Curriculum Vitae
The Johns Hopkins University School of Medicine

Signature:  7-10-2019

Charlotte J. Sumner, M.D.

DEMOGRAPHIC AND PERSONAL INFORMATION

Current Appointments

2017-present Professor, Department of Neurology, Johns Hopkins University, Baltimore, Maryland
2017-present Professor, Department of Neuroscience, Johns Hopkins University, Baltimore, Maryland

Personal data

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Education and Training

Undergraduate
1987-1991 B.A., Princeton University, Princeton, NJ; Ecology and Evolutionary Biology graduated *magna cum laude*

Doctoral/graduate
1991-1996 M.D., University of Pennsylvania School of Medicine, Philadelphia, PA
1994-1995 Howard Hughes Medical Institute/National Institutes of Health Medical Research Scholar, National Institute of Neurological Disorders and Stroke, Bethesda, MD

Postdoctoral
1996-1997 Intern, Internal Medicine, University of California San Francisco, San Francisco, CA
1997-2000 Resident, Neurology, University of California San Francisco, San Francisco, CA
1999-2000 Chief Resident, Neurology, University of California San Francisco, San Francisco, CA
2000-2001 Fellowship, Neuromuscular Disease, Johns Hopkins University School of Medicine, Baltimore, MD
2001-2006 Fellowship, Neurogenetics, Dr. Kenneth Fischbeck, Neurogenetics Branch, National Institute of Neurological Disorders and Stroke, Bethesda, MD

Professional Experience

2006-2011 Assistant Professor of Neurology, Johns Hopkins University, Baltimore, MD
2010-2011 Assistant Professor of Neuroscience, Johns Hopkins University, Baltimore, MD
2006-2013 Special Volunteer, National Institute of Neurological Disorders and Stroke, Bethesda, MD
2011-2017 Associate Professor of Neurology, Johns Hopkins University, Baltimore, MD
2011-2017 Associate Professor of Neuroscience, Johns Hopkins University, Baltimore, MD
2010-present Co-director Charcot Marie Tooth Disease Clinic
2010-present Co-director Spinal Muscular Atrophy Center

PUBLICATIONS

Original Research

1. **Sumner CJ**, Shinohara T, Durham L, Traub R, Major EO, Amemiya K. Expression of multiple classes of nuclear factor-1 family in the developing human brain: differential expression of two classes of NF-1 genes. J Neurovirol 1996;2:87-100.
2. **Sumner CJ,** Sheth S, Griffin JW, Cornblath DR, Polydefkis MJ. The spectrum of neuropathy in diabetes and impaired glucose tolerance. Neurology, 2003;60:108-111.
3. **Sumner CJ**, Huynh TN, Markowitz JA, Perhac JP, Hill B, Coovert DD, Schussler K, Chen X, Jarecki J, Burghes AHM, Taylor JP, Fischbeck KH. Valproic acid increases SMN levels in spinal muscular atrophy patient-derived cell lines. Ann Neurol 2003;54:647-654.
4. Lunn MR, Root DE, Martino AM, Flaherty SP, Kelley BP, Coovert DD, Burghes AH, thi Man N, Morris GE, Zhou J, Androphy EJ, **Sumner CJ**, Stockwell BR. Indoprofen upregulates the survival motor neuron (SMN) protein through a cyclooxygenase-independent mechanism. Chem Biol 2004;11:1489-1493.
5. Puls I, Oh SJ, **Sumner CJ** ,Wallace K, Holzbaur E, Mann EA, Floeter MK, Kennedy WR, Wendelschafer-Crabb G, Vortmeyer A, Powers R, Bidus K, Fischbeck KF, Ludlow CL. Distal spinal and bulbar muscular atrophy caused by dynactin mutation. Ann Neurol 2005;57:687-694.
6. Kernochan LE, Woodling NS, Avila AM, Huynh TN, Fischbeck KH, **Sumner, CJ**. The role of histone acetylation in survival motor neuron gene expression. Hum Mol Genet 2005;14(9):1171-1182.
7. Gupta A, **Sumner CJ**, Castor M, Maslanka S, Sobel J. Botulism Type F in the United States, 1981 to 2002. Neurology 2005;65:1694-1700.
8. Levy J,* **Sumner CJ,*** Tokito M, Ligon LA, LaMonte B, Wallace KE, Harmison G, Puls I, Fischbeck KH, Holzbaur, ELF. A G59S dynactin mutation that causes motor neuron degeneration induces loss of function and protein aggregation. J Cell Biol 2006;172:733-745. (*The first two authors contributed equally to this study)
9. **Sumner CJ**, Kolb SJ, Harmison GG, Jeffries NO, Schadt K, Finkel RS, Dreyfuss G, Fischbeck KH. SMN mRNA and protein levels in peripheral blood: Biomarkers for SMA clinical trials. Neurology 2006;66:1067-73.
10. Kolb S, Gubitz AK, Olszewski RF, Ottinger E, **Sumner CJ**, Fischbeck KH, Dreyfuss G. A novel cell immunoassay to measure survival of motor neurons protein in blood cells. BMC Neurol 2006;6:6.
11. Di Prospero N, **Sumner CJ**, Taylor JP, Fischbeck KH. Safety, tolerability, and pharmacokinetics of high dose Idebenone in patients with Friedreich's ataxia. Arch Neurol 2007;64:803-8.
12. Avila AM, Burnett BB, Taye AA, Gabenella F, Knight M, Hartenstein P, Cizman Z, DiProspero NA, Fischbeck KH, **Sumner CJ**. Trichostatin A increases SMN expression and survival in spinal muscular atrophy mice. J Clin Invest 2007;117:659-71.
13. Babowska JC, Wang H, Zin B, **Sumner CJ**, Blackstone C. Lack of spartin protein in Troyer syndrome suggests a loss-of-function mechanism. Arch Neurol 2007;65:520-4.
14. Narver HL, Kong L, Burnett BG, Choe DW, Bosch M, Taye AA, Eckhaus MA, **Sumner CJ**. Sustained improvement of SMA mice with trichostatin A plus nutrition. Ann Neurol 2008;64:465-70.
15. Burnett BG, Muñoz E, Tandon A, **Sumner CJ**, Fischbeck KH. Regulation of SMN protein stability. Mol Cell Biol 2009;11:1107-15.
16. Kong L, Wang X, Choe DW, Polley M, Bosch- Marcé, Griffin JW, Rich MM, **Sumner CJ**. Impaired synaptic vesicle release and immaturity of neuromuscular junctions in spinal muscular atrophy mice. J Neurosci 2009;29:842-51.
17. **Sumner CJ**, Wee CD, Warsing LC, Choe DW, Ng AS, Lutz C, Wagner KR. Inhibition of myostatin does not ameliorate disease features of severe SMA mice. Hum Mol Genet 2009;18:3145-52.
18. Palazzolo I, Stack C, Kong L, Taylor JP, Musaro A, Adachi H, Katsuno M, Sobue G, **Sumner CJ**, Fischbeck KH, Pennuto M. Overexpression of IGF-1 in muscle attenuates disease in a mouse model of spinal and bulbar muscular atrophy. Neuron 2009;63:316-28.
19. Landouré G, Zdebik AA, Martinez TL, Burnett BG, Stanescu HC, Shi Y, Taye AA, Kong L, Munns CH, Choo SS, Phelps CB, Paudel R, Houlden H, Ludlow CL, Gaudet R, Kleta R, Fischbeck KH, **Sumner CJ**. Mutations in *TRPV4* cause Charcot-Marie-Tooth disease type 2C. Nat Genet 2010;42:170-4.
20. Zimoń, M, Baets, J, Auer-Grumbach, M, Berciano, J, Garcia, A, Lopez-Laso, E, Merlini, L, Hilton-Jones, D, McEntagart, M, Crosby, A, Barisic, N, Boltshauser, E, Shaw, CE, Landouré, G, Ludlow, CL, Gaudet, R, Houlden, H, Reilly, M., Fischbeck, KH, **Sumner, CJ**, Timmerman, V, Jordanova, A, De Jonghe, P. Dominant mutations in the cation channel gene *transient receptor potential vanilloid 4* cause an unusual spectrum of neuropathies. Brain 2010;133:1798-809.
21. Mentis GZ, Liu W, Blivis D, Drobac E, Crowder ME, Kong L, Alvarez FJ, **Sumner CJ**, O'Donovan MJ. Early functional impairment of sensory-motor connectivity in a mouse model of spinal muscular atrophy. Neuron 2011;69:453-467.

22. Bosch-Marcé M, Wee CD, Martinez TL, Lipkes CE, Choe DW, Kong L, Musaro A, **Sumner CJ**. Increased IGF-1 in muscle modulates the phenotype of severe SMA mice. Hum Mol Genet 2011;20:1844-53.
23. Wadhwa V, Thakkar RS, Maragakis N, Höke A, **Sumner CJ**, Lloyd TE, Carrino JA, Belzberg AJ, Chhabra A. Sciatic nerve tumor and tumor-like lesions-uncommon pathologies. Skeletal Radiol 2012;41:763-74.
24. Landoure G, Sullivan JM, Johnson JO, Munns CH, Shi Y, Diallo O, Gibbs RJ, Gaudet R, Ludlow C, Fischbeck KH, Traynor BJ, Burnett BG, **Sumner CJ**. Exome sequencing identifies a novel TRPV4 mutation in a family with CMT2C. Neurology 2012;79:192-194.
25. Martinez TL, Kong L, Wang X, Osborne MA, Crowder ME, Van Meerbeke JP, Xu X, Davis C, Wooley J, Goldhamer DJ, Lutz CM, Rich MM, **Sumner CJ**. SMN in motor neurons determines synaptic integrity in spinal muscular atrophy. J Neurosci 2012; 32:8703-8715.
26. Bricceno KV, Sampognaro PJ, Van Meerbeke JP, **Sumner CJ**, Fischbeck KH and Burnett BG. Histone deacetylase inhibition suppresses myogenin-dependent atrogene activation in spinal muscular atrophy mice. Hum Mol Genet 2012;12:4448-4459.
27. Van Meerbeke JP, Gibbs RM, Plasterer HL, Miao W, Feng Z, Lin MY, Rucki AA, Wee CD, Xia B, Sharma S, Jacques V, Li DK,Pellizzoni L, Rusche JR, Ko CP, and **Sumner CJ**. The DcpS inhibitor RG3039 improved motor function in SMA mice. Hum Mol Genet 2013;22:4074-4083
28. Paez-Colasante X, Seaberg B, Martinez TL, Kong L, **Sumner CJ**, Rimer M. Improvement of neuromuscular synaptic defects without enhanced survival and motor function in severe spinal muscular atrophy mice selectively rescued in motor neurons. PLOS One 2013;8:e75866.
29. **Sumner CJ**, d'Ydewalle C, Wooley J, Fawcett KA, Hernandez D, Gardiner AR, Kalmar B, Baloh RH, Gonzalez M, Züchner S, Stanescu HC, Kleta R, Mankodi A, Cornblath DR, Boylan KB, Reilly MM, Greensmith L, Singleton AB, Harms MB, Rossor AM, and Houlden H. Dominant mutation of *FBXO38* causes distal spinal muscular atrophy with calf predominance. Am J Hum Genet 2013;93:976-983.
30. Tisdale S, Lotti F, Saieva L, Van Meerbeke JP, Crawford TO, **Sumner CJ**, Mentis GZ, and Pellizzoni L. SMN is essential for the biogenesis of U7 small nuclear ribonuclearprotein and 3'-end formation of histone mRNAs. Cell Rep 2013;12:11187-1195.
31. Bricceno KV, Martinez TL, Duguez S, Partridge TA, Fischbeck KH, **Sumner CJ** and Burnett BG. Survival motor neuron protein deficiency impairs myotube formation by altering myogenic gene expression and focal adhesion dynamics. Hum Mol Genet 2014;23:4745-57.
32. Wong CO, Lin YQ, Chen K, Chao Y, Duraine L, Yoon WH, Sullivan JM, Broadhead GT**, Sumner CJ**, Lloyd TE, Macleod GT, Bellen HJ, Venkatachalam K. A TRPV channel in Drosophila motor neurons regulates presynaptic resting Ca2+ levels, synapse growth, and synaptic transmission. Neuron 2014;84:764-77.
33. Fridman V, Bundy B, Reilly MM, Pareyson D, Bacon C, Burns J, Day J, Feely S, Finkel RS, Grider T, Kirk CA, Herrmann DN, Laurá M, Li J, Lloyd T, **Sumner CJ**, Muntoni M, Piscosquito G, Ramchandren S, Shy R, Siskind CE, Yum SE, Moroni I, Pagliano E, Zuchner S, Scherer SS, Shy ME, on behalf of the *Inherited Neuropathies Consortium.* CMT Subtypes and Disease Burden in Patients Enrolled in the Inherited Neuropathies Consortium Natural History Study: a Cross Sectional Analysis. J Neurol Neurosurg Psychiatry 2015;86:873-8.
34. Rindt H, Feng Z, Mazzasette C, Glascock JJ, Valdivia D, Pyles N, Crawford TO, Swoboda KJ, **Sumner CJ**, Ko CP, Lorson CL. Astrocytes influence the severity of spinal muscular atrophy. Hum Mol Genet 2015;24:4094-102.
35. Miller N, Feng Z, Cai Q, Yang B, Cantu J, Topczewski J, Crawford TO, Ko CP, **Sumner CJ**, Ma L, and Ma YC. Non-aggregating tau phosphorylation contributes to motor neuron degeneration in spinal muscular atrophy. J Neurosci 2015;35:6038-6050.
36. Sanmaneechai O, Feely S, Scherer SS, Herrmann DN, Burns J, Muntoni F, Li J, Siskind CE, Day JW, Laura M, **Sumner CJ**, Lloyd TE, Ramchandren S, Shy RR, Grider T, Bacon C, Finkel RS, Yum SW, Moroni I, Piscosquito G, Pareyson D, Reilly MM, Shy ME; Inherited Neuropathies Consortium - Rare Disease Clinical Research Consortium (INC-RDCRC). Gentoype-phenotype characteristics and baseline natural history of heritable neuropathies caused by mutations of MPZ gene. Brain 2015;138:3180-3192.
37. Sullivan J, Zimanyi CM, Aisenberg W, Bears B, Chen D-H, Day JW, Bird TD, Siskind CE, Gaudet R, **Sumner CJ**. Novel mutations highlight the key role of the ankyrin repeat domain in TRPV4-mediated neuropathy. Neurol Genet 2015;1:e29.
38. Chhabra A, Carrino JA, Farahani SJ, Thawait GK, **Sumner CJ**, Wadhwa V, Chaudhry V, Lloyd TE. Whole Body MR Neurography: Prospective Feasibility Study in Polyneuropathy and Charcot Marie Tooth Disease. J Magn Reson Imaging. 2016;44:1513-1521

