



THE NMD INFO



NEWS

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A publication of the Rehabilitation Research and Training Center for the Study of Neuromuscular Diseases
The Department of Physical Medicine and Rehabilitation, School of Medicine, University of California, Davis, 95616

Letter from the Director

I speak for all of us at the Rehabilitation Research and Training Center in Neuromuscular Diseases (RRTC/NMD) when I tell you that we are excited about what we intend to accomplish during the next five-year funding period. Through a comprehensive program of research, training, and information services we will address priorities that were established by our funding agency, the National Institute for Disability and Rehabilitation Research (NIDRR), a division of the Office of Special Education and Rehabilitation Services. Working collaboratively, our five-year plan was shaped by policy makers with knowledge of disability issues, neuromuscular disease (NMD) researchers, and the larger NMD community—patients, their families and other caretakers, physicians, physical therapists, and vocational rehabilitation counselors. We focused on a key concept—relevance. The current NIDRR grant award of \$4 million dollars, after twenty years of NIDRR funding for this project,

demonstrates that we listened and responded effectively to the national dialogue of the disability community.

The mission of the RRTC/NMD 2003-2008 is to enhance the quality of lives of persons with neuromuscular diseases. This means that not only do we work to provide knowledge that will improve the physical condition of individuals with neuromuscular diseases, but we also provide information that will help individuals disabled by NMD to reach their highest possible level of functional independence, the stepping stone to having choices in life. Therefore, rehabilitation is more than arms, legs, and neurons. Ultimately it centers on restoring to a disabled person options in education, employment, family life, and community participation.

The research studies and program initiatives of our center bring together a multidisciplinary team of professionals from

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The mission of the Research and Training Center is to improve the lives of individuals with neuromuscular diseases by developing and evaluating new strategies that address lifelong needs for research-based medical care and counseling, psychosocial well-being, education, and independent living.

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Rehabilitation and Research Training Center in Neuromuscular Diseases 2003-2008

The purpose of the NIDRR Rehabilitation Research and Training Center in Neuromuscular Diseases (RRTC/NMD) is to enhance the health, function and quality of life of persons with neuromuscular diseases (NMD). To achieve this purpose, we have the following goals for 2003-2008:

- 1) Develop a program for multi-center rehabilitation research in NMD;
- 2) Conduct research that continues to address rehabilitation needs, particularly related to exercise, nutrition, pain, secondary conditions, and the quality of life of individuals with neuromuscular diseases;
- 3) Develop and evaluate new or emerging technologies and interventions that provide the information needed to improve employment, community integration, and quality of life outcomes for this population of individuals with disabilities;
- 4) Evaluate health promotion and wellness activities to better understand the factors that enhance the ability of individuals with neuromuscular disease to be physically active and participate in recreational activities; and
- 5) Conduct a comprehensive program of training, dissemination, utilization and technical assistance activities that are well anchored in the research program and address the needs of stakeholders.

Based on a consumer quality of life survey conducted by this RRTC/NMD, consumers with NMD reported that the most significant problems impacting their health, function and quality of life include weakness, breathing problems, pain, fatigue and poor endurance. These problems translate into functional issues,

such as difficulty walking, limitations in their ability to exercise, inability to maintain normal body weight, and problems performing routine activities of daily living. The NIDRR RRTC/NMD research and training program will continue to develop and evaluate interventions directed at these key issues that negatively impact health, function and quality of life. These key interventions include 1) new management strategies for NMD developed through multi-center clinical trials in medical and rehabilitation interventions; 2) application of surgical interventions to improve painful spine deformity; 3) treatment of respiratory insufficiency with pulmonary technologies; and 4) evaluation of activities related to the promotion of health and wellness and interventions to effectively manage disabling pain.

During the course of these research studies, we will develop a clearer understanding of the disease course and impacts of the most prevalent hereditary NMD, as well as effective outcome measurement tools, which are key to testing the usefulness of new medical rehabilitation interventions. The RRTC/NMD 2003-2008 is a National Center of Excellence that will provide consumers with NMD and providers of service with training, technical assistance and access to a wealth of information concerning interventions and rehabilitation approaches to enhance health, function, community integration and quality of life.



