela M, Polvikoski T, Paetau A, Kalimo H, Sulkava R, **Hardy J**, Lai SL, Arepalli S, Hernandez D, Traynor BJ, Singleton A, Tienari PJ, Myllykangas L. APOE and A beta PP Gene Variation in Cortical and Cerebrovascular Amyloid-beta Pathology and Alzheimer's Disease: A Population-Based Analysis. *J ALZHEIMERS DIS* 26(2):377-385 2011. PMID 21654062

Sinclair CD, Morrow JM, Miranda MA, Davagnanam I, Cowley PC, Mehta H, **Hanna MG, **Koltzenburg M**, **Yousry TA**, **Reilly MM**, Thornton JS. Skeletal muscle MRI magnetisation transfer ratio reflects clinical severity in peripheral neuropathies. *J Neurol Neurosurg Psychiatry*. 2011 May 25. [Epub ahead of print] PMID: 21613652

Kullmann DM. Interneuron networks in the hippocampus. *Curr Opin Neurobiol*. 2011 May 31. [Epub ahead of print] PubMed PMID: 21636266.

Malik B, Nirmalanathan N, Bilsland LG, La Spada AR, **Hanna MG, Schiavo G, Gallo JM, **Greensmith L**. Absence of disturbed axonal transport in spinal and bulbar muscular atrophy. *Hum Mol Genet*. 2011 May 1;20(9):1776-86. Epub 2011 Feb11. PMID: 21317158

Krahn M, Illa I, Lévy N, **Bushby K**. 172nd ENMC International Workshop: dysferlinopathies 29-31 January 2010, Naarden, The Netherlands. *Neuromuscul Disord*. 2011 Jul;21(7):503-12. Epub 2011 May 23. PMID: 21602046.

Bushby K. The value of collaboration in improving knowledge on rare diseases. *Can J Neurol Sci*. 2011 May;38(3):387. PMID: 21515491.

Reilich P, Horvath R, Krause S, Schramm N, **Turnbull DM, Trenell M, Hollingsworth KG, Gorman GS, Hans VH, Reimann J, Macmillan A, Turner L, Schollen A, Witte G, Czermin B, Holinski-Feder E, Walter MC, Schoser B, **Lochmüller H**. The phenotypic spectrum of neutral lipid storage myopathy due to mutations in the PNPLA2 gene. *J Neurol*. 2011 May 5. [Epub ahead of print] PMID: 21544567.

Barišić N, Chaouch A, Müller JS, **Lochmüller H**. Genetic heterogeneity and pathophysiological mechanisms in congenital myasthenic syndromes. *Eur J Paediatr Neurol*. 2011 May;15(3):189-96. Epub 2011 Apr 17. PMID: 21498094.

Fratte C, Raman P, Alston CL, Blakely EL, Craig K, Smith C, Evans J, Sellar A, Czermin B, **Hanna MG, Poulton J, Brierley C, Staunton TG, Turnpenny PD, Schaefer AM, **Chinnery PF**, Horvath R, **Turnbull DM**, Gorman GS, Taylor RW. RRM2B mutations are frequent in familial PEO with multiple

mtDNA deletions. *Neurology*. 2011 Jun 7;76(23):2032-4. PMID: 21646632

Scoto M, Cirak S, Mein R, Feng L, Manzur AY, Robb S, Childs AM, Quinlivan RM, Roper H, Jones DH, Longman C, Chow G, Pane M, Main M, **Hanna MG, **Bushby K**, Sewry C, Abbs S, Mercuri E, **Muntoni F**. SEPNI-related myopathies: clinical course in a large cohort of patients. *Neurology*. 2011 Jun 14;76(24):2073-8. PMID: 21670436.

Becker EB, Fogel BL, Rajakulendran S, Dulneva A, **Hanna MG**, Perlman SL, Geschwind DH, Davies KE. Candidate screening of the TRPC3 gene in cerebellar ataxia. *Cerebellum*. 2011 Jun;10(2):296-9. PMID: 21321808

Hall AM, Crawford C, Unwin RJ, **Duchen MR**, Peppiatt-Wildman CM. Multiphoton imaging of the functioning kidney. *J Am Soc Nephrol*. 2011 Jul;22(7):1297-304. Epub 2011 Jun 30.