39. Schartner V, Romero NB, Donkervoort S, Treves S, Munot P, Pierson TM, Dabaj I, Malfatti E, Zaharieva I, Zorzato F, Brochier G, Lornage X, Eymard B, Taratuto AL, Böhm J, Gonorazky H, Ramos-Platt L, Feng L, Phadke R, Bharucha-Goebel D, **Sumner CJ**, Bui MT, Lacene E, Beuvin M, Labasse C, Schneider R, Thompson J, Boland A, Deleuze JF, Matthews E, Pakleza AN, Sewry CA, Franzini-Armstrong C, Biancalana V, Quijano-Roy S, Muntoni F, Fardeau M, Bönnemann CG, Laporte J. *DHPR (CACNA1S)* congenital myopathy. Acta Neuropathologica. 2017;133:517-533.
40. D'Ydewalle C, Ramos DM, Pyles NJ, Ng SY, Gorz M, Pilato CM, Ling K, Kong L, Ward AJ, Rubin LL, Rigo F, Bennett CF, **Sumner CJ**. The antisense transcript SMN-AS1 regulates SMN expression and is a novel therapeutic target for spinal muscular atrophy. Neuron 2017; 93:66-79.
41. Vasavda C, Zaccor NW, Scherer PC, **Sumner CJ**, Synder SH. Measuring G-protein coupled receptor signaling via radiolabeled GTP binding. Journal Visual Experiments 2017; 124.
42. Scherer PC, Zaccor NW, Neumann N, Vasavda C, Barrow R, Ewald A, Rao F, **Sumner CJ**, Synder SH. TRPV1 is a physiological regulator of mu-opioid receptors. Proceedings of the National Academy of Sciences 2017; 114(51):13561-13566.
43. Tao F, Beecham GW, Rebelo A, Blanton S, Moran JJ, Lopez-Anido C, Svaren J, Abreu L, Herrmann D, Day J, **Sumner CJ**, Lloyd T, Li J, Yum S, Bass F, Choi B-O, Pareyson D, Scherer SS, Reilly MM, Inherited Neuropathy Consortium, Shy ME, Zuchener S. Variation in SIPA1L2 is correlated with phenotype modification in CMT type IA. Annals of Neurology, 2019;85:316-330.
44. Tao F, Beecham GW, Rebelo AP, Blanton SH, Moran JJ, Lopez-Anido C, Svaren J, Abreu L, Rizzo D, Kirk CA, Wu X, Feely S, Verhamme C, Saporta MA, Herrmann DN, Day JW, **Sumner CJ**, Lloyd TE, Li J, Yum SW, Taroni F, Baas F, Choi BO, Pareyson D, Scherer SS, Reilly MM, Shy ME, Züchner S; Inherited Neuropathy Consortium. Modifier gene candidates in Charcot Marie Tooth disease type IA: a case only genome wide association study. Journal of Neuromuscular Disorders, 2019;6:201-211.
45. Darras BT, Crawford TO, Finkel RS, Mercuri E, De Vivo DC, Oskoui M, Tizzano E, Ryan MM, Muntoni F, Zhao G, Staropoli J, McCampbell A, Petrilo M, Stebbins C, Fradette S, Farwell W, **Sumner CJ**. Phosphorylated neurofilament heavy chain plasma levels in infants with SMA: analyses from the ENDEAR study. Annals of Clinical and Translational Neurology, 2019;6:932-944.
46. Pilato CM, Park J-H, Kong L, d'Ydewalle C, Valdivia D, Chen K, Griswold-Prenner I, **Sumner CJ**. Motor neuron loss in SMA is not associated with somal stress activated JNK/cJun signaling. Human Molecular Genetics, 2019.
47. Sullivan JM, Hellmich UA, Cox E, McCray BA, Aisenberg W, Blackshaw S, Gaudet R, and **Sumner CJ**. Characterization of the TRPV4 ankyrin repeat domain interactome and its disruption by neuropathy-causing mutations using a human protein microarray. In preparation.
48. Auslander N, Ramos D, Karathia H, **Sumner CJ**, Ruppin E. GENDULF: A novel computational algorithm to identify modifiers of monogenetic diseases. Genomic Medicine, 2019, *Under review*.
49. Guise AJ, Chauhan R , Hao LT, Zelaya I, Meziani A, Kong L, Valdivia D, Brandimarte ECUD, Akten B, d'Ydewalle C, Wesseling H, Cai1 B, Tang S, Crawford TO, He Z, Sahin M, King RW, Rubin LL, Lutz CM, Coppola G, Hemberg M, Beattie CE, **Sumner CJ**, Steen H, Steen JA. Proteogenomic analysis identifies APC/C-mediated degradation of SMN in neurons. Cell, *Under review*, 2019.
50. Zaccor NW, **Sumner CJ**, Snyder SH. The nonselective cation channel TRPV4 inhibits angiotensin II receptors. Journal of Biological Chemistry, *Under revision*, 2019.
51. Mohassel P, Liewluck T, Hu Y, Ezzo D, Ogata T, Saade D, Neuhaus S, Bolduc V, Zou Y, Donkervoort S, Medne L, **Sumner CJ**, Dyck PJB, Wierenga KJ, Tennekoon G, Finkel RS, Chen J, Winder TL, Staff NP, Foley AR, Koch M, Bönnemann CG. Dominant collagen XII mutations cause a distal myopathy. Annals of Clinical and Translational Neurology, 2019, *Accepted*.
52. Ramos D, d'Ydewalle C, Valdivia D, Hatem CL, Gabbeta V, Dakka A, Taylor SJ, Zaworski PG, Swoboda KJ, Prior TW, Snyder PJ, Naryshkin N, Paushkin S, Crawford TO, **Sumner CJ**. Normal developmental, disease and post-therapy SMN expression: implications for treating SMA patients. Journal of Clinical Investigation, 2019, *Accepted*.
53. Fridman V, Sillau S, Acsadi Gyula, Bacon C, Bray K, Burns J, Day J, Feely S, Finkel RS, Grider T, Gutmann L, Herrmann DN, Kirk CA, Knause SA, Laura M, Lewis RA, Li J, Lloyd TE, Moroni I, Muntoni F, Pagliano E, Pareyson D, Pisciotta C, Piscosquito G, Ramchandran S, Saporta M, Sadjadi R, Shy RR, Siskind CE, **Sumner CJ**, Wilcox J, Walk D, Yum SW, Zuchner S, Scherer SS, Reilly MM, Shy ME. A longitudinal study of CMT1A using Rasch analysis based on CMT neuropathy and examination scores. Neurology, 2019, *Accepted*.
54. Sullivan JM, Motley WW, Johnson JO, Aisenberg W, Gaudet R, Traynor BJ, and **Sumner CJ**. A dominant mutation in the notch ligand JAG1 as a novel cause of Charcot-Marie-Tooth disease type 2C. Journal Clinical Investigation, *Under revision 2019*.

55. Woolums BM, McCray BA, Sung H, Tabuchi M, Sullivan JM, Takle K, Yang Y, Mamah C, Aisenberg WH, Saavedra P, Larin BS, Robinson DN, Xiang Y, Wu MN, **Sumner CJ***, Lloyd TE* (*co-corresponding). TRPV4 disrupts mitochondrial transport and causes axonal degeneration via CaMKII-dependent increases of intracellular Ca^{2+}. Neuron, 2019, *submitted.*

Review Articles

1. **Sumner, CJ**. Therapeutics development in spinal muscular atrophy. Neurotherapeutics 2006;3:235-245.
2. **Sumner CJ**. Molecular mechanisms of spinal muscular atrophy. J Child Neurol 2007;22:979-89.
3. Burnett BG, Crawford TO, **Sumner CJ**. Emerging treatment options for spinal muscular atrophy. Curr Treat Options Neurol 2009;11:90-101.
4. Wee C, Kong L, **Sumner CJ**. The genetics of spinal muscular atrophies. Curr Opin Neurol 2010;23:450-8.
5. Van Meerbeke J and **Sumner CJ**. Progress and promise: The current status of spinal muscular atrophy therapeutics. Discov Med 2011;12:291-305.
6. **Sumner, CJ.** Spinal muscular atrophy, John Griffin, and mentorship. JPeripher Nerv Syst 2012;Suppl 3:52-6.
7. Schindler A, **Sumner C**, Hoover-Fong J. TRPV4 associated Disorders. GeneReviews 2014 (http://www.ncbi.nlm.nih.gov/books/NKB1116/)
8. d'Ydewalle C and **Sumner CJ**. SMA therapeutics: Where do we stand? Neurotherapeutics 2015;12:303-16.
9. Landouré G, Samassékon O, Traoré M, Meileur KG, Oumar GC, Burnett BB, **Sumner CJ**, Fischbeck KH. Genetics and genomic medicine in Mali: challenges and future perspectives. Mol Genet Genomic Med.2016;4:126-34.
10. **Sumner CJ**, Crawford TO. Two breakthrough gene targeted treatments for spinal muscular atrophy: challenges remain. Journal of Clinical Investigation, 2018;2018: 3219-3227.
11. Ravi B, Antonellis A, **Sumner CJ**, Leiberman A. Genetic approaches to the treatment of inherited neuromuscular disease. Human Molecular Genetics, 2019.

Case Reports

1. **Sumner CJ**, Golden JA, Hemphill C. Should thrombolysis be contraindicated in patients with cerebral ateriovenous malformations? Crit Care Med 2002;30:2359-2362.
2. **Sumner CJ**, Fischbeck KH. Jaw drop in Kennedy's disease. Neurology 2002;59:1471-1472.
3. **Sumner CJ**, Newman M, Hereema A, Jay C. Apparent relapse of a spinal mycobacterial infection after highly active retroviral therapy. Neurology 2003;61:139-140.
4. Freund B, Hayes L, Rivera Lara L, **Sumner CJ**, Chaudhry V, Chatham-Stephens K, Blythe D, Brooks R, Probasco JC. Adult botulism colonization mimicking brain death. Muscle and Nerve 2017; Accepted.

Books

1. Spinal Muscular Atrophy: Disease Mechanisms and Therapy. Editors: **Sumner CJ**, Paushkin S, and Ko CP. Elsevier/Academic Press. 2016.

Book Chapters

1. Atwood W, **Sumner CJ**, Major EO. Molecular mechanisms of the pathogenesis of progressive multifocal leukoencephalopathy: a JC virus induced demyelinating disease of the human brain. In: Viral Pathogenesis in the Nervous System, edited by Kurara T. Sarkon, Tokoyo, Japan, 1995:133-146.
2. **Sumner CJ,** Fischbeck. Kennedy's Disease. In: Motor Neuron Disorders*,* edited by Shaw PJ, Strong M. Butterworth-Heinmann, Woburn MA, 2002:425-434.
3. Piccioni, F, **Sumner CJ**, Fischeck CJ. Androgen receptor and spinal and bulbar muscular atrophy. In: Zinc Finger Proteins*,* edited by Iuchi S and Kuldell N. Landes Bioscience, Georgetown TX, 2004.
4. **Sumner CJ**. Inflammatory Neuropathies: Guillan-Barré syndrome and chronic inflammatory demyelinating polyradiculoneuropathy. In: Treatment of Pediatric Neurologic Disorders, edited by Singer HS, Crawford TO, Kossoff EH, Hartman AL. Marcel Dekker, Inc. New York, NY, 2004.
5. **Sumner CJ**, Fischbeck KH. Spinal Muscular Atrophy. In: Neurobiology of Disease, edited by Gilman S, Elsevier. San Diego CA, 2006.
6. Sullivan JM, Lloyd TE, **Sumner CJ**. Hereditary channelopathies caused by TRPV4 mutations. In: Pathologies of Calcium Channels, edited by Weiss N and Koschak A. Springer, Berlin, Germany, 2014:413-440.

7. Perez-García MJ, Kong L, **Sumner CJ**, and Tizzano E. Developmental aspects and pathological findings in SMA. In: Spinal muscular atrophy : disease mechanisms and therapy, edited by Sumner CJ, Paushkin S, and Ko CP. Elsevier, USA, 2016.
8. Wooley J, Crowder ME, Pyles NJ, **Sumner CJ.** Spinal Muscular Atrophy. In: ***Neurodegeneration***, edited by Zbigniew K. Wszolek, Anthony Schapira, Ted Dawson, and Nick Wood, Johns Wiley and Sons, Hoboken, NJ, 2017.

Editorials

1. Burnett, BG and **Sumner CJ**, Targeting splicing in spinal muscular atrophy. Annals of Neurology 2008;60:3-6.
2. Sullivan J, Landouré G, Gaudet R, and **Sumner CJ**. TRPV4 neuropathy-causing mutations localize to the convex face of the ankyrin repeat domain Neurology 2014;83:1991.
3. Brandsema JF and **Sumner CJ**. Spinal Muscular Atrophy: Further Expanding the Clinician's Armamentarium. Neurology, *In press*, 2019.