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Project 1: Longitudinal Study of the Relationship between Impairment, Activity Limitation, Participation and Quality of Life in Persons with Confirmed Duchenne Muscular Dystrophy (DMD)

Investigator: Craig M. McDonald, M.D. and Cooperative International Neuromuscular Research Group (CINRG)

This project will be conducted in conjunction with an international group, Cooperative International Neuromuscular Research Group (CINRG), to provide a thorough longitudinal description of Duchenne muscular dystrophy. With the large number of patients available through affiliation with CINRG we will be able to conduct a characterization of DMD that would not be possible with the limited number of boys with DMD at a single center. The study will develop standard outcome measures that can be used as benchmarks in the disease process; will examine the occurrence of secondary conditions that typically occur in these boys; and will evaluate the quality

of life of the boys as the disease progresses. Through this thorough characterization, an evidence-based standard of care will be established for use by clinicians who treat boys with DMD and areas requiring further research will be identified. This will not only benefit the patients, but will provide valuable information to the physicians for use in the treatment of boys with DMD.



Project 2. Evaluate Outcomes of Surgical and Medical Rehabilitation Technologies: Impact of Treatment on Health, Wellness and Community Integration

Investigator: Craig M. McDonald, M.D. and Cooperative International Neuromuscular Research Group (CINRG)

In conjunction with the longitudinal study, there will be a concurrent study that will evaluate the effect of surgery to correct spinal deformity in the boys with DMD

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centers involved in physical and rehabilitation medicine from across the country through our new association with CINRG, Center for International Neuromuscular Disease Research. Our extended network of research partnerships also includes the UC Davis School of Medicine, the University of Washington Department of Rehabilitation Medicine, nonprofit organizations such as the Muscular Dystrophy Association, and the many professionals who serve in an advisory capacity to this RRTC.

We are excited as we are beginning the

initial work on the projects that will continue our goal of providing new insights and information that will work toward a better understanding of neuromuscular diseases and providing methods and information that will enhance the health, function, community integration and quality of life of those affected by NMD.

Craig M. McDonald, M.D.

Director
Rehabilitation Research and Training
Center for the Study of Neuromuscular
Diseases

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and in those with spinal muscular atrophy II. In addition to the surgical procedures, the boys will either receive the current standard postoperative care or will receive additional pulmonary support postoperatively (cough assistance and non-invasive pulmonary support). We will examine the impact of the surgery and intervention on activity levels, self care ability, health and wellness and community integration. The results of this project will be evidence-based conclusions that will be incorporated into the standard-of-care for spinal deformity in Duchenne muscular dystrophy and spinal muscular atrophy II initially in the CINRG GROUP and ultimately in all Muscular Dystrophy Association clinics and by clinicians treating individuals with Duchenne muscular dystrophy and spinal muscular atrophy.

Project 3. Community Pulmonary Technologies in NMD: Impact on Hospitalizations, Pain, Community Integration and Quality of Life

Investigator: Joshua O. Benditt, M.D.

Respiratory failure due to respiratory muscle weakness is one of the major causes of death and is a significant contributor to morbidity and diminished quality of life in boys with DMD. Ventilatory support in the form of noninvasive (without tracheostomy) positive pressure ventilation (NPPV) at night is now widespread in application and appears to increase survival and reduce respiratory complications. When compared with standard tracheostomy ventilation, 24-hour-a-day noninvasive respiratory support has been suggested as a treatment that can prolong survival once

24-hour-a-day noninvasive respiratory support has been suggested as a treatment that can prolong survival in boys with DMD

the maximum benefit from using nocturnal NPPV has been reached and that its use can improve the quality of life. This treatment has not been studied prospectively and is not in widespread use despite reports that it may be of significant benefit. The goals of this project are to determine whether a program of 24-hour-a-day NPPV support for patients with Duchenne muscular dystrophy will result in a) improved quality of life and decreased pain; b) decreased hospitalizations; c) increased community integration; and d) increased survival. This study will also result in the development of a set of training materials for patients/families and healthcare providers that will be designed to allow use of these technologies outside of "expert centers".

Project 4. Promotion of Health and Wellness Through the Development and Evaluation of New Technologies to Assess Physical Activity including Recreational Activities and Nutritional Energy Balance

Investigators: D.D. Kilmer, M.D., Project Director and Susan Aitkens, M.S., Ming Sun, Ph.D., Investigators

For persons with physical limitations due to disease or injury, an important goal is the preservation of or improvement in physical abilities. Yet very little is known about the activity patterns of persons with disabilities. It is generally accepted that such individuals are at the forefront of sedentary existence. As new evidence emerges demonstrating the burden of sedentary existence and obesity on the health of the disabled population, studies such as this to relate

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this burden to physical activity, energy expenditure and compliance with health recommendations are vital. An important component of this project will be the adaptation of a new physical activity monitoring system, the Intelligent Device for Energy Expenditure and Activity (IDEEA), to accommodate persons with disabilities, including those in wheelchairs. The development and application of this system will for the first time allow the assessment of the free-living activities of persons with disabilities and will allow comparison of their activities to established health recommendations.

