

Inherited Neuropathies Consortium

Book Chapters

1. Scherer SS. Genes and Inherited Neuropathies. *Companion to Peripheral Neuropathy*. Philadelphia, PA: Saunders Elsevier; 2010:335-342.
2. Murphy SM, Reilly M. Hereditary amyloid neuropathy. *Autonomic Failure: a textbook of clinical disorders of the autonomic nervous system*. New York: Oxford University Press; 2012.
3. Scherer SS, Feltri ML, Wrabetz L. Genetic Mutations Affecting Myelin Formation. In: Kettenmann H, Ransom BR, eds. *Neuroglia*. New York, NY: Oxford University Press; 2012:798-808.
4. Shy M. Peripheral Neuropathies. *Goldman's Cecil Medicine: Expert Consult Premium Edition*. Philadelphia, PA: Elsevier; 2012:2396-2409.
5. Murphy SM, Laura M, Reilly MM. DNA testing in hereditary neuropathies. *Handbook of clinical neurology*. Vol 115. 2013/08/13 ed2013:213-232. PMID: 23931782
6. Rossor AR, MM. Charcot-Marie-Tooth Disease. In: Hilton-Jones D, Turner M, eds. *Oxford Textbook of Neuromuscular Disorders*. Oxford, UK: Oxford University Press; 2014:61-74.
7. Brennan K, Shy M. Hereditary Neuropathies in Late Childhood and Adolescence. In: Darras B, Jones H, Ryan M, De Vivo D, eds. *Neuromuscular Disorders of Infancy, Childhood and Adolescence: A Clinicians Approach*. 2nd ed: Elsevier Inc; 2015:319-339.
8. Saporta M, Shy M. Peripheral Neuropathies. In: Zigmond M, Coyle J, Rowland L, eds. *Neurobiology of Brain Disorders: Biological Basis of Neurological and Psychiatric Disorders*. 1st ed: Academic Press; 2015:167-188.

Abstracts Presented at Conferences

1. Hall CA, Bacon CJ, Shy ME, Inherited Neuropathies Consortium, Rare Diseases Clinical Research Network Data Management and Coordinating Center. The Rare Diseases Clinical Research Network Contact Registry for the Inherited Neuropathies Consortium. Paper presented at: Charcot-Marie-Tooth Association, 5th International CMT Consortium Meeting; Jun. 25-27, 2013; Antwerp, Belgium.
2. Hainline C, Rizzo D, Shy ME, Inherited Neuropathies Consortium, Rare Diseases Clinical Research Network Data Management and Coordinating Center. Enhancements to the RDCRN Contact Registry for the Inherited Neuropathies Consortium. Poster presented at Peripheral Nerve Society Annual Meeting; Jul. 8-12, 2017; Sitges, Spain.

Conference Proceedings

1. Sanmaneechai O, Feely S, Finkel R, et al. Natural History Baseline Phenotype and Genotype of Hereditary Motor Sensory Peripheral Neuropathies Caused by Mutation in the Myelin Protein

Zero. Paper presented at: 2013 Peripheral Nerve Society Biennial Meeting; June 29–July 3, 2013; Saint-Malo, France.

Journal Articles

1. Dimos JT, Rodolfa KT, Niakan KK, Weisenthal LM, Mitsumoto H, Chung W, Croft GF, Saphier G, Leibel R, Goland R, Wichterle H, Henderson CE, Eggan K. Induced pluripotent stem cells generated from patients with ALS can be differentiated into motor neurons. *Science*. Aug 29 2008;321(5893):1218-1221. PMID: 18669821
2. Houlden H, Laura M, Ginsberg L, et al. The phenotype of Charcot-Marie-Tooth disease type 4C due to SH3TC2 mutations and possible predisposition to an inflammatory neuropathy. *Neuromuscul. Disord*. Apr 2009;19(4):264-269. PMID: 19272779
3. Siskind C, Feely SM, Bernes S, Shy ME, Garbern JY. Persistent CNS dysfunction in a boy with CMT1X. *J. Neurol. Sci*. Apr 15 2009;279(1-2):109-113. PMID: 19193385
4. Shy M. Ascorbic acid for treatment of CMT1A: the jury is still out. *Lancet Neurol*. Jun 2009;8(6):505-507. PMID: 19427270
5. Katona I, Wu X, Feely SM, et al. PMP22 expression in dermal nerve myelin from patients with CMT1A. *Brain*. Jul 2009;132(Pt 7):1734-1740. PMID: 19447823, PMCID: PMC2724915
6. Ramdharry GM, Day BL, Reilly MM, Marsden JF. Hip flexor fatigue limits walking in Charcot-Marie-Tooth disease. *Muscle Nerve*. Jul 2009;40(1):103-111. PMID: 19405092, PMCID: PMC3734534
7. Ramchandren S, Shy ME, Finkel RS. Quality of life in children with CMT type 1A. *Lancet Neurol*. Oct 2009;8(10):880-881; author reply 881. PMID: 19747650
8. Hedges DJ, Burges D, Powell E, Almonte C, Huang J, Young S, Boese B, Schmidt M, Pericak-Vance MA, Martin E, Zhang X, Harkins TT, Zuchner S. Exome sequencing of a multigenerational human pedigree. *PLoS ONE*. December 2009;4(12):e8232. PMID: 20011588, PMCID: PMC2788131
9. Reilly MM, Shy ME. Diagnosis and new treatments in genetic neuropathies. *J. Neurol. Neurosurg. Psychiatry*. Dec 2009;80(12):1304-1314. PMID: 19917815
10. Burns J, Ryan MM, Ouvrier RA. Quality of life in children with Charcot-Marie-Tooth disease. *J. Child Neurol*. Mar 2010;25(3):343-347. PMID: 19713553
11. Huang J, Wu X, Montenegro G, et al. Copy number variations are a rare cause of non-CMT1A Charcot-Marie-Tooth disease. *J. Neurol*. May 2010;257(5):735-741. PMID: 19949810, PMCID: PMC2865568
12. Burns J, Ramchandren S, Ryan MM, Shy M, Ouvrier RA. Determinants of reduced health-related quality of life in pediatric inherited neuropathies. *Neurology*. Aug 24 2010;75(8):726-731. PMID: 20733147, PMCID: PMC2931653
13. Martin ER, Kinnamon DD, Schmidt MA, Powell EH, Zuchner S, Morris RW. SeqEM: an adaptive genotype-calling approach for next-generation sequencing studies. *Bioinformatics*. Nov 15 2010;26(22):2803-2810. PMID: 20861027, PMCID: PMC2971572

