CURRICULUM VITAE
John West Day, MD, PhD

BUSINESS ADDRESS

Professor of Neurology
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Stanford University School of Medicine
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Stanford, CA 94305
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EDUCATION

1969 – 1973 BA, Physics, Oberlin College, Oberlin, Ohio
1974 – 1977 M.D., Medicine, University of Minnesota, Minneapolis, Minnesota
1977 – 1982 Ph.D., Neuroscience, Albert Einstein College of Medicine, Bronx, New York
M.V.L. Bennett, Thesis Advisor

POSTDOCTORAL TRAINING

1982 – 1983 Montefiore Hospital, Bronx, New York Internship in Internal Medicine
1983 – 1986 University of California, San Francisco Resident in Neurology
1985 – 1986 Chief Resident of Neurology, Robert A. Fishman, Department Chairman
1986 – 1988 University of California, San Francisco, California
Clinical Neurophysiology/Electromyography, Richard Olney, Fellowship Director

ACADEMIC APPOINTMENTS

1986 – 1991 Department of Neurology, University of California, San Francisco, Assistant Professor
1992 – 1997 Department of Neurology, University of Minnesota, Assistant Professor
1996 – 2011 Graduate Program in Neuroscience, University of Minnesota, Full Member
1997 – 2001 Department of Neurology, University of Minnesota, Associate Professor
2002 – 2011 Department of Neurology, University of Minnesota, Professor
2006 – 2011 Department of Pediatrics, University of Minnesota, Professor
2008 – 2011 Department of Genetics, Cell Biology, Development, University Of Minnesota, Professor
2011 – Department of Neurology, Stanford University, Professor (Acting)

ACADEMIC ADMINISTRATIVE APPOINTMENTS

1996 – 2003 Director, Center for Muscle Disorders, University of Minnesota
1997 – 2003 Medical Director, Clinical Neuroscience Research Unit
1999 – 2001 Associate Head for Clinical Affairs, Neurology Department, U of MN
1999 – 2011 Institute of Human Genetics, Executive Board, University of Minnesota
2003 – 2011 Director, Paul and Sheila Wellstone Muscular Dystrophy Center, U of MN

CLINICAL/HOSPITAL APPOINTMENTS

1994 – 2011 Director, Muscular Dystrophy Clinic, University of Minnesota
1996 – 1999 Director of Clinical Electrophysiology Laboratories, University of Minnesota

1997 – 2011 Medical Director, Neuromuscular Histology Laboratory
1999 – 2011 Medical Director, Muscular Dystrophy and ALS Clinic, University of Minnesota
1999 – 2003 Medical Director, University of Minnesota Ataxia Clinic
1999 – 2001 Chief of Service, Neurology Service, Fairview University Medical Center
2005 – 2011 Director, Neuromuscular Clinic, Gillette Pediatric Specialty Care
2011 – Director, Neuromuscular Program, Stanford University School of Medicine

CONSULTING AND ADVISORY POSITIONS

1996 – 2011 Myasthenia Gravis Foundation, Minnesota Chapter, Medical Advisory Board
1998 – 2001 Muscular Dystrophy Association, Minnesota Chapter, Board Member
2002 – 2008 National Ataxia Association, Board Member
2004 – 2008 National Ataxia Association, Clinic-Research Liaison
2004 – 2010 Muscular Dystrophy Association, Medical Advisory Committee
2005 Muscular Dystrophy Advisory Committee, NIH/NINDS/NIAMS
2005 – 2007 PTC Therapeutics, Chairman PTC124 Data Management Review Committee
2005 – current United Council for Neurological Subspecialties, Neuromuscular Path Adv Committee
2006 – 2008 Asklepios Biotherapeutics, Duchenne Gene Therapy Advisory Committee
2006 – current Myotonic Dystrophy Foundation, National Advisory Committee
2008 – 2011 NIH-CINCH, Chairman Data and Safety Management Board
2008 – 2011 NIH Genetics of Health and Disease (GHD) Study Section Member
2009 NIH/NIAMS Advisory Committee on Muscular Dystrophy Research
2009 – 2011 Chair, NIH DSMB, CINCH cooperative mexilitene trial for non-dystrophic myotonia
2010 – current Amer Acad of Neurology, Guidelines Committee for myotonic dystrophy treatment
2011 Chair, NIH DSMB, International trial of prednisone for Duchenne muscular dystrophy

SCIENTIFIC ADMINISTRATIVE AND ORGANIZATIONAL ACTIVITIES

Journal Referee: Annals of Neurology, Brain Research, Muscle and Nerve, Neurology, Pediatric Neurology, Journal of Neurology, Neurosurgery and Psychiatry, Journal Neuroscience, Human Molecular Genetics, American Journal Human Genetics
1994-1996 Abstract Reviewer, American Academy of Neurology Annual Meeting
1994-1995 Chair: Pathogenesis of ALS, American Academy of Neurology
1996 Course Director: American Association of Electrodiagnostic Medicine
2004-current Scientific Advisory Committee, National Registry for DM and FSHD
2005 Section Co-Chairman, NIH Burden of Muscle Disease Workshop, Washington, D.C.
2005 Organizer, Director, First International Ataxia Investigators' Meeting, Tampa, FL
2008 Ad Hoc Member, NIH Genetics of Health and Disease Study Section
2008 Organizer, Director, 2nd International Ataxia Investigators' Meeting, Las Vegas, NV

GRANT SUPPORT

Source	Period	Title
NIH	7/78-6/81	Iontophoretic Study of Hatchetfish Giant Synapse
NIH	7/86-6/89	Molecular Basis of Opiate Receptor Physiology
Epilepsy Foundation	7/89-6/90	Cellular Physiol of Epilepsy-Permissive Neurons
Univ. MN Grad. School	7/95-6/96	Characterization of a Form of Myotonic Dystrophy
U MN Grad Sch, Dir.	7/96-6/98	Interdisciplinary Center for Muscle Disease
NIH, Co-Inv.	7/97-6/00	Genetic Mapping of a Novel Disease
NIH, Co-I	6/1/97-5/31/00	Cloning and characterizing a second DM locus
Centeon Pharm., PI	12/96-11/99	Effects of Immunoglobulin on Myasthenia Gravis
FDA Orphan Drug, PI	10/1/97-9/30/00	Effects of Immunoglobulin on Myasthenia Gravis
MDA, Co-PI	1/1/98-12/32/00	Effects of Immunoglobulin on Myasthenia Gravis
U MN Grd Sch, Dir.	7/98-6/01	Interdisciplinary Center for Muscle Disease, Direct
MDA, Co-PI	12/98-11/01	Genetic and Clinical Characterization of DM2
NAF, Co-Inv	1/1/00-12/31/00	Murine model untranslated CTG repeats in SCA8
NAF, PI	1/1/00-12/31/00	Clinical and Genetic Characterization of SCA5
MMF, PI	4/1/00-9/30/01	MRI Brain Imaging in SCA8
NIH, Co-PI	4/1/00-5/31/12	Muscle Training Grant
NIH, Co-I	6/1/00-5/31/05	Genetic mapping of novel DM locus
NIH, Co-I	6/1/00-5/31/05	Molecular and Genetic Characterization of SCA8
ALS Assoc, PI	9/1/01-8/31/02	Genetic mapping of a novel form of ALS
ALS Assoc, PI	1/1/03-12/31/03	Genetic mapping of a novel form of ALS
MMF, PI	3/1/2003-2/29/2004	MRI and genetic characterization of SCA5
AHC, Co-PI	4/14/04-4/13/07	Novel genetic analyses of neuromuscular disorders
NASH Fund, PI	3/1/04-6/30/05	Development of Clinical Testing Program for DMD
MMF, PI	3/1/04-2/28/05	Clinical trials in Duchenne muscular dystrophy
DOD, PI	1/1/2005-12/31/06	Cord Blood Transplantation into Skeletal Muscle
NASH Fund, PI	3/1/04-6/30/05	Development of Metabolic Support for DMD
NIH, Co-I	6/1/05-5/31/09	Transgenic Model of DM2
NIH, Co-I	4/1/06-3/31/11	Clinical and Molecular Characterization of SCA8
NASH Found, Co-I	1/1/2007-1/1/2009	TAT-Utrophin treatment of DMD
NIH, R01 PI	4/1/07-3/31/12	CNS Involvement in DM1 and DM2
Insmed, Site PI	3/1/2008-2/28//2009	IPLEX treatment of DM1
PTC, Site PI	3/1/2009-2/28/2009	PTC124 treatment of DMD point mutations
Genzyme, Site PI	2/1/2008-1/31/2007	Myozyme treatment of adult-onset Pompe
NIH, P01 Proj PI	4/1/08-3/31/13	CNS Involvement in Childhood DM1
NIH, PI (R13)	4/1/2008	International Ataxia Investigators' Meeting
NASH Found, PI	10/1/2007-9/30/2012	Cell-based treatment of DMD
MDA, PI	8/1/2008-7/31/2011	DMD Clinical Research Network
NIH P30, PI	6/1/2009-5/31/2014	MDCenter Core Laboratories
NIH, R01 Co-I	1/2007-12/2011	Dystrophin phenotype-genotype correlations
NIH , RC2 Co-I	9/2009-9/2011	Facioscapulohumeral dystrophy iPS cells
MDA, Site PI	6/2009-5/2012	Outcome measures in infants with Duchenne
MDA, Site PI	6/2009-5/2012	Duchenne cardiomyopathy natural history
PTC, Site PI	3/2008-2/2012	Open-label nonsense-mediated dystrophin study
Marigold Foundation	6/2011-5/2013	Executive function outcome measures for DM