Mazzone E, Bianco F, Martinelli D, Glanzman AM, Messina S, Sanctis RD, Main M, Eagle M, Florence J, Krossschell K, Vasco G, Pelliccioni M, Lombardo M, Pane M, Finkel R, **Muntoni F**, Bertini E, Mercuri E. Assessing upper limb function in nonambulant SMA patients: Development of a new module. *Neuromuscul Disord*. 2011 Jun;21(6):406-12. PMID: 21421316

Dowling JJ, Lillis S, Amburgey K, Zhou H, Al-Sarraj S, Buk SJ, Wraige E, Chow G, Abbs S, Leber S, Lachlan K, Baralle D, Taylor A, Sewry C, **Muntoni F**, Jungbluth H. King-Denborough syndrome with and without mutations in the skeletal muscle ryanodine receptor (RYR1) gene. *Neuromuscul Disord*. 2011 21(6):420-7. PMID: 21514828

Robb SA, Sewry CA, Dowling JJ, Feng L, Cullup T, Lillis S, Abbs S, Lees MM, Laporte J, Manzur AY, Knight RK, Mills KR, Pike MG, Kress W, Beeson D, Jungbluth H, Pitt MC, **Muntoni F**. Impaired neuromuscular transmission and response to acetylcholinesterase inhibitors in centronuclear myopathies. *Neuromuscul Disord*. 2011 Jun;21(6):379-86. PMID: 21440438

Mayhew A, Cano S, Scott E, Eagle M, **Bushby K, **Muntoni F**; on behalf of The North Star Clinical Network for Paediatric Neuromuscular Disease. Moving towards meaningful measurement: Rasch analysis of the North Star Ambulatory Assessment in Duchenne muscular dystrophy. *Dev Med Child Neurol*. 2011 53(6):535-542. PMID: 21410696

Godfrey CA, Foley R, Clement E, **Muntoni F**. Dystroglycanopathies; blurring boundaries. *Curr Opin Genet Dev*. 2011 Jun;21(3):278-85. PMID: 21397493

Alston CL, He L, Morris AA, Hughes I, Goede CD, **Turnbull DM**, McFarland R, Taylor RW. Maternally inherited mitochondrial DNA disease in consanguineous families. *Eur J Hum Genet*. 2011 Jun 29. [Epub ahead of print] PMID: 21712854.

Romaniuk L, **Turnbull DM**, Whittaker RG. Hyperventilation during the EEG is safe in mitochondrial disease. *Clin Neurophysiol*. 2011 Jun;122(6):1270-1. Epub 2010 Dec 15. PMID: 21111674.

Sultan SM, Allen E, Cooper RG, Agarwal S, Kiely P, Oddis CV, Vencovsky J, Lundberg IE, Dastmalchi M, **Hanna MG**, Isenberg DA. Interrater reliability and aspects of validity of the myositis damage index. *Ann Rheum Dis*. 2011 Jul;70(7):1272-6. PMID: 21622773

Cirak S, Arechavala-Gomez V, Guglieri M, Feng L, Torelli S, Anthony K, Abbs S, Garralda ME, Bourke J, Wells DJ, Dickson G, Wood MJ, Wilton SD, **Straub V, Kole R, Shrewsbury SB, Sewry C, **Morgan JE**, **Bushby K**, **Muntoni F** (2011). 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. *Lancet*. 2011 Jul 22. [Epub ahead of print] PMID: 21784508

Reid CA, **Kullmann DM**. GABAA receptor mutations in epilepsy (commentary on Lachance-Touchette et al.). *Eur J Neurosci*. 2011 Jul;34(2):235-6. doi: 10.1111/j.1460-9568.2011.07792.x. PubMed PMID: 21762463.

