

CURRICULUM VITAE

The Johns Hopkins University School of Medicine

THOMAS OWEN CRAWFORD, M.D.

June, 2015

DEMOGRAPHIC AND PERSONAL INFORMATION

Current Appointments

2013 - Professor, The Johns Hopkins University School of Medicine, Department of Neurology
1997 - Joint Appointment, Department of Pediatrics
1999 - Co-Director, Muscular Dystrophy Clinic
1995 - Neurologist, Ataxia Telangiectasia Clinical Center

Personal Data:

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Education and Training:

Undergraduate

1972-76 B.A., Yale College, New Haven, Connecticut, Psychology and Religion

Doctoral

1976-80 M.D. University of Southern California School of Medicine, Los Angeles, California

Postdoctoral

1980-81 Pediatric Internship, Los Angeles County/University of Southern California Medical Center (LAC/USCMC) Pediatric Pavilion, Los Angeles, California
1981-83 Pediatric Residency, LAC/USCMC Pediatric Pavilion
1983-84 Pediatric Chief Resident, LAC/USCMC Pediatric Pavilion
1984-87 Child Neurology Residency, Childrens Hospital of Los Angeles and the LAC/USCMC Department of Neurology, Los Angeles, California
1987-88 Post-doctoral Research Fellowship, Laboratory of Dr. John W Griffin, Department of Neurology The Johns Hopkins Hospital

Professional Experience:

1984 – 1987 Clinical Instructor in Pediatrics, University of Southern California, Los Angeles, California
1988 – 1990 Instructor in Neurology, Department of Neurology, The Johns Hopkins Hospital, Baltimore, MD
1990 – 1997 Assistant Professor of Neurology
1991 – 1997 Assistant Professor of Pediatrics
1996 – 2009 Staff Physician, Mt Washington Pediatric Hospital, Baltimore, MD
1997 – 2013 Associate Professor of Neurology and Pediatrics

RESEARCH ACTIVITIES

Peer Reviewed Original Science Publications

1. **Crawford TO**, Mitchell WG, Snodgrass SR. Lorazepam in childhood status epilepticus and serial seizures: Effectiveness and tachyphylaxis. *Neurology* 1987;37:190-195.
2. Mitchell WG, **Crawford TO**. Diagnosis and treatment of intraparenchymal cerebral cysticercosis in children. *Pediatrics* 1988;82:76-82.
3. **Crawford TO**, Mitchell WG, Fishman LS, Snodgrass SR. Very high dose phenobarbital for refractory status epilepticus in children. *Neurology* 1988;38:1035-1040.

4. Koliatsos VE, **Crawford TO**, Price DL. Axotomy induces nerve growth factor receptor immunoreactivity in spinal motor neurons. *Brain Res* 1991;549:297-304.
5. Levine B, Hardwick JM, Trapp BD, **Crawford TO**, Bollinger RC, Griffin DE. *In vitro* and *in vivo* antibody-mediated clearance of alphavirus infection from neurons. *Science* 1991;254:856-860.
6. Chaudhry V, **Crawford TO**, DeRossett, SE. Thermal sensitivity of demyelinating neuropathy. *Muscle Nerve* 1993;16:301-306.
7. Power C, Kong PA, **Crawford TO**, Wesselingh S, Glass JD, McArthur JC, Trapp BD. Cerebral white matter changes in acquired immunodeficiency syndrome dementia: Alterations in the blood-brain barrier. *Ann Neurol* 1993;34:339-350.
8. Hsieh S-T, **Crawford TO***, Griffin JW. Neurofilament distribution and organization in the myelinated axon of the peripheral nervous system. *Brain Res* 1994;642:316-326. (* corresponding author)
9. Torres JA, Sponseller P, Kim TS, Kuncel RW, **Crawford TO**. Radiographic and histologic characterization of Tc/tw5 mice: an animal model of lumbosacral agenesis/myelomeningocele. *Dev Med Child Neurol* 1994;36:698-706.
10. Hsieh S-T, Kidd GJ, **Crawford TO**, Xu Z, Lin W-M, Trapp BD, Cleveland DW, Griffin JW. Regional modulation of neurofilament organization by myelination in normal axons. *J Neurosci* 1994;14:6392-6401.
11. Thompson TG, DiDonato CJ, Simard LR, Ingraham SE, Burghes AHM, **Crawford TO**, Rochette C, Mendell JR, Wasmuth JJ. A novel cDNA detects homozygous microdeletions in greater than 50% of type 1 spinal muscular atrophy patients. *Nat Genet* 1995;9:56-62.
12. Roy N, Mahadevan MS, McLean M, Shutler G, Yaraghi Z, Farahani R, Baird S, Besner-Johnston A, Lefebvre C, Kang X, Salih M, Aubry H, Tamai K, Guan X, Ioannou P, **Crawford TO**, de Jong PJ, Surh L, Ikeda J-E, Korneluk RG, MacKenzie A. The gene for neuronal apoptosis inhibitory protein is partially deleted in individuals with spinal muscular atrophy. *Cell* 1995;80:167-178.
13. **Crawford TO**, Hsieh S-T, Schryer BL, Glass JD. Prolonged axonal survival in transected nerves of C57BL/Ola mice is independent of age. *J Neurocytol* 1995;24:333-340.
14. Wong PC, Marszalek J, **Crawford TO**, Hsieh S-T, Griffin JW, Cleveland DW. Increasing neurofilament subunit NF-M expression reduces axonal NF-H, inhibits radial growth, and results in neurofilamentous accumulation in human motor neurons. *J Cell Biol* 1995;130:1413-1422.
15. Corse AM, Chaudhry V, **Crawford TO**, Kuncel RW, Griffin JW. Sensory nerve pathology in multifocal motor neuropathy. *Ann Neurol* 1996;39:319-325.
16. Mahattanakul W, **Crawford TO**, Griffin JW, Goldstein JM, Cornblath DR. Cyclosporine-A therapy in chronic inflammatory demyelinating polyneuropathy. *J Neurol Neurosurg Psychiat* 1996;60:185-187.
17. Xu Z, Marszalek JR, Lee MK, Wong PC, Folmer J, **Crawford TO**, Hsieh S-T, Griffin JW, Cleveland DW. Subunit composition of Neurofilaments specifies axonal diameter. *J Cell Bio* 1996;133:1061-1069.
18. Roe TF, **Crawford TO**, Huff KR, Costin G, Kaufman FR, Nelson MD. Brain infarction in children with diabetic ketoacidosis. *J Diabetes Complicat* 1996;10:100-108.
19. Pegoraro E, Mancias P, Swerdlow SH, Raikow RB, Garcia C, Marks H, **Crawford T**, Carver V, Di Cianno B, Hoffman EP. Congenital muscular dystrophy with primary laminin $\alpha 2$ (merosin) deficiency presenting as inflammatory myopathy. *Ann Neurol* 1996;40:782-791.
20. Jung D, Duclos F, Apostol B, Straub V, Lee JC, Allamand V, Venzke DP, Sunada Y, Moomaw CR, Leveille CJ, Slaughter CA, **Crawford TO**, McPherson JD, Campbell KP. Characterization of δ -Sarcoglycan: a novel component of the oligomeric sarcoglycan complex involved LGMD. *J Biol Chem* 1996;271:32321-32329.
21. Marszalek JR, Williamson TL, Lee MK, Xu Z, Hoffman PN, Beecher MW, **Crawford TO**, Cleveland DW. Neurofilament subunit NF-H modulates axonal diameter by selectively slowing neurofilament transport. *J Cell Biol* 1996;135:711-724.
22. Ohno K, Quiram PA, Milone M, Wang H-L, Harper MC, Pruitt JN III, Brengman JM, Pao L, Fischbeck KH, **Crawford TO**, Engel AG. Congenital myasthenic syndromes due to heteroallelic nonsense/missense mutations in the acetylcholine receptor ϵ subunit gene: Identification and functional characterization of 6 new mutations. *Hum Mol Genet* 1997;6:753-766.
23. Coovert DD, Le TT, McAndrew PE, Strasswimmer J, **Crawford TO**, Mendell JR, Coulson SE, Androphy EJ, Prior TW, Burghes AH The survival motor neuron protein in spinal muscular atrophy. *Hum Mol Genet* 1997;6:1205-14.
24. Flanigan KM, **Crawford TO**, Griffin JW, Goebel HH, Kohlschütter A, Ranells J, Camfield P, Ptáček LJ. Localization of the giant axonal neuropathy gene to chromosome 16q24. *Ann Neurol* 1997;43:143-148.