CERTIFICATION AND LICENSURE

1984-1994; 2011 – Medical license, California, 52734
1988 Diplomat, American Board of Psychiatry and Neurology - Certificate 30137
1989 Diplomat, American Board of Electrodiagnostic Medicine - Certificate 543
1992 – current Medical license, Minnesota, 35127
1996 Diplomat, American Board of Psych & Neurol, special qualifications Clin Neurophysiol
1999 Diplomat, American Academy of Neurology, Qualification in Neuromuscular Pathology
2011 ACGME Neuromuscular Medicine Certification Exam

MILITARY SERVICE: None

HONORS AND AWARDS

1978 Grass Foundation Fellow in Neurophysiology, Marine Biological Lab, Woods Holes, MA
1978-1981 NIH/NRSA Postdoctoral Fellowship
1985 Alpha Omega Alpha
1985 Distinguished Teaching Award, University of California San Francisco
1986 Clinical Investigator Development Award, NINCDS, NIH
1996 Distinguished Teaching Award, University of Minnesota Medical School
2001 Distinguished Teaching Award, University of Minnesota Medical School
2002 Leon Poliachik Humanitarian Award, University of Minnesota ALS Clinic
2003 Distinguished Teaching Award, University of Minnesota Medical School
2005 Outstanding Teaching Award, University of Minnesota Medical School
2005 Distinguished Teaching Award, University of Minnesota Medical School
2007 All University Post-Baccalaureate Teaching Award, University of Minnesota
2007 Lifetime Member Academy of Distinguished Teachers, University of Minnesota
2010 Recognized among Best Physicians in Minnesota, Twin Cities Magazine

MEMBERSHIPS AND OFFICES IN PROFESSIONAL SOCIETIES

1978-current Society for Neuroscience
1978-current American Association for the Advancement of Science
1984-current American Academy of Neurology
1989-current American Association of Electrodiagnostic Medicine
2001-current American Association of Human Genetics
2001-current World Muscle Society
2006-curre nt American Neurological Association, Fellow

TEACHING AND MENTORING ACTIVITIES

STUDENT TEACHING:

1986,"Outpatient Neurology," Fourth Year Medical Students, University of California, San Francisco
1987-1991 "Introduction to Clinical Medicine/Neurology," Second Year Medical Students, UCSF
1988-1991 "Stroke," Physical Therapy Students, University of California, San Francisco,
1990-1991 "Headache and Pain," Clinical Pharmacology, Fourth Year Medical Students, UCSF
1991 "The Neurological Exam," 2nd Year Medical Students, UCSF
1992-2011 "Neuromuscular Disease," Second Year Medical Students, University of Minnesota
1993-2001 “Clinical Correlations - Myasthenia Gravis," First Year Medical Students, U of MN

1993-1996 "Coma" Internal Medicine Clerkship, University of Minnesota, twice yearly
1993-2011 "Pain and Pain Management" Second Year Medical Students, University of Minnesota
1993-2011 "Headache and Subarachnoid Hemorrhage" Second Year Medical Students, U of MN
1995 "Congenital myasthenia" Neuroscience Students Itasca Summer Program, U of MN
1992-1999 "Disorders of neuromuscular transmission" Neuroscience Grad Stud, U of MN
1992-1999 Site Coordinator, Neurology Clerkship University of Minnesota: Organize schedule and orient students for each 4 week rotation. Discuss expectations with faculty and help them plan tutorial sessions. Help organize and motivate house staff to participate in teaching. Grade exams and evaluate students each rotation.
1996-1999 Neurology Clerkship Director for all participating hospitals, University of Minnesota
1998-2011 Nervous System and Muscle Disease, Pathophysiology Course Director, principle 2nd Year Medical Student Clinical Neuroscience Course, U of MN
1996-1999 Director, "Introduction to Neuromuscular Junction", a 1-week laboratory course, Neuroscience Students Itasca Summer Program, University of Minnesota,
1997-2011 "Introduction to Neurology", OT/PT students, University of Minnesota,
1999 "A Clinical Approach to Pain" Human Behavior Course 1st Year Med Students U of MN
2000-2001 "The myotonic dystrophies", Biology 3700, University of Minnesota
2000-2011 "Introduction to muscle disorders", Biochemistry 5444 University of Minnesota
2000-2011 "Normal neuromuscular transmission", Biochemistry 5444 University of Minnesota
2000-2011 "Disorders of sarcolemmal ion channels", Biochemistry 5444 University of Minnesota
2000-2011 "Muscular dystrophies", Biochemistry 5444 University of Minnesota
2006-2010 "Introduction to Cranial Nerves", 1st year medical student Neuroscience Course, University of Minnesota
2011 "Genetic bases of muscular dystrophies", Biology 3700, University of Minnesota

RESIDENT TEACHING:

1985-1991 Neurology Resident Neuroscience Course in Neurophysiology, UCSF
1992 "Neurological Emergencies-Headache" Neurology Residents, U of MN
1992 "Neurological Emergencies-Acute Weakness" Neurology Residents, U of MN
1992-1995 Introduction to Electromyography", Neurology Resident 4 lectures, 4 times/yr, U of MN
1993 "Diseases of Muscle", Neurology Resident Neurosci, Symp Organizer/Lecturer, U of MN
1993-2004 "Introduction to Electromyography", PM&R Resident, U of MN
1993-1996 "Myasthenia Gravis", Ophthalmology Resident Education Series, U of MN
1994, 2002, 2004"Metabolic Encephalomyopathies", Neurology Resident Neuroscience Series U of MN
1994 "Neuromuscular Junction", Neurol Resident Neurosci Symp Organizer/Lecturer, U of MN
1994-2010 "The Neurological Exam", Neurology Resident Neuroscience Series, U of MN
1999, 2004 "Myopathies", Neurology Resident Neuroscience Series, U of MN (4 lectures)
1995, 1996, 1997 Intro to Neuromuscular Disorders, Medicine Resident Education, U of MN
1994, 1996, 1997 "Myopathies", PM&R Resident Education Series, U of MN
1996, 1997 "Neuromuscular Junction Abnormalities", PM&R Resident Education Series U of MN
1996-2008 "Introduction to Electromyography", Neurosurgery Resident Education Series, U of MN
2005-2008 "Basic Neurophysiology", Neurol Resident Neurosci Symp Organizer/Lecturer, U of MN

FELLOWSHIP TEACHING:

1986-1991 Neuromuscular Fellowship, Participating Faculty, UCSF
1991-2911 Neuromuscular Fellowship, Participating Faculty, U of MN

1996-2002 ACGME Neuromuscular/Clinical Neurophysiology Fellowship, Director, U of MN
2008-2010 Muscular Dystrophy Clinical/Research Fellowship, Director, U of MN