14. Reilly MM, Shy ME, Muntoni F, Pareyson D. 168th ENMC International Workshop: outcome measures and clinical trials in Charcot-Marie-Tooth disease (CMT). *Neuromuscul. Disord.* Dec 2010;20(12):839-846. PMID: 20850975
15. Saporta MA, Grskovic M, Dimos JT. Induced pluripotent stem cells in the study of neurological diseases. *Stem cell research & therapy.* 2011;2(5):37. PMID: 21936964, PMCID: PMC3308034
16. Smith LJ, Murphy SM, Holmes P, Reilly MM, Reiniger L, Thom M, Lunn MP. A painful right leg. *BMJ.* 2011;342:d1009. PMID: 21411806
17. Amato AA, Reilly MM. The death panel for Charcot-Marie-Tooth panels. *Ann. Neurol.* Jan 2011;69(1):1-4. PMID: 21280068
18. Saporta AS, Sottile SL, Miller LJ, Feely SM, Siskind CE, Shy ME. Charcot-Marie-Tooth disease subtypes and genetic testing strategies. *Ann. Neurol.* Jan 2011;69(1):22-33. PMID: 21280073, PMCID: PMC3058597
19. Patzko A, Shy ME. Update on Charcot-Marie-Tooth disease. *Curr. Neurol. Neurosci. Rep.* Feb 2011;11(1):78-88. PMID: 21080241, PMCID: PMC3685483
20. Russo M, Laura M, Polke JM, et al. Variable phenotypes are associated with PMP22 missense mutations. *Neuromuscul. Disord.* Feb 2011;21(2):106-114. PMID: 21194947
21. Zuchner S, Dallman J, Wen R, Beecham G, Naj A, Farooq A, Kohli MA, Whitehead PL, Hulme W, Konidari I, Edwards YJ, Cai G, Peter I, Seo D, Buxbaum JD, Haines JL, Blanton S, Young J, Alfonso E, Vance JM, Lam BL, Pericak-Vance MA. Whole-exome sequencing links a variant in DHDDS to retinitis pigmentosa. *Am. J. Hum. Genet.* Feb 11 2011;88(2):201-206. PMID: 21295283, PMCID: PMC3035708
22. Montenegro G, Powell E, Huang J, Speziani F, Edwards YJ, Beecham G, Hulme W, Siskind C, Vance J, Shy M, Zuchner S. Exome sequencing allows for rapid gene identification in a Charcot-Marie-Tooth family. *Ann. Neurol.* Mar 2011;69(3):464-470. PMID: 21254193, PMCID: PMC3066289
23. Murphy SM, Polke J, Manji H, Blake J, Reiniger L, Sweeney M, Houlden H, Brandner S, Reilly MM. A novel mutation in the nerve-specific 5'UTR of the GJB1 gene causes X-linked Charcot-Marie-Tooth disease. *J. Peripher. Nerv. Syst.* Mar 2011;16(1):65-70. PMID: 21504505
24. 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 p.Ile112Thr mutation. *Neuromuscul. Disord.* Mar 2011;21(3):223-226. PMID: 21256749
25. Reilly MM, Murphy SM, Laura M. Charcot-Marie-Tooth disease. *J. Peripher. Nerv. Syst.* Mar 2011;16(1):1-14. PMID: 21504497
26. Pareyson D, Reilly MM, Schenone A, et al. Ascorbic acid in Charcot-Marie-Tooth disease type 1A (CMT-TRIAAL and CMT-TRAUK): a double-blind randomised trial. *Lancet Neurol.* Apr 2011;10(4):320-328. PMID: 21393063, PMCID: PMC3154498
27. Katona I, Zhang X, Bai Y, et al. Distinct pathogenic processes between Fig4-deficient motor and sensory neurons. *Eur. J. Neurosci.* Apr 2011;33(8):1401-1410. PMID: 21410794
28. Shy ME. Inherited peripheral neuropathies. *Continuum (Minneapolis, Minn.).* Apr 2011;17(2 Neurogenetics):294-315. PMID: 22810821

29. 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*. May 17 2011;76(20):1690-1696. PMID: 21508331, PMCID: PMC3100135
30. Scherer SS. CMT2A: the name doesn't tell the whole story. *Neurology*. May 17 2011;76(20):1686-1687. PMID: 21508332
31. Siskind CE, Murphy SM, Ovens R, Polke J, Reilly MM, Shy ME. Phenotype expression in women with CMT1X. *J. Peripher. Nerv. Syst.* Jun 2011;16(2):102-107. PMID: 21692908
32. Saporta MA, Katona I, Zhang X, Roper HP, McClelland 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.* Jun 2011;68(6):814-821. PMID: 21670407, PMCID: PMC3711535
33. McCorquodale DS, 3rd, Montenegro G, Peguero A, et al. Mutation screening of mitofusin 2 in Charcot-Marie-Tooth disease type 2. *J. Neurol.* Jul 2011;258(7):1234-1239. PMID: 21258814, PMCID: PMC3125445
34. Polke JM, Laura M, Pareyson D, et al. Recessive axonal Charcot-Marie-Tooth disease due to compound heterozygous mitofusin 2 mutations. *Neurology*. Jul 12 2011;77(2):168-173. PMID: 21715711, PMCID: PMC3140074
35. Almodovar JL, Ferguson M, McDermott MP, Lewis RA, Shy ME, Herrmann DN. In vivo confocal microscopy of Meissner corpuscles as a novel sensory measure in CMT1A. *J. Peripher. Nerv. Syst.* Sep 2011;16(3):169-174. PMID: 22003930
36. Murphy SM, Herrmann DN, 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.* Sep 2011;16(3):191-198. PMID: 22003934, PMCID: PMC3754828
37. Shy ME, Patzko A. Axonal Charcot-Marie-Tooth disease. *Curr. Opin. Neurol.* Oct 2011;24(5):475-483. PMID: 21892080
38. Miller LJ, Saporta AS, Sottile SL, Siskind CE, Feely SM, Shy ME. Strategy for genetic testing in Charcot-Marie-disease. *Acta Myol.* Oct 2011;30(2):109-116. PMID: 22106713, PMCID: PMC3235845
39. Siskind CE, Shy ME. Genetics of neuropathies. *Semin. Neurol.* Nov 2011;31(5):494-505. PMID: 22266887
40. Hutton EJ, Carty L, Laura M, Houlden H, Lunn MP, Brandner S, Mirsky R, Jessen K, Reilly MM. c-Jun expression in human neuropathies: a pilot study. *J. Peripher. Nerv. Syst.* Dec 2011;16(4):295-303. PMID: 22176144
41. Scherer SS. The debut of a rational treatment for an inherited neuropathy? *J. Clin. Invest.* Dec 2011;121(12):4624-4627. PMID: 22045569, PMCID: PMC3226011
42. Holzbaur EL, Scherer SS. Microtubules, axonal transport, and neuropathy. *N. Engl. J. Med.* Dec 15 2011;365(24):2330-2332. PMID: 22168648, PMCID: PMC3776444
43. Rossor AM, Kalmar B, Greensmith L, Reilly MM. The distal hereditary motor neuropathies. *J. Neurol. Neurosurg. Psychiatry.* Jan 2012;83(1):6-14. PMID: 22028385
44. 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*. Jan 2012;83(1):29-32. PMID: 21613652