In addition a simple Activity Logging System (ActLog) will be developed that will allow individuals to record their physical activity and their associated perceived exertion during their activities throughout the day. It will calculate the estimated energy expenditure for the activities as they are recorded.

Once validated, these two activity-monitoring systems will be used to determine the activity profile of persons with disability and to assess compliance with recommended physical activity standards. This information in conjunction with assessment of diet will allow determination of energy balance, the essential determinant of weight gain or loss. This information will ultimately allow dietary and activity recommendations to be made to the



disabled that will align them with a healthy life style.

Project 5. Promotion of Health and Wellness Through Community Recreation and Exercise: Impact of Impairment, Pain, Self-efficacy and Environmental Barriers

Investigators: Mark Jensen, Ph.D., R. Ted Abresch, M.S., Co-Project Directors; C.M. McDonald M.D., R.A. Emmons Ph.D., G.T. Carter M.D.

The broad objective of this study is to better understand the factors that contribute to health, wellness, and community integration in persons with slowly progressive NMD. To gain this understanding a survey will be conducted of adults with myotonic muscular dystrophy type 1, facioscapulohumeral muscular dystrophy, Charcot-Marie-Tooth muscular dystrophy and a group of adult controls without NMD. From the results we will ascertain how the frequency of participation in health and wellness activities in adults with slowly progressive NMDs compares to participation by the controls. We will evaluate how participation in these activities relates to other measures of their ability to function in daily activities. These results will allow us to identify factors that impact health promotion practices and recreational opportunities in persons with NMD and identify those activities that are most likely to succeed in promoting participation. We will then be able to develop a model of health promotion that can serve as a basis for the future development of health promotion interventions and we will develop a health promotion guide for

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persons with NMDs that can improve their health, wellness and function. This information will also form the basis of treatment interventions that will improve their health, wellness and function.

NIDRR Priority: Training and Information Services.

Another issue of major importance identified by members of the NMD community is a lack of information on neuromuscular disease and disability-related resources, programs, and services. When persons with a neuromuscular disease say they "lack information," they generally mean two things: they have searched without success for relevant facts that will help them understand their situation and they assume their health care providers know those facts but have failed to share them. When health care professionals say they lack information, they generally mean that they are unaware of clinically applicable information for performance of their specialized task. In both cases the central problem is the same—information on these complex, relatively rare diseases and the long-term life course each takes is neither centralized nor widely disseminated among nonspecialists.

To address the need for information both for those with NMD and doctors who treat them we will develop a series of informative "how to" and informative videos. These videos will be appropriate for use to train both health professionals and those with NMD and their families/support persons in basic procedures such as transfer techniques, pulmonary



options, physical therapy procedures (stretching, contracture prevention, gait analysis), expectations for a diagnostic work-up and other useful subjects.

We will also disseminate appropriate, up-to-date information several ways. Twice a year we will produce a newsletter that will provide updates on recent developments in neuromuscular disease research, informative articles on areas of interest to those with NMD and a resource list. This newsletter will have both a web-based version and a print version. The web-based version will also provide links to other web sites that can be visited to obtain further information.

We will maintain a web site (NMDinfo.net – under construction) that will be kept current with our ongoing research projects, will be updated as new information is available, such as clinical trials for NMD, and will provide links to web sites that will provide information that will work to improve the health and wellness of those with NMD.

Another goal in the area of information services is to provide experts who will make presentations at meetings of patient organizations for various neuromuscular diseases such as Duchenne muscular dystrophy and Charcot-Marie-Tooth dystrophy. We plan to attend one national meeting each year. In this way we will be able to target the information presented to the problems as they apply to a single neuromuscular disease and address those problems that affect neuromuscular diseases in general.

Summary: These ambitious goals for the five-year period, 2003-2008, will result in the changes in the treatment of those with NMD.