TEACHING CONFERENCES:
1985 "Thunderclap Headache - An Unruptured Cerebral Aneurysm" Neurology Grand Rounds, UCSF
1986 "Normocalcemic Tetany" Neurology Grand Rounds, UCSF
1989 "Electrophysiological Characterization of Spasticity" Neurology Grand Rounds, UCSF
1990 "Ectopic Impulse Generation and Ephaptic Transmission in Neuropathies" Neuro Grd Rds, UCSF
1991 "Adenosine Modulation of Transmission at the Neuromuscular Junction" Neuro Grd Rds, UCSF
1992 "Hypokalemic Periodic Paralysis" Clinical Neurosciences Conference, U of MN
1993 "Multifocal Demyelinating Neuropathies", Neuromuscular Conference, U of MN
1993 "Radiation Amyotrophy", Neuromuscular Conference, U of MN
1994 "Periodic Paralysis - Quantitative Assessment", Neuromuscular Conference, U of MN
1994 "Mitochondrial Myopathies – Clinical/Basic Correlations" Clinical Neurosciences Conf, U of MN
1995 "Temporal Aspects of Conduction Block", Clinical Neurosciences Conf, U of MN
1997 "Paraneoplastic Disorders", Clinical Neurosciences Conference, U of MN
2000 "Spinocerebellar Ataxia type 8", Clinical Neurosciences Conference, U of MN
2001 "Myotonic Dystrophy type 2", Clinical Neurosciences Conference, U of MN
2001 "Myotonic Dystrophy type 2", Department of Neuroscience Colloquium, U of MN
2002 "Myotonic Dystrophy type 2", Dept of Gen, Cell Biol, Dev. Seminar Series, U of MN
2003 "Myotonic Dystrophy type 2", Department of Neurology Seminar Series, U of MN
2004 "Myotonic Dystrophy type 2", Department of Psychiatry Seminar Series, U of MN
2005 "The Myotonic Dystrophies", Department of Internal Medicine Grand Rounds, U of MN
2005 "Developmental Defects in Myotonic Dystrophy", Dept. of Pediatrics Grand Rounds, U of MN
2004 "Novel degenerative disease: FSHD", Department of Neurology Seminar Series, U of MN
2005 "Duchenne Muscular Dystrophy Treatment", Department of Neurology Seminar Series, U of MN
2006 "Myotonic Dystrophy and the Central Nervous System", Clinical Neurosciences, U of MN
2008 "Management of Muscular Dystrophy Patients", Gillette Healthcare Neuroscience Conference
2010 "Congenital Muscular Dystrophies", Gillette Healthcare Neuroscience Conference
2011 "Genetics of Muscular Dystrophy", Human Genetics Course, U of MN

MEDICAL STUDENTS:
Medical Student Advisees:
1992: Thanh Dang
1999: William Malcolm, Melissa Mark
2000: L.T. Nguyen
2002: Joshua Ramsmeyer
2004: Kristina E Catrine, Brian A Hunter, Amanda J Morehouse, Derek Johnson,
2005: Sarah Powel
2006: Manish C. Champaneria, Catherine Ehlen, Jonathan Faust, Brooke Glessing, Barton Iverson, Mollie Johnston, Justine Pidcock, Adrienne E. Soucy, Adam Todd
2007: Thuy An Hoang, Danah M Holman, Matthew W Miller, Sara A Rademacher, Laura C.Speltz, Christopher M Walker

2009: Elisabeth Hurliman, John Cunningham, Nancy Henry-Socha, Yi-Lo Yu, Stephanie Aamodt, Maureen Handoko, Angela Hewitt, Cristina Albott
2010 Mohammad Gol, Roy Bryan Jr., Jose Mejia, Yun Lin

GRADUATE STUDENTS & POSTDOCS:

Graduate Students:

1995-1999 Jon Falkenberg (with Iaizzo, Physiology)
1999-2003 Christina Liquori (with Ranum, MCDBG)
2002-2005 Anne Mosemiller (with Ranum, Neuroscience)
2003-2007 Jamie Malikowski (with Ranum, MCDBG)
2003-2007 Randy Daughters (with Ranum, MCDBG)
2005-2007 Karen Armbrust (with Ranum, Neuroscience)

Postdoctoral Trainees:

1993-1995 Bula Bhattacharyya, (with Gomez)
1998-1999 Mark Leppert (with Ranum)
1998-2001 Melinda Moseley (with Ranum)
2002-2005 Yoshio Ikeda (with Ranum)
2006-2008 Peter Karachunski (MDA Training Fellowship)

PhD Thesis Committees:

1998 Tara Ward, Veterinary School Pathobiology
1998 Jack Grinband, Neuroscience and Biochemistry
1999 Ben Rosner, Mechanical Engineering
1999 Jon Falkenberg, Physiology
1999 Jack Surek, Biochemistry
2001 Matthew Hagen, Neuroscience
2007 Chiao-Nan Chen, Physical Therapy
2007 Eric Richardson, Biomedical Engineering
2007 Justin Barnes, Neuroscience
2008 Daniel Franc, Neuroscience
2010 Blake Edwards, Neuroscience
2011 Justin Barnes, Neuroscience
2011 Yun Lin, Neuroscience

EXTRAMURAL INVITED PRESENTATIONS

1985, 1986 "Introduction to Neurology" Lecture series for Nurse Practitioners, UCSF
1986 "Acute Management of Cerebral Infarct," Symposium Organizer, UCSF
1987 "Clinical Electrophysiology," Dept. of Orthopedics, UCSF
1990 "Advances in Pain and Pain Management," Amer Col Phys Ann Mtg, SF, CA
1991 "Normocalcemic Tetany," Neurology Grand Rounds, University of Texas, Houston
1991 "Inherited Neuronal Hyperexcitability," Neurology Grand Rounds, University of Utah
1991 "Adenosine Modulation of Muscle Acetylcholine Receptors," Department of Neurology, U of MN
1993 "Advances in Pre-Synaptic Physiology," Amer Assoc Clin Neurophy Ann Mtg, UCSF
1994 "Temporal Aspects of Conduction Block," Amer Assoc Electrodiag Med Ann Mtg, San Francisco
1994-2004 "Clinical Features of Myasthenia Gravis", MG Foundation, MN Chapter Ann Mtg (6 talks)
1995 "An Update on Ocular Myasthenia Gravis", MG Foundation, MN Chapter Ann Mtg