45. McLaughlin HM, Sakaguchi R, Giblin W, et al. A recurrent loss-of-function alanyl-tRNA synthetase (AARS) mutation in patients with Charcot-Marie-Tooth disease type 2N (CMT2N). *Hum. Mutat*. Jan 2012;33(1):244-253. PMID: 22009580, PMCID: PMC3240693
46. Murphy SM, Davidson GL, Brandner S, Houlden H, Reilly MM. Mutation in FAM134B causing severe hereditary sensory neuropathy. *J. Neurol. Neurosurg. Psychiatry*. Jan 2012;83(1):119-120. PMID: 21115472, PMCID: PMC3721196
47. Patzko A, Shy ME. Charcot-Marie-Tooth disease and related genetic neuropathies. *Continuum (Minneapolis, Minn.)*. Feb 2012;18(1):39-59. PMID: 22810069
48. Montenegro G, Rebelo AP, Connell J, et al. Mutations in the ER-shaping protein reticulon 2 cause the axon-degenerative disorder hereditary spastic paraplegia type 12. *J. Clin. Invest*. Feb 1 2012;122(2):538-544. PMID: 22232211, PMCID: PMC3266795
49. 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. *J. Neurol. Neurosurg. Psychiatry*. Feb 2012;83(2):232-233. PMID: 21205983, PMCID: PMC3719382
50. Davidson G, Murphy S, Polke J, et al. Frequency of mutations in the genes associated with hereditary sensory and autonomic neuropathy in a UK cohort. *J. Neurol*. Aug 2012;259(8):1673-1685. PMID: 22302274, PMCID: PMC3752368
51. Morrow JM, D'Sa S, Page RA, Hilali MA, Lunn MP, Reilly MM. Rituximab responsive multiple radiculopathies and cranial nerve palsies in association with chronic lymphocytic leukaemia. *J. Neurol*. Mar 2012;259(3):571-573. PMID: 21887515
52. Norton N, Robertson PD, Rieder MJ, Zuchner S, Rampersaud E, Martin E, Li D, Nickerson DA, Hershberger RE. Evaluating pathogenicity of rare variants from dilated cardiomyopathy in the exome era. *Circ. Cardiovasc. Genet*. Apr 1 2012;5(2):167-174. PMID: 22337857, PMCID: PMC3332064
53. Lloyd TE, Machamer J, O'Hara K, et al. The p150(Glued) CAP-Gly domain regulates initiation of retrograde transport at synaptic termini. *Neuron*. Apr 26 2012;74(2):344-360. PMID: 22542187, PMCID: PMC3353876
54. Burns J, Ouvrier R, Estilow T, Shy R, Laura M, Pallant JF, Lek M, Muntoni F, Reilly MM, Pareyson D, Acsadi G, Shy ME, Finkel RS. Validation of the Charcot-Marie-Tooth disease pediatric scale as an outcome measure of disability. *Ann. Neurol*. May 2012;71(5):642-652. PMID: 22522479, PMCID: PMC3335189
55. Harms MB, Ori-McKenney KM, Scoto M, Tuck EP, Bell S, Ma D, Masi S, Allred P, Al-Lozi M, Reilly MM, Miller LJ, Jani-Acsadi A, Pestronk A, Shy ME, Muntoni F, Vallee RB, Baloh RH. Mutations in the tail domain of DYNC1H1 cause dominant spinal muscular atrophy. *Neurology*. May 29 2012;78(22):1714-1720. PMID: 22459677, PMCID: PMC3359582
56. Miller LJ, Patzko A, Lewis RA, Shy ME. Phenotypic presentation of the Ser63Del MPZ mutation. *J. Peripher. Nerv. Syst*. Jun 2012;17(2):197-200. PMID: 22734905, PMCID: PMC3731745

57. Rossor AM, Davidson GL, Blake J, Polke JM, Murphy SM, Houlden H, Innes A, Kalmar B, Greensmith L, Reilly MM. A novel p.Glu175X premature stop mutation in the C-terminal end of HSP27 is a cause of CMT2. *J. Peripher. Nerv. Syst.* Jun 2012;17(2):201-205. PMID: 22734906
58. Rossor AM, Murphy S, Reilly MM. Knee bobbing in Charcot-Marie-Tooth disease. *Practical neurology.* Jun 2012;12(3):182-183. PMID: 22661351, PMCID: PMC3736802
59. Jaffer F, Murphy SM, Scoto M, Healy E, Rossor AM, Brandner S, Phadke R, Selcen D, Jungbluth H, Muntoni F, Reilly MM. BAG3 mutations: another cause of giant axonal neuropathy. *J. Peripher. Nerv. Syst.* Jun 2012;17(2):210-216. PMID: 22734908
60. Voermans NC, Kleefstra T, Gabreels-Festen AA, et al. Severe Dejerine-Sottas disease with respiratory failure and dysmorphic features in association with a PMP22 point mutation and a 3q23 microdeletion. *J. Peripher. Nerv. Syst.* Jun 2012;17(2):223-225. PMID: 22734911
61. Murphy SM, Laura M, Fawcett K, Pandraud A, Liu YT, Davidson GL, Rossor AM, Polke JM, Castleman V, Manji H, Lunn MP, Bull K, Ramdharry G, Davis M, Blake JC, Houlden H, Reilly MM. Charcot-Marie-Tooth disease: frequency of genetic subtypes and guidelines for genetic testing. *J. Neurol. Neurosurg. Psychiatry.* Jul 2012;83(7):706-710. PMID: 22577229, PMCID: PMC3736805
62. Saporta MA, Shy BR, Patzko A, Bai Y, Pennuto M, Ferri C, Tinelli E, Saveri P, Kirschner D, Crowther M, Southwood C, Wu X, Gow A, Feltri ML, Wrabetz L, Shy ME. MpzR98C arrests Schwann cell development in a mouse model of early-onset Charcot-Marie-Tooth disease type 1B. *Brain.* Jul 2012;135(Pt 7):2032-2047. PMID: 22689911, PMCID: PMC3381724
63. Arthur-Farraj PJ, Murphy SM, Laura M, Lunn MP, Manji H, Blake J, Ramdharry G, Fox Z, Reilly MM. Hand weakness in Charcot-Marie-Tooth disease 1X. *Neuromuscul. Disord.* Jul 2012;22(7):622-626. PMID: 22464564, PMCID: PMC3657175
64. Murphy SM, Ovens R, Polke J, Siskind CE, Laura M, Bull K, Ramdharry G, Houlden H, Murphy RP, Shy ME, Reilly MM. X inactivation in females with X-linked Charcot-Marie-Tooth disease. *Neuromuscul. Disord.* Jul 2012;22(7):617-621. PMID: 22483671, PMCID: PMC3657177
65. Osterloh JM, Yang J, Rooney TM, Fox AN, Adalbert R, Powell EH, Sheehan AE, Avery MA, Hackett R, Logan MA, MacDonald JM, Ziegenfuss JS, Milde S, Hou YJ, Nathan C, Ding A, Brown RH, Jr., Conforti L, Coleman M, Tessier-Lavigne M, Zuchner S, Freeman MR. dSarm/Sarm1 is required for activation of an injury-induced axon death pathway. *Science.* Jul 27 2012;337(6093):481-484. PMID: 22678360
66. Abrams CK, Scherer SS. Gap junctions in inherited human disorders of the central nervous system. *Biochim. Biophys. Acta.* Aug 2012;1818(8):2030-2047. PMID: 21871435, PMCID: PMC3771870
67. Shy ME. Lessons from London. *J. Neurol. Neurosurg. Psychiatry.* Aug 2012;83(8):767-768. PMID: 22696588, PMCID: PMC3721145
68. Burns J, Ouvrier R, Estilow T, Shy R, Laura M, Eichinger K, Muntoni F, Reilly MM, Pareyson D, Acsadi G, Shy ME, Finkel RS. Symmetry of foot alignment and ankle flexibility in paediatric Charcot-Marie-Tooth disease. *Clin. Biomech.* Aug 2012;27(7):744-747. PMID: 22424781, PMCID: PMC3389135
69. Davidson G, Murphy S, Polke J, Laura M, Salih M, Muntoni F, Blake J, Brandner S, Davies N, Horvath R, Price S, Donaghy M, Roberts M, Foulds N, Ramdharry G, Soler D, Lunn M, Manji H, Davis M, Houlden H, Reilly M. Frequency of mutations in the genes associated with hereditary

sensory and autonomic neuropathy in a UK cohort. *J. Neurol.* Aug 2012;259(8):1673-1685. PMID: 22302274, PMCID: PMC3752368