RESOURCES
Specifically for Neuromuscular Diseases

RRTC/Neuromuscular Diseases

RRTC/Neuromuscular Diseases
 University of California, Davis
 Med: PM&R TB 191
 Davis, CA 95616
 Phone: Toll Free: 866-508-9656;
 530-752-3447 or 530-752-2903
 E-mail: nmdinfo@ucdavis.edu

The RRTC is part of a nationwide organization of research centers funded by NIDRR. Its mission is to improve the lives of individuals with neuromuscular diseases and their families. The organi-

zation conducts clinical and research studies related to neuromuscular disease. Furthermore, it has several information services and ongoing training programs designed to meet the information needs of the NMD community.

Web site: <http://www.nmdinfo.net>

Besides detailed information on the organization, the website also provides lectures, disease information, links, and other helpful resources regarding neuromuscular disease.

Cooperative International Neuromuscular Research Group (CINRG)

Children's Research Institute
 111 Michigan Ave, NW
 Washington, DC 20010
 Phone: (202) 884-3813
 Fax: (202) 884-6014
 e-mail: lmorton@cnmcresearch.org

The mission of CINRG is, through the conduct of collaborative research and the use of state of the art technologies, to investigate the most promising treatments for the Duchenne Muscular Dystrophy and related neuromuscular disorders, to

improve communication between researchers, physicians, patients and their families, and to establish internationally recognized standards of care for people who suffer from neuromuscular diseases.

Web site: <http://www.cnmcresearch.org/cinrg/index.asp>

The web site presents, an overview of the CINRG program, information on clinical trials and a listing of the sites that are taking part in the CINRG program.

Muscular Dystrophy Association (MDA)

Muscular Dystrophy Association- USA
 National Headquarters
 3300 E. Sunrise Drive
 Tucson, AZ 85718
 Phone: 800-572-1717

Muscular Dystrophy Association (MDA) is a voluntary health agency - a dedicated partnership between scientists and concerned citizens aimed at dominant neuromuscular diseases. MDA

combats neuromuscular diseases through programs of worldwide research, comprehensive medical and community services, and professional and public health education.

Web site: <http://www.mdausa.org>

The MDA website provides extensive information regarding clinical and community services, publications, and research related to muscular dystrophy.

RESOURCES
Specific Neuromuscular Diseases

The FSH Society (Fascioscapulohumeral Dystrophy)

FSH Society Administrative Office:
 Carol A. Perez, Executive Director
 3 Westwood Road
 Lexington, MA 02420
 (781) 860-0501
 carol.perez@fshsociety.org
 The FSH Society is a nonprofit corporation that addresses issues and needs related to FacioScapuloHumeral muscular Dystrophy (FSHD). The Society promotes clinical research and provides

support for those living with FSHD. It also distributes information about the disease to increase public awareness and foster communication among interested parties.

Web site: www.fshsociety.org/

Web site contains general information about the society and the disease. One can also find a bulletin board, a chat room, and many of the society's publications.

Parent Project Muscular Dystrophy

Executive Office:
 1012 North University Boulevard
 Middletown, Ohio 45042
 Pat Furlong, President
 e-mail: pat@parentprojectmd.org
 Phone: 513-424-0696
 Toll-free: 800-714-5437
 Fax: 513-425-9907
 Parent Project Muscular Dystrophy mobilizes people in the United States and Worldwide in collaborative effort to en-

able people with Duchenne and Becker Muscular Dystrophy to survive, thrive and fully participate within their families and communities into adulthood and beyond.

Web site: <http://parentprojectmd.org>

The web site contains up-to-date information on treatment, research and clinical trials for DMD and Becker's dystrophy, information providing emotional support for families and other resources.

Charcot-Marie-Tooth Disease

The Charcot-Marie-Tooth Association
 2700 Chestnut Street
 Chester, PA 19013-4867
 Phone: 800-606-CMTA (2682)
 CMTAssoc@aol.com
 The CMT Assoc. educates, provides support groups, funds research, and promotes public awareness of CMT.
Web: <http://www.charcot-marie-tooth.org/>
 The web site Provides patient support, educational materials, reports on and promotes research, and provides a database (database requires registration).

CMTWorld.org
 P.O. Box 601 Hillsburgh - ON
 N0B 1Z0 Canada
 Phone: +1 (519)855-6376
info@cmtworld.org

CMTWorld, a program of the Canadian Disability Resource Council, provides research and health management information and is dedicated to support those with CMT.

Web site: www.cmtworld.org/index.php

The website provides general information on CMT, research reports, products review, and CMT treatments.