Other Media

2011 **Sumner CJ**, Burghes AH "Influence of housing, feeding, and handling conditions on SMA mouse performance" Treat NMD Neuromuscular Network Experimental Protocols for SMA mice (www.treat-nmd.eu/sma/research-resources/preclinical-standards/sma-sops/).

2011 **Sumner CJ** "Nutrition and exercise in spinal muscular atrophy" Fight SMA (www.youtube.com/watch?v=8DgmILAIJ_E).

2011 **Sumner CJ "**Learn about spinal muscular atrophy," DNA Learning Center Cold Spring Harbor Laboratory (http://www.learnaboutsma.org/therapies/7.html).

2012 **Sumner CJ** The Good Fight 2013 (http://vimeo.com/68715115).

2014 Schindler A, Sumner CJ, Hoover-Fong J "TRPV4 associated Disorders" GeneReviews (http://www.ncbi.nlm.nih.gov/books/NKB1116/).

2015 **Sumner CJ** Fight SMA Annual Webcast (http://vimeo.com/92044761).

2018 **Sumner CJ** "Gene-targeted therapies for spinal muscular atrophy": Society for Neuroscience Webinar: *Gene Therapy to Address Unmet Needs in Neurology* (https://web.sfn.org/OMP/Articles/Scientific-Research/2018/Webinar-Gene-Therapy-to-Address-Unmet-Needs-in-Neurology).

2019 Sumner CJ. "Gene-targeted therapies for spinal muscular atrophy," Expert Talks, Neurodiem. http://www.neurodiem.com/topic/neuromuscular.

2019 Chiriboga CA, Castro DP, Schroth M, **Sumner CJ**, Swoboda KJ. Collaborating in the care of spinal muscular atrophy: a multidisciplinary approach to timely screening, diagnosis, and management. A supplement to Neurology Reviews and Pediatric News.

FUNDING

EXTRAMURAL Funding

Current

9/1/19-8/31/24 **TRPV4 links the blood-neural barrier to motor neuron dysfunction**
R01 NS115475
Sponsor: NINDS/NIH
Total direct costs: TBD
Principal investigator: Sumner CJ
Effort: 25%
The main goal of this project is to characterize the role of TRPV4 in regulating the blood neural barrier and consequences for motor neuron function.

8/1/19-7/31/22 **A key role for TRPV4 in neurodegeneration via control of the blood-nerve barrier.**
629305
Sponsor: Muscular Dystrophy Association
Total direct costs: $300,000
Principal investigator: Sumner, CJ

	The main goal of this project is the characterize mice harboring mutations of the TRPV4 cation channel
3/1/18-2/28/20	**Neurofilaments as markers of neurodegeneration in SMA** Sponsor: Cure SMA Total direct costs: $200,000 Principal investigator: Sumner, CJ The goal of this project is to evaluate the potential of blood NF-L and NF-H levels as biomarkers in SMA mice and patients.
4/1/18-3/31/23	**Axonal developmental pathology in SMA** R01 NS062869 Sponsor: NINDS/NIH Total direct costs: TBD Principal Investigator: Sumner CJ Effort: 25% The main goal of this project is to characterize developmental abnormalities that drive early SMA pathogenesis and timing of therapeutic reversibility.
2/1/18-1/31/20	**Testing the potential of *SMN-AS1* as a therapeutic target in SMA** Foundation grant Sponsor: Cure SMA Total direct costs: $150,000 Effort: 3% The main goal of this project is to characterize mice and primary neurons from mice with targeted knock-out of the SMN-AS1 locus in order to determine the effect of absence of SMN-AS1 on SMN expression.
10/1/10-12/31/20	**lnRNAs in spinal muscular atrophy** Sponsor: Spinal Muscular Atrophy Research Team (SMART) Total direct costs: $550,000 Principal Investigator: Sumner CJ Effort: 5% The goal of this grant is to characterize long noncoding RNAs that regulate SMN expression.
5/1/11-present	**Genetics and Natural History of CMT** Sponsor: Muscular Dystrophy Association and CMT Association Total direct costs: $13,000/year Principal Investigator: Shy ME (University of Iowa) Role: Site Co-PI Effort: 0% The objective of this project is to carry out natural history and genetic studies in patients with inherited neuropathy as part of the Inherited Neuropathy Consortium.
1/1/14–present	**Peripheral Neuropathy Research Registry** Sponsor: The Foundation for Peripheral Neuropathy Total direct costs: $67,500 Principal Investigator: Hoke, Ahmet Role: Co-investigator Effort: 1% This project enrolls patients with phenotypic and molecular correlates of neuropathic pain in patients with peripheral neuropathy.

1/1/15-12/31/20 **Investigation of inherited motor neuron diseases**
Sponsor: Ujala Foundation
Total direct costs: $225,000
Principal Investigator: Sumner CJ
Effort: 1%

2/15/16-2/14/21 **Targeting a long noncoding RNA for the treatment of SMA**
R01 NS096770
Sponsor: NINDS/NIH
Total direct costs: $1,648,314
Principal Investigator: Sumner CJ
Effort: 40%
The main goal of this project is to investigate the role of a SMN associated lncRNA in the regulation of SMN gene expression and to assess its potential as a therapeutic target in SMA.

4/1/16-3/31/19 **Research Supplement to Promote Diversity**
R01 NS096770-01S1
Sponsor: NINDS/NIH
Total direct costs: $167,175
Principal Investigator: Sumner, CJ
Mentee: Daniel Ramos (Ph.D. graduate student)

Previous Grants/Contracts

5/1/16-4/30/18 **Preclinical development of a JNK drug candidate to alter disease progression in SMA**.
Disease foundation grant
Sponsor: Cure SMA
Total direct costs: $300,000
Principal Investigator: Sumner, CJ
Effort: 2%
The main goal of this project is to evaluate whether JNKs are activated in SMA mice and to assess whether novel JNK inhibitors can ameliorate the disease phenotype.

3/1/16-2/28/18 **Assessing the reversibility of proximal axon abnormalities in SMA mice.**
Disease foundation grant
Sponsor: Cure SMA
Total direct costs: $140,000
Principal Investigator: Sumner, CJ
Effort: 3%
The main goal of this project is to determine whether proximal axonal abnormalities in SMA are due to impaired development and when this pathology can be reversed by SMA therapeutics.

11/1/09-12/31/17 **Collection and analysis of human SMA tissues**
Sponsor: Spinal Muscular Atrophy Foundation
Total Direct Costs: $980,981
Principal Investigator: Sumner, CJ
Effort: 5%
The main purposes of this project are to establish a tissue repository of human SMA tissues and to understand the pathology and biochemistry of human SMA.

8/1/04-6/30/09 **Regulation of the survival motor neuron gene**

K22 NS048199-01
Sponsor: NIH/NINDS
Total direct costs: $600,000
Principal Investigator: Sumner, CJ
Effort: 73%
The main goal of this project was to investigate the role of epigenetic determinants in the regulation of the *SMN* gene in order to advance therapy for the spinal muscular atrophy.

3/1/07-2/28/09

Targeting the muscle and neuromuscular junction for SMA therapeutics
Families of SMA Research Award
Sponsor: Families of Spinal Muscular Atrophy
Total Direct Costs: $160,000
Principal Investigator: Sumner, CJ
Effort: 6%
The main goal of this project was to investigate the role of muscle in the pathogenesis of SMA and to examine the ability of histone deacetylase inhibitors and IGF-1 to ameliorate disease manifestations.

8/1/07-7/31/12

Targeting muscle in spinal muscular atrophy
HHMI Physician-Scientist Early Career Award
Sponsor: Howard Hughes Medical Institute
Total Direct Costs: $375,000
Principal Investigator: Sumner, CJ
Effort: 1%
The main goal of this project was to investigate whether there is impaired development of muscle in SMA and whether therapeutic strategies that speed muscle development lessen SMA disease manifestations.

5/15/09-4/30/15
(one year NCE)

Muscle and neuromuscular junctions in spinal muscular atrophy
R01 NS062869
Sponsor: NINDS/NIH
Total direct costs: $1,156,726 (Including subcontract)
Principal Investigator: Sumner CJ
Effort: 40%
The main goal of this project was to investigate the roles of impaired myogenesis and neuromuscular junction synaptogenesis in the pathogenesis of the spinal muscular atrophy.

6/3/09-6/2/10

The electrophysiological correlates of spinal muscular atrophy
NINDS New Initiative
Sponsor: NINDS (Intramural program)/NIH
Total Direct Costs: $250,000
Co-Principal Investigators: Sumner, CJ, O'Donovan, M, Fischbeck, KH
Effort: 1%
The main goal of this project was to characterize the morphological and electrophysiological abnormalities of spinal circuits in spinal muscular atrophy mice and evaluate the effects of drug therapies.

11/1/09-10/31/12

Evaluation of histone deacetylase inhibitors and combination therapy in SMA mice
Sponsor: Spinal Muscular Atrophy Foundation
Total Direct Costs: $210,136
Principal Investigator: Sumner, CJ

Effort: 5%

10/1/10-3/31/12 **Quinazolines in spinal muscular atrophy**
Sponsor: Repligen
Total Direct Costs: $100,000
Principal Investigator: Sumner, CJ
Effort: 1%
The main goal of this project was to investigate the therapeutic effects of quinazoline compounds in SMA mice.

12/1/12-11/30/14 **Motor axon development in spinal muscular atrophy**
Families of SMA Research Award
Sponsor: Families of Spinal Muscular Atrophy
Total Direct Costs: $160,000
Principal Investigator: Sumner, CJ
Effort: 1%
The main goal of this project was to investigate impairments of motor axon radial growth in SMA and the contribution of SMN-deficient Schwann cells to this process.

2/15/14-1/31/16 **A model of TRPV4 channelopathy**
R21 1NS087579
Sponsor: NINDS/NIH
Total direct costs: $324,156
Principal investigator: Sumner CJ
Effort: 10%
The goals of this grant were to characterize a novel mutant knockin model of TRPV4 channelopathy and test whether neuropathy causing mutations result in a gain of channel function in primary neurons.

8/1/13-7/31/16 **Characterization of TRPV4 peripheral neuropathy in animal models**
Muscular Dystrophy Association 2777551
Sponsor: Muscular Dystrophy Association
Total direct costs: $272,727.30
Principal investigator: Sumner CJ
Effort: 5%
The objectives of this project are to generate and characterize *Drosophila* and mouse models of TRPV4 channelopathy.

INTRAMURAL Funding

Previous

7/1/15-6/30/16 **Targeting a SMN lncRNA as a treatment for spinal muscular atrophy**
Accelerated Translation Incubator Program
Sponsor: Johns Hopkins ICTR
Total Direct Costs: $95,000
Principal investigator: Sumner CJ
Effort: 0%
The objective of this project is to evaluate the ability of SMN lncRNA targeted ASOs to increase SMN expression in cultured cells and *in vivo*.

4/1/09-3/31/10 **Evaluation of histone deacetylase inhibitors in spinal muscular atrophy mice**
Accelerated Translation Incubator Program
Sponsor: Johns Hopkins Institute for Clinical and Translational Research (ICTR)

Total Direct Costs: $100,000
Principal Investigator: Sumner, CJ
Effort: 1%
The main goal of this project was to identify novel HDAC inhibitors that show efficacy and minimal long term toxicity in SMA mice.

CLINICAL ACTIVITIES

Clinical Focus

- Neuromuscular diseases including spinal cord, spinal root, motor neuron, peripheral nerve, neuromuscular junction, and muscle disease.
- Special emphasis on inherited neuromuscular diseases including inherited motor neuron (spinal muscular atrophies), peripheral nerve (Charcot Marie Tooth disease) and muscle (muscular dystrophy) disorders.
- General inpatient neurology.

Certification

Medical Licensure

2000-2007 California Medical License number A65378 (Inactive)
2006-present Maryland Medical License number D0065606 (Active)

Specialty Board Certification

2001 American Board of Psychiatry and Neurology, Adult Neurology.
2011 American Board of Psychiatry and Neurology, Adult Neurology Maintenance of Certification.

Clinical Service Responsibilities

2007-present Muscular Dystrophy Association Neuromuscular Clinic ½ day per week
2007-present Neuromuscular consult attending one month per year
2007-present Neurology inpatient/consult attending one month per year
2009-present Charcot Marie Tooth Clinic ½ day two times per month

Clinical Program Building/Leadership

2007-present Specialty Program for clinical care of patients with spinal muscular atrophy. This is a multidisciplinary clinic to provide optimized standard of care to children (Dr. Thomas Crawford) and adults (Sumner) with SMA with attention to transitions from the pediatric to adult setting.
2007-present Muscular Dystrophy Association Clinic Principal Provider.
2010-present Co-Founder and Co-Director, Charcot-Marie-Tooth Clinic, Department of Neurology, Johns Hopkins School of Medicine. This is a multi-disciplinary clinic that includes 3 neurologists, physical and occupational therapists, genetics counselor, and orthotist.

EDUCATIONAL ACTIVITIES

Educational focus

- Medical student, neurology resident, and neuromuscular fellow clinical teaching in outpatient and inpatient settings.
- Undergraduate, graduate student (MD and PhD), and postdoctoral fellow teaching in laboratory setting.
- Classroom teaching of undergraduate, School of Public Health, and medical school students as well as neurology residents.
- Patient education in information sessions and support groups.