25. Cabana MD, **Crawford TO**, Winkelstein JA, Christensen JR, Lederman HM. Consequences of the delayed diagnosis of ataxia-telangiectasia. *Pediatrics* 1997;102: 98-100.
26. Lin C-L G, Bristol LA, Jin L, Dykes-Hoberg M, **Crawford T**, Clawson L, Rothstein JD, Aberrant glutamate transporter mRNA species in amyotrophic lateral sclerosis. *Neuron* 1998;20:589-602.
27. Holland NR, **Crawford TO**, Hauer P, Cornblath DR, Griffin JW, McArthur JC. Small-fiber sensory neuropathies: clinical course and neuropathology of idiopathic cases. *Ann Neurol* 1998;44:47-59.
28. Pegoraro E, Marks H, Garcia CA, **Crawford T**, Mancias P, Connolly AM, Fanin M, Martinello F, Trevisan CP, Angelini C, Stella A, Scavina M, Munk RL, Servidei S, Bonemann C, Bertorini T, Acsadi G, Thompson CE, Gagnon D, Hoganson G, Carver V, Zimmerman RA, Hoffman EP. Laminin alpha2 muscular dystrophy: genotype/phenotype studies of 22 patients. *Neurology* 1998;51:101-110.
29. Yin X, **Crawford TO**, Li C, Roder J, Tu P-h, Lee VM-Y, Griffin JW, Trapp BD. Myelin-associated glycoprotein modulates axonal caliber and neurofilament organization. *J Neurosci* 1998;18:1953-62.
30. Rao MV, Houseweart MK, Williamson TL, **Crawford TO**, Folmer J, Cleveland DW. Neurofilament-dependent radial growth of motor axons and axonal organization of neurofilaments does not require the neurofilament heavy subunit (NF-H) or its phosphorylation. *J Cell Biol* 1998;143:171-81.
31. Gleeson JG, Minnerath SR, Fox JW, Allen KM, Luo RF, Hong SE, Berg MJ, Kuzniecky R, Reitnauer PJ, Borgatti R, Mira AP, Guerrini R, Holmes GL, Rooney CM, Berkovic S, Scheffer I, Cooper EC, Ricci S, Cusmai R, **Crawford TO**, Leroy R, Andermann E, Wheless JW, Dobyns WB, Ross ME, Walsh CA. Characterization of mutations in the gene *doublecortin* in patients with double cortex syndrome. *Ann Neurol* 1999;45:146-53.
32. **Crawford TO**, Sladky JT, Hurko O, Besner-Johnston A, Kelley RI. Abnormal fatty acid metabolism in childhood spinal muscular atrophy. *Ann Neurol* 1999;45:337-343.
33. Sheikh KA, Sun J, Liu Y, Kawai H, **Crawford TO**, Proia RL, Griffin JW, Schnaar RL. Mice lacking complex gangliosides develop Wallerian degeneration and myelination defects. *Proc. Natl. Acad. Sci. USA* 1999; 96:7532-7537.
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35. Lewis RF, Lederman HM, **Crawford TO**. Oculomotor abnormalities in ataxia-telangiectasia. *Ann Neurol* 1999;46:287-295.
36. Lefton-Greif MA, **Crawford TO**, Winkelstein JA, Loughlin GM, Koerner GB, Zahurak M, Lederman HM. Oropharyngeal dysphagia and aspiration in patients with ataxia telangiectasia. *J Pediatr* 1999;136:225-231.
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39. Johnston JJ, Kelley RI, **Crawford TO**, Morton DH, Agarwala R, Koch T, Schäffer AA, Francomano CA, Biesecker LG. A novel nemaline myopathy in the Amish caused by a mutation in troponin T1. *Am J Hum Genet*, 2000;67:814-821.
40. Bizzi A, Ulug AM, **Crawford TO**, Passe T, Bugiani M, Bryan RN, Barker PB. Quantitative proton MR spectroscopic imaging in acute disseminated encephalomyelitis. *Am J Neuroradiol* 2001;22:1125-1130.
41. Katz ES, Gauda E, **Crawford T**, Ogunlesi F, Lefton-Greif MA, McGrath-Morrow S, Marcus CL. Respiratory flutter syndrome: an under-recognized cause of respiratory failure in neonates. *Am J Respir Crit Care Med* 2001;164:1161-1164.
42. Lewis RF, **Crawford TO**. Slow target-directed eye movements in Ataxia Telangiectasia. *Invest. Ophthalmol. Vis Sci* 2002;43:686-691.
43. Lunn MPT, **Crawford TO**, Hughes RA, Griffin JW, Sheikh KA. Anti-myelin associated glycoprotein antibodies alter neurofilament spacing. *Brain* 2002;125:904-911.
44. Farr AK, Shalev B, **Crawford TO**, Lederman HM, Winkelstein JA, Repka MX. Ocular Manifestations of Ataxia Telangiectasia. *Am J Ophthalmol* 2002;134:891-896.
45. Lin DDM, **Crawford TO**, Barker PB. Proton magnetic resonance spectroscopy in the diagnostic evaluation of suspected mitochondrial diseases. *Am J Neuroradiol* 2003;24:33-41.
46. Chaudhry V, Chaudhry M, **Crawford TO**, Simmons-O'Brien E, Griffin JW. Toxic neuropathy in patients with pre-existing neuropathy. *Neurology* 2003;60:337-340.
47. Hoke A, Ho T, **Crawford TO**, LeBel C, Hilt D, Griffin. Glial cell line-derived neurotrophic factor alters axon Schwann cell units and promotes myelination in unmyelinated nerve fibers. *J Neurosci* 2003;23:561-567.