Statland JM, Wang Y, Richesson R, Bundy B, Herbelin L, Gomes J, Trivedi J, Venance S, Amato A, **Hanna MG**, Griggs R, Barohn RJ; Cinch Consortium. An interactive voice response diary for patients with non-dystrophic myotonia. *Muscle Nerve*. 2011 Jul;44(1):30-5. PMID: 21674518

- **Sinclair CD, Morrow JM, **Hanna MG**, **Reilly MM**, **Yousry TA**, Golay X, Thornton JS. Correcting radiofrequency inhomogeneity effects in skeletal muscle magnetisation transfer maps. *NMR Biomed*. 2011 Jul 27. [Epub ahead of print] PMID: 21796708
- Bönnemann CG, Rutkowski A, Mercuri E, **Muntoni F**; for the CMD Outcomes Consortium. 173rd ENMC international workshop: Congenital muscular dystrophy outcome measures 5-7 March 2010, Naarden, The Netherlands. *Neuromuscular Disorders*, 2011 Jul;21(7):513-522. PMID: 21641800
- Young C, Brutkowski W, Lien CF, Arkle S, **Lochmüller H**, Zabłocki K, Górecki DC. P2×7 purinoceptor alterations in dystrophic mdx mouse muscles: Relationship to pathology and potential target for treatment. *J Cell Mol Med*. 2011 Jul 27.[Epub ahead of print] PMID: 21794079.
- Mantyh PW, **Koltzenburg M**, Mendell LM, Tive L, Shelton DL. Antagonism of nerve growth factor-TrkA signaling and the relief of pain. *Anesthesiology*. 2011 Jul;115(1):189-204. PMID: 21602663
- Whittaker RG, **Turnbull DM**, Whittington MA, Cunningham MO. Impaired mitochondrial function abolishes gamma oscillations in the hippocampus through an effect on fast-spiking interneurons. *Brain*. 2011 Jul;134(Pt 7):e180; author reply e181. Epub 2011 Mar 4. Review. PMID: 21378098.
- Swalwell H, Kirby DM, Blakely EL, Mitchell A, Salemi R, Sugiana C, Compton AG, Tucker EJ, Ke BX, Lamont PJ, **Turnbull DM**, McFarland R, Taylor RW, Thorburn DR. Respiratory chain complex I deficiency caused by mitochondrial DNA mutations. *Eur J Hum Genet*. 2011 Jul;19(7):769-75.. Epub 2011 Mar 2. PMID: 21364701
- Apabhai S, Gorman GS, Sutton L, Elson JL, Plötz T, **Turnbull DM**, Trenell MI. Habitual physical activity in mitochondrial disease. *PLoS One*. 2011;6(7):e22294. Epub 2011 Jul 22. PMID: 21799815
- Trabzuni D, Ryten M, Walker R, Smith C, Imran S, Ramasamy A, Weale ME, **Hardy J**. Quality control parameters on a large dataset of regionally dissected human control brains for whole genome expression studies. *J Neurochem* 17 Aug 2011 PMID 21848658
- Matthews E, Miller JA, Macleod MR, Ironside J, Ambler G, Labrum R, Sud R, Holton JL, **Hanna MG**. Sodium and chloride channelopathies with myositis: Coincidence or connection? *Muscle Nerve*. 2011 Aug;44(2):283-8. Epub 2011 Jun 22. PMID: 21698652
- Muntoni F**, Wood MJ. Targeting RNA to treat neuromuscular diseases. *Nat Rev Drug Discov*. 2011 Aug 1;10(8):621-37. PMID: 21804598
- Colombo E, Romaggi S, Blasevich F, Mora M, Falcone C, **Lochmüller H**, Morandi L, Farina C. The neurotrophin receptor p75NTR is induced on mature myofibres in inflammatory myopathies and promotes myotube survival to inflammatory stress. *Neuropathol Appl Neurobiol*. 2011 Aug 18. [Epub ahead of print] PMID: 21851375.
- Chaouch A, Müller JS, Guergueltcheva V, Dusl M, Schara U, Rakocević-Stojanović V, Lindberg C, Scola RH, Werneck LC, Colomer J, Nascimento A, Vilchez JJ, Muelas N, Argov Z, Abicht A, **Lochmüller H**. A retrospective clinical study of the treatment of slow-channel congenital myasthenic syndrome. *J Neurol*. 2011 Aug 7. [Epub ahead of print] PMID: 21822932.
- Reilich P, Krause S, Schramm N, Klutzny U, Bulst S, Zehetmayer B, Schneiderat P, Walter MC, Schoser B, **Lochmüller H**. A novel mutation in the myotilin gene (MYOT) causes a severe form of limb girdle muscular dystrophy 1A (LGMD1A). *J Neurol*. 2011 Aug;258(8):1437-44. Epub 2011 Feb 20. PMID: 21336781.
- Lehtokari VL, Pelin K, Herczegfalvi A, Karcagi V, Pouget J, Franques J, Pellissier JF, Figarella-Branger D, von der Hagen M, Huebner A, Schoser B, **Lochmüller H**, Wallgren-Pettersson C. Nemaline myopathy caused by mutations in the nebulin gene may present as a distal myopathy. *Neuromuscul Disord*. 2011 Aug;21(8):556-62. Epub 2011 Jul 2. PMID: 21724397.
- **Nadarajah VD, van Putten M, Chaouch A, Garrood P, **Straub V**, **Lochmüller H**, Ginjaar HB, Aartsma-