70. Michell AW, Gaitatzis A, Burge J, Reilly MM, Kapoor R, Koltzenburg M. Isolated motor conduction block associated with infliximab. *J. Neurol.* Aug 2012;259(8):1758-1760. PMID: 22349873, PMCID: PMC3125445
71. Pitceathly RD, Murphy SM, Cottenie E, Chalasani A, Sweeney MG, Woodward C, Mudanohwo EE, Hargreaves I, Heales S, Land J, Holton JL, Houlden H, Blake J, Champion M, Flinter F, Robb SA, Page R, Rose M, Palace J, Crowe C, Longman C, Lunn MP, Rahman S, Reilly MM, Hanna MG. Genetic dysfunction of MT-ATP6 causes axonal Charcot-Marie-Tooth disease. *Neurology.* Sep 11 2012;79(11):1145-1154. PMID: 22933740, PMCID: PMC3525307
72. Ramdharry GM, Day BL, Reilly MM, Marsden JF. Foot drop splints improve proximal as well as distal leg control during gait in Charcot-Marie-Tooth disease. *Muscle Nerve.* Oct 2012;46(4):512-519. PMID: 22987691
73. Zimon M, Baets J, Almeida-Souza L, De Vriendt E, Nikodinovic J, Parman Y, Battaloglu E, Matur Z, Guergueltcheva V, Tournev I, Auer-Grumbach M, De Rijk P, Petersen BS, Muller T, Fransen E, Van Damme P, Loscher WN, Barisic N, Mitrovic Z, Previtali SC, Topaloglu H, Bernert G, Beleza-Meireles A, Todorovic S, Savic-Pavicevic D, Ishpekova B, Lechner S, Peeters K, Ooms T, Hahn AF, Zuchner S, Timmerman V, Van Dijck P, Rasic VM, Janecke AR, De Jonghe P, Jordanova A. Loss-of-function mutations in HINT1 cause axonal neuropathy with neuromyotonia. *Nat. Genet.* Oct 2012;44(10):1080-1083. PMID: 22961002
74. Scherer SS, Kleopa KA. X-linked Charcot-Marie-Tooth disease. *J. Peripher. Nerv. Syst.* Dec 2012;17 Suppl 3:9-13. PMID: 23279425, PMCID: PMC3779456
75. Estilow T, Kozin SH, Glanzman AM, Burns J, Finkel RS. Flexor digitorum superficialis opposition tendon transfer improves hand function in children with Charcot-Marie-Tooth disease: case series. *Neuromuscul. Disord.* Dec 2012;22(12):1090-1095. PMID: 22944171, PMCID: PMC3883304
76. Fawcett KA, Murphy SM, Polke JM, Wray S, Burchell VS, Manji H, Quinlivan RM, Zdebik AA, Reilly MM, Houlden H. Comprehensive analysis of the TRPV4 gene in a large series of inherited neuropathies and controls. *J. Neurol. Neurosurg. Psychiatry.* Dec 2012;83(12):1204-1209. PMID: 22851605
77. Patzko A, Bai Y, Saporta MA, Katona I, Wu X, Vizzuso D, Feltri ML, Wang S, Dillon LM, Kamholz J, Kirschner D, Sarkar FH, Wrabetz L, Shy ME. Curcumin derivatives promote Schwann cell differentiation and improve neuropathy in R98C CMT1B mice. *Brain.* Dec 2012;135(Pt 12):3551-3566. PMID: 23250879, PMCID: PMC3577101
78. Ramdharry GM, Thornhill A, Mein G, Reilly MM, Marsden JF. Exploring the experience of fatigue in people with Charcot-Marie-Tooth disease. *Neuromuscul. Disord.* Dec 2012;22 Suppl 3:S208-213. PMID: 23182641
79. Kleopa KA, Abrams CK, Scherer SS. How do mutations in GJB1 cause X-linked Charcot-Marie-Tooth disease? *Brain Res.* Dec 3 2012;1487:198-205. PMID: 22771394, PMCID: PMC3488165
80. Tesson C, Nawara M, Salih MA, et al. Alteration of fatty-acid-metabolizing enzymes affects mitochondrial form and function in hereditary spastic paraplegia. *Am. J. Hum. Genet.* Dec 7 2012;91(6):1051-1064. PMID: 23176821, PMCID: PMC3516610

81. Saporta MA, Shy ME. Inherited peripheral neuropathies. *Neurol Clin.* 2013;31(2):597-619. PMID: 23642725, PMCID: PMC3646296
82. Vester A, Velez-Ruiz G, McLaughlin HM, et al. A loss-of-function variant in the human histidyl-tRNA synthetase (HARS) gene is neurotoxic in vivo. *Hum. Mutat.* Jan 2013;34(1):191-199. PMID: 22930593, PMCID: PMC3535524
83. Stevens JC, Murphy SM, Davagnanam I, Phadke R, Anderson G, Nethisinghe S, Bremner F, Giunti P, Reilly MM. The ARSACS phenotype can include supranuclear gaze palsy and skin lipofuscin deposits. *J. Neurol. Neurosurg. Psychiatry.* Jan 2013;84(1):114-116. PMID: 23123642
84. Martin E, Schule R, Smets K, et al. Loss of function of glucocerebrosidase GBA2 is responsible for motor neuron defects in hereditary spastic paraplegia. *Am. J. Hum. Genet.* Feb 7 2013;92(2):238-244. PMID: 23332916, PMCID: PMC3567271
85. Komyathy K, Neal S, Feely S, Miller LJ, Lewis RA, Trigge G, Siskind CE, Shy ME, Ramchandren S. Anterior tibialis CMAP amplitude correlations with impairment in CMT1A. *Muscle Nerve.* Apr 2013;47(4):493-496. PMID: 23456782, PMCID: PMC3608739
86. Kennerson ML, Yiu EM, Chuang DT, et al. A new locus for X-linked dominant Charcot-Marie-Tooth disease (CMTX6) is caused by mutations in the pyruvate dehydrogenase kinase isoenzyme 3 (PDK3) gene. *Hum. Mol. Genet.* Apr 1 2013;22(7):1404-1416. PMID: 23297365, PMCID: PMC3596851
87. Cottenie E, Menezes MP, Rossor AM, et al. Rapidly progressive asymmetrical weakness in Charcot-Marie-Tooth disease type 4J resembles chronic inflammatory demyelinating polyneuropathy. *Neuromuscul. Disord.* May 2013;23(5):399-403. PMID: 23489662
88. Saporta MA, Shy ME. Inherited peripheral neuropathies. *Neurol. Clin.* May 2013;31(2):597-619. PMID: 23642725, PMCID: PMC3646296
89. Burns J, Menezes M, Finkel RS, et al. Transitioning outcome measures: relationship between the CMTpedS and CMTNSv2 in children, adolescents, and young adults with Charcot-Marie-Tooth disease. *J. Peripher. Nerv. Syst.* Jun 2013;18(2):177-180. PMID: 23781965, PMCID: PMC3714225
90. Murphy SM, Ernst D, Wei Y, et al. Hereditary sensory and autonomic neuropathy type 1 (HSANI) caused by a novel mutation in SPTLC2. *Neurology.* Jun 4 2013;80(23):2106-2111. PMID: 23658386, PMCID: PMC3716354
91. Oates EC, Rossor AM, Hafezparast M, et al. Mutations in BICD2 cause dominant congenital spinal muscular atrophy and hereditary spastic paraplegia. *Am. J. Hum. Genet.* Jun 6 2013;92(6):965-973. PMID: 23664120, PMCID: PMC3675232
92. Reilly MM. Obstructive sleep apnoea, restless leg syndrome and Charcot-Marie-Tooth disease type 1: important associations. *J. Neurol. Neurosurg. Psychiatry.* Jun 11 2013. PMID: 23757421
93. Boukhris A, Schule R, Loureiro JL, et al. Alteration of ganglioside biosynthesis responsible for complex hereditary spastic paraplegia. *Am. J. Hum. Genet.* Jul 11 2013;93(1):118-123. PMID: 23746551, PMCID: PMC3710753
94. Marquez-Infante C, Murphy SM, Mathew L, et al. Asymmetric sensory ganglionopathy: a case series. *Muscle Nerve.* Jul 2013;48(1):145-150. PMID: 23744601
95. Hawke F, Chuter V, Burns J. Impact of nocturnal calf cramping on quality of sleep and health-related quality of life. *Qual. Life Res.* Aug 2013;22(6):1281-1286. PMID: 23011494