48. Kerr DA, Lladó J, Shamblott MJ, Maragakis NJ, Irani DN, **Crawford TO**, Krishnan C, Dike S, Gearhart JD, Rothstein JD. Human embryonic germ cell derivatives facilitate motor recover of rats with diffuse motor neuron injury. *J. Neurosci* 2003;23: 5131-5140.
49. Jin J-P, Brotto MA, Hossain MM, Huang Q-Q, Brotto LS, Nosek TM, Morton DH, **Crawford TO**. Truncation by Glu₁₈₀ nonsense mutation results in complete loss of slow skeletal muscle troponin T in a lethal nemaline myopathy: function of the muscle fiber type-specific troponin T isoforms. *J Biol Chem* 2003;278:26159-26165.
50. Garcia ML, Lobsiger CS, Shah SB, Deerinck TJ, Crum J, Young D, Ward CM, **Crawford TO**, Gotow T, Uchiyama Y, Ellisman MN, Calcutt NA, Cleveland DW. NF-M is an essential target for the myelin-directed “outside in” signaling cascade that mediates radial axonal growth. *J Cell Biol* 2003;163:1011-1020.
51. Nowak-Wegrzyn A, **Crawford TO**, Winkelstein JA, Carson KA, Lederman HM. Immunodeficiency and infections in ataxia-telangiectasia. *J Pediatr* 2004;144:505-511.
52. Agrawal, PB, Strickland CD, Midgett C, Morales A, Newburger D, Poulos MA, Tomczak KK, Ryan MM, Iannaccone ST, **Crawford TO**, Laing NG, Beggs AH. Heterogeneity of nemaline myopathy cases with skeletal muscle actin (ACTA1) gene mutations. *Ann Neurol* 2004;56:86-96.
53. Wang X, Huang Q-Q, Breckenridge MT, Chen A, **Crawford TO**, Morton DJ, Jin J-P. Cellular Fate of Truncated Slow Skeletal Muscle Troponin T Produced by Glu₁₈₀ Nonsense Mutation in Amish Nemaline Myopathy. *J Biol Chem* 2004;280:13241-13249.
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56. Hudspeth MP, Holden KR, **Crawford TO**. The “slurp” test: Bedside evaluation of bulbar muscle fatigue. *Pediatrics* 2006;118:e530-533.
57. **Crawford TO**, Skolasky RL, Fernandez R, Rosquist KJ, Lederman HM. Survival probability in ataxia telangiectasia. *Arch Dis Child* 2006;91:610-1.
58. Lin D, **Crawford T**, Lederman H, Barker P. Proton MR Spectroscopic Imaging in Ataxia-Telangiectasia. *Neuropediatrics* 2006;37:241-246.
59. Krosschell KJ, Maczulski JA, **Crawford TO**, Scott C, Swoboda KJ. A modified Hammersmith functional motor scale for use in multi-center research on spinal muscular atrophy. *Neuromuscul Dis* 2006;16:417-426.
60. Pidcock FS, Krishnan C, **Crawford TO**, Salorio C, Trovato M, Kerr DA. Acute transverse myelitis in childhood: center based analysis of 47 cases. *Neurology* 2007;68:1474-1480.
61. Mandir AS, Jilfiker J, Thomas G, Minahan RE, **Crawford TO**, Williams MA, Rigamonti D. Extrapyrmidal signs in normal pressure hydrocephalus: an objective assessment. *Cerebrospinal Fluid Res* 2007; 4:7.
62. Alterman N, Fattal-Valevski A, Moyal L, **Crawford TO**, Lederman HM, Ziv Y, Shiloh Y. Ataxia-Telangiectasia: mild presentation despite null *ATM* mutation and severe cellular phenotype. *Am J Med Genet* 2007;143:1827-1934.
63. Gómez-Curet I, Robinson KG, Funanage VL, **Crawford TO**, Scavina M, Wang W. Robust quantification of the SMN gene copy number by real-time TaqMan PCR. *Neurogenetics* 2007;8:271-8.
64. McGrath-Murrow S, Lefton-Greif M, Rosquist K, **Crawford T**, Kelly A, Zeitlin P, Carson KA, Lederman LM. Pulmonary Function in Adolescents with Ataxia Telangiectasia. *Pediatr Pulmonol* 2008;43:59-66.
65. Gavrilina TO, McGovern VL, Workman E, **Crawford TO**, Gogliotti RG, Didonato CJ, Monani UR, Morris GE, Burghes HM. Neuronal SMN expression corrects spinal muscular atrophy in severe SMA mice while muscle specific SMN expression has no phenotypic effect. *Hum Mol Genet* 2008;17:1063-1075.
66. Jurynek MJ, Xia R, Mackrill JJ, Gunther D, **Crawford T**, Flanigan KM, Abramson JJ, Howard MT, Grunwald DJ. Selenoprotein N is required for ryanodine receptor calcium release channel activity in human and zebrafish muscle. *Proc Natl Acad Sci U.S.A.* 2008;105:12485-12490.
67. Swoboda KJ, Scott CB, Reyna SP, Prior TW, LaSalle B, Sorenson SL, Wood J, Acsadi G, **Crawford TO**, Kissel JT, Krosschell KJ, D'Anjou G, Bromberg MB, Schroth MK, Chan GM, Elsheikh B, Simard LR. Phase II open label study of valproic acid in spinal muscular atrophy. *PLoS ONE* 2009;4:e5268.
68. Workman E, Saieva L, Carrel TL, **Crawford TO**, Liu D, Lutz C, Beattie CE, Pellizzoni L, Burghes AH. A SMN missense mutation complements SMN2 restoring snRNPs and rescuing SMA mice. *Hum Mol Genet* 2009;18:2215-2229.
69. Shaikh AG, Marti S, Tarnutzer AA, Palla A, **Crawford TO**, Straumann D, Taylor AM, Zee DS. Gaze fixation deficits and their implication in ataxia-telangiectasia. *J Neurol Neurosurg Psychiatry* 2009;80:858-864.

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71. Swoboda KJ, Scott CB, **Crawford TO**, Simard LR, Reyna SP, Krossschell KJ, Acsadi G, Elsheik B, Schroth MK, D'Anjou G, LaSalle B, Prior TW, Sorenson S, Maczulski JA, Bromberg MB, Chan GM, Kissel JT, Project Cure Investigator's Network. SMA Carni-Val trial part I: Double-blind, randomized, placebo-controlled trial of L-Carnitine and Valproic Acid in spinal muscular atrophy. *PLoS ONE* 2010;5: e12140.
72. Lewelt A, Scott C, Sakonju A, Kissel J, **Crawford TO**, Acsadi G, D'Anjou G, Elsheikh B, Reyna S, Schroth M, Maczulski JA, Elovic E, Swoboda K. Compound muscle action potential and motor function in children with spinal muscular atrophy. *Muscle Nerve* 2010;42:703-8.
73. Finanger Hedderick ELF, Simmers JL, Soleimani A, Andres-Mateos E, Marx R, Files DC, King L, **Crawford TO**, Corse AM, Cohn RD. Loss of sarcolemmal nNOS is common in acquired and inherited neuromuscular disorders. *Neurology* 2011;76:960-7.
74. Bilchick KC, Plitt D, Salerno M, Plitt D, Dori Y, **Crawford TO**, Drachman D, Thompson WR. Prevalence and distribution of regional scar in dysfunctional myocardial segments in Duchenne muscular dystrophy based on myocardial tagging and echocardiographic assessments of circumferential strain. *J Cardiovasc Magn Reson* 2011;13:20.
75. Shaikh AG, Marti S, Tarnutzer AA, Palla A, **Crawford TO**, Straumann D, Carey JP, Nguyen KD, Zee DS. Ataxia-telangiectasia: a "disease model" to understand the cerebellar control of vestibular reflexes. *J Neurophysiol*, 2011;105:3034-41.
76. Kissel JT, Scott CB, Reyna SP, **Crawford TO**, Simard LR, Krossschell KJ, Acsadi G, Elsheik B, Schroth MK, D'Anjou G, LaSalle B, Prior TW, Sorenson S, Maczulski JA, Bromberg MB, Chan GM, Swoboda KJ. SMA CarniVal trial part II: A prospective, single-armed trial of l-carnitine and valproic acid in ambulatory children with spinal muscular atrophy. *PLoS ONE* 6(7) e21296, 2011.
77. Lefton-Greif MA, **Crawford TO**, McGrath-Morrow S, Carson KA, Lederman HM. Safety and caregiver satisfaction with gastrostomy in patients with ataxia telangiectasia. *Orphanet J Rare Dis*, 2011;6:23.
78. McElroy MJ, Shaner AC, **Crawford TO**, Thompson GH, Rishi V, Kadakia RV, Behrooz A, Akbarnia BA, Skaggs DL, Emans JB, Sponseller PD. Growing rods for scoliosis in spinal muscular atrophy: Structural effects, complications, and hospital stays. *Spine*, 2011 36:1305-11.
79. Shen X-M, **Crawford TO**, Brengman J, Acsadi G, Iannacone S, Karaca E, Khoury C, Mah JK, Edvardson S, Bajzer Z, Rodgers D, Engel AG. Mechanisms of Pathogenicity of Missense Mutations in Human Choline Acetyltransferase. *Hum Mutat* 2011; 32:1259-67.
80. Dzieciatkowska M, Qi G, You J, Bemis KG, Sahn H, Lederman HM, **Crawford TO**, Gelbert LM, Rothblum-Oviatt C, Wang M. Proteomic characterization of cerebrospinal fluid from ataxia-telangiectasia (A-T) patients using a LC/MS-based label-free protein quantification technology. *Int J Proteomics*, 2011; ID 578903.
81. Lockman JL, Iskander AJ, Bembea M, **Crawford TO**, Lederman HM, McGrath-Morrow S, Easley RB. Anesthetic and perioperative risk in the patient with ataxia-telangiectasia. *Pediatr Anaesth*, 2011; 22: 256-262.
82. **Crawford TO**, Paushkin SV, Kobayashi DT, Forrest SJ, Joyce CL, Finkel RS, Kaufmann P, Swoboda KJ, Tiziano FD, Lomastro R, Li RH, Trachtenberg F, Plasterer T, Chen KS. Evaluation of SMN protein, transcript, and copy number in the Biomarkers for Spinal Muscular Atrophy (BforSMA) clinical study. *PLoS One* 7(4) e33572, 2012.
83. Finkel RS, **Crawford TO**, Kaufmann P, Swoboda KJ, Juhasz P, Li X, Li RH, Trachtenberg F, Tiziano FD, Lomastro R, Paushkin S, Kobayashi DT, Forrest S, Chen KDS, Joyce C, Plasterer T, Bfor SMA Trial Group. Spinal muscular atrophy biomarker discovery project: candidate proteins, metabolites, and transcripts. *PLoS One* 7(4) e35462, 2012.
84. Lochman JL, Iskander AJ, Bembea M, **Crawford TO**, Lederman HM, McGrath-Morrow S, Easley RB. The critically ill patient with ataxia-telangiectasia: A case series. *Pediatr Crit Care Med* 2012; 13:256-262.
85. Chew S, Balasubramanian R, Chan W-M, Kang PB, Andrews C, Webb BC, MacKinnon SE, Balasubramanian R, Chan W-M, Kang PB, Andrews C, Webb BC, MacKinnon SE, Oystreck DT, Rankin J, **Crawford TO**, Geraghty M, Pomeroy SL, Crowley WF, Jabs EW, Hunter DG, Grant PE, Engle EC. A novel syndrome caused by the E410K amino acid substitution in the neuronal β - tubulin isotype 3. *Brain*, 2013; 136:522-535.
86. Kobayashi DT, Shi J, Stephen L, Ballard KL, Dewey R, Mapes J, Chung B, McCarthy K, Swoboda KJ, **Crawford TO**, Li R, Plasterer T, Joyce C and the Biomarkers for SMA Study Group. SMA-MAP: A plasma protein biomarker panel for spinal muscular atrophy. *PLoS One* 8(4) e60113, 2013.
87. Shaikh AG, Zee DS, Mandir AS, Lederman HM, **Crawford TO**. Disorders of upper limb movements in ataxia telangiectasia. *PLoS ONE*, 8(6): e67042, 2013.