1996, “Immunosuppressive Treatment of Inflammatory Neuropathies”, Neurology Grand Rounds, U NB
1997 "PROMM, Similarities with Myotonic Dystrophy” Grand Rounds, Stanford Medical School
1997 “Immunosuppressive Treatment of Inflammatory Neuropathies”, Spec Lecture, Stanford Med Sch
1997 “A Novel form of Myotonic Dystrophy in a Minnesota Family”, ENMC, Netherlands
1998 “Clinical features of Myotonic Dystrophy type 2”, IX Interntl Conf on Neuromus Dis, Adelaide
1998 “A Novel form of Myotonic Dystrophy”, Grand Rounds, OHSU, Portland
1999 “The DM2 locus in PROMM”, 2nd Intl Myotonic Dystrophy Consortium, North Carolina
2000 “Clinical Features of DM2”, European Neuromuscular Consortium (ENMC), Netherlands
2000 “Clinical and Genetic Features of DM2”, 3rd Intl Myotonic Dystrophy Consortium, Kyoto
2001 “Clinical and Genetic Features of DM2”, Special Invited Lecture, Wurzburg, Germany
2002 “The Myotonic Dystrophies, International Neuromuscular Congress”, Vancouver
2002 “Recent Advances in Myotonic Dystrophies, University Virginia, Charlottesville
2003 “Clinical and Molecular Features of DM2”, European Neuromuscular Consortium, Netherlands
2003 “Clinical and Molecular Features of DM2”, 4th Intl Myotonic Dystrophy Consortium, Glasgow
2003 “Stem Cells in Muscular Dystrophy”, Parents Project for Muscular Dystrophy, Cincinnati
2003 “RNA toxicity in the Myotonic Dystrophies”, UCLA Neurology Grand Rounds, Los Angeles, CA
2003 “RNA toxicity in the Myotonic Dystrophies”, Mayo Neurology Grand Rounds, Rochester, MN
2004 “RNA toxicity of the Myotonic Dystrophies,” U IA Neurology Grand Rounds, Iowa City, IA
2004 “Pathophysiology of the Myotonic Dystrophies”, World Muscle Soc Keynote, Goteborg, Sweden
2006 “RNA Mediated Neuromuscular Disease”, Int’l Neuromuscular Congress Keynote, Istanbul
2006 “Clinical Features of Myotonic Dystrophy”, Int’l Neuromuscular Congress Symposium, Istanbul
2007 “Clinical Features of Myotonic Dystrophy”, Neurology Grand Rounds, Mass Gen Hosp, Boston
2008 “Genetics of ALS”, ACNS Annual Meeting, Savannah, Georgia
2008 “Myotonic Dystrophy: Unusual Mechanisms underlie a Complex Phenotype”, Neurology Grand Rounds, Stanford University, Palo Alto
2008 “Understanding the RNA Pathogenesis of Myotonic Dystrophy”, Department of Genetics, UCSD, San Diego, CA
2009 “CNS Aspects of Myotonic Dystrophy”, Keynote Address, International Myotonic Dystrophy Consortium, Wurzburg, Germany
2010 “Neuromuscular Disorders Come Into Focus: Impending Treatments for Muscular Dystrophy”, Keynote Address, Annual Stanford University Neuroscience Conference, SF, CA
2011 “Central Nervous System Effects of Myotonic Dystrophy”, Keynote Address, International Myotonic Dystrophy Consortium, Clearwater, FL
2011 “How far to go in diagnosing neuromuscular disorders: relevance of genetic testing for sporadic and familial disease”. Annual Stanford University Neuroscience Conference, SF, CA
2012 “Clinical Insights Into the Pathogenesis of Myotonic Dystrophy”, St. Louis University School of Medicine, St. Louis, MO
2012 “Defining CNS Aspects of Myotonic Dystrophy”, Rochester University, Rochester, NY

PUBLICATIONS

1. Aljure E, **Day JW**, Bennett MVL. Postsynaptic depression of Mauthner cell-mediated startle reflex, a possible contributor to habituation. *Brain Res* (1980) 188:261-268. *Annotation: This collaborative work stemmed from an observation I made of novel command neuron stimulation that allowed behavioral experiments by Dr. Aljure, with which I assisted, on an intact fish model developed by Dr. Bennett. Dr. Aljure and I wrote the initial draft of the manuscript.*

2. **Day JW**, Hall DH, Hall LM, Bennett MVL. Alpha-bungarotoxin labeling and acetyl-cholinesterase localization at the Mauthner fiber giant synapse in the hatchetfish. *J Neuroscience* (1983) 3:272-279. *Annotation: This collaborative effort resulted from histological experiments that I conceived and performed under the guidance of Drs. Hall, on a teleost central nervous system synaptic model developed by Dr. Bennett. I wrote the manuscript.*

3. **Day JW**, Huse WD, Bennett MVL. Time course of miniature postsynaptic potentials at the Mauthner fiber giant synapse of the hatchetfish. *Brain Res* (1985) 325:115-128. *Annotation: This collaborative effort resulted from physiological experiments that I conceived and performed, with assistance from my colleague Dr. Huse, on a teleost central nervous system synaptic model developed by Dr. Bennett. I wrote the manuscript.*

4. Huse WD, **Day JW**, Bennett MVL. Postsynaptic currents at the Mauthner fiber giant synapse of the hatchetfish. *Brain Res* (1985) 325:129-141. *Annotation: This collaborative effort resulted from experiments that I participated in with Dr. Huse, on a teleost CNS model synapse developed by Dr. Bennett. I reviewed and edited the manuscript.*

5. **Day JW**, Raskin NH, Thunderclap headache: symptom of unruptured cerebral aneurysm. *The Lancet* (November 29, 1986) 1247-1248. *Annotation: This case report resulted from my observation and definitive evaluation of a patient. I wrote the manuscript and collaborated with Dr. Raskin to coin the term.*

6. Adams M, Rhyner P, **Day JW**, DeArmond S, Smuckler EA. Whipple's disease confined to the central nervous system. *Ann Neurol* (1987) 21(1):104-108. *Annotation: This case report stems from a novel patient that I identified and definitively evaluated with both clinical and post-mortem testing, which was primarily reported by the pathologists. I reviewed and edited the manuscript.*

7. **Day JW**, Parry GJ. Normocalcemic tetany abolished by calcium infusion. *Ann Neurol* (1990) 27(4):438-440. *Annotation: This collaborative effort with Dr. Parry led to identification of a novel apparently genetic disorder with neuronal hyperexcitability. I performed the electrophysiological evaluation with Dr. Parry and wrote the manuscript.*

8. Pitchford S, **Day JW**, Gordon A, Mochly-Rosen D. Nicotinic acetylcholine receptor desensitization is regulated by activation-induced extracellular adenosine accumulation. *J Neurosci* (1992) 12 (11):4540-4544. *Annotation: This collaborative effort resulted from laboratory work I did with Dr. Pitchford, pursuing observations made by Dr. Mochly-Rosen. I generated cell cultures used in the experiments, participated in data evaluation. I reviewed and edited the manuscript initially prepared by Drs. Pitchford and Mochly-Rosen.*

9. Gomez CM, Bhattacharyya B, Charnet P, **Day JW**, Labarca C, Wollman RW, Lambert EH. A transgenic mouse model of the slow-channel syndrome. *Muscle Nerve* (1996) 19:79-87. *Annotation: This collaborative work was performed on mutant acetylcholine receptor transgenic mice generated by Dr. Gomez in which I personally performed the patch-clamp recording with the post-doctoral fellow, Dr. Bhattacharyya working in my lab. I analyzed the data and reviewed and edited the manuscript initially drafted by Dr. Gomez.*

10. Brass TJ, Loushin MKH, **Day JW**, Iaizzo PA. An improved method for muscle force assessment in neuromuscular disease. *J Med Engineering and Technology* (1996) 20(2): 67-74. *Annotation: This collaborative work resulted from a force-assessment device conceived and constructed by Dr. Iaizzo, in which I personally identified and studied all subjects. I reviewed and edited the manuscript initially drafted by Drs. Brass and Iaizzo.*

11. Gomez CM, Maselli R, Gundeck JE, Chao M, **Day JW**, Tamamizu S, Lasalde JA, McNamee M, Wollman RL. Slow-channel transgenic mice: a model of postsynaptic organellar degeneration at the neuromuscular junction. *J Neurosci* (1997) 17(11):4170-4179. *Annotation: This collaborative work was performed on mutant acetylcholine receptor transgenic mice generated by Dr. Gomez in which I personally performed the patch-clamp recording and analysis. I reviewed and edited the manuscript initially drafted by Dr. Gomez.*

12. Bhattacharyya BJ, Gomez CM, **Day JW**, Gundeck JE, Leonard S, Wollmann RL. Desensitization of mutant acetylcholine receptors in transgenic mice reduces the amplitude of neuromuscular synaptic currents. *Synapse (*1997) 27(4):367-77. *Annotation: This collaborative work was performed on mutant acetylcholine receptor transgenic mice generated by Dr. Gomez in which I personally performed and analyzed the patch-clamp recording with the post-doctoral fellow, Dr. Bhattacharyya, working in my lab. I reviewed and edited the manuscript initially drafted by Dr. Gomez.*

13. Gomez CM, Maselli R, Williams JM, Bhattacharyya BB, Wollmann RL, **Day JW**. Genetic manipulation of AChR responses suggests multiple causes of weakness in slow-channel syndrome. *Annals of the New York Academy of Sciences* (1998) 841:167-80. *Annotation: This collaborative work was performed on mutant acetylcholine receptor transgenic mice generated by Dr. Gomez in which I personally performed and analyzed the patch-clamp recording with the post-doctoral fellow, Dr. Bhattacharyya, working in my lab. I reviewed and edited the manuscript initially drafted by Dr. Gomez.*