Rus AM, van Ommen GJ, den Dunnen JT, 't Hoen PA. Serum matrix metalloproteinase-9 (MMP-9) as a biomarker for monitoring disease progression in Duchenne muscular dystrophy (DMD). *Neuromuscul Disord*. 2011 Aug;21(8):569-78. Epub 2011 Jul 2. PMID: 21724396.

Park U, Vastani N, Guan Y, Raja SN, **Koltzenburg M**, Caterina MJ. TRP vanilloid 2 knock-out mice are susceptible to perinatal lethality but display normal thermal and mechanical nociception. *J Neurosci*. 2011 Aug 10;31(32):11425-36. PMID: 21832173

Elstner M, **Turnbull DM**. Transcriptome analysis in mitochondrial disorders. *Brain Res Bull*. 2011 Aug 3. [Epub ahead of print] PMID: 21856382.

Muses S, Morgan JE, Wells DJ (2011). A new extensively characterised conditionally immortal muscle cell-line for investigating therapeutic strategies in muscular dystrophies. *PLoS One*. 2011;6(9):e24826. Epub 2011 Sep 14. PMID: 21935475

Ackroyd MR, Whitmore C, Prior S, Kaluarachchi M, Nikolic M, Mayer U, **Muntoni F, **Brown SC**. Fukutin-related protein alters the deposition of laminin in the eye and brain. *J Neurosci*. 2011 Sep 7;31(36):12927-35. PMID:21900571

Kullmann DM. What's wrong with the amygdala in temporal lobe epilepsy? *Brain*. 2011 Oct;134(Pt 10):2800-1. Epub 2011 Sep 14. PubMed PMID: 21921018.

Fletcher EV, **Kullmann DM**, Schorge S. Alternative Splicing Modulates Inactivation of Type 1 Voltage-gated Sodium Channels by Toggling an Amino Acid in the First S3-S4 Linker. *J Biol Chem*. 2011 Oct 21;286(42):36700-8. Epub 2011 Sep 2. PubMed PMID: 21890636.

Morrow JM, D'Sa S, Page RA, Hilali HA, Lunn MP, **Reilly MM**. Rituximab responsive multiple radiculopathies and cranial nerve palsies in association with chronic lymphocytic leukaemia. *J Neurol* 2011: Sep 2 (epub ahead of print). PMID 21887515

Murphy SM and **Reilly MM**. Amyloid Neuropathies. *ACJNR* 2011: 11; 16-19.