88. Shaikh AG, Marti S, Tamutzer AA, Palla A, **Crawford TO**, Zee DS, Straumann D. Effects of 4-aminopyridine on nystagmus and vestibulo-ocular reflex in ataxia-telangiectasia. *J Neurol*, 2013;260:2728-2735.
89. Tisdale S, Lotti, F, Saieva L, Van Meerbeke JP, **Crawford TO**, Sumner CJ, Mentis GZ, Pellizzoni L. SMN is essential for the biogenesis of U7 small nuclear ribonucleoprotein and 3'-end formation of histone mRNAs. *Cell Reports* 2013; 5:1187-1195.
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91. Lin DDM, Barker PB, Lederman HM, **Crawford TO**. Cerebral abnormalities in adults with ataxia telangiectasia. *Am J Neuroradiol*, 2014; 35:119-123.
92. McGrath-Morrow S, Lederman HM, Aherrera AD, Lefton-Greif MA, **Crawford TO**, Ryan R, Wright J, Collaco JM. Pulmonary function in children and young adults with ataxia-telangiectasia. *Pediatr Pulmonol*, 2014; 49:84-90.
93. Volkow ND, Tomasi D, Wang G-J, Studentsova Y, Margus B, **Crawford TO**. Brain glucose metabolism in adults with ataxia-telangiectasia and their asymptomatic relatives. *Brain* 2014;137:1753-61.
94. Mirski KT, **Crawford TO**. Motor and cognitive delay in Duchenne muscular dystrophy: Implication for early diagnosis. *J Pediatr*, 2014; 165:1008-1010.
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96. Miller N, Feng Z, Edens B, Yang B, Shi H, Sze C, Hong B, Su S, Cantu J, Topczewski J, **Crawford TO**, Ko C-P, Sumner C, Ma L, Ma Y-C. Non-aggregating tau phosphorylation by Cyclin-Dependent Kinase 5 contributes to motor neuron degeneration in spinal muscular atrophy" *J Neurosci* 2015; 35:6038-6050.
97. Rindt H, Feng Z, Mazzasette C, Glascock JJ, Valdivia D, Pyles N, **Crawford TO**, Swoboda KJ, Patitucci TN, Ebert AD, Sumner CJ, Ko C-P, Lorson CL. Astrocytes influence the severity of spinal muscular atrophy. *Hum Mol Genet* 2015; 24:4094-410
98. Stabley DL, Harris AW, Holbrook J, Chubbs NJ, Lozo KW, **Crawford TO**, Swoboda KJ, Funanage VL, Wang W, Mackenzie W, Scavina M, Sol-Church K, Butchbach MER. *SMN1* and *SMN2* copy numbers in cell lines derived from patients with spinal muscular atrophy as measured by array digital PCR *Molecular Genetics and Genomic Medicine* in press 2015

Inventions, Patents, Copyrights *None*

EXTRAMURAL SPONSORSHIP

Current Grants

- | | |
|------------------|---|
| 9/1/95 – 8/31/13 | “Ataxia Telangiectasia Clinical Center”
Ataxia Telangiectasia Children’s Project
\$164,692
Role: Neurology consultant; 15%; To develop and maintain an international clinical center for patients with A-T, to develop collaborative clinical and laboratory research in A-T |
| 7/1/02 – 6/30/13 | Muscular Dystrophy Clinical Center
Muscular Dystrophy Association (MDA)
\$149,954
Role: Clinic Co-Director; 10% effort |

Pending Grants

- | | |
|------------------|---|
| 11/1/12-10/31/14 | “Collection and analysis of human SMA tissues”
Spinal Muscular Atrophy Foundation
\$224,703
PI: Charlotte Sumner |
|------------------|---|

Role: Case review and coordination, collection and dissection of tissues; 10%; Role in collecting and distributing human SMA and control tissues from surgical and autopsy sources as a resource for answering specific questions about SMA pathology

Previous Grants

- 12/1/89 – 11/20/94 “Peripheral regulation of motor neuron gene expression”
NS01412-01
NIH, NINDS: Clinical Investigator Development Award: National Institutes of Health 1 KO8
\$340,400
Role: PI, effort: 80%; Notes: Experiments directed to the exploration of the influence of muscle on gene expression within innervating motor neurons
- 7/1/95 – 6/30/96 “FK366 Phase IIb study: Efficacy in diabetic neuropathy”
Fujisawa USA
\$12,400
Role: pathology reviewer; Contract to provide expert services reviewing study subject’s nerve
- 1/1/97 - 12/31/99 “Abnormal fatty acid metabolism and expression of SMN in spinal muscular atrophy (SMA)”
MDA grant 2378
\$150,000
Role: PI, effort: 30%
- 1/1/99 – 12/31/99 “Supplemental Dietary Glutamine in the Treatment of SMA”
Families of Spinal Muscular Atrophy (FSMA)
\$55,200
Role: PI; Notes: Funding was awarded, but the budget was cut below what was thought necessary to conduct the project. Grant was returned.
- 9/1/00 – 8/31/01 “Neurologic pathophysiology of ataxia telangiectasia”
Ataxia Telangiectasia Children’s Project (ATCP)
Role: PI, effort: 10%
- 7/1/01 – 6/30/06 HIV Sensory neuropathy: age, risk factors, and immunopathogenesis
NIH NS 43011
Co P.I. Justin McArthur
Role: Pathology review and morphometry; 5% effort
- 2/1/03 – 1/3/08 “Survival Motor Neuron Genes in Spinal Muscular Atrophy”
NIH RO1 NS038650
\$16,000 subcontract from Ohio State University
PI: Arthur Burghes
Role: Pathologist and morphometry, 5%
- 2003 – 2005 Families of Spinal Muscular Atrophy, Chesapeake Chapter
SMA book fund
Role: PI; 20% effort
- 1/1/04 – 12/31/04 “Multicenter trial on Development of Outcome Measures for SMA”
PROCRECR
Families of SMA (FSMA)
\$57,419
Role: PI, 30% effort
- 7/1/05 – 10/31/07 “Project Cure, CarniVal Type II” (Treatment Trial)
PROCR2