14. Koob MD, **Day JW**, Benzow KA, Bird TD, Moseley ML, Ranum LPW. Rapid cloning of expanded trinucleotide repeat sequences from genomic DNA. *Nature Genetics* (1998) 18(1):72-5. *Annotation: This collaborative work stemmed from clinically defined, but genetically unclassified ataxia and neuromuscular patients available both through my clinic and through contributions from collaborators of Dr. Ranum. I provided a clinical evaluation of subjects, and helped assess significance of the expansions detected and cloned by this novel method developed by Drs. Koob and Ranum. I reviewed and edited the manuscript initially written by Drs. Koob and Ranum.*

15. Wick MJ, Matthias-Hagen VL, **Day JW**, Gomez CM, McGlennen RC. Long range PCR-based diagnosis of Friedreich ataxia in the clinical molecular diagnosis laboratory. *Molecular Diagnosis* (1998) 3:3-9. *Annotation: This collaborative work stemmed from a large number of clinically defined, but genetically unclassified ataxia patients available both through my and Dr. Gomez*'s *clinics. Dr. Wick was able to confirm the diagnosis of Friedreich's ataxia genetically using long-range PCR. I reviewed and edited the manuscript initially written by Dr. Wick.*

16. Ranum LPW. Rasmussen PF. Benzow KA. Koob MD. **Day JW**. Genetic mapping of a second myotonic dystrophy locus. *Nature Genetics* (1998) 19(2):196-8. *Annotation: This collaborative effort stemmed from a large family I identified and characterized as having a novel disorder, and for which I obtained support and actively participated in weekly discussions of genetic characterization and clinical evaluation. Dr. Ranum was the molecular geneticist. The manuscript was initially authored and edited by me with Dr. Ranum.*

17. **Day JW**, Roellofs R, Leroy B, Pech I, Amos J, Benzow K, Ranum L. Clinical and genetic characteristics of a five generation family with a novel form of myotonic dystrophy. *Neuromuscular Disorders* (1999) (1):19-27. *Annotation: This collaborative effort stemmed from a large family I identified, initial family members having seen Dr. Roellofs shortly before his death, and which I characterized as having a novel disorder, and for which I obtained support and participated in weekly assessments of genetic characterization and ongoing clinical evaluation for several years while we identified the locus. Dr. Ranum provided molecular genetics expertise. The manuscript was initially authored and edited by me with Dr. Ranum.*

18. Koob MD, Moseley M, Schut L, Bird T, Benzow KA, **Day JW**, Ranum LP. Identification of an untranslated CTG expansion in SCA8. *Nature Genetics* (1999) 21(4):379-84. *Annotation: A family harboring this novel repeat expansion was isolated (using methods described in ref 14) and I evaluated many affected family members along with Drs. Schut and Bird. I participated in weekly evaluation of the genetic testing and clinical evaluations. I edited and reviewed the manuscript initially drafted by Drs. Koob and Ranum.*

19. Li LY, Kelkar P, Exconde RE, **Day JW**, Parry GJ. Adult-onset "infant" botulism: an unusual cause of weakness in the intensive care unit. *Neurology* (1999) 53(4):891. *Annotation: I clinically evaluated this patient including performing electromyographic characterization that helped establish the diagnosis. I reviewed and edited the manuscript originally drafted by Drs. Parry and Li.*

20. International Myotonic Dystrophy Consortium. New nomenclature and DNA testing guidelines for myotonic dystrophy type 1 (DM1). The International Myotonic Dystrophy Consortium (IDMC). *Neurology* (2000) 54(6):1218-21. *Annotation: This collaborative statement came in part from a discussion I organized at the second meeting of the International Myotonic Dystrophy Consortium. Dr. Ashizawa authored the manuscript which I reviewed and edited.*

21. **Day JW**, Schut LJ, Moseley ML, Ranum LPW. Spinocerebellar ataxia type 8 (SCA8) Clinical features of a large family. *Neurology* (2000) 55:649-657. *Annotation: This collaborative work stemmed from the clinical characterization of the largest family we identified harboring the SCA8 expansion (ref 18). I interviewed and examined family members with Dr. Schut and participated in weekly assessments of on-going genetic and clinical characterization. I authored the initial manuscript and reviewed and edited it with the co-authors.*

22. Koenigbauer, UF, Eastland T, **Day JW**. Clinical illness due to parvovirus B19 infection after infusion of solvent/detergent-treated pooled plasma. *Transfusion* (2000) 40:1203-1206. *Annotation: This case report stemmed from a patient I treated with plasmapheresis who had subsequent infection as documented by Dr. Eastland. I reviewed and edited the manuscript initially drafted by Drs. Koenigbauer and Eastland.*

23. Moseley ML, Schut LJ, Bird TD, Koob MD, **Day JW**, Ranum LP. SCA8 CTG repeat: en masse contractions in sperm and intergenerational sequence changes may play a role in reduced penetrance. *Human Molecular Genetics* (2000) 9(14):2125-30. *Annotation: This collaborative work stemmed from my clinical characterization and obtaining of clinical specimens from many SCA8 males. I participated in weekly assessment and design of the on-going clinical and genetic characterization. I reviewed and edited the manuscript initially authored by Drs. Moseley and Ranum.*

24. Liquori CL, Ricker K, Moseley ML, Jacobsen JF, Kress W, Naylor SL, **Day JW***, Ranum LPW*. Myotonic Dystrophy Type 2: Caused by a CCTG Expansion in Intron 1 of *ZNF9*. *Science* (2001) 293:864-867. *Annotation: This collaborative effort stemmed many family I identified and characterized, as well as those characterized by Dr. Ricker, and for which I obtained support and participated in weekly evaluation and design of on-going genetic testing and clinical evaluation leading to identification of the mutation. The manuscript was initially authored and edited by Dr. Ranum and me.*

25. **Day JW**, Sakamoto C, Lehmann-Horn, Iaizzo PA. Force assessment in periodic paralysis after electrical muscle stimulation. *Mayo Clinic Proceedings* (2002) 77(3):232-40. *Annotation: This collaborative work stemmed from multiple periodic paralysis subjects I identified and characterized clinically with novel test design in part using the device previous developed (ref 10). I wrote the initial draft, edited by Drs. Iaizzo and myself.*

26. Wolfe GI, Barohn RJ, Foster BM, Jackson CE, Kissel JT, **Day JW**, Thornton CA, Nations SP, Bryan WW, Amato AA, Freimer ML, Parry GJ, Mendell JR. Randomized, controlled trial of intravenous immunoglobulin in myasthenia gravis. *Muscle and Nerve* (2002) 26:549-552. *Annotation: This multicenter investigation of IVIg treatment of myasthenia was spearheaded by Dr. Wolfe; I was the site PI of the Minnesota site, participating in conferences regarding study design, completion and data analysis as well as reviewing and editing the manuscript drafted by Dr. Wolfe.*

27. Koc ON, **Day JW**, Nieder M, Gerson SL, Lazarus HM, Krivit W. Allogeneic mesenchymal stem cell infusion for treatment of metachromatic leukodystrophy (MLD) and Hurler syndrome. *Bone Marrow Transplantation* (2002) 30:215-222. *Annotation: This collaborative work stemmed from our experience in developing and performing cell-based treatment of these disorders, with my role being design and completion of the clinical and electrophysiological characterization of the neurological response to treatment. I reviewed and edited the manuscript initially drafted by Dr. Krivit.*

28. **Day JW**, Ricker K, Jacobsen J, Rasmussen L, Dick K, Kress W, Schneider C, Koch M, Beilman G, Harrison A, Dalton J, Ranum L. Myotonic dystrophy type 2: molecular, diagnostic and clinical spectrum. *Neurology* (2003) 60:657-664. *Annotation: This collaborative work stemmed from the many families I identified and characterized, as well as those characterized by Dr. Ricker, and for which I helped generate support and co-led weekly evaluation and design of on-going genetic testing and clinical evaluation. I authored the initial manuscript which was edited and revised with Drs. Ricker and Ranum.*

29. Muley SA, **Day JW**. Autoimmune Rippling Muscle. *Neurology* (2003) 61:869–870. *Annotation: This case report was of a patient initially identified by Dr. Muley, in which I participated in the clinical evaluation and interpretation as well as in writing and editing the manuscript.*