Murphy SM, Khan U, Alifrangis C, Hazell S, Hrouda D, Blake J, Ball J, Gabriel C, Markarian P, Rees J, Karim A, Seckl MJ, Lunn MP, **Reilly MM**. Anti Ma2-associated myeloradiculopathy: expanding the phenotype of anti-Ma2 associated paraneoplastic syndromes. *JNNP* 2011 (epub ahead of print) PMID 21205983

Owen N, Zhou H, Malygin AA, Sangha J, Smith LD, **Muntoni F**, Eperon IC. Design principles for bifunctional targeted oligonucleotide enhancers of splicing. *Nucleic Acids Research* 2011; 1;39(16):7194-208. PMID: 21602265

Klein A, Jungbluth H, Clement E, Lillis S, Abbs S, Munot P, Pane M, Wraige E, Schara U, **Straub V, Mercuri E, **Muntoni F**. Muscle magnetic resonance imaging in congenital myopathies due to ryanodine receptor type 1 gene mutations. *Arch Neurol*. 2011 Sep;68(9):1171-9. PMID: 21911697

Godfrey C, Clement E, Abbs S, **Muntoni F**. Exclusion of WWP1 mutations in a cohort of dystroglycanopathy patients. *Muscle & Nerve*, 2011; 4; 388-392. PMID: 21996799

Wood AJ, Müller JS, Jepson CD, Laval SH, **Lochmüller H, **Bushby K**, Barresi R, **Straub V**. Abnormal vascular development in zebrafish models for fukutin and FKRP deficiency. *Hum Mol Genet*. 2011 Sep 27. [Epub ahead of print] PMID: 21926082.

Ivanidze J, Hoffmann R, **Lochmüller H**, Engel AG, Hohlfeld R, Dornmair K. Inclusion body myositis: laser microdissection reveals differential up-regulation of IFN- γ signaling cascade in attacked versus nonattacked myofibers. *Am J Pathol*. 2011 Sep;179(3):1347-59. PMID: 21855683

Kachramanoglou C, Carlstedt T, **Koltzenburg M**, Choi D. Self-Mutilation in Patients After Nerve Injury May Not Be Due to Deafferentation Pain: A Case Report. *Pain Med*. 2011 Sep 21. [Epub ahead of print] PMID: 21939498.

****Muntoni F**, Torelli S, Wells DJ, **Brown SC**. Muscular dystrophies due to glycosylation defects: diagnosis and therapeutic strategies. *Curr Opin Neurol*. 2011 Oct;24(5):437-42. PMID: 21825985

Rajakulendran S, Parton M, Holton JL, **Hanna MG**. Clinical and pathological heterogeneity in late-onset partial merosin deficiency. *Muscle Nerve*. 2011 Oct;44(4):590-3. PMID: 21922472

Guergueltcheva V, Müller JS, Dusl M, Senderek J, Oldfors A, Lindbergh C, Maxwell S, Colomer J, Mallebrera CJ, Nascimento A, Vilchez JJ, Muelas N, Kirschner J, Nafissi S, Kariminejad A, Nilipour Y, Bozorgmehr B, Najmabadi H, Rodolico C, Sieb JP, Schlotter B, Schoser B, Herrmann R, Voit T, Steinlein OK, Najafi A, Urtizbera A, Soler DM, **Muntoni F, Hanna MG, Chaouch A, Straub V, Bushby K, Palace J, Beeson D, Abicht A, Lochmüller H. Congenital myasthenia syndrome with tubular aggregates caused by GFTP-1 mutations. 2011. *J Neurology*. Oct 6 epub ahead of print. PMID 21975507

Rossor AM, Kalmar B, **Greensmith L, Reilly MM. The distal hereditary motor neuropathies. *JNNP* 2011; Oct 25 (epub ahead of print) PMID 22028385

Murphy SM, Herrmann DM, McDermott MP, Scherer SS, Shy ME, **Reilly MM, Pareyson D**. Reliability of the CMT neuropathy score (second version) in Charcot Marie Tooth disease. *J Peripher Nerv Syst* 2011: 16; 191-198. PMID 22003934

Riviere JB, Ramalingham S, Lavastre V, Shekarabi M, Holbert S, Lafontaine J, Srour M, Merner N, Rochefort D, Hince P, Gaudet R, Mes-Masson AM, Baets J, Houlden H, Brais B, Nicholson G, Van Esch H, Nafissi S, De Jonghe P, **Reilly MM, Timmerman V, Dion PA, Rouleau PA**. KIF1A, an axonal transporter of synaptic vesicles, is mutated in hereditary sensory and autonomic neuropathy type 2. *Am J Hum Genet* 2011: 89: 219-230. PMID 21820098

Nethisinghe S, Clayton L, Vermeer S, Chapple JP, **Reilly MM, Bremner F, Giunti P**. Retinal imaging in Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay. *Neuro-ophthalmology* 2011: 35: 197-201.