- Families of Spinal Muscular Atrophy (FSMA)
\$127,749
Role: PI, 15% effort
- 10/1/05 – 9/30/07 “High resolution MRI, Eye movement tracking, and tremor analysis in Ataxia Telangiectasia”
279-2056
Ataxia Telangiectasia Children’s Project
\$62,500
Co-Pis: David Zee, Thomas Crawford
Role: Co-PI; 10%
- 2/1/08 – 9/30/09 “Project Cure, CarniVal 2 for infants” (Treatment Trial)
PROCRCR0809
Families of Spinal Muscular Atrophy (FSMA)
\$151,800
Role: PI, 9% effort
- 10/1/08 – 9/30/09 “Biomarkers for Spinal Muscular Atrophy”
104691
SMA Foundation, Inc.
\$47,205
Role: PI, 12.5% effort
- 11/1/09 – 10/31/12 “Collection and analysis of human SMA tissues”
SMA Foundation, Inc.
\$ 310,225
PI: Charlotte Sumner
Role: Case review and coordination, collection and dissection of tissues; 10%; Role in collecting and distributing human SMA and control tissues from surgical and autopsy sources as a resource for answering specific questions about SMA pathology
- 12/1/09 – 11/31/11 “Improving Care for Children with Muscular Dystrophies”
Dana and Albert Broccoli Foundation
100,000
Role: PI, 10% effort

Research Program Building / Leadership *None*

EDUCATIONAL ACTIVITIES

Educational Publications

Peer Reviewed, original, educational publications

1. **Crawford TO**, Clinical evaluation of the floppy infant. *Pediatr Ann* 1992;21:348-54.
2. **Crawford TO**, Pardo CA. The neurobiology of childhood spinal muscular atrophy. *Neurobio of Dis* 1996;3:97-110.
3. **Crawford TO**. From enigmatic to problematic: the new molecular genetics of childhood spinal muscular atrophy. *Neurology* 1996;46:335-40.
4. Chance PF, Ashizawa T, Hoffman EP, **Crawford TO**, Molecular basis of neuromuscular diseases., *Phys Med Rehabil Clin N Am* 1998;9:49-81.
5. **Crawford TO**. Concern about the design of clinical trial for SMA. *Neuromuscul Disord* 2004;14:456-460.
6. Wang CH, Finkel RS, Bertini ES, Schroth M, Simonds A, Wong B, Aioysius A, Morrison L, Main M, **Crawford TO**, Treja A. Consensus statement for standard of care in spinal muscular atrophy. *J Child Neurol* 2007;22:1027-1049.

7. Swoboda KJ, Kissel JT, **Crawford TO**, Bromberg MB, Acsadi G, D'Anjou G, Krosschell KJ, Reyna SP, Schroth MK, Scott CB, Simard LR. Perspectives on clinical trials in spinal muscular atrophy. *J Child Neurol* 2007;22:957-66.
8. Burnett BG, **Crawford TO**, Sumner CJ. Emerging treatment options for spinal muscular atrophy. *Curr Treat Options in Neurol* 2009;11:90-101.
9. Gitlin JM, Fischbeck K, **Crawford TO**, Cwik V, Fleischman A, Gonye K, Heine D, Hobby K, Kaufmann P, Keiles S, MacKenzie A, Musci T, Prior T, Lloyd-Puryear M, Sugarman EA, Terry SF, Urv T, Wang C, Watson M, Yaron Y, Frosst P, Howell RR. Carrier testing for spinal muscular atrophy. *Genet Med*, 2010; 12:621-22.
10. McGrath-Morrow, SA, Gower WA, Rothblum-Oviatt C, Brody AS, Langston C, Fan LL, Lefton-Greif MA, **Crawford TO**, Troche M, Sandlund JT, Auwaerter PG, Easley B, Loughlin GM, Carroll JL, Lederman HM. Evaluation and Management of Pulmonary Disease in Ataxia-Telangiectasia. *Pediatr Pulmonol* 2010; 45:847-859.

Invited Reviews

1. **Crawford TO**. Ataxia-telangiectasia. *Sem Ped Neurol* 1998;5:287-294.

Editorials

1. Mitchell WG, **Crawford TO**. Lorazepam is the treatment of choice for status epilepticus. *J Epilepsy* 1990;3:7-10.

Case Reports

1. Ruderman JW, Pomerance JJ, Hodgman JE, **Crawford T**. Congenital septicemia due to *Pseudomonas aeruginosa*. *Clin Pediatr (Phila)* 1983;22:630-642.
2. Zions LE, Osterkamp JA, **Crawford TO**, Harvey JP Jr. Congenital annular bands in identical twins. A case report. *J Bone Joint Surg* 1984;66:450-3.
3. Mitchell WG, **Crawford TO**. Intraventricular alpha 2b interferon for SSPE. *Neurology* 1987;37:1884.
4. Hsu CD, Feng TI, **Crawford TO**, Johnson TR, Unusual fetal movement in congenital myotonic dystrophy. *Fetal Diagn Ther* 1993;8:200-2.
5. Mogayzel PJ, Colombani PM, **Crawford TO**, Yang SC. Bilateral diaphragm paralysis following lung transplantation and cardiac surgery in a 17 year old. *J Heart Lung Transplant* 2002;21:710-712.
6. Choo-Kang LR, Ogunlesi FO, McGrath-Morrow SA, **Crawford TO**, Marcus CL. Recurrent pneumothoraces associated with nocturnal noninvasive ventilation in a patient with muscular dystrophy. *Pediatr Pulmonol* 2002;34:73-78.
7. Arav-Boger R, **Crawford T**, Steere AC, Halsey NA. Cerebellar ataxia as the presenting manifestation of Lyme disease. *Pediatr Infect Dis J* 2002;21:353-356.
8. Wachtel LE, **Crawford TO**, Dhossche DM, Reti IM. Electroconvulsive therapy for pediatric malignant catatonia with cerebellar dysgenesis. *Ped Neurol* 2010;43:427-30.
9. Wagner MW, Poretti A, Wang T, **Crawford TO**, Huisman TAGM, Bosemani T. Susceptibility-weighted imaging in Cockayne syndrome. *J Pediatr* 2014;165:416.
10. Wolf DS, Golden WC, Hoover-Fong J, Applegate C, Cohen BA, Germain-Lee EL, Goldberg MF, **Crawford TO**, Gauda EB. High-dose glucocorticoid therapy in the management of seizures in neonatal incontinentia pigmenti: A case report. *J Child Neurol* 2015;30:100-6

Letters

1. **Crawford TO**, Skolasky RL. The Relationship of *SMN* to ALS (letter) *Annals of Neurology* 2002;52:857-858.
2. **Crawford TO**, Comi A, Freeman JM, Kossoff EH, Singer H, Vining EPG, Yohay K. Re: *Practice Parameter: Evaluation of the child with global developmental delay*, *Neurology* 2003;61:1315.

Books Edited

1. Singer HS, Kossoff EH, Hartman AL, **Crawford TO** eds. *Treatment of Pediatric Neurologic Disorders*. Taylor and Francis Group, Boca Ratan Florida 2005.

Book Chapters

1. Kuncel RW, **Crawford TO**, Rothstein JD, Drachman DB: Motor neuron diseases. In: Asbury AK, McKhann GM, McDonald WI eds: *Diseases of the Nervous System*, Second Edition. W.B. Saunders Co, Philadelphia PA., 1991.
2. Griffin JW, Hoffman PN, **Crawford TO**: Pathophysiology of neuronal and axonal degenerations. In: Asbury AK, McKhann GM, McDonald WI eds: *Diseases of the Nervous System*, Second Edition. W.B. Saunders Co, Philadelphia PA., 1991.
3. **Crawford TO**, Respiratory failure and other complications of neuromuscular disease. In: Holbrook, PR ed; *Textbook of Pediatric Critical Care*. W.B. Saunders, Co. Philadelphia PA, 1993.

