

## MUSCULAR DYSTROPHY

### WHAT IS THE PUBLIC HEALTH ISSUE?

Duchenne muscular dystrophy (DMD) affects about 1 in 4,000 males and is the most common form of muscular dystrophy in children. In the absence of newborn screening, DMD is usually diagnosed around children 3 to 6 years of age. Early signs include failure to walk by 18 months, frequent falling, difficulty getting up from a sitting or lying position, and a waddling gait. As muscle deterioration progresses, children with DMD become unable to walk around 12 years of age. The disease is fatal in the teens or early twenties, due to severe respiratory and heart problems. A milder form of the disease, Becker muscular dystrophy, is caused by mutations in the same gene. The combined spectrum is referred to as DBMD. Standard birth-defects monitoring systems in the United States do not detect children with DBMD because children do not have recognizable signs or symptoms at birth. Consequently, existing birth-defects monitoring systems would need to be supplemented with additional activities to find all cases of DBMD.

In addition to Muscular Dystrophy Association clinics and other muscular dystrophy clinics, cases may be ascertained through state or regional muscular dystrophy chapters or associations, hospitals, private physicians, and diagnostic laboratories. The lack of a uniform standard of care for DBMD results in inter-clinic variation in treatment options. A long-term follow-up study of children with DBMD is necessary to evaluate different treatment options and to develop and evaluate standards of care. In addition, these activities will serve as the basis for a population-based assessment of the impact of DBMD.

### WHAT HAS CDC ACCOMPLISHED?

- Convened meetings with stakeholders to identify key epidemiologic research questions as well as research needs related to families with DBMD.
- Developed a plan for monitoring activities and awarded cooperative agreements to Arizona, Colorado, Iowa, and New York.
- Financed a survey of carrier females, women who carry the mutation for DBMD, but do not have DBMD.
- Funded the Children's National Medical Center in Washington, D.C., to develop materials and conduct a family needs assessment.
- Funded the University of Iowa to develop a system that can pool data across sites and conduct a qualitative study of family needs.
- Hired a parent consultant and public health genetic counselor to assist in various aspects of the program.
- Supported the efforts of existing state partners to enable them to conduct annual interviews of families participating in the family needs assessment project.
- Work with grantees to develop data collection strategies for DBMD.

### WHAT ARE THE NEXT STEPS?

- Add an additional state to the surveillance network (depending on objective review panel findings).
- Continue to work closely with stakeholders, keeping them updated as new activities become operational.
- Convene national expert meetings on newborn screening for DBMD and on treatment.
- Fund one to two states to conduct a feasibility planning study for newborn screening for DBMD.
- Work with grantees to develop optimal case finding activities.

For additional information on this or other CDC programs, visit [www.cdc.gov/program](http://www.cdc.gov/program)

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