Peters MJH, Van Nes SI, Vanhoutte EK, Bakkers M, Van Doorn PA, Merkies IS, and Faber CG; on behalf of the PeriNomS Study group. Revised normative values for grip strength with the Jamar dynamometer. *JPNS* 2011: 16: 47-50. PMID 21504502

Uusimaa J, Jungbluth H, Fratter C, Crisponi G, Feng L, Zeviani M, Hughes I, Treacy EP, Birks J, Brown GK, Sewry CA, McDermott M, **Muntoni F, Poulton J**. Reversible infantile respiratory chain deficiency is a unique, genetically heterogeneous mitochondrial disease. *J Med Genet*. 2011 Oct;48(10):660-8. PMID: 21931168

Sarkozy A, Windpassinger C, Hudson J, Dougan CF, Lecky B, Hilton-Jones D, Eagle M, Charlton R, Barresi R, **Lochmüller H, Bushby K, Straub V. Phenotypic heterogeneity in British patients with a founder mutation in the FHL1 gene. *Eur J Hum Genet*. 2011 Oct;19(10):1038-44. Epub 2011 Jun 1. PMID: 21629301.

Chamova T, Florez L, Guergueltcheva V, Raycheva M, Kaneva R, **Lochmüller H, Kalaydjieva L, Tournev I**. ANO10 c.1150_1151del is a founder mutation causing autosomal recessive cerebellar ataxia in Roma/Gypsies. *J Neurol*. 2011 Oct 19. [Epub ahead of print] PMID: 22008874.

Greaves LC, Reeve AK, Taylor RW, **Turnbull DM**. Mitochondrial DNA and Disease. *J Pathol*. 2011 Oct 12. [Epub ahead of print] PMID: 21989606.

Greaves LC, Barron MJ, Campbell-Shiel G, Kirkwood TB, **Turnbull DM**. Differences in the accumulation of mitochondrial defects with age in mice and humans. *Mech Ageing Dev*. 2011 Oct 12. [Epub ahead of print] PMID: 22015485.

Mutsaers CA, Wishart TM, Lamont DJ, Riessland M, Schreml J, Comley LH, Murray LM, Parson SH, **Lochmüller H, Wirth B, Talbot K, Gillingwater TH**. Reversible molecular pathology of skeletal muscle in spinal muscular atrophy. *Hum Mol Genet*. 2011 Nov 15;20(22):4334-44. Epub 2011 Aug 12. PMID: 21840928.

Janiczek RL, Gambarota G, Sinclair CD, **Yousry TA, Thornton JS, Golay X, Newbould RD**. Simultaneous T(2) and lipid quantitation using IDEAL-CPMG. *Magn Reson Med*. 2011 Nov;66(5):1293-302. Epub 2011 May 20. PMID: 21604292.

Matthews E, Davis M, Griggs RC, Haworth E, **Hanna MG**. Acetazolamide efficacy in hypokalemic periodic paralysis and the predictive role of genotype. *Neurology*. 2011. Nov 29;77(22):1960-4. [Epub ahead of print] PMID: 22094484.

Burge J and **Hanna MG**. Novel insights into the pathomechanisms of skeletal muscle channelopathies. *Current Opinion in Neurotherapeutics*. 2011 Nov 15. [Epub ahead of print]. PMID: 22083238

In Press

Hudson H, Yu-Wai-Man P, Griffiths PG, Horvath R, Carelli V, Zeviani M, **Chinnery PF**. Variation in MAPT is not a contributing factor to the incomplete penetrance in LHON. *Mitochondrion* 2011: In press. PMID:21397051

Traba J, Szabadkai G, del Arco, A., **Duchen, MR** and Satrústegui, J (2011) SCaMC-1 mediated adenine nucleotide import augments mitochondrial Ca²⁺ buffering capacity and confers resistance to oxidative stress induced cell death in cancer cells, *Cell Death and Differentiation*, in press.