Teaching

Classroom instruction

JHMI

2006-present	Lecturer, Johns Hopkins University School of Medicine, Department of Neurology Resident lecture series (annually): "Spinal Muscular Atrophies"
2008-present	Lecturer, Johns Hopkins School of Public Health, Department of Molecular Microbiology and Immunology, Introduction to the Biomedical Sciences (annually): "Spinal muscular atrophy: Clinical features, genetics, and therapeutics development"
2010-'14,'17-'19	Lecturer, Johns Hopkins University School of Medicine medical student neuroscience curriculum (annually) "Peripheral neuropathies"
2010-2012	Lecturer, Johns Hopkins University School of Medicine First year medical students neuroscience curriculum Teaching the neurological history and examination
2011, 2012	Lecturer, Johns Hopkins Multidisciplinary Pain Medicine Fellows, "A Neuromuscular Clinician's Approach to a Patient with Neuropathic Pain."
3/6/2012	Lecturer, Kennedy Kreiger Institute Neurodevelopmental Fellows Lecture Series, "Developing treatment for inherited motor neuron diseases."
2/12,'13,'15	Lecturer, Johns Hopkins Department of Neuroscience future graduate students, "Alterations of neural circuitry in inherited motor neuron diseases."
9/25/12-pres	Lecturer, Johns Hopkins University Undergraduate Neuroscience Course: Diseases and Disorders of the Nervous system (annually). "Spinal muscular atrophy: clinical features, genetics, and progress towards treatment."
10/2/12	Mentor, Johns Hopkins School of Medicine OWISM Speed Mentoring: "Academic Success."
6/20/13	Lecturer, Johns Hopkins Department of Neurology Summer Student Program "Is spinal muscular atrophy now a treatable disease?"
8/07/13	Lecturer, Johns Hopkins School of Medicine MD/PhD program Herlong Rounds.
11/05/13	Lecturer, Johns Hopkins School of Medicine Department of Neurology Neuromuscular Fellows "Spinal muscular atrophies/hereditary motor neuropathies"
7/24/14	Lecturer, Johns Hopkins School of Medicine Neurology Summer Student Conference: "Developing treatment for spinal muscular atrophy"
4/30/15& 5/1/18	Small group instructor, "Diseases of neurodevelopment", Johns Hopkins Department of Neuroscience Graduate Student Seminar Course: Neurodevelopment.
10/14/16	Johns Hopkins Department of Neurology Residents "Meet Your Mentors" Panel.
11/9/16	Invited speaker, "The patients we remember," Johns Hopkins Medical Student SIGN Group.
4/26/17, 5/2/18,5/1/19	Invited seminar, "The career path of neurologist clinician scientist," Johns Hopkins University School of Medicine 1st year medical students.
11/8/17	Lecturer, "Charcot Marie Tooth Disease," Johns Hopkins Department of Neurology Resident Teaching Conference.

Regional/National

4/1/02	Lecturer, "Neurogenetics Cases." Neurogenetics Course, American Academy of Neurology Annual Meeting, Denver, CO
4/5/03	Lecturer, "Neurogenetics Cases," Neurogenetics Course, American Academy of Neurology Annual Meeting, Honolulu, HI
2/10/10	Lecturer, Mayo Clinic, Department of Neurology Resident lecture series: "Inherited neuromuscular disease cases"
10/14/11	Lecturer, Sinai Hospital Physical and Rehabilitation Medicine Resident Teaching series, "Charcot Marie Tooth Disease."
4/3/15	Lecturer, "Neuromuscular cases" Carver College of Medicine Department of Neurology Neuromuscular Cases Conference, Grand Rapids, Iowa.
11/5/16	Discussant, "Desert with experts," A Women's Journey Conference, Baltimore, MD.
11/10/17	Invited Speaker, "Antisense oligonucleotides for spinal muscular atrophy," Society for Neuroscience Neurobiology of Disease Symposium, Washington DC.
10/10/18	Invited Spearker, "Update on hereditary motor neuropathies," American Academy of Neuromuscular and Electrodiagnostic Medicine Annual Meeting, Washington DC.
5/22/19	"Treatments for SMA: A breakthrough in neurodegeneration translational research," UCSF Department of Neurology Residency Conference, San Francisco, CA.

5/23/19 "Therapeutics development for Charcot Marie Tooth disease," UCSF Department of Neurology Neuromuscular fellow conference, San Francisco, CA.

6/14/19 Plenary Speaker, "Treatments for SMA: a breakthrough in neurodegenerative research," CCRC Symposium for MD/PhD students, Bethesda MD.

Clinical instruction

2006-present Muscular Dystrophy Association Clinic: Attending physician supervision/teaching of medical students, residents, post-doctoral fellows in the outpatient clinic one morning per week.

2006-present Neurology Consultation and Inpatient Neurology Service: Attending physician supervision/teaching of medical students and residents during rounds on the neurology inpatient and consultation service at Johns Hopkins Bayview Hospital one month per year.

2006-present Neuromuscular Diseases Neuromuscular Service: Attending physician supervision/teaching of clinical fellow during rounds on the neuromuscular consultation service at Johns Hopkins Hospital one month per year.

2012-present Charcot Marie Tooth Clinic: Attending physician supervision/teaching of medical students, residents, post-doctoral fellows in the outpatient clinic one morning per month.

CME

6/12/09 "Inherited Neuropathies", Johns Hopkins Neuromuscular Division and Department of Neurology Update in Neuromuscular Disease, Baltimore, MD.

6/20/12 "SMN Biology and Therapeutic Strategies", Families of Spinal Muscular Atrophy Course: Interdisciplinary Perspectives on Spinal Muscular Atrophy: Defining Your Role. Bloomington, MN.

6/13/14 "SMN Biology and Therapeutic Strategies", Interdisciplinary perspectives on spinal muscular atrophy: defining your role. Washington, DC.

Workshops/seminars

National

6/6/04 Lecturer, "Neuroanatomy and the neurological examination" National Society of Genetic Counselors, Washington, DC.

10/7/06 Lecturer, American Neurological Association Residents Career Advisory Seminar

4/10/07 Lecturer, FELCOM NIH Fellows Career Development Seminar, "Getting and keeping a job after NIH fellowship,"

4/23/08 Lecturer, Fight SMA meeting informational workshop for SMA patients, "Nutrition and SMA"

3/18/08 Lecturer, Annual Howard Hughes Medical Institute Meeting of Medical Fellows and Research Scholars, "Pathway to Becoming a Physician-Scientist"

6/21/08 Lecturer, Families of SMA Patient Conference, "How the biology of SMA Guides Therapeutic Approaches"

7/16/08 Lecturer, NINDS summer student seminar "Becoming a Physician-Scientist"

6/21/09 Lecturer, Families of SMA Patient Conference, "Progress in Basic Research Panel"

4/26/10 Lecturer, Fight SMA meeting informational workshop for SMA patients, "Living with SMA"

9/24/10 Lecturer, Maryland/Pennsylvania CMT Patient Support Group, "Inherited Neuropathies"

6/26/11 Lecturer, Families of SMA Patient Conference, "Major findings from the 15th SMA research meeting and next steps."

10/13/13 Lecturer, American Neurological Association 2013 "Meeting choosing your career path: the sky is the limit the perspective of a basic scientist"

11/12/14 Lecturer, American Neurological Association 2014 "Meeting choosing your career path: the sky is the limit the perspective of a basic scientist"

11/13/14 Panelist, "Enhancing diversity in academic neurology," American Neurological Association Meeting 2014.

9/26/15 Lecturer, American Neurological Association 2015 Early Career Faculty Development Course "Finding happiness: Panel discussions in lab, translational and clinical research.

11/9/16 Panel Discussant, Elsevier and JHU Welch Library/JHU Professional Development and Career Office Scholarly Publishing Symposium.

6/29/17 Panel Discussant, "Basic research funding in SMA," Cure SMA Annual Meeting, Orlando FL.

11/26/18 Panel Discussant, "Diversity in Science" in Ethics and Society Course, Johns Hopkins Neuroscience Graduate Student Program.

International

7/8/17 Invited Lecturer, 2017 Peripheral Nerve Society Meeting Teaching Course, "Innervation and denervation in health and disease," Sitges, Spain.

Mentoring

Predoctoral Mentees/Advisees

2002 Stephen Perhac, B.S., Undergraduate research, NINDS. Awarded Exceptional NINDS Summer Student Award. Current position: Anesthesiologist in Seattle Washington. Shared publication #3.

2002-2003 Thanh Huynh, B.A., Undergraduate research HHMI/NIH research scholarship. Awarded the AAN Saul R. Korey Medical Student Essay Award in Experimental Neurology. Current position: Clinical Instructor in Pulmonology and Critical Care Medicine, University of California Los Angeles. Shared publication #3.

2003-2004 Nathan Woodling, B.S. Candidate, Undergraduate research NINDS, Awarded Exceptional NINDS Summer Student Award and The Levon O. Parker Scholarship. Current Position: Completed PhD in Neuroscience at Stanford and now a postdoctoral fellow at the University of College London in Dr. Laboratory of Linda Patridge. Shared publication #6.

2003-2004 Lauren Kernochan, B.A., Undergraduate research HHMI/NIH research scholarship. Awarded the AAN Saul R. Korey Medical Student Essay Award in Experimental Neurology. Current position: Completed Residency in Pathology at the University of Washington and now a practicing anatomical pathologist in Santa Barbara California. Shared publication #6.

2004-2005 Melissa Russo, B.S., Undergraduate research HHMI/NIH research scholarship. Awarded the AAN Saul R. Korey Medical Student Essay Award in Experimental Neurology. Current position: Completed residency in Obstetrics and Gynecology and now a fellow in maternal-fetal medicine at Johns Hopkins University School of Medicine. Shared publication: #6.

2006-2008 Dong Choe, B.S., Predoctoral research, Department of Neurology, JHU. Current position: Completed medical school at Albert Einstein College of Medicine of Yeshiva University and now an Emergency Medicine Residency Stony Brook University Hospital. Shared publications: #14, 16, 17, 22.

2007 Khadeijah Palmer-Rhodes, High school summer research.

2008-2012 Tara Martinez, Graduate student, Department of Molecular Microbiology and Immunology, Johns Hopkins University. Current Position: US patent office. Shared publications: #19, 22, 25, 28, 31.

2008-2010 Claribel Wee, Predoctoral research, Department of Neurology, JHU. Current position: Neurology Resident: Upstate Medical University State University of New York. Shared publications: #17, 22.

2008 Marianne Stazza, Undergraduate research, JHU. Current position: Graduate student Drexel University.

2009-2010 Celeste Lipkes, Undergraduate research, JHU. Current position: Medical Student Virginia Commonwealth University. Shared publication: #22.

2009-2010 Jonathan F. Thorndike, Predoctoral research, JHU. Current position: Medical Student Brown University.

2009 Jaclyn S. Lopez, Undergraduate research, JHU.

2010-2012 Heloisa Carvalho, Undergraduate research, JHU. Current position: Medical Student Boston University.

2010-2011 Katie Davis, Predoctoral research, JHU.

2010-present James Van Meerbeke, B.A. Predoctoral research, Department of Neurology, JHU. Current position: Medical Student University of Maryland. Symposium presentation American Academy of Neurology Meeting 2012. Shared publications: #25, 26, 27, 30, review #5.

2010-2012 Melissa Crowder, B.A. Predoctoral research, Department of Neurology, JHU. Poster presentation at Families of SMA meeting 2012. Current position: Neurology resident Dartmouth Medical School. Shared publications: #21, 25.

2010-2011 Lauren Woo, B.A. Predoctoral research, JHU. Current position: Medical Student at University of Massachusetts Medical School.

2010-2011 Kristen Klepac, B.A. Predoctoral research, JHU. Current position: Medical Student University of California San Francisco.

2010-2011 Emmanuel Ohuabunwa, Undergraduate research, JHU. Current position: Medical Student Yale University.

2010-2014	Xixi Xu, Undergraduate research and Masters Student, Johns Hopkins University. Symposium presentation at Families of SMA meeting 2014. Current position: Medical Student at Boston University.
2010-2014	Paul Sampognaro, Medical Student Research, Johns Hopkins School of Medicine. Current position: Neurology resident at the University of California San Francisco. Poster presentation Medical Student Research Day. Shared publication: #26.
2011-2013	Diana Villaneuva, Undergraduate research, University of Maryland. Current position: Biopharmaceutical Manufacturing Engineer Glaxo Smith Kline.
2011-2014	Adam Miller, Undergraduate research JHU and then research technologist. Current position: Medical Student University of Wisconsin.
2011-2012	Joe Wooley, Predoctoral research, JHU. Current position: Medical Student Case Western Reserve School of Medicine. Shared publication: #29.
2011	Emily Bergbower, Johns Hopkins Cellular and Molecular Medicine Graduate Student rotation.
2012	Aggie Rudicki, Johns Hopkins Cellular and Molecular Medicine Graduate Student rotation. Shared publication: #29.
2012	Emily Kuehn, Johns Hopkins Department of Neuroscience Graduate Student rotation.
2012-2016	Noah Pyles, Undergraduate student rotation from Bowdoin College and Research technologist. Shared publication: #34.
2012-2014	Sharmaine Ross, Undergraduate Student Research, Johns Hopkins University. Current position: Masters student at Columbia University.
2012-2013	Kristy Carranza, Undergraduate Student Research, Johns Hopkins University. Current position: Masters student Boston University.
2012	Chanel Matney, Johns Hopkins Department of Neuroscience Graduate Student rotation.
2012-2014	Rhiannon Desideri, Predoctoral research as research technologist. Current position: Veterinary Student Cornell Univeristy.
2012-2017	David Valdivia, Predoctoral research as research technologist. Shared publication: #34. Current position: Medical student University of Miami.
2012	Jacqueline Pham, Johns Hopkins Cellular and Molecular Medicine Graduate Student Rotation.
2012-2015	Mario Gorz, Undergraduate Student Research and Research Technologist. Current Position: Medical student SUNY Downstate.
2013&2014	Celeste Pilato, Undergraduate Summer Student Research from Dickinson College.
2013	Nicole Bonsavage, Undergraduate Student Research, Johns Hopkins University.
2013	Shi (Andy) Huang, Undergraduate Student Research, Johns Hopkins University.
2013	Rachael Cohen, Johns Hopkins School of Medicine Cellular and Molecular Medicine Graduate Student Rotation.
2013	Jonathan Ling, Johns Hopkins School of Medicine Pathobiology Graduate student rotation.
2014-present	Daniel Ramos, Johns Hopkins School of Medicine Department of Neuroscience graduate student. Awards/Grants: 1) National Science Foundation Graduate Student Award Honorable Mention. 2) Research Supplement to Promote Diversity R01 NS096770-01S1. 3) Kirschstein-NRSA predoctoral fellowship award.
2014	Purnima Padmanabhan, Johns Hopkins School of Medicine Department of Neuroscience graduate student rotation.
2014	Anais Kessler, Johns Hopkins Packard Center High School Summer Student.
2014	Gideon Loevinson, Johns Hopkins School of Medicine MD/PhD student rotation.
2014-2015	Breanna Bears, Research technologist. Current Position: Medical Student University of Maryland.
2014-2017	Zachary Fusfeld, Undergraduate Student Research, Johns Hopkins University.
2014-2015	Doris Valenzuela-Araujo, Undergraduate Student Research, Johns Hopkins University. Current position: Medical Student Ohio State University.
2014-2017	Jennifer Huh, Undergraduate Student Research and Research technologist, Johns Hopkins University. Current position: Medical student University of Chicago.
2014-present	William Aisenberg, Graduate Student, Cellular and Molecular Medicine, Johns Hopkins School of Medicine. Awards/Grants: 1) Richard P. Bunge Prize for outstanding poster presentation at 6th CMTR Meeting 2016, 2) Travel grant to attend the 6th CMTR Meeting 2016, 3) F32 NIH Award. Shared publication #37.
2015-2017	Billy Kim, Undergraduate Student Research, Johns Hopkins University.