30. Walk D, Li LYJ, Parry GJP, **Day JW**. Rapid Resolution of Quadriplegic CIDP by Combined Plasmapheresis and IVIg. *Neurology* (2004) 62 155-156. *Annotation: This paper reported a novel treatment I designed, administered and evaluated and which was also successfully applied in a patient identified by Dr. Walk. I drafted the initial manuscript and oversaw its revision.*

31. Liquori CL, Ikeda Y, Weatherspoon M, Ricker K, Schoser BGH, Dalton JC, **Day JW**, Ranum LPW. Myotonic Dystrophy Type 2: Human Founder Haplotype and Evolutionary Conservation of the Repeat Tract. *Am J Hum Genet* (2003) 73:849-862. *Annotation: This collaborative effort stemmed many families I identified and characterized with this disorder, as well as those characterized by Dr. Ricker; I helped organize and administer support and co-led weekly evaluation and design of on-going genetic testing and clinical evaluation leading to identification of the mutation. I reviewed and edited the manuscript initially authored by Drs. Ranum and Liquori.*

32. Savkur RS, Philips AV, Cooper TA, Dalton JC, Moseley ML, Ranum LP, **Day, JW**. Insulin receptor splicing alternation in myotonic dystrophy type 2. *Am J Hum Genet* (2004) 74(6):1309-13. *Annotation: I established this collaborative effort to characterize molecularly the disorder I helped identify, with testing by Dr. Savkur, in Dr. Cooper's laboratory, using specimens I obtained from clinically and genetically characterized individuals. I authored the initial manuscript and oversaw its editing and revision.*

33. Schoser BG, Ricker K, Schneider-Gold C, Hengstenberg C, Durre J, Bultmann B, Kress W, **Day JW**, Ranum LP. Sudden cardiac death in myotonic dystrophy type 2. *Neurology* (2004) 63(12):2402-4. *Annotation: This collaborative work stemmed from subjects identified by me and Drs. Ricker and Schoser. I revised and edited the manuscript initially drafted by Dr. Schoser.*

34. Ikeda Y., Dalton JC, Moseley ML, Gardner KL, Bird TD, Ashizawa T, Seltzer WK, Pandolfo M, Milunsky A, Potter NT, Shoji M, Vincent JB, **Day JW,** Ranum, LPW. Spinocerebellar ataxia type 8: molecular genetic comparisons and haplotype analysis of 37 ataxia families. *Am J Hum Genet*. (2004) 75:3-16. *Annotation: This collaborative work stemmed from the large number of subjects identified harboring an SCA8 expansion collected by Drs. Ikeda, Bird, Ashizawa, Pandolfo, Potter, Shoji, Vincent and myself, in addition to those submitted from other sources to Dr. Ranum for analysis. I co-led the weekly evaluation and design of on-going genetic testing and clinical evaluation of this topic. I reviewed and edited the manuscript initially drafted by Dr. Ranum.*

35. Meisler MH, Trudeau MM, Dalton JC, **Day JW**, Ranum LP. Gene symbol: SCN8A. Disease: Ataxia. Accession #Hd0520. *Hum Genet* (2006) 118(6):776. *Annotation: This collaborative work stemmed from SCN8A mutations identified on genetic screening that Dr. Meisler had submitted to Dr. Ranum. I evaluated and collected specimens from the family to characterize the phenotype associated with this mutation. I reviewed and edited the manuscript initially drafted by Dr. Meisler.*

36. Trudeau MM, Dalton JC, **Day JW**, Ranum LP, Meisler MH. Heterozygosity for a protein truncation mutation of sodium channel SCN8A in a patient with cerebellar atrophy, ataxia, and mental retardation. *J Med Genet* (2006) 43(6):527-30. *Annotation: This collaborative work stemmed from SCN8A mutations identified on genetic screening that Dr. Meisler had submitted to Dr. Ranum. I evaluated and collected specimens from the family to characterize the phenotype associated with this mutation. I reviewed and edited the manuscript initially drafted by Dr. Meisler.*

37. Margolis JM, Schoser BG, Moseley ML, **Day JW**, Ranum LP. DM2 intronic expansions: evidence for CCUG accumulation without flanking sequence or effects on ZNF9 mRNA processing or protein expression. *Hum Mol Genet* (2006) 15(11):1808-15. *Annotation: This collaborative effort stemmed from families I identified and characterized, using specimens I obtained, and for which I actively participated in weekly evaluation and design of on-going genetic testing and evaluation. I reviewed and edited the manuscript initially authored by Drs. Ranum and Margolis.*

38. Ikeda Y, Dick KA, Weatherspoon MR, Gincel D, Armbrust KR, Dalton JC, Stevanin G, Durr A, Zuhlke C, Burk K, Clark HB, Brice A, Rothstein JD, Schut LJ, **Day JW,** Ranum LPW. Spectrin mutations cause spinocerebellar ataxia type 5. *Nature Genet* (2006) 38: 184-190. *Annotation: This collaborative effort stemmed from my characterization, along with Dr. Schut, of this family, initially identified by Dr. Ranum. I generated support and actively participated in the weekly evaluation and design of on-going genetic testing and clinical evaluation leading to identification of the mutation. I edited the manuscript initially authored by Dr. Ranum.*

39. Moseley ML, Zu T, Ikeda Y, Gao W, Mosemiller AK, Daughters RS, Chen G, Weatherspoon MR, Clark HB, Ebner TJ, **Day JW**, Ranum LPW. Transgenic mouse model helps define the molecular pathophysiology of spinocerebellar ataxia type 8. *Nature Genet* (2006) 38:758-69. *Annotation: This collaborative work characterized the transgenic model developed by Drs. Ranum and Moseley. I actively participated in the weekly evaluations of model generation and characterization to assess its comparability to the clinical disorder. I reviewed and edited the manuscript initially drafted by Drs. Moseley and Ranum.*

40. Saito T, Amakusa Y, Kimura T, Yahara O, Aizawa H, Ikeda Y, **Day JW**, Ranum LP, Ohno K, Matsuura T. Myotonic dystrophy type 2 in Japan: ancestral origin distinct from Caucasian families. *Neurogenetics (*2008) 9(1):61-3. *Annotation: This family was identified and collected by Dr. Matsuura. I collaborated with him to compare the clinical phenotype and genetic results with our previously reported work on the same disorder. Dr. Matsuura wrote the initial draft of the paper, which I reviewed and edited.*

41. **Day, JW.** Congenital muscular dystrophy in a new age. *Neurology* (2008) 71(5): 308-9. *Annotation: I wrote this editorial.*

42. Krueger KAD, Tsuji S, Fukuda Y, Takahashi Y, Goto J, Dalton JC, Miller MB, **Day JW**, Ranum LPW. SNP haplotype mapping in a small ALS family. *Plos One* (2009) 4(5):e3687. *Annotation: Dr. Ranum initially identified the family, which I collected, clinically characterized and assisted with its genetic characterization. Dr. Krueger initially drafted the manuscript, which I reviewed and edited.*

43. Arikian A, Boutelle K, Peterson CB, Dalton, JC, **Day JW**, Crow SJ. Targeting Parents for the Treatment of Pediatric Obesity in Boys with Duchenne Muscular Dystrophy. *Eat Weight Disord* (2010) 15(3):161-5. *Annotation: I initiated this collaboration with Dr. Crow, identified, characterized and recruited all subjects, and helped design and modify the protocol for the study during monthly discussions. Dr. Arikian drafted the manuscript, which I reviewed and edited.*

44. Flanigan KM, Dunn DM, von Niederhausern A, Soltanzadeh P, Gappmaier E, Howard MT, Sampson JB, Mendell JR, Wall C, King WM, Pestronk A, Florence JM, Connolly AM, Mathews KD, Stephan CM, Laubenthal KS, Wong BL, Morehart PJ, Meyer A, Finkel RS, Bonnemann CG, Medne L, **Day JW**, Dalton JC, Margolis MK, Hinton VJ; United Dystrophinopathy Project Consortium, Weiss RB. Mutational spectrum of DMD mutations in dystrophinopathy patients: application of modern diagnostic techniques to a large cohort. *Hum Mutat* (2009) 30(12):1657-66. *Annotation: Our involvement in this collaborative work stemmed from the large number of genetically and clinically defined Duchenne specimens we collected, several demonstrating novel genetic mechanisms; I enrolled and evaluated all subjects at our site. This genetic mutation prevalence study depended upon having a large set of comparably defined data, which we as a group discussed on quarterly conference calls, and at annual face-to-face meetings. I reviewed and edited the paper initially drafted by Drs. Flanigan and Weiss.*

