Prevalence of Duchenne/Becker Muscular Dystrophy Among Males Aged 5--24 Years --- Four States, 2007

Muscular dystrophies are a group of genetic diseases characterized by progressive skeletal muscle weakness and muscle cell death with replacement of muscle cells by fibrosis and fat (1). The most common muscular dystrophy in children is Duchenne muscular dystrophy (DMD), which predominantly affects males (2). Historically, DMD has resulted in loss of ambulation between ages 7 and 13 years and death in the teens or 20s (3). The average age at diagnosis is 5 years, despite earlier onset of symptoms (4). Becker muscular dystrophy is similar to DMD but has later onset and slower, more variable progression of symptoms. Birth prevalence of DMD has been estimated at 1 in 3,500 (2.9 per 10,000) male births and Becker muscular dystrophy at 1 in 18,518 (0.5 per 10,000) male births (5). To estimate the population-based prevalence of Duchenne/Becker muscular dystrophy (DBMD) and describe selected clinical outcomes, CDC and investigators from the Muscular Dystrophy Surveillance Tracking and Research Network (MD STARnet) analyzed data for males born during 1983--2002 that were reported to the MD STARnet from four participating states. This report summarizes those findings, which indicated overall state-specific prevalences on January 1, 2007, of 1.3--1.8 per 10,000 males aged 5--24 years. Among MD STARnet subjects, more than 90% of males with DBMD aged ≥15 years used wheelchairs. Nearly 60% of males with DBMD born during 1983--1987 had survived through 2007, emphasizing the need to develop and implement programs that address lifelong needs of males with DBMD.

Since 2004, MD STARnet has conducted named population-based surveillance of DBMD in four states (Arizona, Colorado, Iowa, and 12 counties* in western New York) for males born on or after January 1, 1982 (6). Cases are ascertained by reviewing medical records of patients in hospitals or specialty clinics using the International Classification of Diseases, Ninth Revision, Clinical Modification (ICD-9) nonspecific muscular dystrophy code (359.1). Trained abstractors screen medical records to determine if the patient might have DBMD. Medical records of potential cases are abstracted using a standardized tool to collect data on clinical signs and symptoms, family history, screening and diagnostic tests, medical treatments, clinical outcome (e.g., death and mobility), and case demographics. Clinical reviewers apply MD STARnet standard case definition criteria (6)† to classify each case as definite, probable, or possible. Cases meeting the definite and probable definitions are included in this report.

The analyses in this report used two different time frames. For the calculation of prevalence rates and the use of wheelchairs among males with DBMD identified by MD STARnet, January 1, 2007, was used. December 31, 2007, was used to report deaths and survival. For each of the four sites, residency was defined as having a home address in the corresponding state and at least one clinic visit during 2005--2007. Denominators used for rate calculations were the male resident population from the U.S. Census Bureau's 2007 population estimates (8). For the analysis of wheelchair use, an indicator of mobility impairment, any use of a wheelchair, scooter, or stroller (excluding use of a stroller before age 7 years) was considered to be wheelchair use. Because of the progressive nature of DBMD, wheelchair use was assumed to continue after initiation.

To describe the death and survival of males with DBMD, MD STARnet subjects born during 1983--2002 and who ever resided in the four geographic areas were included in the analysis. Known deaths that occurred through December 31, 2007, were identified from clinic reports. Investigators used state death certificate files, the National Death Index, and/or Social Security files to ascertain additional deaths. Known deaths were included in the analysis regardless of the state where they occurred. Subjects with no report of death were classified as survivors (i.e., males with a clinic visit during 2005--2007) or lost to follow-up (i.e., no clinic visit during that period and no report of death).

On January 1, 2007, 452 definite or probable cases of DBMD in males born during 1983--2002 were identified in the four states. Of these, 46 (10%) died before the end of 2006 and 57 (13%) were lost to follow-up. The remaining 349 males with definite or probable DBMD were used to calculate point prevalence in 2007 (Table 1). Overall DBMD prevalence rates per 10,000 males aged 5--24 years ranged from 1.3 (Arizona) to 1.8 (western New York). Age- and state-specific prevalences per 10,000 males ranged from 0.9 (Iowa) to 1.9 (western New York) for males aged 5--9 years; 1.4 (Colorado) to 2.5 (Iowa) for males aged 10--14 years; 1.6 (Arizona) to 2.5 (Colorado) for males aged 15--19 years; and 0.8 (Arizona) to 1.1 (western New York) for males aged 20--24 years. For the 349 males with DBMD at the beginning of 2007, the age-specific percentages for those who used wheelchairs were 29% at age 5--9 years and >90% at age ≥15 years (Table 2).