2015-2017 Celeste Pilato, Research technologist. Shared publication #39, #40 (Current position: medical student).
2015-present Maria Tejera, Undergraduate Student Research, Johns Hopkins University.
2015-present Nicholas Zaccor, Johns Hopkins School of Medicine Department of Neuroscience MD/PhD Graduate student. Shared publication #41.
2016-present Michelle Santangelo, Undergraduate Student Research, Johns Hopkins University.
2016-present Jae Hong Park, Research technologist.
3/17-5/17 Kirsten Maulding, Cellular and Molecular Medicine Graduate Student Rotation.
6/17-present Pamela Saavedra, Research technologist.
6/17&18 -8/17&18 John Maragakis, High school summer student.
6/17-8/17 Lale Sude Gucer, International medical student rotation (from Turkey).
9/17-present Chloe Grzyb, Undergraduate Student Research, Johns Hopkins University
9/17-present Scotty McGaugh, Undergraduate Student Research, Johns Hopkins University
9/17-present Cera Hassain, Postgraduate Research
5/19-present Michelle Harran, Department of Neuroscience Graduate Student.

Postdoctoral Mentees/Advisees

2007-present Lingling Kong, Ph.D. Postdoctoral fellow (2007-2012), Research Associate (2012-present). Multiple meeting presentations. Shared publications: #14, 16, 18, 19, 21, 22, 25, 28, 40.
2007-2010 Marta Bosch-Marcé, Ph.D. Current position: ORISE Fellow, Section of Cell Biology, Laboratory of Cellular Hematology, DH/OBRR/CBER, US Food and Drug Administration. Shared publications: #16, 22.
2011-present Jeremy Sullivan, Ph.D. Postdoctoral fellow (2011-2012), Research Associate (2012-present). Shared publications: #32, 37, Editorial #2.
2012 Tara Martinez, Ph.D. Current position: Biotech Patent Examiner at the US Patent and Trademark Office. Shared publications: #19, 22, 25, 28, 31.
2012-2017 Constantin d'Ydewalle, Ph.D. Awards/Grants: 1) Fight SMA Foundation and Gwendolyn Strong Foundation Emerging Investigator Award. 2) MDA Career Development Award (08/01/2015-07/31/2018). Shared publications: #29, 40 Review #8. Current position: Senior scientist Johnson and Johnson, Belgium.
2013-2015 Sonja Scholz, M.D., Ph.D. Neurology resident. Junior mentor for R25 proposal.
2013-2015 Lindsey Hayes, M.D., Ph.D. Neurology resident. Junior mentor for R25 proposal. Pharmacodynamic biomarker development for antisense oligonucleotide therapy in ALS.
2014-present Payam Mohassel, M.D. Neurogenetics Fellow, NIH. Postdoctoral fellowship mentorship committee.
2014-present Peggy Lazerow, M.D. Neurology resident. Research mentor for residency research project: "SMN levels in CSF of children."
2016-present Brett McCray, M.D., Ph.D. Neuromuscular/CMT Fellow. Awarded Inherited Neuropathy Consortium Fellowship (8/16-7/17), Johns Hopkins Clinician-Scientist Award, K23 NIH Career Development Award.
5/2019-present Bhavya Ravi, PhD, Postdoctoral fellow.

Thesis Committees

2007 Katie Provost: Johns Hopkins Department of Molecular Microbiology and Immunology Graduate Student Preliminary Oral Exam Committee
2007 Danielle Large: Johns Hopkins Department of Cellular and Molecular Medicine Graduate Student Preliminary Oral Exam Committee
2009-2013 Caitlin Engelhard: Department of Neuroscience Graduate Thesis Committee.
2011-2014 Saniya Fayzullina: Johns Hopkins Department of Pathology/ Pathobiology Graduate Program Thesis Committee.
2001-2014 Elizabeth McDonald: Johns Hopkins McKusick-Nathans Institute of Genetic Medicine/ Human Genetics Graduate Program Thesis Committee.
2011 Michael Ayairs: Johns Hopkins Pathobiology Graduate Program Preliminary Oral Examination Committee
2011 Jennifer Albertz: Johns Hopkins Cellular and Molecular Medicine Graduate Student Preliminary Oral Exam Committee

2012-2016 Clint Cave: Johns Hopkins Department of Neuroscience Graduate Student Thesis Committee
2012 Melissa Bowerman: University of Ottawa Graduate Program in Cellular and Molecular Medicine External Examiner Thesis Defense Committee.
2012 Alisa Mo: Johns Hopkins Department of Neuroscience Graduate Student Oral Examination Committee.
10-4-2012 Rosie Jiang Johns Hopkins Pathobiology Graduate Program Oral Examination Committee.
11-6-12 Amy Anderson Johns Hopkins Cellular and Molecular Medicine Oral Examination Committee.
2013-2015 Kyle Metz Johns Hopkins Molecular Medicine and Immunology Graduate Program Thesis Committee.
2013-15 Bipasha Mukherjee-Clavin MD/PhD thesis committee chair.
11-6-13 Michael Topper Cellular and Molecular Medicine Graduate Student Preliminary Oral Exam Committee.
11-19-13 Leslie Kirby Cellular and Molecular Medicine Graduate Student Preliminary Oral Exam Committee.
2014-2017 Jonathan Ling, Johns Hopkins Pathobiology Graduate Program Thesis Committee.
11-12-14 Jackie Douglas, Johns Hopkins Cellular and Molecular Medicine Oral Examination Committee.
11-14-14 Michael Hwang, Johns Hopkins Cellular and Molecular Medicine Oral Examination Committee.
11-20-14 Deepthi Ashok, Johns Hopkins Cellular and Molecular Medicine Oral Examination Committee.
11-21-14 Justin Lee, Columbia University Department of Neuroscience Graduate Program External Examiner Thesis Defense Committee.
2015-pres Tim Wang, Johns Hopkins School of Public Health Molecular Medicine and Immunology Graduate Program Thesis Committee.
2015-pres Brian Woolums, Johns Hopkins Department of Pharmacology Graduate Student Program Thesis Committee.
10-20-15 Drew Bell, Johns Hopkins Pathobiology Graduate Student Program Oral Examination Committee.
5-10-16 Daniel Giovinazzo, Johns Hopkins Department of Neuroscience Graduate Student Program Oral Examination Committee.
11-1-16 Yazmin Gonzalez, Johns Hopkins Cellular and Molecular Medicine Oral Examination Committee.
11-9-16 Kyla Britson, Johns Hopkins Cellular and Molecular Medicine Oral Examination Committee.
11-10-16 Alexandra Murray, Johns Hopkins Cellular and Molecular Medicine Oral Examination Committee.
2016-2018 Aneesh Donde, Johns Hopkins Pathobiology Graduate Student Program Thesis Committee, Chair.
2018 Lawrence Van Helleputte, University of Leuven PhD Thesis Committee Member, Leuven Belgium.
10-30-18 Jenne Glatzer, Johns Hopkins Cellular and Molecular Medicine Oral Examination Committee.
11-16-18 Sarah Nathan, Johns Hopkins Cellular and Molecular Medicine Oral Examination Committee.

Educational Program Building/Leadership
None

Educational Demonstration Activities to External Audiences
None

RESEARCH ACTIVITIES

Research Focus

- Inherited peripheral nerve and motor neuron diseases: clinical and genetic characterization, dissection of molecular and cellular mechanisms, and identification and validation of therapeutic strategies.

Research Program Building

2006-present Co-Founder and Co-Director of the Johns Hopkins translational research program in spinal muscular atrophy. Bench to bedside effort focused on disease mechanisms, preclinical assessment of novel therapeutics, formation of SMA tissue repository, and clinical trials.
2006-present Co-Founder and co-Director of bench to bedside research program focused on identifying novel genetic causes and mechanisms of Charcot Marie Tooth disease.
10/11-present A principal investigator in The Michael S. and Karen G. Ansari ALS Center for Cell Therapy and Regeneration Research at Johns Hopkins.
1/12-present Site Co-lead Investigator for The Inherited Neuropathy Consortium (INC)/Rare Disease Clinical Research Consortium (RDCRC)-an international network of clinical investigators dedicated to developing the infrastructure necessary to evaluate therapies for patients with heritable peripheral neuropathies

Research Demonstration Activities

4/15/15 & 4/13/16	"Research in neurology and neuroscience." Practicum for Johns Hopkins School of Medicine medical students to experience laboratory based neuroscience research.

Inventions, Patents, Copyrights

4/15	Modulation of SMN expression, Rigo F, C. CF Bennett, van Outyrve d'Ydewalle C, Sumner CJ. Assignee Name and Address: ISIS Pharmaceuticals, Carlsbad, CA, Provisional patent submitted.
6/16	Combinations for the Modulation of SMN expression, Rigo F, C. CF Bennett, van Outyrve d'Ydewalle C, Sumner CJ. Assignee Name and Address: ISIS Pharmaceuticals, Carlsbad, CA, Provisional patent submitted

Technology Transfer Activities
None

SYSTEM INNOVATION AND QUALITY IMPROVEMENT ACTIVITIES
None

ORGANIZATIONAL ACTIVITIES

Institutional Administrative Appointments

2007-2014	Department of Neurology Residency Selection Committee
2010-present	Johns Hopkins School of Medicine M.D./Ph.D. Student Selection Committee
2010-present	Departments of Neurology and Neurosurgery Appointments and Promotions Committee
2010-present	Member, Pathobiology Graduate Student Program
2010-present	Member, Neuroscience Graduate Student Program
2010-2011	Department of Neurology Fellowship Director
2011-present	Member, Cellular and Molecular Medicine Graduate Student Program
2011-2014	Chair, Department of Neurology Residency Selection Committee
2014-2015	Member, Johns Hopkins School of Medicine Research Council
2014-present	Member, Department of Neurology Executive Council

Editorial Activities

Editorial Board Appointments

2013-2017	Associate Editor, *Experimental Neurology Journal*
2014-present	Editorial Advisory Board Member, *Annals of Clinical and Translational Neurology*
2016-present	Editorial Advisory Board Member, *Neuromuscular Disorders*
2015-2016	Book editor, Spinal Muscular Atrophy: Disease Mechanisms and Therapy. Editors: **Sumner CJ**, Paushkin S, and Ko CP. Elsevier/Academic Press. 2016.
2017-present	Editorial Advisory Board Member, *Experimental Neurology Journal*
2017-present	Associate Editor, *Journal of Clinical Investigation*

Journal Peer Review Activities
Acta Neuropathologica
American Journal of Medical Genetics
Annals of Neurology
Annals of Clinical and Translational Neurology
Brain
Cell Stem Cell
eLife
Embo Molecular Medicine
European Journal of Human Genetics

European Journal of Neurology
Experimental Neurology
Experimental Physiology
Human Molecular Genetics
Journal of Cell Biology
Journal of Clinical Investigation
Journal of Neurological Sciences
Journal of Neuromuscular Diseases
Journal of Neuropathology and Experimental Neurology
Journal of Neuroscience
Muscle and Nerve
Nature Communications
Nature Genetics
Nature Methods
Nature Neuroscience
Nature Reviews Neuroscience
Neurology
Neurology Genetics
Neuromuscular Disorders
Neuron
Neuroscience Letters
Neurotherapeutics
PLOS Genetics
PLOS One
Proceedings of the National Academy of Sciences
Science
Science Translational Medicine