96. Siskind CE, Panchal S, Smith CO, et al. A review of genetic counseling for Charcot Marie Tooth disease (CMT). *J. Genet. Couns.* Aug 2013;22(4):422-436. PMID: 23604902
97. Lewis RA, McDermott MP, Herrmann DN, et al. High-Dosage Ascorbic Acid Treatment in Charcot-Marie-Tooth Disease Type 1A: Results of a Randomized, Double-Masked, Controlled Trial. *JAMA neurology.* Aug 1 2013;70(8):981-987. PMID: 23797954, PMCID: PMC3752369
98. Tousignant R, Trepanier A, Shy ME, Siskind CE. Genetic testing practices for CMT1A. *Muscle Nerve.* Aug 20 2013. PMID: 23963961
99. Johnson NE, Heatwole CR, Ferguson M, Sowden JE, Jeanat S, Herrmann DN. Patient Identification of the Symptomatic Impact of Charcot-Marie-Tooth Disease Type 1A. *Journal of clinical neuromuscular disease.* Sep 2013;15(1):19-23. PMID: 23965405, PMCID: PMC3752697
100. Eschbach J, Sinniger J, Bouitbir J, et al. Dynein mutations associated with hereditary motor neuropathies impair mitochondrial morphology and function with age. *Neurobiol. Dis.* Oct 2013;58:220-230. PMID: 23742762, PMCID: PMC3748180
101. Klein CJ, Duan X, Shy ME. Inherited neuropathies: clinical overview and update. *Muscle Nerve.* Oct 2013;48(4):604-622. PMID: 23801417
102. Landoure G, Zhu PP, Lourenco CM, et al. Hereditary Spastic Paraplegia Type 43 (SPG43) is Caused by Mutation in C19orf12. *Hum. Mutat.* Oct 2013;34(10):1357-1360. PMID: 23857908, PMCID: PMC3819934
103. Rossor AM, Polke JM, Houlden H, Reilly MM. Clinical implications of genetic advances in Charcot-Marie-Tooth disease. *Nature reviews. Neurology.* Oct 2013;9(10):562-571. PMID: 24018473
104. Pareyson D, Piscoquito G, Moroni I, Salsano E, Zeviani M. Peripheral neuropathy in mitochondrial disorders. *Lancet Neurol.* Oct 2013;12(10):1011-1024. PMID: 24050734
105. Sagnelli A, Piscoquito G, Pareyson D. Inherited neuropathies: an update. *J. Neurol.* Oct 2013;260(10):2684-2690. PMID: 24061768
106. Pitceathly RD, Taanman JW, Rahman S, et al. COX10 Mutations Resulting in Complex Multisystem Mitochondrial Disease That Remains Stable Into Adulthood. *JAMA neurology.* Oct 7 2013. PMID: 24100867
107. Gonzalez M, Nampoothiri S, Kornblum C, et al. Mutations in phospholipase DDHD2 cause autosomal recessive hereditary spastic paraplegia (SPG54). *Eur. J. Hum. Genet.* Nov 2013;21(11):1214-1218. PMID: 23486545, PMCID: PMC3798837
108. Gonzalez M, McLaughlin H, Houlden H, et al. Exome sequencing identifies a significant variant in methionyl-tRNA synthetase (MARS) in a family with late-onset CMT2. *J. Neurol. Neurosurg. Psychiatry.* Nov 2013;84(11):1247-1249. PMID: 23729695, PMCID: PMC3796032
109. Mandarakas M, Hiller CE, Rose KJ, Burns J. Measuring Ankle Instability in Pediatric Charcot-Marie-Tooth Disease. *J. Child Neurol.* Nov 2013;28(11):1456-1462. PMID: 23696628

110. Sumner CJ, d'Ydewalle C, Wooley J, et al. A dominant mutation in FBXO38 causes distal spinal muscular atrophy with calf predominance. *Am. J. Hum. Genet.* Nov 7 2013;93(5):976-983. PMID: 24207122, PMCID: PMC3824115
111. Bonifert T, Karle KN, Tonagel F, et al. Pure and syndromic optic atrophy explained by deep intronic OPA1 mutations and an intralocus modifier. *Brain.* 2014;137(Pt 8):2164-2177. PMID: 24970096, PMCID: PMC4107747
112. Herrmann DN, Horvath R, Sowden JE, et al. Synaptotagmin 2 mutations cause an autosomal-dominant form of lambert-eaton myasthenic syndrome and nonprogressive motor neuropathy. *Am. J. Hum. Genet.* 2014;95(3):332-339. PMID: 25192047, PMCID: PMC4157148
113. Lam BL, Zuchner SL, Dallman J, et al. Mutation K42E in dehydrodolichol diphosphate synthase (DHDDS) causes recessive retinitis pigmentosa. *Adv. Exp. Med. Biol.* 2014;801:165-170. PMID: 24664694
114. Mannil M, Solari A, Leha A, et al. Selected items from the Charcot-Marie-Tooth (CMT) Neuropathy Score and secondary clinical outcome measures serve as sensitive clinical markers of disease severity in CMT1A patients. *Neuromuscul Disord.* 2014;24(11):1003-1017. PMID: 25085517
115. Timmerman V, Strickland AV, Zuchner S. Genetics of Charcot-Marie-Tooth (CMT) Disease within the Frame of the Human Genome Project Success. *Genes.* 2014;5(1):13-32. PMID: 24705285, PMCID: PMC3978509
116. Wen R, Dallman JE, Li Y, et al. Knock-down DHDDS expression induces photoreceptor degeneration in zebrafish. *Adv. Exp. Med. Biol.* 2014;801:543-550. PMID: 24664742
117. Foley AR, Menezes MP, Pandraud A, et al. Treatable childhood neuronopathy caused by mutations in riboflavin transporter RFVT2. *Brain.* Jan 2014;137(Pt 1):44-56. PMID: 24253200, PMCID: PMC3891447
118. Laura M, Hutton EJ, Blake J, et al. Pain and small fiber function in charcot-marie-tooth disease type 1A. *Muscle Nerve.* Jan 7 2014. PMID: 24395492
119. Mudge AJ, Bau KV, Purcell LN, et al. Normative reference values for lower limb joint range, bone torsion, and alignment in children aged 4-16 years. *J. Pediatr. Orthop. B.* Jan 2014;23(1):15-25. PMID: 23852035
120. Synofzik M, Gonzalez MA, Lourenco CM, et al. PNPLA6 mutations cause Boucher-Neuhauser and Gordon Holmes syndromes as part of a broad neurodegenerative spectrum. *Brain.* Jan 2014;137(Pt 1):69-77. PMID: 24355708, PMCID: PMC3891450
121. Esteves T, Durr A, Mundwiller E, et al. Loss of association of REEP2 with membranes leads to hereditary spastic paraplegia. *Am. J. Hum. Genet.* Feb 6 2014;94(2):268-277. PMID: 24388663, PMCID: PMC3928657
122. Ramchandren S, Jaiswal M, Feldman E, Shy M. Effect of pain in pediatric inherited neuropathies. *Neurology.* Mar 4 2014;82(9):793-797. PMID: 24477108, PMCID: PMC3945655

