CDC Duchenne/Becker Muscular Dystrophy (DBMD) Activities Update 2005

Background on DBMD:

What is muscular dystrophy?

Muscular dystrophy is a group of genetic disorders characterized by progressive muscle weakness. There are many forms of muscular dystrophy. These forms of muscular dystrophy:

- Affect people at different ages.
- Affect different muscles.
- Have different genetic causes.

What is Duchenne and Becker muscular dystrophy?

Duchenne muscular dystrophy (DMD) is the most common form of muscular dystrophy in children.

- It is most often found when a child is 3 to 6 years of age.
- Early signs can be walking at an older age, falling often, and having problems getting up when sitting or lying.
- Muscles get weaker over time and, when they are about 12 years of age, children with DMD no longer are able to walk.
- DMD is usually fatal in the teens or early 20s, most often due to breathing or heart problems, or both.
- A milder form of DMD is called **Becker muscular dystrophy (BMD**). Together, these two disorders are called Duchenne/Becker muscular dystrophy (DBMD)
- DBMD almost always affects males because the gene associated with DBMD is on the X chromosome. (*Males only have one X chromosome*).
- A female can be a carrier for DBMD, which means she will have one copy of the gene associated with the disorder on one of her X chromosomes. (*Females have two X chromosomes*). Females with one copy of the gene associated with DBMD do not usually have muscular dystrophy, but they may have mild muscle weakness.

What questions about DBMD is CDC working on?

Public health departments and health care providers need better information about Duchenne and Becker muscular dystrophy (DBMD) in order to help people get the services they need. The questions that need to be answered are:

- How common is DBMD?
- Is the rate of DBMD equally common in all racial and ethnic groups?
- What are the early signs and symptoms of DBMD?
- Does the type of care received make a difference in the lives of people affected by the disorder?
- What medical and social services are families getting?
- Do different groups of people get different care?

Current CDC projects and update on activities

Muscular Dystrophy Surveillance Tracking and Research Network (MD STARnet)

Background

Researchers in Arizona, Colorado, Iowa, and New York have set up a tracking system for Duchenne/Becker muscular dystrophy (DBMD). Information will be gathered on a regular basis from medical records and interviews covering topics such as:

- Demographic data (e.g., race and ethnicity).
- Types of treatments that have been received.
- Types of clinics in which the care was received.
- Any medical problems associated with DBMD.

Goal

To find everyone with DBMD in the four states and get information that can be used by health care providers and policy makers to improve care for people with DBMD and their families.

How will people with DBMD be found?

A number of different sources will be used to find people with DBMD, including:

- Neuromuscular clinic medical records (such as those from Muscular Dystrophy Association (MDA) clinics).
- Hospital records.
- Emergency rooms.
- Pathology laboratories.
- Orthopedists.
- Muscular dystrophy organizations, including Parent Project Muscular Dystrophy (PPMD) and MDA

Project update August 2005

- Medical record data gathering began in the four states in April 2004.
- A caregiver interview has been developed, and families will be invited to take part in January 2006.
- The state of Georgia will be added to MD STARnet in fall 2005.
- An independent organization was hired in June 2005 to conduct quality assurance and control for data gathering methods for MD STARnet.

Family Needs Assessment Projects

Why the projects are needed?

Health departments and health care providers have to understand the needs of families affected by Duchenne/Becker muscular dystrophy (DBMD) so that resources can be identified. CDC is sponsoring two projects to identify what services families with DBMD need:

- **National Initiative for Families With Duchenne (NIFD)**
- **Needs of Families and Patients With Muscular Dystrophy (NFPMD)**

National Initiative for Families with Duchenne

National Initiative for Families With Duchenne (NIFD)

Background

CDC is working with researchers at the Children's National Medical Center in Washington, D.C., and Columbia University in New York on a survey of parents of children with Duchenne/Becker muscular dystrophy (DBMD) in the United States and Puerto Rico. The survey will include a large number of families from many backgrounds. Information collected from the survey will include:

- What services families are getting.
- What services families need.
- What problems families have in getting the services they need.
- How families feel about newborn screening for DBMD.

In addition, directors of clinics that provide care for people with DBMD will be asked about the services that are provided in their areas. This information will help state health departments make decisions about other programs related to these disorders.

Goal

- To improve services for families with DBMD.

Project update August 2005

- The survey has been developed and approved by the appropriate Institutional Review Boards (IRBs).
- Families and physicians will be invited to take part beginning in fall of 2005.

NFPMD project *Needs of Families and Patients With Muscular Dystrophy (NFPMF)*

Background

Researchers at the University of Iowa are funded by CDC to identify the needs of families with Duchenne muscular dystrophy (DMD) and childhood-onset Becker muscular dystrophy (BMD). The researchers are talking with people in families affected by Duchenne/Becker muscular dystrophy (DBMD) in Iowa, including:

- Young men with DMD, or childhood-onset BMD, who are older than 14 years of age.
- The parents, brothers, and sisters of young men with DBMD.

Goals

- Identify and rank the needs of individuals with and families affected by DMD and childhood-onset BMD at different stages in their lives.
- Identify factors that can affect whether families can get services and resources they need.
- Find out how being told about DMD and childhood-onset BMD affects the person with DBMD and his immediate family.
- Find out how the family feels about newborn screening.