Other Peer Review Activities
None

Advisory Committee, Review Groups/Study Sections

2004	NIH Clinical Trials in Spinal Muscular Atrophy Workshop, Bethesda MD
2005-2007	Spinal Muscular Atrophy Foundation, Scientific Advisory Board
2006-present	Spinal Muscular Atrophy International Coordinating Committee and Biomarker Subcommittee
2007	American Academy of Neurology Annual Meeting Peripheral Neuropathy Abstract Review Committee
2007-2008	Association Française Contre Les Myopathies (ad hoc reviewer)
2007-2008	Telethon Italy (ad hoc reviewer)
2007-2008	American Neurological Association Annual Meeting Career Development Committee
2008	Welcome Trust (ad hoc reviewer)
2008-2009	NINDS SMA Project RFP Review Committee
2009-2013	NINDS SMA Project Advisory Committee
2010-present	Cure Spinal Muscular Atrophy Translational Research Advisory Council
2010	NIH Skeletal muscle and exercise physiology study section (ad hoc reviewer)
2010	Action Medical Research England and Wales (ad hoc reviewer)
2010	American Academy of Neurology Annual Meeting Peripheral Neuropathy Abstract Review Committee
2010	Muscular Dystrophy Association (ad hoc reviewer)
2010	The Johns Hopkins Musculoskeletal Pilot and Feasibility Program Grant Review Committee
2010	NIH Therapy Development Workshop in SMA, Working Group 2: Animal Models in SMA and Their Use
2011-present	The Robert Packard Center for ALS Research at Johns Hopkins Operating Committee Member
2011-present	Ortho McNeil Janssen Brain Science Institute Joint Steering Committee Member
2011	Johns Hopkins Institute of Translational and Clinical Research ATIP Grant Review
2011	Johns Hopkins Department of Neurology K and first time R01 grant review

2011	NINDS/NIH Rare Disease Clinical Research Network (U54) Review Committee
2011	American Academy of Neurology Annual Meeting Peripheral Neuropathy Abstract Review Committee
2011	American Academy of Neurology Annual Meeting Anterior Horn Cell Abstract Review Committee
2011	NINDS neuroNEXT SMA Special Emphasis Panel (G54) Review Committee
2012-2016	Neurofibromatosis Therapeutic Acceleration Program (NTAP) Scientific Advisory Committee.
2012	Association Française Contre Les Myopathies (ad hoc reviewer) Trampoline Grant
2012	NIH/Chronic Dysfunction and Integrative Neurodegeneration (CDIN) Study Section (as hoc reviewer)
2012	Prinses Beatrix Fonds Netherlands (ad hoc reviewer).
2012-13	Johns Hopkins OWISM Leadership Program Applicant Review Committee.
2012-2014	FSMA/Calibr Joint Steering Committee
2013-present	SMA Foundation Muscle Advisory Board
2013	Muscular Dystrophy Association (ad hoc reviewer)
2013	Telethon Italy (ad hoc reviewer)
2013	ANA Annual Meeting Abstract Review Committee
2013-2019	NIH/Chronic Dysfunction and Integrative Neurodegeneration (CDIN) Study Section Member.
2014	SMA Europe (ad Hoc grant reviewer)
2014	Foundation Thierry Latran (ad Hoc grant reviewer)
2014, 2015	ANA Annual Meeting Abstract Review Committee
2014	Johns Hopkins Department of Neurology K and first time R01 grant review
7/2014	Johns Hopkins School of Medicine SYNERGY Grant Review Committee
7/15/2014	ALS iPS Personalized Therapeutics Discovery and Big Data Meeting, Packard Center, Johns Hopkins School of Medicine, Baltimore, MD
11/14-present	Neurobiology of Disease Workshop Committee at the Society for Neuroscience
2015-2017	Amyotrophic Lateral Sclerosis Association Grant Review Committee
2015	Reviewer research program at University of Leuven, Belgium
2015	Neurological Foundation of New Zealand, ad hoc grant reviewer
10/15/15	Cure SMA Board Meeting, Chicago, IL
10/15	Swiss Foundation of Muscle Disease, ad hoc grant reviewer
11/13/15	Medical Advisory Board Avexis, Chicago, IL
2015-present	Medicine and Science Committee, Cure SMA Board Advisory Committee
1/28-30/16	Inherited Neuropathies Consortium-Rare Disease Clinical Research Consortium Planning Meeting
4/22/16	ALS Association Grant Review Committee
2016-present	Muscular Dystrophy Association Scientific Advisory Board.
12/2016	AFM-TELETHON France, Ad hoc reviewer.
2016	ZRG1 MOSS R-02 NIH Skeletal muscle structure and function special emphasis panel reviewer.
12/2-12/3/16	Biogen Nuture Extension Study Advisory Board.
2/17	European Science Foundation ad hoc grant reviewer
4/21/17	Medical Advisory Board, Avexis, Boston MA.
5/20/17	Muscular Dystrophy Association MVP program ad hoc grant reviewer.
7/1/17	Roche Olexisome Advisory Board, Orlando, FL.
7/2017	Peripheral Nerve Society Annual Meeting Awards Committee
2/22/18	Pfizer Scientific Advisory Committee Meeting
6/13/18	Biogen SMA in Adults Medical Advisory Board, Tx.
6/16/18	Cure SMA Board Meeting, Chicago, Il.
7/21-22/18	Avexis Medical Advisory Board Meeting, Chicago, Il
9/22/18	Genentech Medical Advisory Meeting. Boston, MA.
10/15-128/18	Roche iAdvise Biomarker Advisory Panel.
2018-pres	Charcot Marie Tooth Research Foundation Scientific Advisory Committee
2019	Johns Hopkins University Young Investigator Day Awards Committee
2019-pres	Charcot Marie Tooth Research Foundation Scientific Advisory Board Member
2019-pres	Cure SMA Scientific Advisory Board
2018-pres	Spinraza Individual Patient Humanitarian Access Program Medical Expert Committee (MEC)
2019	Board Member, Peripheral Nerve Society.

Professional Societies

1999-present	American Academy of Neurology, member.
2006-present	Society of Neuroscience, member.
2010-present	Peripheral Nerve Society, member.
2011-present	American Neurological Association, member.
2017-present	World Muscle Society, member.
2017-present	American Society for Clinical Investigation

Conference Organizer

2010	Peripheral Nerve Society 2011 Meeting Organizational Committee
2012-13	Muscular Dystrophy Association Annual Scientific Meeting Organizing Committee
2013	American Neurological Association Interactive Lunch Workshop Task Force
2013-2016	American Neurological Association Meeting Scientific Program Advisory Committee
2013-2014	Meeting Organizer, New Directions in Biology of Disease of Skeletal Muscle Conference.
11/14-3/15	Muscular Dystrophy Association Annual Scientific Meeting Organizing Committee
2016-2017	Peripheral Nerve Society 2017 Meeting Scientific Program Committee
2018-2019	Peripheral Nerve Society 2019 Meeting Scientific Program Committee
2019-20	Peripheral Nerve Society 2020 Meeting Scientific Program Co-Chair

Session Chair

4/14/05	Session Chair, Peripheral Neuropathy, American Academy of Neurology Annual Meeting
4/17/08	Session Chair, Acquired and Genetic Neuropathies, American Academy of Neurology Annual Meeting
4/14/10	Session Chair, Anterior horn cell: Spinal muscular atrophy, American Academy of Neurology Annual Meeting
6/29/11	Peripheral Nerve Society Inherited Neuropathies Session Chair
6/30/11	Charcot Marie Tooth Association Neurobiology Session Chair
4/25/12	Session Chair, Anterior horn cell: Genetics and Molecular Mechanisms, American Academy of Neurology.
4/23/13	Muscular Dystrophy Association Annual Scientific Meeting Session Chair "Preclinical therapeutic studies in disease model mice"
6/13/13	Session Moderator: "Enhancing the Predictive Ability of Preclinical Drug Studies," Families of Spinal Muscular Atrophy Annual Research Meeting.
6/20/14	Moderator, Virology and Immunology Section II, Three Decades of Research in PML and Disorders affecting the CNS, NIH, Bethesda, MD.
10/12/14	Session Organizer and Co-Chair Plenary Symposium: "Novel Concepts in Pain Generation and Management," American Neurological Association Annual Meeting, Baltimore, MD.
3/13/15	Session Organizer and Co-Chair: Motor neuron/nerve/NMJ disorders-Animal models, preclinical studies, biomarkers, 2015 Muscular Dystrophy Association Scientific Meeting.
7/1/15	Session Co-Chair: Genetic Neuropathy, Peripheral Nerve Society Meeting, Quebec, Cananda.
9/28/15	Session Organizer and Co-Chair: The life and death of axons in neurological disease symposium, American Neurological Association Meeting, Chicago, IL.
9/8/16	Session Chair: "Pathomechanisms of CMT," 6th International Charcot-Marie-Tooth and Related Neurology Consortium (CMTR) Meeting, Venice, Italy.
7/9/17	Session Chair: "Hot Topics Symposium," Peripheral Nerve Society Meeting, Sitges, Spain.
7/21/18	Session Chair: "Inherited Neuropathies," Peripheral Nerve Society Meeting, Baltimore, MD.

Consultantships

2009-2010	Genzyme/Isis Medical Advisory Board
2009-present	SAIC Consultant
2011	Bristol-Myers Squibb Company Consultant
2012	Repligen Incorporated Consultant
2012	Biogen Idec Consultant
2012	BioLife

2013-2015, 2018 Pfizer
2013-present Avexis
2014, 2016,
2018 Biogen Idec
2018 Cytokinetics
2014-present IONIS (ISIS) Pharmaceuticals
2014-present Roche
2014-present PTC Therapeutics
2019 Proneurotech

RECOGNITION

Awards/Honors

1991 Special Award for an Undergraduate Thesis of Unusual Originality, Princeton University
1994-96 The Howard Hughes Institute Fellowship for Medical Studies Award, Howard Hughes Medical Institute
1996 The Dr. O. H. Pepper Prize, University of Pennsylvania School of Medicine
2000 The Teaching Award for Cherished House staff, University of California, San Francisco Class of 2000
2001 Award for Excellence in Teaching, Johns Hopkins Department of Neurology
Class of 2001
2002-04 NINDS Competitive Fellowship Award, NIH
2004-07 Spinal Muscular Atrophy Foundation/American Academy of Neurology Young Investigator Award in Spinal Muscular Atrophy
2008 NINDS Group Merit Award
2011 Fellow, American Neurological Association
2011-2012 Johns Hopkins School of Medicine Leadership Program for Women Faculty
2015 Johns Hopkins University Catalyst Award
2017 Osler Attending Preceptor Program
2018 American Society for Clinical Investigation Member
2018 Richard P. Bunge Prize, Peripheral Nerve Society Annual Meeting
2018 Interurban Clinical Club Member
2019 AAN Annual Meeting Abstract of Distinction
2019 University of California San Francisco Neurology Bright Visiting Teaching Scholar

Invited Talks/Panels

<u>JHMI/Regional</u>

4/10/02 "Dopamine responsive dystonia," NINDS Grand Rounds, Bethesda, MD.
10/20/03 "Histone deacetylase inhibitors as treatment for spinal muscular atrophy," NHGRI Fellows Seminar Bethesda, MD.
11/15/03 "Histone deacetylase inhibitors as treatment for spinal muscular atrophy," Joint NIH/Johns Hopkins Neurogenetics Seminar, Baltimore, MD.
9/30/04 "Drug candidates for SMA," NINDS Workshop on Clinical Trials in Spinal Muscular Atrophy, Bethesda, MD.
6/08/04 "Clinical and Molecular Features of Spinal Muscular Atrophy," NINDS Grand Rounds, Bethesda, MD.
3/08/05 "Amyotrophic lateral sclerosis," Demystifying Medicine NIH Lecture Series Bethesda, MD.
6/10/05 "Clinical and pathophysiological features of distal spinal and bulbar muscular atrophy with vocal fold paresis," Johns Hopkins University Neuroscience, Baltimore, MD.
10/11/05 "Molecular therapeutic targets for inherited motor neuron disease," Johns Hopkins University Department of Neurology, Baltimore, MD.
5/23/07 "Therapeutics Development for Spinal Muscular Atrophy," Johns Hopkins Department of Neurology Young Investigators Seminar Series, Baltimore, MD.
10/15/07 "Spinal muscular atrophy: molecular pathogenesis and therapeutics development," Howard Hughes Medical Institute Research Scholars Program Lecture series, Bethesda, MD.

11/3/08 "Translating genetics into treatment for the motor neuron disease spinal muscular atrophy," Johns Hopkins University Clinical Neuroscience Seminar, Baltimore, MD.

9/30/09 "Spinal muscular atrophy: Molecular pathogenesis and therapeutics development." Molecular Pathology Seminar Series. Johns Hopkins Department of Pathology. Baltimore, MD.

10/10/09 "Spinal muscular atrophy: Molecular pathogenesis and therapeutics development." Johns Hopkins Department of Neurology 40th Anniversary. Baltimore, MD.

2/18/10 "Neuromuscular disease in 2010," Johns Hopkins Department of Neurology Grand Rounds, Baltimore, MD.

12/15/10 "Spinal Muscular Atrophies and Regeneration Strategies," Brain Science Institute Nerve Regeneration Interest Group, Johns Hopkins University, Baltimore, MD.

10/13/10 "Motor neurons, genes, and synapses," Johns Hopkins Brain Sciences Institute Brain Night Seminar series.

1/8/11 "Understanding disruptions of neural circuitry in inherited motor nerve diseases," The Friends of the Axon, the Schwann Cell, and Jack Griffin: Celebration of Peripheral Nerve at Johns Hopkins, Baltimore, MD.

9/9/11 "Disruptions in neural circuitry in spinal muscular atrophies." Johns Hopkins Department of Neuroscience Retreat. St. Michael's, MD.

11/5/11 "Disruptions in neural circuitry in spinal muscular atrophy." Johns Hopkins MSTP/M.D. Ph.D. Annual Retreat, Hershey, PA.

11/27/11 "Disruptions of neural connectivity in spinal muscular atrophies." Johns Hopkins Department of Neurology Clinical Neuroscience Seminar, Baltimore, MD.

5/24/12 "Mechanisms of inherited motor neuron diseases," Johns Hopkins-NINDS Joint Research Seminar, Baltimore, MD.