123. Fischmann A, Morrow JM, Sinclair CD, et al. Improved anatomical reproducibility in quantitative lower-limb muscle MRI. *J. Magn. Reson. Imaging*. Apr 2014;39(4):1033-1038. PMID: 24123788
124. Liu YT, Hersheson J, Plagnol V, et al. Autosomal-recessive cerebellar ataxia caused by a novel ADCK3 mutation that elongates the protein: clinical, genetic and biochemical characterisation. *J. Neurol. Neurosurg. Psychiatry*. May 2014;85(5):493-498. PMID: 24218524, PMCID: PMC3995328
125. Revuelta GJ, Montilla J, Benatar M, et al. An (1)(8)F-FDG PET study of cervical muscle in parkinsonian anterocollis. *J. Neurol. Sci*. May 15 2014;340(1-2):174-177. PMID: 24725739, PMCID: PMC4305331
126. Parker B, Alexander R, Wu X, et al. Detection of Copy Number Variation by SNP-Allelotyping. *J. Neurogenet*. Jun 2 2014:1-5. PMID: 24830919
127. Caballero Oteyza A, Battaloglu E, Ocek L, et al. Motor protein mutations cause a new form of hereditary spastic paraplegia. *Neurology*. Jun 3 2014;82(22):2007-2016. PMID: 24808017, PMCID: PMC4105256
128. Morrow JM, Sinclair CD, Fischmann A, et al. Reproducibility, and age, body-weight and gender dependency of candidate skeletal muscle MRI outcome measures in healthy volunteers. *Eur. Radiol*. Jul 2014;24(7):1610-1620. PMID: 24748539, PMCID: PMC4046083
129. Johnson NE, Sowden J, Dilek N, et al. Prospective Study of Muscle Cramps in Charcot-Marie-Tooth Disease. *Muscle Nerve*. Jul 5 2014. PMID: 25042364
130. Fridman V, Oaklander AL, David WS, et al. Natural History and Biomarkers in Hereditary Sensory Neuropathy Type 1. *Muscle Nerve*. Jul 10 2014. PMID: 25042817
131. Finkel RS, McDermott MP, Kaufmann P, et al. Observational study of spinal muscular atrophy type I and implications for clinical trials. *Neurology*. Aug 26 2014;83(9):810-817. PMID: 25080519, PMCID: PMC4155049
132. Sadjadi R, Reilly MM, Shy ME, et al. Psychometrics evaluation of Charcot-Marie-Tooth Neuropathy Score (CMTNSv2) second version, using Rasch analysis. *J. Peripher. Nerv. Syst*. Sep 2014;19(3):192-196. PMID: 25400013, PMCID: PMC4303498
133. Brewer MH, Ma KH, Beecham GW, et al. Haplotype-specific modulation of a SOX10/CREB response element at the Charcot-Marie-Tooth disease type 4C locus SH3TC2. *Hum. Mol. Genet*. Oct 1 2014;23(19):5171-5187. PMID: 24833716, PMCID: PMC4168306
134. Hershey LA, Perlmutter JS. Smoking and Parkinson disease: where there is smoke there may not be fire. *Neurology*. Oct 14 2014;83(16):1392-1393. PMID: 25217061
135. Dortch RD, Dethrage LM, Gore JC, Smith SA, Li J. Proximal nerve magnetization transfer MRI relates to disability in Charcot-Marie-Tooth diseases. *Neurology*. Oct 21 2014;83(17):1545-1553. PMID: 25253751, PMCID: PMC4222857

136. Griffin LB, Sakaguchi R, McGuigan D, et al. Impaired function is a common feature of neuropathy-associated glycyl-tRNA synthetase mutations. *Hum. Mutat.* Nov 2014;35(11):1363-1371. PMID: 25168514, PMCID: PMC4213347
137. Beutler AS, Kulkarni AA, Kanwar R, et al. Sequencing of Charcot-Marie-Tooth disease genes in a toxic polyneuropathy. *Ann. Neurol.* Nov 2014;76(5):727-737. PMID: 25164601, PMCID: PMC4388308
138. Gonzalez MA, Feely SM, Speziani F, et al. A novel mutation in VCP causes Charcot-Marie-Tooth Type 2 disease. *Brain.* Nov 2014;137(Pt 11):2897-2902. PMID: 25125609, PMCID: PMC4208462
139. Johnson NE, Heatwole CR, Dilek N, et al. Quality-of-life in Charcot-Marie-Tooth disease: The patient's perspective. *Neuromuscul. Disord.* Nov 2014;24(11):1018-1023. PMID: 25092060, PMCID: PMC4253871
140. Cottenie E, Kochanski A, Jordanova A, et al. Truncating and missense mutations in IGHMBP2 cause Charcot-Marie Tooth disease type 2. *Am. J. Hum. Genet.* Nov 6 2014;95(5):590-601. PMID: 25439726, PMCID: PMC4225647
141. Horga A, Pitceathly RD, Blake JC, et al. Peripheral neuropathy predicts nuclear gene defect in patients with mitochondrial ophthalmoplegia. *Brain.* Dec 2014;137(Pt 12):3200-3212. PMID: 25281868, PMCID: PMC4240292
142. Brennan KM, Bai Y, Pisciotta C, et al. Absence of Dystrophin Related Protein-2 disrupts Cajal bands in a patient with Charcot-Marie-Tooth disease. *Neuromuscul. Disord.* 2015;25(10):786-793. PMID: 26227883, PMCID: PMC4920059
143. Gutmann L, Shy M. Update on Charcot-Marie-Tooth disease. *Curr. Opin. Neurol.* 2015;28(5):462-467. PMID: 26263471
144. Jerath NU, Crockett CD, Moore SA, et al. Rare Manifestation of a c.290 C>T, p.Gly97Glu VCP Mutation. *Case reports in genetics.* 2015;2015:239167. PMID: 25878907, PMCID: PMC4386706
145. Nolano M, Manganelli F, Provitera V, et al. Small nerve fiber involvement in CMT1A. *Neurology.* 2015;84(4):407-414. PMID: 25540311, PMCID: PMC4336000
146. Philippakis AA, Azzariti DR, Beltran S, et al. The Matchmaker Exchange: a platform for rare disease gene discovery. *Hum. Mutat.* 2015;36(10):915-921. PMID: 26295439, PMCID: PMC4610002
147. Thal DR, Zuchner S, Gierer S, et al. Abnormal Paraplegin Expression in Swollen Neurites, tau- and alpha-Synuclein Pathology in a Case of Hereditary Spastic Paraplegia SPG7 with an Ala510Val Mutation. *Int. J. Mol. Sci.* 2015;16(10):25050-25066. PMID: 26506339, PMCID: PMC4632789
148. McColgan P, Viegas S, Gandhi S, et al. Oculoleptomeningeal Amyloidosis associated with transthyretin Leu12Pro in an African patient. *J. Neurol.* Jan 2015;262(1):228-234. PMID: 25488473, PMCID: PMC4289971