During 2007, the number of deaths increased from 46 to 58. Of the 12 additional deaths, 10 were counted in the prevalence analysis and...
two had been categorized as lost to follow-up on January 1, 2007. Death records indicated that the two males who had been categorized as lost to follow-up died in 2007. Of the 58 deaths, 54 (93%) were attributable to DBMD. Of the remaining four, one resulted from poisoning, one was attributed to an unintentional injury, and two resulted from unknown causes. Among subjects born during 1983--2002, survival ranged from 58% for the oldest males (1983--1987) to 100% for youngest group (1998--2002) (Table 3).

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**Editorial Note:**

This report describes the first population-based assessment of DBMD prevalence in the United States. The analysis used data from the four states participating in MD STARnet, a surveillance system for DBMD that is unique because it is population-based, ascertains DBMD cases using a nonspecific ICD-9 code augmented by medical record review, and follows cases longitudinally. The results indicate an overall site-specific prevalence at January 1, 2007, of 1.3--1.8 per 10,000 males aged 5--24 years. Comparable studies for this population are not available. However, newborn screening for males with DMD range from 1 in 7,730 (1.3 per 10,000) to 1 in 3,871 (2.6 per 10,000) (8).

In all four states, lower prevalences were observed consistently for the youngest and oldest groups, compared with the middle two age groups. The lower prevalences for males aged 5--9 years (born during 1998--2002) might reflect diagnostic delays previously reported by MD STARNet (4). The case classification used by MD STARNet rigorously defines probable or definite cases, but might not identify all cases of DBMD. This might be especially true for males aged 20--24 years (born during 1983--1987) because widespread use of genetic diagnostic tests did not begin until the early 1990s. Additionally, this group has the lowest survival.

Data show that some males (29%) who have DBMD experience impaired mobility by the end of their first decade of life and more than 90% used wheelchairs by the end of their second decade of life. Although not showing age distribution of wheelchair use, a previous study reported loss of ambulation at a mean age of 9.4 years and dependency on an electric wheelchair at a mean age of 14.6 years (9). More public health programs that promote early identification of motor delays leading to diagnosis of DBMD at an earlier age would provide opportunities for prescribing mobility aids and initiating therapeutic interventions to support prolonged ambulation.

The analysis of survival among MD STARNet subjects indicated that all males born during 1993--2002 (corresponding to ages 5--9 years) were surviving at the end of 2007, and 60% of those born during 1983--1987 (corresponding to ages 20--24 years) were surviving. A 2002 British study, which only included males who have DMD, reported mean survival of 19--25 years, and survival was related to receipt of respiratory therapy (10). As males with DBMD increasingly survive into adulthood, practitioners who care for adults are increasingly challenged to manage patients who have rare disorders, such as DMD, who historically did not survive to adulthood. Programs are needed to address their changing medical, psychosocial, educational, and vocational needs. For example, public health programs that promote earlier diagnosis of DMD might allow earlier prescription of mobility aids and earlier initiation of therapeutic interventions to support prolonged ambulation.

The findings in this report are subject to at least five limitations. First, approximately 13% of MD STARNet subjects were lost to follow-up. Cases lost to follow-up were excluded from analyses because neither residency nor survival could be confirmed. If cases lost to follow-up were included in the analyses, prevalence rates could increase and the percentage of survivors would increase or decrease. Second, as for all surveillance systems, some cases might not be ascertained, which could lower prevalence rates. Circumstances reducing case ascertainment include frequent delays in diagnosis (4), problems locating the records of males born more than 20 years ago (records were archived or lost), and out of state medical care. Third, current case classification, which rigorously defines probable or definite cases, might underrepresent true identification of all cases of DBMD, especially for males born during 1983--1987. Fourth, the variable follow-up period was too brief to observe long-term survival for all males with DBMD. Finally, because the analysis was conducted in just four states, these findings are not nationally representative.

DBMD results from both familial and new genetic mutations; therefore, any newborn male can be affected. Accurate population based prevalence estimates of DBMD provide information to policy makers, health-care providers, and payers for the planning and provision of services.