6/16/12 "Reversing synaptic dysfunction in motor neuron disease." Scientific symposium celebrating Guy McKhann, MD and the Induction of the Inaugural McKhann Scholar. Baltimore, MD.

12/13/12 "Is spinal muscular atrophy now a treatable disease?" Johns Hopkins Department of Neurology Grand Rounds, Baltimore, MD.

1/8/13 "Pathogenesis of inherited motor neuron and peripheral nerve disease," Johns Hopkins Department of Neuroscience Lab Lunch Seminar Series, Baltimore, MD.

11/19/15 "Therapeutics development for spinal muscular atrophy: forging a path for other neurogenetic diseases." Johns Hopkins University School of Medicine Neurology Grand Rounds, Baltimore, MD.

12/9/15 "An update on Charcot Marie Tooth disease" Johns Hopkins University School of Medicine Pediatric Neurology Grand Rounds, Baltimore, MD.

10/28/16 "Inherited motor neuron and peripheral nerve diseases," Johns Hopkins Neuromuscular division retreat, Annapolis, MD.

11/30/17 "A remarkable journey: SMA therapeutics development," Johns Hopkins Department of Neurology Grand Rounds, Baltimore, MD.

12/6/18 "Spinal muscular atrophy: a breakthrough in translational neurodegeneration research," Partnering Toward Discovery Seminar Series, Johns Hopkins University School of Medicine Seminar Series, Baltimore MD.

National

10/21/02 "Cell culture models of ALS and SMA," ALS Association Young Investigator Workshop, Lafayette Hill, PA.

3/9/04 "Histone deacetylase inhibitors as treatment for SMA," SMA Foundation: What is the Molecular Basis of Neuron Loss? Cold Spring Harbor, NY.

12/15/04 "Molecular therapeutic targets for inherited motor neuron disease" University of California Los Angeles Department of Neurology, Los Angeles, CA.

2/2/05 "Therapeutics development for SMA," Emory University Center for Neurodegenerative Diseases, Atlanta, GA.

3/10/05 "Molecular therapeutic targets for inherited motor neuron disease," Jefferson University Department of Neurology, Philadelphia, PA.

5/10/05	"Molecular therapeutic targets for inherited motor neuron disease," Columbia University Pediatric Neurology Grand Rounds, New York City, NY.
9/01/05	"Molecular therapeutic targets for inherited motor neuron disease," University of California, San Francisco School of Medicine Neurology Grand Rounds, San Francisco, CA.
12/02/05	"Spinal muscular atrophies: progress toward treatment," Emory University Neurology Grand Rounds, Atlanta, GA.
9/19/06	"Histone deacetylase inhibitors in SMA" Spinal Muscular Atrophy Foundation, Banbury, NY.
10/18/06	"Molecular mechanisms of SMA," Neurobiology of Disease in Children Symposium, 2006 Child Neurology Society Meeting, Pittsburgh, PA.
10/24/06	"Histone deacetylase inhibitors for spinal muscular atrophy," Spinal Muscular Atrophy Project, Kent Island, MD.
5/7/07	"Trichostatin A and SMA," SMA Project Steering Committee Meeting, Boston, MA.
9/21/07	"Therapeutics development for spinal muscular atrophy," ALS Packard Center Board of Governors Meeting, New York, NY.
9/29/07	"Spinal muscular atrophy biology and therapeutics development: an overview," Spinal Muscular Atrophy International Coordinating Committee Summit on Drug Development, Speaker panel discussion "Prioritizing Preclinical Compounds" Bethesda, MD.
10/30/07	"Spinal muscular atrophy: Natural history and therapeutics development," Genzyme Corporation Framingham, MA.
1/25/08	"SMA therapeutics development: From the bedside to the bench and back again," PTC Therapeutics South Plainfield, NJ.
3/01/08	"HDAC inhibitors in SMA mice," Spinal Muscular Atrophy Foundation, Half Moon Bay, CA.
4/29/08	"SMA therapeutics development: From the bedside to the bench and back again," NINDS Grand Rounds Bethesda, MD.
10/21/08	"Immaturity of the motor unit and the effect of histone deacetylase inhibition in SMA," Spinal Muscular Atrophy Foundation Banbury, NY.
10/28/08	"Synaptic dysfunction in SMA" NINDS Grand Rounds, NIH, Bethesda, MD.
3/21/09	"Histone deacetylase inhibitors in spinal muscular atrophy," Spinal Muscular Atrophy Foundation Scientific Advisory Board Meeting, Half Moon Bay, CA.
5/8/09	"Spinal muscular atrophy" Louisiana State University Health Sciences Center Neuromuscular Symposium New Orleans, LA.
10/9/09	"Histone deacetylase inhibitors for the treatment of spinal muscular atrophy," Medical Seminar Series, Burke Medical Research Institute, White Plains, NY.
2/20/10	"Translating genetics into treatment for spinal muscular atrophy," Visiting Professor, Mayo Clinic Department of Neurology Grand Rounds, Rochester, NY.
4/19/10	"Developing treatment for spinal muscular atrophy," Translational Science Research Seminar, Nemours Alfred I. DuPont Hospital for Children. Wilmington, DE.
6/10/10	"Synaptic abnormalities in SMA mice," Electrophysiology Seminar Series, Wright State University, Dayton, Ohio.
10/28/10	"Animal models of SMA and their appropriate use in therapy development", NIH Therapy Development Workshop in SMA, Bethesda, MD.
11/11/10	"TRPV4 and CMT2C/dSMA," Charcot-Marie-Tooth Association Workshop: Defining therapeutic approaches to CMT2, San Diego, CA.
10/28/11	"Genetics of Spinal Muscular Atrophies." Northeast ALS Consortium Annual Meeting Genetics Symposium. Clearwater, FL.
11/14/11	"Disruptions of motor neurons, their axons, and target muscles in human spinal muscular atrophy," SMA Satellite Meeting: Pretzels and endplates: Motor neuron pathology and the role of SMN in motor neuron development. Society for Neuroscience Meeting, Washington DC.
7/11/12	"Pathogenic Aspects of Spinal Muscular Atrophy." Biogen Idec. Boston, MA.
10/9/12	"The role of the SMA determining gene SMN in synaptic and axonal maturation and maintenance: Molecular mechanism of axon degeneration meeting, Howard Hughes Medical Institute, Janelia Farm Research Campus, VA.
11/7/12	"Motor neurons, genes, and synaptic connectivity: Identifying therapeutics for inherited motor neuron diseases." Nemours/A.I. DuPont Hospital Research Seminar Series. Wilmingon, DE.

1/31/13 "Defining cellular consequences of SMN deficiency in SMA mice and humans." ISIS pharmaceuticals, San Diego, CA.

2/14/13 "HDAC inhibitors and RG3039: Review of use in spinal muscular atrophy." Northeast ALS consortium Round Table, Boston, MA.

4/23/13 "Spinal muscular atrophy mouse models" Muscular Dystrophy Association Research Meeting, Washington, DC.

5/9/13 "Disruptions of neural connectivity in inherited motor neuron diseases." Jackson Laboratories Scientific Seminar Series, Bar Harbor, ME.

7/20/13 "Is muscle an important target for SMA therapeutics." Muscle Advisory Board Meeting, Spinal Muscular Atrophy Foundation, NYC, NY.

10/4/13 "The roles of SMA genes in axonal development, connectivity, and maintenance." University of Pennsylvania School of Medicine Institute of Regenerative Medicine Neuroscience Seminar Series, Philadedelphia, PA.

10/16/13 "Developing treatment for spinal muscular atrophy." Yale University School of Medicine Department of Neurology Grand Rounds, New Haven, CT.

5/8/14 "Pathology of severe SMA in humans." Roche/SMAF/PTC Academic Collaborators Meeting, New York, NY.

6/10/14 "Developing treatment for spinal muscular atrophy." Shriners Hospital Pediatric Research Center Temple University, Philadelphia PA.

7/18/14 "The ongoing journey of therapeutics development for spinal muscular atrophy." University of California San Francisco Department of Neurology Memory and Aging Center Grand Rounds, UCSF, San Franscisco CA.

10/7/14 "The ongoing journey of therapeutics development for spinal muscular atrophy." Massachusetts General Hospital Department of Neurology Neuroscience Seminar, Boston MA

12/10/14 "The interplay of motor neuron development and degeneration in SMA" Department of Pathology, Anatomy and Cell Biology Grand Rounds, Thomas Jefferson University, Philadelphia PA.

1/20/15 "Developing treatment for spinal muscular atrophy" Department of Neurology Pediatric Neurology Grand Rounds, NYU School of Medicine, New York City, New York.

4/02/15 "Dissecting mechanisms of inherited motor neuron disease" Carver College of Medicine Department of Biochemistry Research Seminar, Grand Rapids, Iowa.

6/10/15 "Are we on the verge of treatment for spinal muscular atrophy?" University of California Los Angeles School of Medicine Department of Neurology Grand Rounds, Los Angeles, CA.

1/20/16 "Therapeutics development for spinal muscular atrophy: forging a path for other neurogenetic diseases." University of Maryland Neurology Grand Rounds, Baltimore, MD.

3/30/16 "Therapeutics development for spinal muscular atrophy: forging a path for other neurogenetic diseases." University of Michigan Neurology Grand Rounds, Ann Arbor, Michigan.

4/29/16 "Pathomechanisms of inherited motor neuron diseases." RANA Therapeutics, Boston, MA.

5/6/16 "Muscle as a therapeutic target in SMA." SMA Foundation, New York City, NY.

10/26/16 "What are the therapeutic opportunities in spinal muscular atrophy." Cytokinetics, South San Francisco, CA.

11/8/16 "Successes and challenges in developing therapeutics for spinal muscular atrophy." Wake Forest Department of Neurology Grand Rounds, Winston Salem, NC.

12/1/16 "Successes and challenges in developing therapeutics for spinal muscular atrophy." Department of Neurology Grand Rounds University of Buffalo, Buffalo, NY.

4/24/17 "Nusinersen in Infants Diagnosed with Spinal Muscular Atrophy (SMA): Study Design and Initial Interim Efficacy and Safety Findings from the Phase 3 International ENDEAR Study" in Contemporary Clinical Issues Plenary Symposium, American Academy of Neurology Annual Meeting, Boston MA.

5/10/17 "Developing therapeutics for inherited motor neuron disease." The Janice Massey Lectureship Department of Neurology Grand Rounds, Duke University, Durham, NC.

6/16/17 "The successes and challenges of therapeutics development for spinal muscular atrophy," Keynote Lecture 3rd Annual Shriner's/Temple Symposium on Neural Repair, Philadelphia, PA.

10/23/17 "Impaired motor neuron development precedes degeneration in SMA," Cell Biology of ALS: Emerging themes from human genetics. Banbury, NY.

11/3/17 "Can lncRNAs be targeted in SMA?" Keynote speaker, Annual Noncoding RNA Meeting, University of Alabama, Birmingham, AL.

12/7/17 "Impaired motor neuron development precedes degeneration in SMA," Neuroscience Department Seminar Series, Case Western Reserve University, Cleveland OH.

12/8/17 "Optimizing therapeutics for spinal muscular atrophy," Department of Neurology, University Hospitals, Cleveland OH.

2/1/18 "Is SMA a developmental disease," Human Gene Therapy Center, Nationwide Children's Hospital, Columbus, OH.

2/27/18 "How do we optimize treatment of SMA?" National Institute of Neurological Disorders and Stroke Grand Rounds, Bethesda, MD.

6/14/18 "Is there a developmental component to SMA?" Cure SMA Annual International Meeting, Dallas, TX.

6/28/18 "Impaired motor axon development in motor neuron disease," Cell Biology of the Neuron Gordon Conference, Waterville Valley, NH.

11/30/18 "Optimizing SMA Therapeutics," PTC Therapeutics, South Plainfield, NJ.

1/24/19 "Spinal muscular atrophy: a breakthrough in translational neurodegeneration research," University of Washington Grand Rounds, Seattle, WA.

2/27/19 "Timing, drug delivery, biomarkers: optimizing therapeutic efficacy in SMA," Biogen, Boston, MA.

4/15/19 "Therapeutics development for axonal CMT," MDA Clinical and Scientific Meeting, Orlando, FL.

5/24/19 "Optimizing treatments for spinal muscular atrophy," Neurology Grand Rounds Stanford University, Palo Alto, CA.

6/29/19 "Developmental and temporal needs of SMN protein," Cure SMA Annual Meeting, Anaheim CA.

7/23/19 "Therapeutic targeting of RNA in spinal muscular atrophy," Amyotrophic lateral sclerosis and other motor neuron diseases Gordon Conference, Snow Mountain, VT.

9/13/19 "Opportunities and challenges of SMA therapeutics," Hershey Department of Neurology Grand Rounds, Hershey, PA.

9/24/19 "Gene targeting therapy for SMA: a breakthrough for neurodegenerative disease," Northwestern Department of Neurology Grand Rounds, Chicago, IL.

<u>International</u>

11/17/09 "Preclinical models-When to use animal models relative to human trials," Animal Models Assessment Panel Discussion. Bringing Down the Barriers-Translational Medicine in Inherited Neuromuscular Diseases. Treat NMD/NIH International Conference, Brussels, Belgium.

3/15/10 "Synaptic dysfunction in SMA" The Ottawa Conference on New Directions in Skeletal Muscle Biology and Disease of Muscle, Ottawa, Canada.

3/29/10 "Influence of housing, feeding, and handling conditions on SMA mice," Creating Standard Operating Procedures for SMA mice, Treat NMD Neuromuscular Network Conference, Zurich, Switzerland.

4/3/12 "Disruption of synaptic connectivity in spinal muscular atrophy." University of Ottawa Research Seminar, Ottawa, Canada.