Girard, M, Larivière, R, Parfitt, DA, Deane, E, Gaudet R, Blondeau F, Prenosil G, Vermeulen EGM, **Duchen MR**, Gehring K, McKinney RA, Brais B, Chapple JP and McPherson PS, Mitochondrial dysfunction and Purkinje cell loss in autosomal recessive spastic ataxia of Charlevoix-Saguenay *PNAS*, in press.

Rajakulandren S and **Hanna MG**. Neuronal calcium channelopathies and neurological disease. *Current Opinion in Neurotherapeutics in press*

Plotz P, Porto S, Holton J, Matthews E, **Hanna MG**. Necrotising myositis and anti sRNP antibodies. *Neurology in press*.

Rayan D, Sweeney MG, Haworth A, Davis M, **Hanna MG**. Exon deletion/duplication in the muscle chloride channel can cause myotonia congenita. *Neurology in press*

Tan V, Rayan D, **Hanna MG**, Bostock H. Muscle velocity recovery cycles is a sensitive tool to identify patients with Anderson Tawil muscle channelopathy. *Journal of Neurology Neurosurgery and Psychology in press*

Turner C, Hilton-Jones D, Parton M, Miller A and **Hanna MG**. MRC Centre workshop on translational research opportunities in Myotonic Dystrophy Neuromuscular Disorders *in press*

Howarth A, Ryan D, Davis M, **Hanna MG**. Genetic variation across muscle channels and risk of muscle disease. *Neurogenetics in press*

Sweeney MG, Pitceathly R, **Hanna MG**. Genetic variome and prediction of mtDNA variation pathogenicity. *Neurogenetics in press*

Read J, Kinali M, **Muntoni F**, Weaver T, Garralda ME. Siblings of young people with Duchenne muscular dystrophy - A qualitative study of impact and coping. *Eur J Paediatr Neurol*. 2010 Aug 19.

Godfrey C, Clement E, Abbs S, **Muntoni F**. Exclusion of WWP1 mutations in a cohort of dystroglycanopathy patients. *Muscle & Nerve*, accepted for publication.

Ambegaonkar G, Manzur AY, Robb SA, Kinali M, Muntoni F. The multiple phenotypes of Arthrogryposis multiplex congenita with reference to the neurogenic variant. *Eur J Paediatr Neurol*. 2011 Feb 21

Logan CV, Lucke B, Pottinger C, Abdelhamed ZA, Parry DA, Szymanska K, Diggle CP, van Riesen A, Morgan JE, Markham G, Ellis I, Manzur AY, Markham AF, Shires M, Helliwell T, Scoto M, Hübner C, Bonthron DT, Taylor GR, Sheridan E, **Muntoni F**, Carr IA, Schuelke M, Johnson CA. Mutations in MEGF10, a regulator of satellite cell myogenesis, cause early onset myopathy, areflexia, respiratory distress and dysphagia (EMARDD). *Nat Gen*, accepted for publication.

Scott E, Eagle M, Mayhew A, Freeman J, Main M, Sheehan J, Manzur A, **Muntoni F**; The North Star Clinical Network for Paediatric Neuromuscular Disease. Development of a Functional Assessment Scale for Ambulatory Boys with Duchenne Muscular Dystrophy. *Physiother Res Int*. 2011 Sep 23. doi: 10.1002/pri.520.

