

Stanford




John W. Day, MD, PhD

Professor of Neurology, of Pediatrics (Genetics) and, by courtesy, of Pathology at the Stanford University Medical Center

Neurology & Neurological Sciences

 NIH Biosketch available Online

 Curriculum Vitae available Online

CLINICAL OFFICES

- **Stanford Neuroscience Health Center**

213 Quarry Rd Rm 2851

MC 5957

Palo Alto, CA 94304

Tel (650) 723-6469

Fax (650) 497-0091

- **Child Neurology**

730 Welch Rd

2nd Fl

Palo Alto, CA 94304

Tel (650) 723-0993

Fax (650) 723-7299

ACADEMIC CONTACT INFORMATION

- **Alternate Contact**

Angelica Martinez - Neuromuscular Administrative Assistant

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Bio

CLINICAL FOCUS

- Neurology

ACADEMIC APPOINTMENTS

- Professor - Med Center Line, Neurology & Neurological Sciences
- Professor - Med Center Line, Pediatrics - Medical Genetics
- Professor - Med Center Line (By courtesy), Pathology
- Member, Maternal & Child Health Research Institute (MCHRI)

ADMINISTRATIVE APPOINTMENTS

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

HONORS AND AWARDS

- Grass Foundation Fellow in Neurophysiology, Marine Biological Lab, Woods Holes, MA (1978)

- Distinguished Teaching Award, University of California San Francisco (1985)
- Clinical Investigator Development Award, NINCDS, NIH (1986)
- Distinguished Teaching Award, University of Minnesota Medical School (1996)
- Distinguished Teaching Award, University of Minnesota Medical School (2001)
- Leon Poliachik Humanitarian Award, University of Minnesota ALS Clinic (2002)
- Distinguished Teaching Award, University of Minnesota Medical School (2003)
- Outstanding Teaching Award, University of Minnesota Medical School (2005)
- Distinguished Teaching Award, University of Minnesota Medical School (2005)
- All University Post-Baccalaureate Teaching Award, All University Post-Baccalaureate Teaching Award (2007)
- All University Post-Baccalaureate Teaching Award, University of Minnesota (2007)
- Recognized among Best Physicians in Minnesota, Twin Cities Magazine (2010)

PROFESSIONAL EDUCATION

- Fellowship: UCSF Dept of Neurology (1987) CA
- Residency: UCSF Dept of Neurology (1986) CA
- Professional Education: Albert Einstein College of Medicine Office of the Registrar (1982) NY
- Medical Education: University of Minnesota School of Medicine Registrar (1977) MN
- Residency, UCSF , Department of Neurology (1986)
- Board Certification: Neuromuscular Disease, American Board of Psychiatry and Neurology (2011)
- Internship: Montefiore Medical Center - Albert Einstein College of Medicine (1983) NY
- Board Certification: Neurology, American Board of Psychiatry and Neurology (1988)
- Ph.D, Albert Einstein College of Medicine , Neuroscience (1982)
- M.D, University of Minnesota , Medicine (1977)
- BA, Oberlin College , Physics (1973)

LINKS

- Dr. Day's Lab: <https://med.stanford.edu/day-lab.html>
- Get a Second Opinion: <https://stanfordhealthcare.org/second-opinion/overview.html>

Research & Scholarship

CURRENT RESEARCH AND SCHOLARLY INTERESTS

Our Neuromuscular Division organizes a comprehensive effort to combat and conquer diseases of the peripheral nerves and muscles, including the muscular dystrophies (myotonic, Duchenne, limb girdle, facioscapulohumeral, and congenital muscular dystrophies), motor neuron disorders (ALS and SMA), neuromuscular junction disease (MG, CMS), and peripheral neuropathies (CMT, CIDP). While keeping the patients and families foremost in mind, our research seeks to: define and understand genetic causes; clarify the molecular and cellular consequences of genetic change; determine the multisystemic features that are underappreciated but clinically significant consequence of these diseases; develop and improve methods for managing and treating each disease.

We have identified the genetic cause of several neuromuscular disorders, most notably myotonic dystrophy type 2, which we continue to study to advance understanding of all forms of myotonic dystrophy. We have also contributed to genetic understanding of Duchenne muscular dystrophy, and other muscle and ataxic disorders. We are continuing to investigate the epigenetic and molecular consequences of these diseases through investigation of patient-derived specimens.