149. Safka Brozkova D, Deconinck T, Griffin LB, et al. Loss of function mutations in HARS cause a spectrum of inherited peripheral neuropathies. *Brain*. 2015;138(Pt 8):2161-2172. PMID: 26072516, PMCID: PMC4840952
150. Sanmaneechai O, Feely S, Scherer SS, et al. Genotype-phenotype characteristics and baseline natural history of heritable neuropathies caused by mutations in the MPZ gene. *Brain*. 2015;138(Pt 11):3180-3192. PMID: 26310628, PMCID: PMC4643641
151. Schmidt WM, Rutledge SL, Schule R, et al. Disruptive SCYL1 Mutations Underlie a Syndrome Characterized by Recurrent Episodes of Liver Failure, Peripheral Neuropathy, Cerebellar Atrophy, and Ataxia. *Am. J. Hum. Genet.* 2015;97(6):855-861. PMID: 26581903, PMCID: PMC4678415
152. Saporta MA, Dang V, Volfson D, et al. Axonal Charcot-Marie-Tooth disease patient-derived motor neurons demonstrate disease-specific phenotypes including abnormal electrophysiological properties. *Exp. Neurol.* Jan 2015;263:190-199. PMID: 25448007, PMCID: PMC4262589
153. Zimon M, Battaloglu E, Parman Y, et al. Unraveling the genetic landscape of autosomal recessive Charcot-Marie-Tooth neuropathies using a homozygosity mapping approach. *Neurogenetics*. Jan 2015;16(1):33-42. PMID: 25231362
154. Mead S, Reilly MM. A new prion disease: relationship with central and peripheral amyloidoses. Nature reviews. *Neurology*. Feb 2015;11(2):90-97. PMID: 25623792
155. Scoto M, Rossor AM, Harms MB, et al. Novel mutations expand the clinical spectrum of DYNC1H1-associated spinal muscular atrophy. *Neurology*. Feb 17 2015;84(7):668-679. PMID: 25609763, PMCID: PMC4336105
156. Ernst D, Murphy SM, Sathiyandan K, et al. Novel HSAN1 mutation in serine palmitoyltransferase resides at a putative phosphorylation site that is involved in regulating substrate specificity. *Neuromolecular Med.* Mar 2015;17(1):47-57. PMID: 25567748, PMCID: PMC4326654
157. Baets J, Duan X, Wu Y, et al. Defects of mutant DNMT1 are linked to a spectrum of neurological disorders. *Brain*. Apr 2015;138(Pt 4):845-861. PMID: 25678562
158. Jerath NU, Shy ME. Hereditary motor and sensory neuropathies: Understanding molecular pathogenesis could lead to future treatment strategies. *Biochim. Biophys. Acta*. Apr 2015;1852(4):667-678. PMID: 25108281
159. Sanmaneechai O, Swenson A, Gerke AK, Moore SA, Shy ME. Inclusion body myositis and sarcoid myopathy: coincidental occurrence or associated diseases. *Neuromuscul. Disord.* Apr 2015;25(4):297-300. PMID: 25599912
160. Pelayo-Negro AL, Carr AS, Laura M, Skorupinska M, Reilly MM. An observational study of asymmetry in CMT1A. *J. Neurol. Neurosurg. Psychiatry*. May 2015;86(5):589-590. PMID: 25313262, PMCID: PMC4413808

161. Motley WW, Griffin LB, Mademan I, et al. A novel AARS mutation in a family with dominant myeloneuropathy. *Neurology*. May 19 2015;84(20):2040-2047. PMID: 25904691, PMCID: PMC4442103
162. Carr AS, Polke JM, Wilson J, et al. MFN2 deletion of exons 7 and 8: founder mutation in the UK population. *J. Peripher. Nerv. Syst.* Jun 2015;20(2):67-71. PMID: 26114802
163. Carr AS, Pelayo-Negro AL, Jaunmuktane Z, et al. Transthyretin V122I amyloidosis with clinical and histological evidence of amyloid neuropathy and myopathy. *Neuromuscul. Disord.* Jun 2015;25(6):511-515. PMID: 25819286
164. Ramchandren S, Shy M, Feldman E, Carlos R, Siskind C. Defining disability: development and validation of a mobility-Disability Severity Index (mDSI) in Charcot-Marie-tooth disease. *J. Neurol. Neurosurg. Psychiatry*. Jun 2015;86(6):635-639. PMID: 25157034, PMCID: PMC4920058
165. Rossor AM, Evans MR, Reilly MM. A practical approach to the genetic neuropathies. *Practical neurology*. Jun 2015;15(3):187-198. PMID: 25898997
166. Tetreault M, Gonzalez M, Dicaire MJ, et al. Adult-onset painful axonal polyneuropathy caused by a dominant NAGLU mutation. *Brain*. Jun 2015;138(Pt 6):1477-1483. PMID: 25818867, PMCID: PMC4542621
167. Pisciotta C, Bai Y, Brennan KM, et al. Reduced neurofilament expression in cutaneous nerve fibers of patients with CMT2E. *Neurology*. Jul 21 2015;85(3):228-234. PMID: 26109717, PMCID: PMC4516296
168. Abrams AJ, Hufnagel RB, Rebelo A, et al. Mutations in SLC25A46, encoding a UGO1-like protein, cause an optic atrophy spectrum disorder. *Nat. Genet.* Aug 2015;47(8):926-932. PMID: 26168012, PMCID: PMC4520737
169. Fridman V, Bundy B, Reilly MM, et al. CMT subtypes and disease burden in patients enrolled in the Inherited Neuropathies Consortium natural history study: a cross-sectional analysis. *J. Neurol. Neurosurg. Psychiatry*. Aug 2015;86(8):873-878. PMID: 25430934, PMCID: PMC4516002
170. Gonzaga-Jauregui C, Harel T, Gambin T, et al. Exome Sequence Analysis Suggests that Genetic Burden Contributes to Phenotypic Variability and Complex Neuropathy. *Cell reports*. Aug 18 2015;12(7):1169-1183. PMID: 26257172, PMCID: PMC4545408
171. Saifee TA, Parees I, Kassavetis P, et al. Tremor in Charcot-Marie-Tooth disease: No evidence of cerebellar dysfunction. *Clin. Neurophysiol.* Sep 2015;126(9):1817-1824. PMID: 25641441
172. Strickland AV, Schabhuhtl M, Offenbacher H, et al. Mutation screen reveals novel variants and expands the phenotypes associated with DYNC1H1. *J. Neurol.* Sep 2015;262(9):2124-2134. PMID: 26100331, PMCID: PMC4573829
173. Jerath NU, Kamholz J, Grider T, Harper A, Swenson A, Shy ME. Coexistence of a T118M PMP22 missense mutation and chromosome 17 (17p11.2-p12) deletion. *Muscle Nerve*. Nov 2015;52(5):905-908. PMID: 26012543, PMCID: PMC4596757

174. Whittaker RG, Herrmann DN, Bansagi B, et al. Electrophysiologic features of SYT2 mutations causing a treatable neuromuscular syndrome. *Neurology*. Dec 1 2015;85(22):1964-1971. PMID: 26519543, PMCID: PMC4664120
175. Albulym OM, Kennerson ML, Harms MB, et al. MORC2 mutations cause axonal Charcot-Marie-Tooth disease with pyramidal signs. *Ann. Neurol*. 2016;79(3):419-427. PMID: 26659848, PMCID: PMC4936275
176. Auer-Grumbach M, Toegel S, Schabhuttl M, et al. Rare Variants in MME, Encoding Metalloprotease Neprilysin, Are Linked to Late-Onset Autosomal-Dominant Axonal Polyneuropathies. *Am J Hum Genet*. 2016;99(3):607-623. PMID: 27588448, PMCID: PMC5011077
177. Cornett KM, Menezes MP, Bray P, et al. Phenotypic Variability of Childhood Charcot-Marie-Tooth Disease. *JAMA neurology*. 2016;73(6):645-651. PMID: 27043305
178. Horga A, Tomaselli PJ, Gonzalez MA, et al. SIGMAR1 mutation associated with autosomal recessive Silver-like syndrome. *Neurology*. 2016;87(15):1607-1612. PMID: 27629094, PMCID: PMC5067545
179. Hu B, Arpag S, Zuchner S, Li J. A novel missense mutation of CMT2P alters transcription machinery. *Ann Neurol*. 2016;80(6):834-845. PMID: 27615052, PMCID: PMC5177458
180. Jerath NU, Gutmann L, Reddy CG, Shy ME. Charcot-marie-tooth disease type 1X in women: Electrodiagnostic findings. *Muscle Nerve*. 2016;54(4):728-732. PMID: 26873881
181. Lassuthova P, Safka Brozkova D, Krutova M, et al. Severe axonal Charcot-Marie-Tooth disease with proximal weakness caused by de novo mutation in the MORC2 gene. *Brain*. 2016;139(Pt 4):e26. PMID: 26912637
182. Lunn MP, Ellis L, Hadden RD, Rajabally YA, Winer JB, Reilly MM. A proposed dosing algorithm for the individualized dosing of human immunoglobulin in chronic inflammatory neuropathies. *J Peripher Nerv Syst*. 2016;21(1):33-37. PMID: 26757367
183. Mademan I, Harmuth F, Giordano I, et al. Multisystemic SYNE1 ataxia: confirming the high frequency and extending the mutational and phenotypic spectrum. *Brain*. 2016;139(Pt 8):e46. PMID: 27197992, PMCID: PMC4958896
184. Manganelli F, Pisciotta C, Reilly MM, et al. Nerve conduction velocity in CMT1A: what else can we tell? *Eur J Neurol*. 2016;23(10):1566-1571. PMID: 27412484, PMCID: PMC5603914
185. Manole A, Chelban V, Haridy NA, et al. Severe axonal neuropathy is a late manifestation of SPG11. *J Neurol*. 2016;263(11):2278-2286. PMID: 27544499, PMCID: PMC5065903
186. Merkel PA, Manion M, Gopal-Srivastava R, et al. The partnership of patient advocacy groups and clinical investigators in the rare diseases clinical research network. *Orphanet J. Rare Dis*. 2016;11(1):66. PMID: 27194034, PMCID: PMC4870759