4. **Crawford TO**, Thompson JD: Familial neuropathies. In: Johnson RT, Griffin JW, eds: *Current therapy in neurologic disease*, Fourth edition. Mosby-Year Book, Inc. St Louis, Missouri. 1993.
5. **Crawford TO**. Neuromuscular disorders. In: Capute AJ, Pasquale PJ, eds: *Neurodevelopmental Disabilities in Infancy and Childhood*, Second Edition. Paul H Brookes Publishing Co. Baltimore, Maryland 1996.
6. Drachman D, **Crawford TO**. Major muscular dystrophies. In: Johnson RT, Griffin JW, eds: *Current Therapy in Neurologic Disease*, Fifth edition. Mosby-Year Book, Inc. St Louis, Missouri. 1996.
7. Griffin JH, **Crawford TO**, McArthur JC: Peripheral neuropathies associated with HIV infection. In Gendelman HE, Lipton SA, Epstein L, Swindells S, eds: *The Neurology of Aids*, Chapman & Hall, New York, NY. 1998.
8. **Crawford TO**. The Floppy Infant. In Bradley WG, Daroff RB, Fenichel GM, Marsden CD, eds: *Neurology in Clinical Practice*, Butterworth-Heinemann, Philadelphia, PA 1999.
9. **Crawford TO**. Spinal Muscular Atrophy: Current Research and Therapy. In Kuncel RW: *Motor Neuron Disease*. WB Saunders, Inc. Philadelphia, PA. 2002.
10. **Crawford TO**, Jones HR. The spinal muscular atrophies and other disorders of the anterior horn cell. In Brown WF, Bolton CF, Aminoff MJ, eds: *Clinical Neurophysiology and Neuromuscular Diseases*, chapter 87. W.B. Saunders, Co. Philadelphia PA 2002.
11. **Crawford TO**. Spinal Muscular Atrophies. In Jones HR, De Vivo DC, Darras BT eds: *Neuromuscular Disorders of Infancy, Childhood and Adolescence: A Clinician's Approach* Chapter 8 Pg 145-166 Butterworth Heinemann, Philadelphia, PA 2003.
12. **Crawford TO**. Infant Botulism. In Jones HR, De Vivo DC, Darras BT eds: *Neuromuscular Disorders of Infancy, Childhood and Adolescence: A Clinician's Approach* Chapter 32 Pg 547-554 Butterworth Heinemann, Philadelphia, PA 2003.
13. **Crawford TO**. The Floppy Infant. In Bradley WG, Daroff RB, Fenichel GM, Jankovic J, eds: *Neurology in Clinical Practice*, Butterworth-Heinemann, Philadelphia, PA 2003.
14. Gatti RA, **Crawford TO**, Mandir AS, Perlman S, Mount HTJ. Ataxia Telangiectasia. In Beal MF, Lang AE, Ludolph A eds: *Neurodegenerative Diseases: Neurobiology Pathogenesis and therapeutics*. Cambridge University Press, Cambridge, 2005.
15. **Crawford TO**. Spinal Muscular Atrophies and Other Disorders of the Anterior Horn Cell. In Holmes GL, Moshé S, Jones HR. *Clinical Neurophysiology of Infancy, Childhood and Adolescence*. Elsevier 2005.
16. Gatti RA, **Crawford TO**, Mandir AS, Perlman S, Mount HTJ. Ataxia telangiectasia. In: Beal MF, Lang AE, Ludolph A eds: *Neurodegenerative diseases: Neurobiology, pathogenesis and therapeutics*. Cambridge University Press, Cambridge 2005.
17. **Crawford TO**, Shiloh Y. Ataxia Telangiectasia. In Brice A, Pulst S-M eds: *Spinocerebellar degenerations: the ataxias and spastic paraplegias*. Butterworth Heinemann Elsevier, Philadelphia 2007.
18. Sponseller PD, **Crawford TO**. *Neuromuscular disorders in Pediatric Orthopedics*. In Sponseller PD, ed Handbook of Pediatric Orthopedics, 2nd Edition. Thieme Medical Publishers, New York 2011.
19. Sakonju A, **Crawford TO**. *Acquired Presynaptic Neuromuscular Junction Disorders*. In Darras BT, Jones HR, Ryan MM, De Vivo DC eds: *Neuromuscular Disorders of Infancy, Childhood and Adolescence: A Clinician's Approach*. 2nd Ed. Chapter 25 Pg 445-455. Elsevier, San Diego CA 2015.

Other Media

1. **Crawford TO**. Infant botulism. In *Neurobase*. Arbor Publishing Co, La Jolla 1994, revised 1997 (CD ROM).
2. Lederman H, **Crawford TO**, eds, Ataxia-Telangiectasia. Handbook for families and caregivers. On line publication at the A-T Children's Project, 1st ed, 2002. www.atcp.org/Handbook%20TOC.htm

Other

1. Featured on and off-screen commentator on 6 videos for instructional course in palliative medicine, *A Vision of Hope: Integration of Palliative Care in Chronic Pediatric Disease*, Project coordinators Rushton C, Geller G, film producer Nigel Noble, funded by HHS (1RC1NR011710-01) and Starvos Niarchos Foundation.

Illustrations

1. Seidel M. Epic Geography, James Joyce's *Ulysses*. (Illustrations by **Crawford TO**) Princeton University Press: Princeton, New Jersey 1976.

Teaching

Classroom Instruction

- 1992 - present Annual participation in 2nd Year Medical Student Neurology/Neuropathology Course (with new curriculum, in 2010 evolved into the Neurology and Special Senses course for first year medical students)
- 1995 - 2008 Annual participation in Neurology Clinical Skills Course
- 2009 - present Director, Neurology Clinical Skills Course

CME Instruction (selected)

- 10/92 Symposium Director, Neuromuscular Disorders, Child Neurology Society

- 10/95 *Brachial Plexus Palsy*: Trends in Pediatrics course, Johns Hopkins
10/96 *Spinal Muscular Atrophy*: Trends in Pediatrics course, Johns Hopkins
10/97 *Evaluation of the Floppy Infant*: Trends in Pediatrics course, Johns Hopkins
10/99 *Ataxia Telangiectasia*: Neurogenetics for the Practitioner, NYU Post-Graduate CME; New York, NY
6/00 *Guillain Barré Syndrome; Neuropathies in Children, Ataxia Telangiectasia, and Spinal Muscular Atrophy*: Current Concepts in Child Neurology, Children's Hospital and Health Center; San Diego, CA
6/01 *Ataxia Telangiectasia*: American Academy of Neurology 53rd Annual Meeting; Philadelphia, PA.
10/01 *Point-Counterpoint: Therapy Services for Developmental Disorders*: Trends in Pediatrics course, Johns Hopkins
10/02 *Update on Neuromuscular Disorders*: Trends in Pediatrics course, Johns Hopkins
10/02 *Neuromuscular Diseases of Children: From Clinical Evaluation to the Genome*: American Academy of Pediatrics; Boston, MA
10/03 *Developmental Delay*: Trends in Pediatrics course, Johns Hopkins
10/05 *Rational Path to Diagnosis of the Floppy Infant*: Child Neurology Society
5/05 *Interpreting the data after an injury: Neurophysiology*. Exploring shoulder dystocia and brachial plexus injury. Multidisciplinary Perspectives. Johns Hopkins CME
10/06 *Spinal Muscular Atrophy: Neurogenetics for the Practitioner* NYU Post-Graduate CME; New York, NY
2009 *Potential Treatments for Spinal Muscular Atrophy*: Muscular Dystrophy Association Clinic Directors' Conference. Las Vegas, NV
2011 *Advances in Therapeutic Development*. Recent Advances in Pediatric Neuromuscular Disease: Spotlight on Spinal Muscular Atrophy (CDC-sponsored continuing education course), Denver, CO
2013 *Overview of Common NM Disorders and Effects on Respiratory Function*: Pulmonary Care of Children with Neuromuscular Disease, American Thoracic Society Annual Meeting, Philadelphia, PA
2014 *Neurophysiology 101*: American Association of Neurologic Surgeons, Baltimore, MD

Non-CME Educational Lectures (Patient Groups)

- 1994- present Families of Spinal Muscular Atrophy yearly family conference, at each a 2-day series of talks and group discussions on SMA basic science and clinical care topics to patients and families
4/00 Ataxia Telangiectasia Family Conference, A-T Children's Project, Chicago, Illinois
5/07 Ataxia Telangiectasia Family Conference, A-T Society, Nottingham, England
5/08 Ataxia Telangiectasia Family Conference, BraShA-T, Brisbane Australia
5/09 Ataxia Telangiectasia Society, Birmingham, U.K.