**Acknowledgments**

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References


* Includes Allegany, Cattaraugus, Chautauqua, Erie, Genesee, Livingston, Monroe, Niagara, Ontario, Orleans, Wayne, and Wyoming counties. The 12 counties represented 14% of males aged 5--24 years in New York.
† Definite cases were symptomatic and had 1) a documented dystrophin mutation, or 2) a muscle biopsy that showed an abnormality of dystrophin with no alternative explanation identified, or 3) an elevated creatine kinase (CK) level, a pedigree compatible with X-linked inheritance, and an affected family member with dystrophin mutation or dystrophin abnormality on muscle biopsy. Probable cases were symptomatic, had an elevated CK level, and had an X-linked pedigree consistent with a dystrophinopathy. Possible cases were symptomatic and had an elevated CK level.

What is already known on this topic?

Most previous prevalence studies of Duchenne/Becker muscular dystrophy (DBMD) have been conducted in clinical populations, and no population-based study has been conducted in the United States.

What is added by this report?

In 2007, the estimated prevalence of DBMD at four U.S. sites was 1.3--1.8 per 10,000 males aged 5--24 years, and a majority of surveillance subjects were surviving into adulthood.

What are the implications for public health practice?

More public health interventions, such as educational and vocational support, and access to and use of health care addressing DBMD patient physical and psychosocial needs are needed to support these patients and their families as they transition from childhood into adulthood.

### TABLE 1. Overall and age-specific prevalence of Duchenne/Becker muscular dystrophy among males, by age group --- Muscular Dystrophy Surveillance Tracking and Research Network (MD STARnet), four states, January 1, 2007

<table>
<thead>
<tr>
<th>Age group (yrs)</th>
<th>Arizona</th>
<th>Colorado</th>
<th>Iowa</th>
<th>Western New York*</th>
</tr>
</thead>
<tbody>
<tr>
<td>5--9</td>
<td>26</td>
<td>232,419</td>
<td>1.1</td>
<td>23</td>
</tr>
<tr>
<td>10--14</td>
<td>35</td>
<td>226,682</td>
<td>1.5</td>
<td>23</td>
</tr>
<tr>
<td>Age group (yrs)</td>
<td>No.</td>
<td>Using wheelchair</td>
<td></td>
<td></td>
</tr>
<tr>
<td>----------------</td>
<td>-----</td>
<td>------------------</td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>No.</td>
<td>(%)</td>
<td></td>
<td></td>
</tr>
<tr>
<td>5--9</td>
<td>72</td>
<td>21 (29)</td>
<td></td>
<td></td>
</tr>
<tr>
<td>10--14</td>
<td>102</td>
<td>84 (82)</td>
<td></td>
<td></td>
</tr>
<tr>
<td>15--19</td>
<td>118</td>
<td>110 (93)</td>
<td></td>
<td></td>
</tr>
<tr>
<td>20--24</td>
<td>57</td>
<td>52 (91)</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

* Excludes stroller use before age 7 years.
§ All residential cases as of January 1, 2007, meeting the definite or probable case definition used by MD STARnet (7).

### TABLE 3. Survival* of males diagnosed with Duchenne/Becker muscular dystrophy (DBMD), by years of birth --- Muscular Dystrophy Surveillance Tracking and Research Network (MD STARnet), four states,† December 31, 2007

<table>
<thead>
<tr>
<th>Years of birth</th>
<th>No.</th>
<th>Lost to follow-up ¶</th>
<th>Deaths</th>
<th>Survival (%)**</th>
</tr>
</thead>
<tbody>
<tr>
<td>1998--2002</td>
<td>76</td>
<td>4</td>
<td>0</td>
<td>(100)</td>
</tr>
<tr>
<td>1993--1997</td>
<td>118</td>
<td>15</td>
<td>1</td>
<td>(99)</td>
</tr>
</tbody>
</table>

* Excludes stroller use before age 7 years.
§ All residential cases as of January 1, 2007, meeting the definite or probable case definition used by MD STARnet (7).
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1988--1992  151  18  20  (85)
1983--1987  107  18  37  (58)

§ Subjects who met the definite or probable case definition used by MD STARnet (7).
¶ Two males with DBMD were lost to follow-up as of January 1, 2007, but had a death record in 2007. These males are included as deaths.
** [(No. - lost to follow-up - deaths) / (No. - lost to follow-up)] x 100.