Project update August 2005

- Interview protocols have been developed, and families will be invited to participate in January 2006.

Palliative Care and Hospice Needs of Families With Children Who Have Duchenne Muscular Dystrophy

What is palliative care?

Palliative care is comprehensive care offered to a person with a progressive illness with the goals of improving quality of life and easing symptoms. Palliative care also includes the end-of-life care that is more commonly known as hospice care.

Background

Unofficial reports indicate that many males with Duchenne muscular dystrophy (DMD) and their families do not receive palliative care services. The reasons why are not known, but could be related to:

- The difficulties in changing from pediatric to adult health care as males with DMD live to be older than 21 years of age.
- The idea that palliative care is only for end-of-life care and indicates giving up of hope.

More research is needed to identify the palliative care of males with DMD and their families, as well as any problems meeting these needs.

Goals

- To identify the palliative care and hospice needs of males with DMD and their families
- To identify the barriers that people with DMD and their families face in thinking about, looking for, or getting palliative and hospice care.

Project Update August 2005

- This project has been awarded to the researchers at MD STARnet Arizona.
- The survey instrument for this project has been developed and will be conducted as part of the interview process that is beginning in January 2006

Health Care Issues for Hispanic Families With DMD

Background

Hispanic families of children with special health care needs face specific barriers to services and care. This study will help begin gathering some insights into the service needs of and barriers to services faced by Hispanic families of children with Duchenne muscular dystrophy (DMD).

- One focus group with seven to nine people was conducted in Spanish to get information about what people think and how they feel about these issues.

Goal

To begin to identify the needs and some of the problems of Hispanic families specific to DMD.

Project Update August 2005

- A focus group was conducted in August 2004. (Report is available on the CDC website. See contact information that follows.)
- In fall 2005, a focus group with non-Hispanic families will be conducted, and the results of the two focus groups will be compared.

Cardiac Health in Female Carriers of Duchenne/Becker muscular dystrophy (DBMD)

Background

Females can be a carrier for Duchenne/Becker muscular dystrophy (DBMD), which means they will have one copy of the gene associated with the disorder, but they do not usually have muscular dystrophy. They may have mild muscle weakness and a small number of carriers develop heart problems 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 medicines that might help reduce the effects of these heart problems.

A large-scale, mailed, self-completed survey will be used to collect information about what female DBMD carriers know or believe about cardiac health care and how they act based on this information. This project will be carried out by CDC and other researchers at Battelle Centers for Public Health Research and Evaluation.

Goals

At the current time, there are no official recommendations for female carriers of DBMD regarding cardiac testing and treatments. Therefore, the goals of this project are:

- To find out what factors affect the use of preventive cardiac health care by female carriers of DBMD.
- To develop effective educational materials about preventive cardiac health care for women who are carriers of DBMD.
- To find ways to let women know about the recommendations once they are available.
- To improve health messages to carriers of other X-linked conditions.

Project Update
August 2005

- Interviews and focus groups with female carriers and health care providers were conducted in spring 2005. (A report from these activities will be available in fall 2005.)
- The survey has been developed and is awaiting approval by the U. S. Office of Management and Budget.
- The survey is expected to be mailed in January 2006.

Newborn Screening for Duchenne Muscular Dystrophy (DMD)

Past Activities:

Issues related to newborn and infant screening for DBMD can be complicated. These issues include how to make sure people understand the screening process and all possible outcomes, and how to make sure everyone can get screening. On March 12, 2004, CDC sponsored a one-day meeting in Atlanta, Georgia, with experts from around the world to look at newborn screening for Duchenne muscular dystrophy (DMD). This expert group formed the Newborn Screening for Duchenne Muscular Dystrophy Workgroup. At the meeting, past and present DMD newborn screening programs were discussed, as well as known and potential risks and benefits of such programs. A full report of the meeting is available on the CDC website (see contact information that follows).

Early Screening and Diagnosis of Duchenne Muscular Dystrophy

Background

CDC funded two research projects to gather more information on issues identified by the Newborn Screening for Duchenne Muscular Dystrophy Workgroup related to newborn and infant screening for DMD.

- The Children's Research Institute in Ohio will offer a test to screen newborns for DMD before they leave the hospital to see if people understand the screening process and all possible outcomes.
- Emory University in Atlanta, Georgia will offer a test to screen infants for DMD through pediatricians' offices and see if it is possible that everyone will have equal access to the screening test.

The two groups will study issues such as:

- How well the informed consent process works.
- What problems false-positive screening results can cause.
- Families' experiences with the screening programs.
- How pediatricians and other clinicians feel about the screening programs.

Project Update August 2005

- Both groups are conducting laboratory studies to validate the screening test.
- Emory University is currently conducting focus groups to determine what type of information is useful to parents in deciding about the infant screening test.
- Pilot newborn screening and infant screening programs will be reviewed by the appropriate ethics committees, and are expected to begin in January 2006.

How to get more information on these projects and other topics on Duchenne and Becker muscular dystrophy (DBMD)	Website: www.cdc.gov/ncbddd/duchenne/index.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