

BIOGRAPHICAL SKETCH

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NAME John W. Day, MD, PhD	POSITION TITLE Professor of Neurology, Pediatrics and Pathology, Stanford University		
eRA COMMONS USER NAME johnday			
EDUCATION/TRAINING <i>(Begin with baccalaureate or other initial professional education, such as nursing, and include postdoctoral training.)</i>			
INSTITUTION AND LOCATION	DEGREE <i>(if applicable)</i>	YEAR(s)	FIELD OF STUDY
Oberlin College, Oberlin, Ohio	BA	1973	Physics
University of Minnesota, Minneapolis, Minnesota	MD	1977	Medicine
Albert Einstein College of Medicine	PhD (NeurSc)	1982	Neuroscience
University of California, San Francisco, California		1983-1986	Neurology Residency
University of California, San Francisco, California		1986-1988	Neuromuscular Fellowship

A. Personal Statement

In the 25 years that I have had direct responsibility in caring for patients with neuromuscular disorders I have developed sufficient diagnostic and management skills to attract a large population of muscular dystrophy patients. Relative to this proposal to investigate the potential for cell-based treatment of Duchenne Muscular Dystrophy (DMD), I have helped form one of the primary referral centers for DMD patients in the Upper Midwest – leading to the designation of the UMN/Gillette Muscular Dystrophy Program as one of five national centers in the Muscular Dystrophy Association's competitively established DMD clinical research network. This status led to the UMN/Gillette site being one of the premier sites for recent DMD clinical studies due to the large population of patients and the comprehensive management and research facilities. Because of this prominent research ability we will be able to recruit the subjects for the studies described in this proposal.

Although my primary position has transitioned to Stanford as of September, 2011, I am returning to MN on a monthly basis to continue expanding and utilizing the neuromuscular resources we have developed for clinical research purposes, which will allow me to facilitate the tissue collection proposed in this grant. Furthermore, we are actively developing a comparable resource at Stanford that I will make available to Dr. Perlingiero for her studies. In short, we are uniquely positioned to support the important studies proposed in the application that will provide important insights for development of cell-based treatments of DMD and all forms of muscular dystrophy.

B. Positions and Honors

Positions and Employment

1986-1991	Assistant Professor of Neurology, University of California, San Francisco
1992-1998	Assistant Professor of Neurology, University of Minnesota
1998-2002	Associate Professor of Neurology, University of Minnesota
2002-2011	Professor of Neurology, University of Minnesota
2011-	Adjunct Professor of Neurology, University of Minnesota
2011-	Professor of Neurology, Pediatrics and Pathology, Stanford University

Other Experience and Professional Memberships

1988	Board Certification American Board of Psychiatry and Neurology
1989	Certification American Board of Electrodiagnostic Medicine
1998	American Board of Psychiatry and Neurology, Special Qualifications in Neurophysiology
1994-2011	Director/Co-Director, Muscular Dystrophy Clinic, University of Minnesota
1994-2011	Director, Neuromuscular Histology Laboratory, University of Minnesota
2003-2011	Director, Paul and Sheila Wellstone Muscular Dystrophy Center, University of Minnesota

2005-2010	Medical Advisory Committee, Muscular Dystrophy Association
2006-2011	Advisory Board, Myotonic Dystrophy Foundation
2009-2011	Chair, DSMB CINCH, DMD NIH sponsored clinical trials

Honors

1985	Alpha Omega Alpha
1985	Outstanding teaching award, University of California, San Francisco
1996-2005	Distinguished Teaching Awards, University of Minnesota (x4)
2005	Outstanding Teaching Award, University of Minnesota
2007	Post-Baccalaureate Distinguished Teaching Award, University of Minnesota

B. Selected peer-reviewed publications (selected, in chronological order)

1. Ranum LP, Rasmussen PF, Benzow KA, Koob MD, **Day JW**. Genetic mapping of a second myotonic dystrophy locus. *Nature Genetics*. 19(2):196-8, 1998.
2. **Day JW**, Roelofs R, Leroy B, Pech I, Amos J, Bensow K, Ranum L. Clinical and genetic characteristics of a five generation family with a novel form of myotonic dystrophy. *Neuromuscular Disorders* 9(1):19-27 1999.
3. Koob MD, Moseley M, Schut L, Bird T, Benzow KA, **Day JW**, Ranum LP. Identification of an untranslated CTG expansion in SCA8. *Nature Genetics*. 21(4):379-84, 1999.
4. **Day JW**, L.J. Schut, M.L. Moseley, and L.P.W. Ranum. Spinocerebellar ataxia type 8 (SCA8) Clinical features of a large family. *Neurology* 55:649-657, 2000.
5. 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* 293:864-867, 2001.
6. **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. *Neurol* 60:657-64, 2003.
7. Savkur RS, Philips AV, Cooper TA, Dalton JC, Moseley ML, Ranum LPW, **Day JW**. Insulin Receptor Splicing Alteration in Myotonic Dystrophy Type 2. *Am. J. Hum. Genet.* 74:1309–1313, 2004.
8. 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 LP. Spectrin mutations cause spinocerebellar ataxia type 5. *Nature Genetics* Feb;38(2):184-90. 2006.
9. 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
10. 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* 9(1):61-3, 2008.
11. 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* 15(3):161-5. 2010.
12. 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* 30(12):1657-66. 2009.
13. de Greef JC, Lemmers RJLF, Camano P, **Day JW**, Desnuelle C, Dunand M, 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* 75(17):1548-54. 2010.
14. 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* 32:299–308. 2011.
15. 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* 21(2):89-96. 2011.

C. Research Support

Ongoing Research Support

R01NS056592 (Day) 4/1/07-3/31/12 1.92 calendar

NIH/NINDS

Structural and Functional CNS Changes in Myotonic Dystrophy Types 1 and 2

This grant provides support for MRI and neuropsychological comparisons of DM1 and DM2 adults.

P01 NS058901 (Ranum overall PI) 4/1/08-3/31/13 1.2 calendar

NIH/NINDS

Myotonic Dystrophy: Molecular Pathophysiology and CNS Effects

Project 3 (PI, Day)

Structural and Functional CNS Changes in Children with Myotonic Dystrophy

This funds MRI and neuropsychological studies of DM1 children, complementing NS056592 study of adults.

Muscular Dystrophy Association (Day, PI through 8/2011) 08/01/2008 – 07/31/2011 0.60 calendar

DMD CRN grant

Duchenne Muscular Dystrophy Clinical Trials Network

Clinical trials infrastructure support for DMD studies.

P30 AR057220 (Day, PI through 8/2011) 06/01/2009 – 05/31/2014 1.2 calendar

NIAMS/NINDS

Muscular Dystrophy Center Core Laboratories

This funds infrastructure support for resources to study muscle function and disease.

Recent Prior Research Support

R01-NS040389 (Ranum) 6/1/00-3/31/09 0.48 calendar

NIH/NINDS (Co-Investigator)

Molecular and Genetic Characterization of SCA8

This grant funds molecular characterization of SCA8, and generation of a mouse model of SCA8.

R01-NS056158 (Ranum) 3/1/07-2/28/09 0.48 calendar

NIH/NINDS (Co-Investigator)

Molecular and Genetic Characterization of SCA5

This grant provides funding for role of Beta-III spectrin in spinocerebellar Ataxia Type 5

UMN-KUL (Day) 11/1/07-6/30/09 0.06 calendar

Minnesota Medical Foundation

Stem Cell Program To Treat Duchenne Muscular Dystrophy

Funding for collaboration of UMN and Katholieke Universiteit, Leuven, for cell based DMD treatment.