Other Instructional and Service Activities

- 1992 - Present Medical Assistance to Yearly MDA camp in St Mary's County
1998 - Present local MDA Labor Day Telethon assistance including on-air interview
2003 - Present Regular Contributor to FSMA "Ask the Experts" Web Page
2008 Discussion on SMA Research with Jerry Lewis (Prepared for MDA National Telethon)

Workshops/Seminars

- 2013 *Becker Muscular Dystrophy Conference*. Chair Tom Crawford, sponsored by Muscular Dystrophy Association. Johns Hopkins Hospital, Baltimore, MD.

Educational Program Building / Leadership *None*

Educational Extramural Funding *None*

CLINICAL ACTIVITIES

Certification

Medical, other state / government licensure:

- 1980 - 1987 California
1987 - present Maryland (00037130)

Boards, other specialty certifications:

- 1978-81 National Board of Medical Examiners, Part I (13 June, 1978), Part 2 (9April, 1980), Part 3 (20 May, 1981); USMRE# 32353864, NBME # 235386
1986 Diplomat American Board of Pediatrics
1990 Diplomat American Board of Psychiatry and Neurology with Special Qualification in Child Neurology (certificate #721)
1997 Diplomat American Board of Psychiatry and Neurology: Sub-specialty Certification in Clinical Neurophysiology (certificate #799)

Clinical Service Responsibilities

1989 - Child Neurology Clinic Attending (1-2 clinics/week)
1989 - MDA Clinic (1-2 clinics / week)
1989 - Child Neurology Ward Attending (2 months / year)
1996 - EMG Attending (1 clinic commitment)
1996 - 01 Child Neurology Clinic Attending, Mt Washington Pediatric Hospital (10% commitment)
2006 - Co-Director MDA Clinic (10% commitment)

Clinical Program Building / Leadership *None*

Clinical Extramural Funding *None*

SYSTEM INNOVATION AND QUALITY IMPROVEMENT ACTIVITIES *None*

ORGANIZATIONAL ACTIVITIES

Institutional Administrative Appointments *None*

Editorial Activities

Editorial Board Appointments

2003-2008 Neuromuscular Disorders

Journal Reviewer

1995-present Annals of Neurology
1996-present Neurology
1998-present Muscle and Nerve
1998-present Pediatric Neurology
2001-present Neuromuscular Disorders
2004 American Journal of Medical Genetics

Occasional: Journal of Pediatrics
Pediatrics
Proceedings of the National Academy of Sciences
Journal of Medical Genetics
Human Genetics
Journal of Pediatric Hematology/Oncology
Archives of Physical Medicine and Rehabilitation

Advisory Committees, Review Groups/Study Sections

1991 Scientific Selection Committee, Child Neurology Society
1992 Symposium Director, Neuromuscular Disorders Child Neurology Society
2000 – Present Medical Advisory Board, Families of Spinal Muscular Atrophy
2002 – Present Scientific Advisory Board, Families of Spinal Muscular Atrophy
2003 Wellstone Muscular Dystrophy Clinical Research Center (MDCRC) Scientific Review Panel, NIH
2003 – 05 Medical Advisor, SMA Foundation
2004 Molecular, Cellular and Developmental Neurosciences Special Emphasis Panel (MDCN-A), NIH
2004 - 2008 Senator Paul D. Wellstone Muscular Dystrophy Cooperative Research Centers Data Safety and committee
2004 - Present Scientific and Medical Advisory Board, Claire Altman Heine Foundation, Inc.
2006 Special Consultant, Barth Syndrome Foundation
2006 SMA Standard of Care: FSMA, SMA Foundation, MDA sponsored Study Group Consultant
2006 - Present Muscular Dystrophy Association Medical Advisory Committee (2006 ad hoc member, standing member beginning 2007, meet twice yearly in March and October)
2007 *Ad hoc* reviewer, Congressionally Directed Medical Research Program (CDMRP) for the US Army Medical and Material Command (USAMRMC)
2010 MDA Clinical Advisory Committee

Board Activities:

1994 - (Occasional) Examiner, American Board of Neurology and Psychiatry

Professional Societies

1990-1996 Fellow American Academy of Pediatrics
1985 - Present Fellow American Academy of Neurology
1987 - Present Fellow Child Neurology Society
1987 - Present Society for Neuroscience
1997 - Present Peripheral Nerve Society
2006 - Present American Neurological Association

Consultantships

1993 Regeneron Pharmaceuticals: CNTF for SMA
1995 University of Michigan: Automated Computerized Morphometry of Nerve
1997 Fujisawa Pharmaceutical Co, Ltd., "A phase IIB study of the effects and safety of FK366 in patients with diabetic neuropathy," Teased fiber analysis of nerve biopsies
1997 Jacobus Pharmaceuticals: 3,4 DiAminopyridine for Congenital Myasthenic Syndromes
2005 CepTor corporation C101 Phase I/II DMD Protocol Development
2006-9 BG Medicine / SMA Foundation Biomarker Scientific Advisory Board
2009 Repligen Corporation, special consultant
2012 Genzyme: special consultant

RECOGNITION

Awards and Honors

1975 John C. Schroeder Award, Yale College
1978 Award for Special Support, USC School of Medicine Minority Medical Students
2001 Frank Ford Award, for excellence in teaching, Department of Neurology
2006 Fellow, American Neurological Association
2012 Neurology Core Clerkship Clinical Faculty Brain Award for Excellence in Teaching
2013 A.B. Baker Teacher Recognition Certificate, American Academy of Neurology
2013 Frank Ford Award, for excellence in teaching, Department of Neurology

Invited Talks, Panels

Visiting Professorships

10/99 Department of Neurology, New Jersey Medical School, Newark, NJ
4/03 Department of Neurology, Vanderbilt University, Nashville, TN
1/06 Department of Neurology, Wayne State University, Detroit MI

Invited Lectures, International

10/95 Minneapolis, MN *SMA Genetics*: International SMA Consortium, MDA sponsored
11/97 Clermond-Ferand, France: *Development of a Scale for Assessing the Neurologic Impairment of A-T*. INSERM 7TH International Ataxia-Telangiectasia Workshop
9/99 Prague, Czech Republic: *Congenital Myasthenic Syndromes, New Developments in Spinal Muscular Atrophy*: International Conference of EMG and Clinical Neurophysiology
10/00 Abu Dhabi, United Arab Emirates: *Neuromuscular Diseases of Children*. Grand Rounds, Emirates Palomar Medical Technology Services
5/01 Nottingham, England: *Nosology and Semiology of A-T*: Ataxia Telangiectasia Society, Nottingham, England
1/02 Naarden, Netherlands: *Therapy for Spinal Muscular Atrophy*: 103rd European Neuromuscular Centre, Naarden, the Netherlands
9/03 Frasier Island, Brisbane, Australia: *Insights into pathogenesis from the clinic*. International Ataxia Telangiectasia Workshop
2/04 Tel Aviv, Israel: *The Neurology of Ataxia Telangiectasia*: Featured speaker at opening of the clinical center for Association for Fighting A-T Disease
9/04 Göteborg, Sweden: *Specific Therapies for SMN-Associated SMA*: World Muscle Society
10/04 Birmingham, England: *Features of Cerebellar and Extrapyrmidal Impairment in A-T*, and *Insights into the Pathogenesis of A-T from Long-term Neurologic Follow Up*. International Conference on the Clinical Science of Ataxia Telangiectasia. University of Birmingham
7/05 Lake Maggiore, Italy: *Development, Degeneration and Downstream Damage; the Neurology of Ataxia Telangiectasia*. The 2005 International Workshop on Ataxia Telangiectasia, ATM and the DNA Damage Response
10/05 London, England: *Neuromuscular Transmission Defects*. 5th International EMG Conference, Improving the Use of Electromyography in Paediatrics

