CDC Duchenne/Becker Muscular Dystrophy Activities: August 2004

Muscular dystrophy is a group of genetic disorders characterized by progressive muscle weakness. There are many forms of muscular dystrophy. Different kinds of muscular dystrophy have different genetic causes, affect people at different ages, and affect different muscles. Duchenne muscular dystrophy (DMD) is the most common form of muscular dystrophy in children. Without newborn screening, DMD is usually diagnosed when a child is 3 to 6 years of age. Early signs can include delay in walking, frequent falling, and difficulty getting up from a sitting or lying position. Muscles continue to weaken over time and, around the time they are 12 years of age, children with DMD become unable to walk. The disease is usually fatal in the teens or early 20s, most often due to breathing or heart problems, or both. A milder form of the disease is known as Becker muscular dystrophy (BMD). Together, these two diseases are called Duchenne/Becker muscular dystrophy (DBMD). The gene for DBMD is on the X chromosome, so DBMD affects males almost exclusively. For more information on the causes of DBMD, see www.cdc.gov/ncbddd/duchenne/causes.htm.

In order to provide people with DBMD and their families with the services they need, public health departments and health care providers need better information about DBMD. Here are some of the questions that need to be answered, and why the answers are important:

- *How common is DBMD and is it equally common in different racial and ethnic groups?*
- It is important to know how many people have DBMD so that public health officials and health care providers can make sure that they have enough resources to provide services to everyone. Also, if there are fewer people with DBMD in a particular area than expected, then there might be children with DBMD who are not being diagnosed (in which case health care providers might need better methods of recognizing and diagnosing the condition so that all people with DBMD can have access to optimal treatment).

- *What are the early signs and symptoms of DBMD?* Knowing the early signs and symptoms of DBMD will help physicians create better methods for earlier diagnosis of DBMD by health care providers.

- *Does the type of care received affect the severity or course of DBMD?* It is important to know if different types of care result in different outcomes so that optimal outcome for all people with DBMD can be ensured by developing and promoting standards of care.

- *Does the type of gene changes affect the severity or course of DBMD?* Knowing the effect of different mutations on the severity or course of DBMD will help families to know what to expect so they can plan for the future.

- *What medical and social services are families receiving?* It is important that all families with DBMD receive all of the services that they need. Therefore, it is helpful to know what services families are receiving and what problems they have in getting needed services.

- *Do different groups of people receive different care?* It is important to find groups of people with DMD who are not receiving the same services as others so that equal access to needed services can be ensured.

Using traditional public health research approaches, the Centers for Disease Control and Prevention (CDC) is working with partners in state health departments and universities to answer these and other questions about DBMD. Current CDC projects include:

Muscular Dystrophy Surveillance Tracking and Research Network:

CDC is working with researchers in Arizona, Colorado, Iowa, and western New York State to set up surveillance systems for Duchenne/Becker muscular dystrophy (DBMD). The project is called the Muscular Dystrophy Surveillance Tracking and Research Network, or MD STARnet. The goal of the project is to find all DBMD patients in the areas by using information from different sources, such as clinic medical records and hospital records. Information about each child's treatments and how he is doing medically will be collected from his medical records. Because many DBMD patients are seen in Muscular Dystrophy Association (MDA) clinics, the researchers are working closely with the MDA clinics in their states. In addition, the researchers will be searching for DBMD patients through other neuromuscular clinics, emergency rooms, pathology laboratories, orthopedists, and other muscular dystrophy associations to ensure that all patients with DBMD are included in the project. The states have worked together to come up with a common system that can be used to find patients and collect information. Families who are identified in these areas will be asked to take part in interviews with public health workers to provide information related to DBMD that might not be found in the medical records.

The types of information that will be collected include basic demographic information (such as race and ethnicity), the types of treatments that have been received, the types of clinics that the care was received in, and any medical problems associated with DBMD. Information will be collected from medical records and interviews on a regular basis.

Status: The group has developed and begun using a computer system for saving and combining the information collected. Abstractors started collecting information from medical records April 2004. The group is now developing the interview questionnaire. Fiscal Year 2004 funds will be used to add another state to the system, add an independent quality assurance and control system, and pilot DNA analysis activities for the project.

Family Needs Assessments:

CDC is sponsoring two projects to identify the service needs of families with DBMD. The results of these projects will help health departments and health care providers understand the needs of families with DBMD so that needed resources can be identified.

National Initiative for Families With Duchenne (NIFD): CDC is working with researchers at the Children's National Medical Center in Washington, D.C., on a survey of parents of children with DBMD in the United States and Puerto Rico. The researchers on this project plan to ask parents of children with DBMD about how they have dealt with having a child with DBMD, what services they can get, what services they need, what problems they have in getting needed services, and their feelings about newborn screening for DBMD. The survey will include a large number of families from many backgrounds, and the results will help state health departments improve services for families with DBMD. In addition, because some of the families in this survey were first diagnosed through pilot newborn screening projects, the researchers will find out whether newborn screening had an impact on how families have dealt with the condition, their use of services, and quality of life. This information will help state health departments that are thinking about using newborn screening for DBMD in the future.

Status: The survey has been developed and approved by appropriate Institutional Review Boards (IRBs, or ethics review committees). Survey mailing is scheduled to begin in September 2004.