11/7/14 "Role of SMN expression in muscle in SMA animal models and humans." Amsterdam, Netherlands. European NeuroMuscular Centre SMA Workshop.

9/26/15 "Targeting a long noncoding RNA as a novel strategy to induce SMN early in SMA pathogenesis." 3rd Ottawa International Conference on Neuromuscular Biology, Disease, and Therapy, Ottawa, Canada.

10/7/16 "From SMN1 disruption to SMA," Spinal muscular atrophy: from genotype to phenotype satellite symposium at 21st International Congress of the World Muscle Society, Granada, Spain.

8/9/17 "Developing therapeutics for inherited motor neuron diseases," The Australian and New Zealand Child Neurology Society Meeting, Brisbane, Australia.

9/1/17 "Future directions in neuromuscular disease research." 4th Ottawa International Conference on Neuromuscular Biology, Disease, and Therapy, Ottawa, Canada.

1/26/18 "SMA as a systemic disease," International Scientific Congress on Spinal Muscular Atrophy, Krakow Poland.

3/28/18 "SMA therapeutics development: bench to bedside and back again," VIB/KU Leuven Research Seminar, Leuven Belgium.

9/24/18 "Failed perinatal sorting of motor axons is associated with impaired myofiber growth in SMA," International conference of muscle wasting: Molecular mechanisms of muscle wasting during aging and disease. Ascona, Switzerland.

12/12/18 "Optimizing therapeutics for SMA," Roche Seminar, Roche, Basel Switzerland.

3/13/19 "Impaired motor axon development dictates the temporal dependency of therapeutic effects in SMA," Molecular and cellular mechanisms of axon degeneration, 7th international meeting, Loch Lomond, Scotland.

6/22/19 "Mechanisms of TRPV4 induced peripheral nerve disease," Peripheral Nerve Society Meeting, Genoa, Italy.

OTHER PROFESSIONAL ACCOMPLISHMENTS

Posters

1. **Sumner CJ**, Avila A, Kernochan LE, Huynh TN, Woodling N, Fischbeck KH. "Epigenetic regulation of the survival motor neuron gene: Identifying molecular targets for SMA therapeutics," 2004 Families of SMA International Research Group Meeting, Schaumburg, IL
2. **Sumner CJ,** Avila AM, Burnett BG, Taye AA, Knight MA, Di Prospero NA, Fischbeck KH. "Trichostatin A treatment after disease onset increases survival of mice with SMA." 2006 American Academy of Neurology Meeting, San Diego, CA.
3. **Sumner CJ,** Avila AM, Burnett BG, Taye AA, Knight MA, Di Prospero NA, Fischbeck KH. Trichostatin A treatment after disease onset increases survival of mice with SMA. 2006 Society for Neuroscience Meeting, Atlanta, GA.
4. Choe DW, Warsin LC, Ng AS, Wagner KR, **Sumner CJ**. "Myostatin inhibition in SMA mice," 2008 Families of SMA Annual Research Meeting, Boston, MA.
5. Narver H, Burnett B, Kong L, Choe DW, Taye AA, Fischbeck KH, **Sumner CJ**. "Early administration of HDAC inhibition prolongs survival of SMA mice." 2007 American Academy of Neurology Meeting, Boston, MA.
6. Mentis GZ, **Sumner CJ**, O'Donovan MJ. "Altered synaptic input and excitability of motor neurons in SMA mice. 2008 Society for Neuroscience Meeting, Washington, DC.
7. Kong L, Wang X, Choe DW, Polley M, Burnett BG, Bosch-Marce M, Griffin JW, Rich MM, **Sumner CJ**. "Impaired synaptic vesicle release and immaturity of neuromuscular junctions in spinal muscular atrophy mice." 2008 Society for Neuroscience Meeting, Washington, DC.
8. Crowder ME, Polley MA, Kong L, Van Meerbeke JP, Wee CD, Murphy K, Griffin JW, Swoboda K, Crawford TO, **Sumner CJ**. Analysis of motor unit pathology in SMA patients. 2011 Society for Neuroscience Meeting, Washington, DC.
9. Kong L, Martinez TL, Wang X, Osborne MA, Crowder ME, Van Meerbeke JP, Xu X, Davis C, Wooley J, Goldhamer DJ, Lutz CM, Rich MM, **Sumner CJ**. "Survival motor neuron protein in motor neurons determines synaptic integrity in SMA." 2012 Society for Neuroscience Meeting, New Orleans, LA.
10. Lombardo J, Kong L, **Sumner CJ**, Harrington MA. Reduced levels of Survival Motor Neuron protein expression alters intrinsic excitability of motor neurons. 2014 Society for Neuroscience Meeting, Washington DC.
11. Aisenberg WH, Sullivan JM, Huh JS, **Sumner CJ**. Exploring the regulation of transient receptor potential vanilloid 4 (TRPV4) by the E3 ubiquitin ligase NEDD4. 2016 6th International Charcot-Marie-Tooth and Related Neuropathy Consortium (CMTR) Meeting, Venice, Italy.
12. Sullivan JM, Aisenberg WH, Woolums B, Huh JS, Lloyd TE, **Sumner CJ**. Dominant TRPV4 mutations in hereditary axonal neuropathies. 2016 6th International Charcot-Marie-Tooth and Related Neuropathy Meeting. Venice, Italy.
13. Sullivan JM, Aisenberg WH, Woolums B, Huh JS, Lloyd TE, **Sumner CJ**. Dominant TRPV4 mutations in hereditary axonal neuropathies. 6th Molecular Mechanisms of Axon Degeneration Meeting. 2016. Bar Harbor, ME.
14. D'Ydewalle C, Ramos DM, Pyles NJ, Ng SY, Gorz M, Pilato CM, Ling K, Kong L, Ward AJ, Rubin LL, Rigo F, Bennett CF, **Sumner CJ**. "Knockdown of a SMN associated lncRNA as a novel therapeutic strategy for SMA," 2016, Annual Meeting of the American Neurological Association, Baltimore MD.

Oral/Podium Presentations

15. Polydefkis M, Sheth SG, **Sumner C**J, Hauer P, Hoke A, Griffin JW, Cornblath DR. "Peripheral neuropathy in impaired glucose tolerance and diabetes." 2002 American Academy of Neurology Meeting, Denver, CO. [Dr. Polydefkis gave the talk]

16. **Sumner CJ**, Griffin JW, Chaudhry V. "The role of intraepidermal nerve fiber density in leprous neuropathy." 2001 Meeting of the American Association of Neuromuscular and Electrodiagnostic Medicine.
17. **Sumner CJ**, Huynh TN, Markowitz JA, Perhac S, Hill B, Coovert DD, Schussler BS, Chen X, Jarecki J, Burghes AH, Taylor JP, Fischbeck KH. "Valproic acid increases SMN in spinal muscular atrophy cell lines," 2003 Families of SMA International Research Group Meeting, Washington, DC.
18. **Sumner CJ**, Huynh T, Markowitz J, Taylor JP, Fischbeck KH. "Histone deacetylase inhibitors as treatment for spinal muscular atrophy." 2003 American Academy of Neurology Meeting, Honolulu, HI.
19. **Sumner CJ.** "Epidermal nerve fibers in spinal and bulbar muscular atrophy," 2003 Kennedy's Disease Association Meeting, New Orleans, LA.
20. **Sumner CJ.** "Histone acetylation and the SMN gene," 2005 Fight SMA/Andrew's Buddies Researcher Annual Meeting, Washington, DC.
21. **Sumner CJ**, Puls I, Levy J, Wallace KE, LaMonte B, Floeter MD, Ludlow CL, Holzbaur ELF, Fischbeck KH. "Clinical and pathophysiologic features of distal spinal and bulbar muscular atrophy with vocal fold paresis due to dynactin mutation." 2005 American Academy of Neurology Meeting, Miami Beach, FL.
22. **Sumner CJ.** "Histone deacetylase inhibitors in SMA." 2006 Fight SMA/Andrew's Buddies Annual Meeting, Washington, DC.
23. **Sumner CJ,** Avila A, Burnett BG, Taye AA, Knight MA, Di Prospero NA, Fischbeck KH. "Trichostatin A increases survival of mice with spinal muscular atrophy," 2006 Families of Spinal Muscular Atrophy Research Meeting, Montreal, Canada.
24. **Sumner CJ.** "SMA and TSA." 2007 Fight SMA/Andrew's Buddies Meeting, Washington, DC.
25. **Sumner CJ.** "Early administration of HDAC inhibition prolongs survival of SMA mice," 2007 Families of Spinal Muscular Atrophy Annual Meeting, Schaumburg, IL
26. **Sumner CJ**. "Therapeutics in SMA mice," 2008 Fight SMA/Andrew's Buddies Meeting, Washington, DC.
27. Kong L, Choe DW, Bosch-Marce M, **Sumner CJ**. "Immaturity of neuromuscular junctions in SMA mice: Implications for therapy." 2008 Families of SMA Annual Research Meeting, Boston, MA.
28. **Sumner CJ.** "Neuromuscular junctions in spinal muscular atrophy," 2009 Fight SMA/Andrew's Buddies Meeting, Washington, DC.
29. Kong L, Wang X, Choe DW, Polley M, Bosch- Marcé, Griffin JW, Rich MM, **Sumner CJ**. "Impaired synaptic vesicle release and immaturity of neuromuscular junctions in spinal muscular atrophy mice." 2009 Families of SMA meeting, Cincinnati, Ohio.
30. Kong, L, Mentis G, O'Donovan M, **Sumner CJ**, "Peripheral and central synaptic dysfunction in the inherited motor neuron disease, spinal muscular atrophy," 2009 Cellular and Network Functions in the Spinal Cord Meeting, Madison, WI.
31. **Sumner, CJ**. "Synaptic function in SMA mice," 2010 Fight SMA/Andrew's Buddies Meeting, Washington, DC.
32. **Sumner CJ**. "Overview of therapeutic strategies in SMA" 2010 Families of Spinal Muscular Atrophy International Research Conference, Santa Clara, CA.
33. **Sumner CJ.** "Cellular mechanisms of SMA disease pathogenesis in mouse and human," 2011 Fight SMA/Andrew's Buddies Meeting, Washington, DC.
34. **Sumner CJ**. "Review of neuronal and muscle phenotypes in humans" 2011 Families of Spinal Muscular Atrophy International Research Conference, Orlando, FL.
35. Sullivan JM, Landoure G, Martinez TL, Burnett BG, Kong L, Sahin B, Gaudet R, Lloyd TE, **Sumner CJ**. "Dominant TRPV4 mutations in hereditary neuropathies." 2011 Peripheral Nerve Society Meeting, Potomac, MD. [Dr. Sullivan gave this presentation]
36. **Sumner CJ.** "SMN is motor neurons determines synaptic integrity in SMA," 2012 Fight SMA/Andrew's Buddies Meeting, Washington, DC.
37. Van Meerbeke JP, Gibbs RG, Plasterer HL, Feng Z, Lin MY, Wee CD, Xia B, Sharma S, Jacques V,Rusche JR, Ko CP, and **Sumner CJ**. "The Therapeutic Effects of RG3039 in Severe Spinal Muscular Atrophy Mice and Normal Human Volunteers." 2012 American Academy of Neurology Meeting, New Orleans, LA. [Mr. VanMeerbeke gave this presentation]
38. **Sumner CJ**. "Animal models of SMA." 2013 Muscular Dystrophy Association Scientific Conference, Washington, DC.
39. **Sumner CJ** "Cellular consequences of SMN deficiency in mice and humans" 2013 Fight SMA/Andrew's Buddies Meeting, Washington, DC.

40. Sullivan JM, Yang M, Le A, Mamah C, Lloyd T, **Sumner CJ**. "Dominant TRPV4 mutations in hereditary axonal neuropathies." 2013 Peripheral Nerve Society Meeting, Saint Malo, France. 6-13-2013. [Dr. Sullivan gave this presentation]
41. **Sumner CJ** "Motor neuron pathology in human SMA." 2013 American Neurological Association Meeting Neuromuscular Interest Group, New Orleans, LA.
42. **Sumner CJ.** "Motor axon development in spinal muscular atrophy." 2014 Fight SMA/Andrew's Buddies Meeting, Washington, DC.
43. Xu X, Desideri R, Kong L, Davis C, Osborne M, Hua Y, Rigo F, Krainer AR, Bennett F, Lutz C, **Sumner CJ**. "Impaired motor axon development in spinal muscular atrophy." 2014 Families of SMA meeting, Washington DC.
44. **Sumner CJ.** "SMA, mouse models, and therapeutics development." 2014 New Directions in Skeletal Muscle Biology Meeting, Chicago IL.
45. Kong L, Xu X, Desideri R, Davis C, Osborne M, Hua Y, Rigo F, Krainer AR, Bennett F, Lutz C, **Sumner CJ**. "Impaired motor axon development in spinal muscular atrophy." 2014 The Joint Symposium of the 4th International Neural Regeneration Symposium and 6th International Spinal Cord Injury Treatment and Trial Symposium and 9th Asia Pacific Symposium on Neural Regeneration, Nanjing, China.[Dr. Kong gave this presentation]
46. **Sumner CJ**. "Impaired proximal axonal development may drive early somal loss in SMA." 2015 Muscular Dystrophy Association Scientific Conference, Washington, DC.
47. **Sumner CJ.** "Impaired proximal axon development in spinal muscular atrophy." 2015 Fight SMA/Andrew's Buddies Meeting, Washington, DC.
48. **Sumner CJ.** "The 'simple' questions of SMA pathogenesis." 2016 Fight SMA Conference, Alexandria, VA.
49. **Sumner CJ.** "Knockdown of a SMN associated lncRNA as a novel therapeutic strategy for SMA," 2016, Annual Meeting of the American Neurological Association Plenary Data Blitz presentation and Neuromuscular Special Interest Group presentation, Baltimore MD.