45. de Greef JC, Lemmers RJLF, Camano P, **Day JW**, Desnuelle C, DunandM, van Engelen BGM, Kiuri-Enari S, Padberg GW, Rosa AL, Sacconi S, Spuler S, Tarnopolsky M, Venance SL, Frants RR, van der Maarel SM, Tawil R. D4Z4 repeat contraction-independent facioscapulohumeral muscular dystrophy type 2 represents a true FSHD subgroup. *Neurology* (2010) 75(17):1548-54. *Annotation: I collected several FSHD patients who were without the common mutation, and contributed them to the efforts of Drs. Van der Maarel and Tawil establishing this novel genetic mechanism. I reviewed and edited the paper that was originally drafted by Dr. Tawil.*

46. Appel SH, Cwik VA, **Day JW**. Trauma, TDP-43, and amyotrophic lateral sclerosis. *Muscle Nerve* (2010) 42(6):851-2. *Annotation: I conceived of and drafted this letter.*

47. Flanigan KM, Dunn DM, von Niederhausern A, Soltanzadeh P, Howard MT, Sampson JB, Swoboda KJ, Bromberg MB, Mendell JR, Taylor L, Anderson CB, Pestronk A, Florence J, Connolly AM, Mathews KD, Wong B, Finkel RS, Bonnemann CG, **Day JW**, McDonald C, Weiss RB. Nonsense mutation-associated Becker muscular dystrophy: interplay between exon definition and splicing regulatory elements within the DMD gene. *Hum Mutat* (2011) 32:299–308. *Annotation: Our involvement in this collaborative work stemmed from the large number of genetically and clinically defined Duchenne specimens we collected; I evaluated and enrolled all subjects at our site. This genotype-phenotype study depended upon having a large set of comparably defined data, which we as a group discussed on quarterly conference calls, and an annual face-to-face meeting. The paper was drafted by Drs. Flanigan and Weiss, which I reviewed and edited.*

48. Wozniak JR, Mueller BA, Ward EE, Lim KO, **Day JW**. White matter abnormalities and neurocognitive correlates in children and adolescents with myotonic dystrophy type 1: a diffusion tensor imaging study. *Neuromuscul Disord* (2011) 21(2):89-96. *Annotation: I conceived of this research program, clinically and genetically characterized all subjects, reviewed progress throughout the investigations, and edited the manuscript first drafted by Dr. Wozniak.*

49. Rau F, Freyermuth F, Fugier C, Villemin JP, Fischer MC, Jost B, Dembele D, Gourdon G, Nicole A, Duboc D, Wahbi K, **Day JW**, Fujimura H, Takahashi MP, Auboeuf D, Dreumont N, Furling D, Charlet-Berguerand N. Misregulation of miR-1 processing is associated with heart defects in myotonic dystrophy. Nat Struct Mol Biol. 2011 Jun 19;18(7):840-5. *Annotation: I identified, clinically characterized, and genetically defined subjects with DM2, and recruited families to contribute autopsy specimens. I discussed the possible testing of DM2 autopsy specimens with Dr. Charlet, whose laboratory was comparing results of miR processing in DM1, suggesting that co-existence in DM2 would help define the molecular pathophysiology. Dr. Charlet's laboratory wrote the manuscript, which I reviewed and edited.*

50. Escolar DM, Hache LP, Clemens PR, Cnaan A, McDonald CM, Viswanathan V, Kornberg AJ, Bertorini TE, Nevo Y, Lotze T, Pestronk A, Ryan MM, Monasterio E, **Day JW**, Zimmerman A, Arrieta A, Henricson E, Mayhew J, Florence J, Hu F, Connolly AM. Randomized, blinded trial of weekend vs daily prednisone in Duchenne muscular dystrophy. Neurology. 2011 Aug 2;77(5):444-52. *Our involvement in this collaborative work stemmed from the large number of genetically and clinically defined Duchenne specimens we collected; I evaluated and enrolled all subjects at our site and enrolled them for this study comparing two treatment regimens. The paper was drafted by Dr. Escolar, which I reviewed and edited.*

51. Blonsky K, Monckton D, Wieringa B, Schoser B, **Day JW**, van Engelen B. 2010 Marigold therapeutic strategies for myotonic dystrophy. Neuromuscul Disord. 2012 Jan;22(1):87-94. *Annotation: This paper detailed the results of a myotonic dystrophy therapeutic directions workshop that I helped chair. The paper was drafted by Dr. van Engelen, which I reviewed and edited.*

52. Ikeda Y, Ranum LP, **Day JW**. Clinical and genetic features of spinocerebellar ataxia type 8. Handb Clin Neurol. 2012;103:493-505. *Annotation: This paper detailed our genetic and clinical characterization of SCA8. I drafted and edited the manuscript, which was also edited by the co-authors.*

53. Dick KA, Ikeda Y, **Day JW**, Ranum LP. Spinocerebellar ataxia type 5. Handb Clin Neurol. 2012;103:451-9. *Annotation: This paper detailed our genetic and clinical characterization of SCA5. The manuscript was drafted by Dr. Dick, which I reviewed and edited.*

54. Franc DT, Muetzel RL, Robinson PR, Rodriguez CP, Dalton JC, Naughton CE, Mueller BA, Wozniak JR, Lim KO, **Day JW**. Cerebral and muscle MRI abnormalities in myotonic dystrophy. Neuromuscul Disord. 2012 Jan 28. *Annotation: I conceived of this research program, clinically and genetically characterized all subjects, obtained funding, reviewed progress throughout the investigations, and drafted and edited the manuscript.*

55. Bushby K, Finkel R, Wong B, Barohn R, Campbell C, Comi GP, Connolly AM, **Day JW**, Flanigan KM, Goemans N, Jones K, Mercuri E, Quinvilan R, Renfroe JB, Russman B, Ryan M, Tulinius M, Voit T, Barth J, Elfring G, Morsy M, Reha A, Peltz S, McDonald C, PTC124-GD-007-DMD Study Group. Ataluren Treatment of Nonsense Mutation Dystrophinopathy. (submitted) *Annotation: Our involvement in this collaborative work stemmed from the large number of genetically and clinically defined Duchenne specimens we collected; I evaluated and enrolled all subjects at our site. This clinical trial of the first definitive treatment of DMD is now under review at NEJM. The paper was drafted by Dr. McDonald, which I reviewed and edited.*

56. Flanigan KM, Ceco E, Lamar KM, Kaminoh Y, Dunn DM, Mendell JR, Pestronk A, Florence JM, Mathews KD, Finkel RS, Swoboda KJ, Howard MT, **Day JW**, McDonald C, McNally EM, Weiss RB. LTBP4 genotype predicts prolonged ambulation in Duchenne Muscular Dystrophy (submitted) *Annotation: Our involvement in this collaborative work stemmed from the large number of genetically and clinically defined Duchenne specimens we collected; I evaluated and enrolled all subjects at our site. This genotype-phenotype study depended upon having a large set of comparably defined data, which we as a group discussed on quarterly conference calls, and an annual face-to-face meeting. The paper drafted by Dr. Flanigan, which I reviewed and edited, is now under review at NEJM.*

57. Karachunski PI, Margolis M, Dalton JC, **Day JW**. Fluctuating LGMD-2I in Twin Brothers: Response to Prednisone. (submitted). *Annotation: I identified, clinically characterized, and defined the unusual genetic cause for this progressive disorder. I have written the manuscript and am overseeing the review process.*

58. Karachunski PI, Margolis J, Dehnbostel J, Dalton JC, Ranum LPW, **Day JW**. Potassium Channelopathy Causes Familial Neuromyotonia with Delayed and Prolonged Trousseau Response. (submitted). *Annotation: I identified, clinically characterized, and defined the genetic for this novel disorder. I have written the manuscript and am overseeing the review process.*

ABSTRACTS:

1. Aljure E, **Day JW**, Bennett MVL. Implied locus of habituation of the Mauthner fiber mediated startle reflex in the hatchetfish. Soc Neurosci Abstr 5:735, 1979.
2. Huse WD, **Day JW**, Bennett MVL. Voltage dependence of synaptic currents at a CNS synapse in the hatchetfish. Soc Neurosci Abstr 5:742, 1979.
3. Hall DH, **Day JW**, Hall LM, Bennett MVL. Alpha-bungarotoxin labeling of an identified central synapse. Soc Neurosci Abstr 7:703, 1981.
4. **Day JW**, Huse WD, Bennett MVL. Two phase decay of MPSPs at the hatchetfish giant synapse. Soc Neurosci Abstr 7:724, 1981.
5. Bennett MVL, **Day JW**. Mauthner fiber reflex: descending activity mediates "habituation" of the response to Mauthner fiber stimulation. Soc Neurosci Abstr (1981) 7:843.
6. **Day JW**, Raskin NH. Sentinel headache without subarachnoid hemorrhage. Neurology 35(suppl 1):268, 1985.
7. **Day JW**. Effect of opiates on the action potential of neuroblastoma cells. Soc Neurosci Abstr 13:769, 1987.
8. Midani H, Parry GJ, **Day JW**. A Case of Inflammatory Neuropathy Misdiagnosed as Charcot-Marie-Tooth. AAEM Annual Meeting, New Orleans, 1993.
9. **Day JW**, Sakamoto C, Lehmann-Horn F, Parry GJ, Iaizzo PA. Assessment of dorsiflexor torque to aid in the diagnosis of periodic paralysis. International Congress on Neuromuscular Diseases, Kyoto, 1994, Muscle Nerve Suppl 1:S209, 1994.
10. Bhattacharyya B, **Day JW**, Gomez CM. Transgenic mice with slow acetylcholine receptor ion channels: reduced miniature endplate current amplitudes may be due to desensitization. Soc Neurosci Abstr 20:713, 1994.
11. **Day JW**, Maselli R, Bhattacharyya B, Gomez CM. Acetylcholine receptor mutations alter electrophysiological function in transgenic mice. Neurology 45 (suppl 4): A283, 1995.
12. Brass TJ, **Day JW**, Iaizzo PA. Stimulated force measurements in neuromuscular diseases. American Academy of Neurology 45:120P, 1995

13. Wrubel B, **Day JW**, Parry GJ. Needle versus surface electrode recording of sensory nerve action potentials. American Academy of Electrodiagnostic Medicine Annual Meeting, Montreal, 1995
14. Gomez CM, Maselli R, **Day JW**, Wollman RW. Calcium overload and postsynaptic vacuolization in transgenic mice as a function of cholinergic receptor ion channel kinetics. Society for Neuroscience Abstracts 21:1490, 1995.
15. Ranum LPW, Benzow KA, Lundgren JK, Koob MD, **Day JW**. A CTG repeat expansion detected by RED in a five generation kindred with a novel form of myotonic dystrophy. Am J Hum Genet, Suppl A: 280., 1996.
16. **Day JW**, Williams JM, Charnet P, Gomez CM. Slow-channel transgenic mice: a model of transsynaptic calcium overload. Soc for Neuroscience Abstracts 23:1134, 1997.
17. **Day JW**, Leroy B, Pech I, Benzow KA, Ranum LPW. Clinical and Physiological Features of a Novel Form of Myotonic Dystrophy. IX Intl. Conf Neuromuscular Disease, Adelaide, 1998.
18. Ranum LPW, Rasmussen PF, Benzow KA, Koob MD, **Day JW**. Genetic characterization of a novel form of myotonic dystrophy. Amer Soc Human Gen, Denver, 1998.
19. Koob MD, Moseley ML, Schut LJ, Bird TD, Benzow KA, **Day JW**, Ranum LPW. A 3' untranslated CTG repeat causes spinocerebellar ataxia type 8 (SCA8). Amer Soc Human Gen, Denver, 1998.
20. Ranum LPW, Rasmussen PF, Benzow KA, Koob MD, **Day JW**. Genetic mapping and clinical characterization of a second form of myotonic dystrophy (DM2). Amer Soc Human Gen, Denver, 1998.
21. Moseley ML, **Day JW**, Schut LJ, Bird TD, Benzow KA, Koob MD, Ranum LPW. Frequency and dramatic instability of the 3'untranslated CTG repeat causing spinocerebellar ataxia type 8 (SCA8). Amer Soc Human Gen, Denver, 1998.
22. Wolfe GI, Barohn RJ, Foster BM, Jackson CB, Kissel JT, **Day JW**, Thornton CA. Intravenous immunoglobulin therapy for generalized myasthenia gravis. Amer Acad Neur, Toronto, 1999
23. **Day JW**, Ricker K, Liquori CL, Durand AC, Ranum LPW. The genetic locus for myotonic dystrophy type 2 (*DM2*) also underlies many cases of PROMM. Second International Myotonic Dystrophy Consortium (IDMC) Conference, Research Triangle Park, North Carolina, 1999.
24. Stelow E, Eastlund DT, **Day JW**. Parvovirus infection following plasma exchange using solvent-detergent treated pooled plasma containing parvovirus. AABB, 1999
25. Ranum LPW, Moseley ML, Leppert MF, van den Engh G, La Spada AR, Koob MD, **Day JW**. Spinocerebellar ataxia type 8: Massive expansions and deletions may cause reduced penetrance. Amer Soc Human Gen, San Francisco, 1999.
26. Koc ON, **Day JW**, Brown D, Andrews P, Peters C, Nieder M, Gerson SL, Lazarus HM, Caplan AI, Laughlin MJ, Raghavan S, Kolodny EH, Krivit W. Results of a phase I clinical trial of allogeneic mesenchymal stem cell (MSC) transplantation in patients with Hurler disease and metachromatic leukodystrophy (MLD). American Society of Hematology, 2000.
27. Moseley M, Jacobsen J, Liquori C, Durand A, Davis L, Ikeda Y, Bird T, Spiegel R, Ashizawa T, Pandolfo M, Potter N, Schaefer F, Milunski A, Kennedy J, Seltzer W, Atwood L, Vincent J, **Day JW**, Ranum LPW. SCA8 CTG expansion: evidence for a common haplotype and highly mutable region on both ataxia and non-ataxia chromosomes. Amer Soc Human Gen, Philadelphia, 2000.
28. Li L, Walk D, **Day JW**. Rapid Response to Combined Plasma Exchange Followed Immediately by Intravenous Immunoglobulin in Refractory CIDP. Peripheral Nerve Society, Innsbruck. 2001.
29. Ranum LPW, Liquori CL, Moseley ML, Jacobsen JF, Phillips A, Savkur R, Kress W, Naylor SL, Cooper T, Ricker K, **Day JW**. Myotonic Dystrophy Type 2 is caused by a CCTG expansion in intron 1 of *ZNF9*. World Muscle Society, Salt Lake City, 2001.

30. Ranum LPW, Liquori CL, Moseley ML, Jacobsen JF, Phillips A, Savkur R, Kress W, Naylor SL, Cooper T, Ricker K, **Day JW**. Myotonic Dystrophy Type 2 is caused by a CCTG expansion in intron 1 of *ZNF9*. 3rd International Myotonic Dystrophy Consortium, Kyoto, 2001.
31. **Day JW**, Ricker K, Dick KA, Rasmussen L, Dalton JC, Jacobsen JF, Liquori CL, Kress W, Schneider C, Moseley ML, Ranum LPW. Clinical and Molecular Correlations in Myotonic Dystrophy Type 2. 3rd International Myotonic Dystrophy Consortium, Kyoto, 2001.
32. Ranum LPW, Liquori CL, Moseley ML, Jacobsen JF, Phillips A, Savkur R, Kress W, Naylor SL, Cooper T, Ricker K, **Day JW**. Myotonic Dystrophy Type 2 is caused by a CCTG expansion in intron 1 of *ZNF9*. Amer Soc Human Gen, San Diego, Am J Human Genetics, 69:211, 2001.
33. Liquori CL, **Day JW**, Ranum LPW. Conserved haplotypes indicate founder mutation in myotonic dystrophy type 2. Amer Soc Human Gen, San Diego, 2001, Am J Human Genetics, 69:424, 2001.
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