We have focused on defining the central nervous system features of neuromuscular disorders, which severely impact patients and families but have been incompletely investigated, explained or managed. Detailed neuropsychological and brain MRI studies are helping to define the developmental and progressive CNS aspects of these conditions, for which we then seek molecular and cellular explanations through cell-based studies of patient-derived specimens.

To assure our research is translatable to clinical practice, we are simultaneously involved in collaborative clinical research on novel treatments for neuromuscular disease, including antisense oligonucleotides and pharmacologic manipulation of muscle function, viral gene therapies and cell-based treatments.

In summary, we work with patients to define neuromuscular disorders more rigorously and understand them more thoroughly, so novel treatments will successfully combat these devastating disorders.

Clinical Research Studies:

2014- "Clinical and Genetic Characterization of Myotonic Dystrophy"- PI: Dr. John Day, MD, PhD

"Clinical and Genetic Characterization of Myotonic Dystrophy-cont.(Sleep Study)"- PI: Dr. John Day, MD, PhD/ Co- Investigators: Dr. Chad Ruoff and Dr. Brian Wandell

2014- "Subject Database and Specimen Repository for Neuromuscular and Neurodegenerative Disorders"- PI: Dr. John Day, MD, PhD

2014-"Insulin Resistance and Insulin Secretion in Patients with Myotonic Dystrophy"- PI: Dr. John Day, MD, PhD/ Co-PI: Dr. Josh Knowles, MD.

2014-"Defining and Managing the Neuropsychological Abnormalities of Myotonic Dystrophy (CHRI protocol on DM)"- PI: Dr. John Day, MD, PhD/ Co-PI: Dr. Tesi Rocha and Karolina Watson, NP

2014- "CHAR0312 Duchenne Muscular Dystrophy Tissue Bank for Exon Skipping",- PI: Dr. John Day, MD, PhD/ Co-PI: Dr. Tesi Rocha.

2014-" A Phase 3 Efficacy and Safety Study of Ataluren (PTC124) in Patients with Nonsense Mutation Dystrophinopathy"- PI: Dr. John Day, MD, PhD/ Co- Investigator-Carly Siskind

2014-Clinical Study of Spinal Muscular Atrophy (PNCR/SMAF protocol)".

CLINICAL TRIALS

- A Study for Participants With Spinal Muscular Atrophy (SMA) Who Previously Participated in Nusinersen (ISIS 396443) Investigational Studies., Recruiting
- Children's Health Research Institute(CHRI), Stanford Lucile Packard Children Hospital (LPCH) Protocol on Myotonic Dystrophy, Recruiting
- Evaluate Safety and Biological Activity of ATYR1940 in Patients With Early Onset Facioscapulohumeral Muscular Dystrophy, Recruiting
- Gene Replacement Therapy Clinical Trial for Patients With Spinal Muscular Atrophy Type 1, Recruiting
- Genetics of Charcot Marie Tooth (CMT) - Modifiers of CMT1A, New Causes of CMT2, Recruiting
- Long-Term Outcomes of Ataluren in Duchenne Muscular Dystrophy, Recruiting
- Multicenter Observational Study of Myotonic Dystrophy Type 1, Recruiting
- Safety and Tolerability of WVE-210201 in Patients With Duchenne Muscular Dystrophy, Recruiting
- Study of SRP-4045 and SRP-4053 in DMD Patients, Recruiting
- A Study to Assess the Efficacy, Safety and Pharmacokinetics of Nusinersen (ISIS 396443) in Infants With Spinal Muscular Atrophy (SMA), Not Recruiting
- Clinical Outcome Study for Dysferlinopathy, Not Recruiting
- Clinical Study of Spinal Muscular Atrophy, Not Recruiting
- Efficacy and Safety of Tideglusib in Congenital Myotonic Dystrophy, Not Recruiting
- Natural History Evaluation of Charcot Marie Tooth Disease (CMT) Types CMT1B, CMT2A, CMT4A, CMT4C, and Others, Not Recruiting

Teaching

STANFORD ADVISEES

Postdoctoral Faculty Sponsor

Tahereh Kamali