- 4/06 Nijmegen, Netherlands: *Neurologic Problems in Children with Ataxia Telangiectasia*. Workshop Nijmegen Breakage Syndrome and Ataxia Telangiectasia
- 6/06 Montreal, Canada: *Advances in Acute and Chronic Inflammatory Neuropathies*. 10th International Child Neurology Congress
- 9/06 Banff, Canada: *Neurology of Ataxia Telangiectasia*. 12th International Ataxia Telangiectasia Workshop
- 9/06 Edinburgh, Scotland: *The Role of EMG in the diagnosis of the Spinal Muscular Atrophies*. International Congress of Clinical Neurophysiology
- 10/07 London, England: *Toxic Neuropathy; What's Happening in SMA*. 6th International EMG Conference, Improving the Use of Electromyography in Paediatrics
- 10/07 Quebec City, Quebec: *Ataxia Telangiectasia*. Child Neurology Society Symposium, Movement disorders at the interface between bench and bedside
- 4/08 Otsu, Japan: *Neurology Goings-On from the Hopkins A-T Clinical Center*. 13th International Ataxia Telangiectasia Workshop
- 5/08 Brisbane, Australia: *Ataxia Telangiectasia; Spinal Muscular Atrophy; The Genetics of Neuromuscular Disease*. (Keynote address and affiliated lectures). Australia-New Zealand Association of Neurology
- 5/08 Brisbane, Australia: *The Neurology of Ataxia Telangiectasia*. Ataxia-Telangiectasia Forum
- 5/08 Trabzon, Turkey: *Hereditary Ataxias, with specific focus on Ataxia Telangiectasia* (Keynote lecture); and *SMA and the SMAs*. X. USUAL ÇOCUK NÖROLOJİSİ KONGRESİ
- 5/09 Birmingham, England: *Assessment of Ataxia Telangiectasia: Clinical Scale Development*. Ataxia Telangiectasia Society Annual Conference
- 10/11 Frankfurt, Germany: *Plenary Address: Clinical Science of Ataxia Telangiectasia: Status Quo and Future Directions*. European A-T Workshop
- 1/12 London, United Kingdom: *Ataxia Telangiectasia*. Keynote address, British Pediatric Neurology Association
- 2/12 New Delhi, India: *Update on neurologic investigations at the Johns Hopkins A-T clinical Center*. 14th International Workshop on Ataxia-Telangiectasia ATW2012
- 6/12 Cambridge, United Kingdom: *Instrumented and non-instrumented assessments*. A-T Clinical Research Conference
- 7/13 Birmingham, U.K.: *What is Static, What is Degenerative and how can we measure each?* International Ataxia Telangiectasia Workshop 2013.

Invited Lectures, National

- 3/98 Bethesda, MD: *The Neurologist's Contribution*. Joint NIH, A-T Children's Project Sponsored Conference on Ataxia Telangiectasia
- 4/99 Bethesda, MD: *Spinal Muscular Atrophy: What Next?* Joint conference sponsored by Andrew's Buddies and NINDS
- 9/99 Las Vegas, NV. Eighth International Workshop for Ataxia Telangiectasia Chairman Workshop on Phenotypes, genotypes and immune status
- 10/99 Newark, NJ: *Ataxia Telangiectasia*. Neurology Grand Rounds, New Jersey Medical School
- 6/00 Columbus, OH: *The Neurology of Spinal Muscular Atrophy*. Spinal Muscular Atrophy Diagnosis, Management and Novel Findings Providing Insight for Treatment Conference, Children's Hospital of Columbus
- 9/00 Rancho Sante Fe, CA: *Summary of Neural Pathogenesis*. Molecular Neurobiology of ATM Scientific Workshop, A-T Children's Project
- 11/00 Cold Springs Harbor, NY: *The Neurologist's Agenda for Research*. Molecular Neurobiology of ATM, Bambury Conference Series
- 7/01 Tareyton, NY: *Non-Cerebellar Neurologic Deficits in A-T: The role of DNA damage response defects in Neurodegenerative Diseases*, A-T Children's Project
- 9/01 Albuquerque, New Mexico: *Spinal Muscular Atrophy*: American Association of Electrodiagnostic Medicine Annual Meeting
- 5/02 Boston, MA: *Congenital Cerebellar Abnormalities in Ataxia Telangiectasia*: Ataxia Telangiectasia International Meeting
- 4/03 Nashville, TN: *Spinal Muscular Atrophy*, Neurology Grand Rounds, Vanderbilt University
- 10/03 Baltimore, MD: *Children's Neuromuscular Disorders*: Neurology Grand Rounds, University of Maryland,
- 3/04 Cold Spring Harbor, NY: *Natural history of SMA and the Design of Clinical Trials*: Spinal Muscular Atrophy: What is the molecular basis of neuronal loss? Bambury Conference Series
- 10/04 Washington, D.C.: *Ataxia Telangiectasia* Georgetown University, Department of Neurology Grand Rounds
- 3/06 Bethesda, MD: *Extra-Cerebellar Characteristics of A-T*. NINDS sponsored Ataxia Telangiectasia Clinical Research Workshop
- 10/06 Washington D.C.: *Spinal Muscular Atrophy and the Spinal Muscular Atrophies*. American Association of Neuromuscular and Electrodiagnostic Medicine
- 9/07 Rochester NY: *Trial of Losartan for Duchenne Muscular Dystrophy*. Muscle Study Group
- 9/07 Rosemont, IL: *Comparison and Development of Quantitative Neurologic Endpoints*. NIH and A-T Children's Project sponsored Neurology Workshop for Ataxia Telangiectasia, Comparison and Development of Neurologic Endpoints

- 3/08 Las Vegas, NV: *Ataxia Telangiectasia*. National Ataxia Foundation Meeting
- 5/08 San Diego, CA: *Differential Diagnosis of the Hypotonic Infant Presenting for Muscle Biopsy*. Symposium on Malignant Hyperthermia
- 5/08 Minneapolis, MN: *Spinal Muscular Atrophy*. Inaugural Pediatric Neurosciences Conference, Gillette Children's Hospital
- 1/09 Las Vegas, NV: *Potential therapies for spinal muscular atrophy*. MDA Clinic Directors Meeting
- 10/09 Washington, D.C.: *Evaluating the child with muscle weakness*. American Academy of Pediatrics National Conference & Exhibition
- 12/09 Baltimore, MD: *Spinal muscular atrophy: A ten million, one hundred, ~ ten, ~one, and 0.1 year perspective*. Johns Hopkins Neurology Grand Rounds
- 4/10 Charleston, MD: *Keynote Address: The Floppy Infant, and one special story*. Pediatric Neuroscience Update, Medical University of South Carolina
- 4/10 Redondo Beach, CA: *Ataxia Telangiectasia: developmental and degenerative features*. International Ataxia Telangiectasia Workshop
- 9/10 Washington, D.C.: *State of the Art in Biomarker Development in Spinal Muscular Atrophy*. NIH/FDA Conference on Antisense Oligonucleotide Therapies in Neuromuscular Disease
- 10/10 San Francisco, CA: *Neuropathology and Semiology of Ataxia Telangiectasia: Neurocircuitry of Ataxia Telangiectasia*. Ataxia Telangiectasia Children's Project Special Conference
- 10/10 Washington, D.C.: *Proof-of-Concept Trial Design and Conduct, Clinical Trial Feasibility and Conduct Issues for SMA*: NIH Therapy Development Workshop in SMA
- 11/11 Savannah, GA: *Ataxia Telangiectasia*. Neurobiology of Disease in Children Symposium, Childhood Ataxia and the CNS
- 11/12 Washington, D.C.: *Challenges in Drug Discovery for Ataxia Telangiectasia*: Neurosciences and Drug Discovery Forum.
- 4/13 Baltimore MD: *Spinal Muscular Atrophy*. Johns Hopkins Department of Obstetrics and Gynecology Grand Rounds
- 6/13 Anaheim, CA.: *Outcome measures comparing mice and human*. FSMA National Conference
- 11/13 Baltimore, MD: *Ataxia Telangiectasia*. Johns Hopkins Neurology Grand Rounds
- 10/14 Hartford, CT: *Ataxia Telangiectasia*. University of Connecticut/Connecticut Children's Medical Center Neurology Grand Rounds and visiting professorship.
- 1/15 Baltimore, MD: *Spinal Muscular Atrophy: A Multidimensional and Very Cool Story*. St Agnes Pediatrics Grand Rounds.

Professional Organization Courses

- 6/11 Orlando, FL: *From Mouse to Man: How Relevant are Findings in SMA Models?* International Conference on SMA (FSMA sponsored)