Needs of Families and Patients with Muscular Dystrophy (NFPMD): CDC is working with researchers at the University of Iowa on a project to identify the needs of families with DMD and childhood-onset BMD. The researchers are talking with individuals with DBMD and their families, specifically, young men with DMD or childhood-onset BMD who are older than 14 years of age, and their parents, brothers, and sisters. The goals of the project are to (1) identify and rank the needs of individuals and families with DMD and childhood-onset BMD at different stages in their lives; (2) identify things that can affect whether families can get services and resources; (3) find out how being told about DMD and childhood-onset BMD affects the individual and his immediate family; and (4) find out how the family feels about newborn screening.

Status: Interview protocols are being developed and should begin in Fall 2004.

Palliative Care and Hospice Needs of Families With Children Who Have Duchenne Muscular Dystrophy: Palliative care is comprehensive care offered to a person with a progressive illness and his or her family, with the goals of improving quality of life and easing symptoms. Palliative care also includes the end-of-life care that is more often thought of as hospice care. Unofficial reports indicate that males with DMD and their families are less likely to use palliative care and hospice services than families with children with other conditions that result in premature death. Because many males with DMD live to be older than 21 years of age, one factor could be the differences in services that are available to pediatric and adult patients. Other factors are also likely to be involved, such as the idea that palliative care is only end-of-life care and a giving up of hope. More research is needed to identify the palliative care and hospice needs of males with DMD and their families, as well as any problems with filling these needs. The two main goals of this project are (1) to identify the palliative care and hospice needs of males with DMD and their families and (2) to identify the barriers that individuals and their families face in considering, seeking, or obtaining palliative and hospice care.

Status: Anticipated award date: September 2004.

Health Care Issues for Hispanic Families With DMD: Hispanic families of children with special health care needs face specific barriers to services and care. The goal of this project is to begin to identify the needs and some of the problems for Hispanic families specific to DMD. The study will help us to start to get some insights into the service needs and barriers to services faced by Hispanic families of children with DMD. One focus group with seven to nine individuals will be conducted in Spanish to collect qualitative information. The information gathered from this initial project will be used to help develop future projects on this topic.

Status: Focus group was conducted August 2004. Anticipated report date: October 2004.

Cardiac Health in Female Carriers of DMD:
Females who are DMD carriers (whether or not they have symptoms) sometimes have heart problems later in adult life that leave them short of breath or unable to do moderate exercise. The chance that a female carrier will develop heart problems is not known. However, such heart problems can be serious and life threatening. While there is no cure, there are a number of medications that might help reduce the effects of these heart problems.

This project will use a large-scale, mailed, self-completed survey to collect information about what female DMD carriers know or believe about cardiac health care and how they act based on this information. The objectives of the project are (1) to find out what things affect the use of preventive cardiac health care by female carriers of DMD and (2) to develop new and workable plans that will increase preventive cardiac health care in this population. At the current time, there are no official recommendations for female carriers of DMD regarding

cardiac testing and treatments. Therefore, the goals of this project are to find ways to let women know about the recommendations once they are available. It is likely that the results of this study can also be used to improve health messages to carriers of other X-linked conditions. This project will be carried out by CDC and researchers at Battelle.

Status: Interviews and focus groups with carrier females and health care providers are scheduled for Fall 2004. The information from these interviews and focus groups will be used to develop the survey instrument.

Newborn Screening for Duchenne Muscular Dystrophy:

Newborn Screening for Duchenne Muscular Dystrophy Workgroup: On March 12, 2004, the CDC sponsored a one-day meeting in Atlanta, Georgia, with experts from around the world to look at newborn screening for DMD. At the meeting, past and present DMD newborn screening programs were discussed, as well as known and potential risks and benefits of such programs

Status: Two reports from the meeting are in preparation. One is a report for the general public, and will be released in September 2004. The second report will be submitted for publication in a peer-reviewed journal. Highlights from the meeting were presented at the NCBDDD Conference in July 2004.

Early Screening and Diagnosis of Duchenne Muscular Dystrophy: To further research on the issues identified by the Newborn Screening for Duchenne Muscular Dystrophy Workgroup, the National Center on Birth Defects and Developmental Disabilities at CDC recently announced funding under a cooperative agreement for research in both infant and newborn screening for DMD (Federal Register: June 24, 2004 (Volume 69, Number 121)). NCBDDD plans to fund two programs. One program will do a trial of newborn screening of DMD and see if the informed consent process works well in the birth hospital. The other program will do a trial of screening during infancy through pediatricians' offices and look at the possibility that everyone will have equal access to the screening test. Both programs will look at how well the informed consent process works, the number of false-positive screening results, the problems that false-positive screening results can cause, what families go through during the screening process, and how pediatricians and other clinicians feel about the screening program. Depending on the applications that are received in response to this announcement, one state might be selected to conduct both newborn and infant screening for DMD, or two separate states might be selected to conduct either newborn or infant screening.

Status: Applications were reviewed in August 2004. Anticipated Award Date: September 2004.

Contact Information:

Website: www.cdc.gov/ncbddd/duchenne/default.htm

E-mail: dmd@cdc.gov

Address: Duchenne/Becker Muscular Dystrophy Program
Division of Human Development and Disability
National Center on Birth Defects and Developmental Disabilities
Centers for Disease Control and Prevention
Mailstop E-88
1600 Clifton Road NE
Atlanta, Georgia 30333