Tosetti M, Linsalata S, Battini R, Volpi L, Cini C, Presciutti O, **Muntoni F**, Cioni G, Siciliano G. Muscle metabolic alterations assessed by 31-phosphorus magnetic resonance spectroscopy in mild becker muscular dystrophy. *Muscle Nerve*. 2011 May 26. doi: 10.1002/mus.22181. [Epub ahead of print]

D Boërio, **L Greensmith** and H Bostock (2011) A model of mouse motor nerve excitability and the effects of polarizing current. *Journal of the Peripheral Nervous System* *In press*

A Smith, S Passey, **L Greensmith**, V Mudera & MP Lewis (2011) Characterisation and optimisation of a simple, repeatable system for the long term in vitro culture of aligned myotubes in 3D *Journal of Cellular Biochemistry*. *In press*

Under Review

Tracey A. Willis, **Kieren G. Hollingsworth**, Anna Coombs, Marie-Louise Sveen, Soren Anderson, Tanya Stojkovic, Michelle Eagle, Anna Mayhew, Paulo Loureiro de Sousa, Liz Dewar, Jasper M. Morrow, Chris D. Sinclair, **John S. Thornton**, **Katie Bushby**, **Hanns Lochmuller**, **Michael G. Hanna**, Jean-Yves Hogrel, Pierre G. Carlier, John Vissing, **Volker Straub** MRI muscle fat quantification in Limb Girdle Muscular Dystrophy 2I is more sensitive to detect disease progression than clinical Assessments of muscle strength and function. *Annals of Neurology*

Pitceathly R, Rahman S, **Hanna MG**. How do deletions in Mitochondrial DNA cause neuromuscular disorders? *Neuromuscular Disorders Review*

In Preparation

Author list to include: Jasper M Morrow, Emma Matthews, Dipa L Raja Rayan, Arne Fischmann, Ibrahim Amer, Christopher JD Sinclair, **John S Thornton**, **Mary M Reilly**, **Tarek A Yousry**, **Michael G Hanna**. Skeletal muscle MRI shows distinct abnormalities in genetically proven non-dystrophic myotonias.

Author list to include: Arne Fischmann, Christopher DJ Sinclair, Jasper M Morrow, **Mary M Reilly**, **Michael G Hanna**, **John S Thornton**, **Tarek A Yousry**. Effects of patient positioning and anatomical localisation upon reproducibility of quantitative MRI of lower limb muscles.

Author list to include: Morrow JM, Sinclair CDJ, Fischmann A, **Reilly MM**, **Hanna MG**, **Thornton JS**, **Yousry TA**. Quantitative MRI of skeletal muscle in healthy volunteers: inter-scan reproducibility and normal inter-subject and between muscle variation.

Author list to include: Morrow JM, Sinclair CDJ, Fischmann A, **Thornton JS**, **Yousry TA**, **Reilly MM**, **Hanna MG**. Validation as an outcome measure of quantitative MRI of skeletal muscle in Charcot-Marie-Tooth disease and inclusion body myositis.

Author list to include: Fischmann A, Morrow JM, **Sinclair CDJ**, **Thornton JS**, **Reilly MM**, **Miller J**, **Yousry TA**, **Hanna MG**. Pattern of Involvement in Inclusion Body Myositis: a new scoring system.

Author list to include: Pitceathly RDS, **Sinclair CDJ**, Ali N, Bremner F, Morrow JM, Davagnanam I, **Rahman S**, Plant G, **Thornton JS**, **Yousry T**, **Hanna MG**. Quantitative MRI of extra-ocular muscles in single mitochondrial DNA deletion disorders.

Author list to include: Tracey A. Willis, Kieren G. **Hollingsworth**, Marie-Louise Sveen, Tanja Stojkovic, Michelle Eagle, Anna Mayhew, Paulo Loureiro, Jasper **Morrow**, Chris D. **Sinclair**, John S. **Thornton**, Katie **Bushby**, Hanns **Lochmüller**, Mike **Hanna**, Pierre G. Carlier, John Vissing, Volker **Straub**. Muscle involvement in Limb Girdle Muscular Dystrophy 2I; a multicentre comparison of semi quantitative and quantitative MRI techniques.

Jaffa F, Reilly M, **Hanna MG**, Houlden H, Next generation sequencing :implications for neuromuscular disease and practice. *Journal of Neurology, Neurosurgery and Psychiatry* *Review*