- 187.** Motley WW, Palaima P, Yum SW, et al. De novo PMP2 mutations in families with type 1 Charcot-Marie-Tooth disease. *Brain*. 2016;139(Pt 6):1649-1656. PMID: 27009151, PMCID: PMC4916861
- 188.** Panosyan FB, Mountain JM, Reilly MM, Shy ME, Herrmann DN. Rydel-Seiffer fork revisited: Beyond a simple case of black and white. *Neurology*. 2016;87(7):738-740. PMID: 27412138, PMCID: PMC4999169
- 189.** Perez-Siles G, Grant A, Ellis M, et al. Characterizing the molecular phenotype of an Atp7a(T985I) conditional knock in mouse model for X-linked distal hereditary motor neuropathy (dHMNX). *Metallomics : integrated biometal science*. 2016;8(9):981-992. PMID: 27293072
- 190.** Piscoquito G, Saveri P, Magri S, et al. Screening for SH3TC2 gene mutations in a series of demyelinating recessive Charcot-Marie-Tooth disease (CMT4). *J Peripher Nerv Syst*. 2016;21(3):142-149. PMID: 27231023
- 191.** Rebelo AP, Abrams AJ, Cottenie E, et al. Cryptic Amyloidogenic Elements in the 3' UTRs of Neurofilament Genes Trigger Axonal Neuropathy. *Am. J. Hum. Genet*. 2016;98(4):597-614. PMID: 27040688, PMCID: PMC4833435
- 192.** Rossor AM, Tomaselli PJ, Reilly MM. Recent advances in the genetic neuropathies. *Curr Opin Neurol*. 2016;29(5):537-548. PMID: 27584852, PMCID: PMC5130159
- 193.** Shy ME. Gene therapy, CMT1X, and the inherited neuropathies. *Proc Natl Acad Sci U S A*. 2016;113(17):4552-4554. PMID: 27078106, PMCID: PMC4855541
- 194.** Shy M. LRSAM1 lessons. *Ann Neurol*. 2016;80(6):821-822. PMID: 28001317
- 195.** Vallat JM, Nizon M, Magee A, et al. Contactin-Associated Protein 1 (CNTNAP1) Mutations Induce Characteristic Lesions of the Paranodal Region. *J Neuropathol Exp Neurol*. 2016;75(12):1155-1159. PMID: 27818385
- 196.** Brewer MH, Chaudhry R, Qi J, et al. Whole Genome Sequencing Identifies a 78 kb Insertion from Chromosome 8 as the Cause of Charcot-Marie-Tooth Neuropathy CMTX3. *PLoS Genet*. Jul 2016;12(7):e1006177. PMID: 27438001, PMCID: PMC4954712
- 197.** Bis DM, Schule R, Reichbauer J, et al. Uniparental disomy determined by whole-exome sequencing in a spectrum of rare motoneuron diseases and ataxias. *Molecular genetics & genomic medicine*. 2017;5(3):280-286. PMID: 28546998, PMCID: PMC5441426
- 198.** Hengel H, Magee A, Mahanjah M, et al. CNTNAP1 mutations cause CNS hypomyelination and neuropathy with or without arthrogryposis. *Neurology Genetics*. 2017;3(2):e144. PMID: 28374019, PMCID: PMC5363873
- 199.** Horga A, Laura M, Jaunmuktane Z, et al. Genetic and clinical characteristics of NEFL-related Charcot-Marie-Tooth disease. *J Neurol Neurosurg Psychiatry*. 2017. PMID: 28501821

- 200.** Jerath NU, Shy ME. Charcot-Marie-Tooth disease type 1C: Clinical and electrophysiological findings for the c.334G>a (p.Gly112Ser) LITAF/SIMPLE mutation. *Muscle Nerve*. 2017. PMID: 28164329
- 201.** Kalmar B, Innes A, Wanisch K, et al. Mitochondrial deficits and abnormal mitochondrial retrograde axonal transport play a role in the pathogenesis of mutant Hsp27 induced Charcot Marie Tooth Disease. *Hum Mol Genet*. 2017. PMID: 28595321
- 202.** Laura M, Singh D, Ramdharry G, et al. Prevalence and orthopedic management of foot and ankle deformities in Charcot-Marie-Tooth disease. *Muscle Nerve*. 2017. PMID: 28632967
- 203.** Manganelli F, Parisi S, Nolano M, et al. Novel mutations in dystonin provide clues to the pathomechanisms of HSAN-VI. *Neurology*. 2017;88(22):2132-2140. PMID: 28468842
- 204.** Panosyan FB, Kirk CA, Marking D, et al. Carpal Tunnel Syndrome in Inherited Neuropathies: A Retrospective Survey. *Muscle Nerve*. 2017. PMID: 28692128
- 205.** Rocha N, Bulger DA, Frontini A, et al. Human biallelic MFN2 mutations induce mitochondrial dysfunction, upper body adipose hyperplasia, and suppression of leptin expression. *eLife*. 2017;6. PMID: 28414270, PMCID: PMC5422073
- 206.** Rossor AM, Morrow JM, Polke JM, et al. Pilot phenotype and natural history study of hereditary neuropathies caused by mutations in the HSPB1 gene. *Neuromuscul Disord*. 2017;27(1):50-56. PMID: 27816334, PMCID: PMC5260843
- 207.** Saporta MA, Shy ME. A human cellular model to study peripheral myelination and demyelinating neuropathies. *Brain*. 2017;140(4):856-859. PMID: 28375459
- 208.** Shy M, Rebelo AP, Feely SM, et al. Mutations in BAG3 cause adult-onset Charcot-Marie-Tooth disease. *J Neurol Neurosurg Psychiatry*. 2017. PMID: 28754666
- 209.** Tomaselli PJ, Rossor AM, Horga A, et al. Mutations in noncoding regions of GJB1 are a major cause of X-linked CMT. *Neurology*. 2017;88(15):1445-1453. PMID: 28283593, PMCID: PMC5386440
- 210.** Tsai PC, Soong BW, Mademan I, et al. A recurrent WARS mutation is a novel cause of autosomal dominant distal hereditary motor neuropathy. *Brain*. 2017. PMID: 28369220
- 211.** Wang DS, Wu X, Bai Y, et al. PMP22 exon 4 deletion causes ER retention of PMP22 and a gain-of-function allele in CMT1E. *Annals of clinical and translational neurology*. 2017;4(4):236-245. PMID: 28382305, PMCID: PMC5376752
- 212.** Panosyan FB, Kirk CA, Marking D, et al. Carpal tunnel syndrome in inherited neuropathies: A retrospective survey. *Muscle Nerve*. 2018;57(3):388-394. PMID: 28692128, PMCID: PMC5762426