

110TH CONGRESS  
2D SESSION

# S. 2618

To amend the Public Health Service Act to provide for research with respect to various forms of muscular dystrophy, including Becker, congenital, distal, Duchenne, Emery-Dreifuss facioscapulohumeral, limb-girdle, myotonic, and oculopharyngeal muscular dystrophies.

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## IN THE SENATE OF THE UNITED STATES

FEBRUARY 8 (legislative day, FEBRUARY 6), 2008

Ms. KLOBUCHAR (for herself, Mr. ISAKSON, Mr. BROWN, Ms. COLLINS, Mr. COLEMAN, and Mr. HARKIN) introduced the following bill; which was read twice and referred to the Committee on Health, Education, Labor, and Pensions

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## A BILL

To amend the Public Health Service Act to provide for research with respect to various forms of muscular dystrophy, including Becker, congenital, distal, Duchenne, Emery-Dreifuss facioscapulohumeral, limb-girdle, myotonic, and oculopharyngeal muscular dystrophies.

1       *Be it enacted by the Senate and House of Representa-  
2       tives of the United States of America in Congress assembled,*

3       **SECTION 1. SHORT TITLE.**

4       This Act may be cited as the “Paul D. Wellstone  
5       Muscular Dystrophy Community Assistance, Research,  
6       and Education Amendments Act of 2008”.

## 1 SEC. 2. FINDINGS.

2 Congress makes the following findings:

3 (1) The muscular dystrophies are devastating  
4 diseases that have a significant impact on quality of  
5 life, not only for the individual who experiences its  
6 painful symptoms and resulting disability, but also  
7 for family members and caregivers.

(3) Myotonic muscular dystrophy is the second most prominent form of muscular dystrophy and the type most commonly found in adults, affecting an estimated 1 in 8,000 people. However, it can affect people of any age, from birth to old age. Described as the most variable disease known in medicine, it is multisystemic and can cause not only muscle atrophy and myotonia, but also serious cardiac, respiratory, endocrine, gastrointestinal, skeletal and central nervous system complications, as well as problems with the eyes, teeth, and hair. As it passes

1 from one generation to the next, it generally worsens  
2 with earlier onset. Congenital myotonic muscular  
3 dystrophy is the most severe form of myotonic muscular  
4 dystrophy affecting infants and causing severe  
5 cognitive delays. It often causes sudden death; how-  
6 ever, others can live for many years with this slowly  
7 degenerative disorder.

8 (4) Facioscapulohumeral muscular dystrophy  
9 (referred to in this section as “FSHD”) is the sec-  
10 ond most prevalent adult muscular dystrophy and  
11 the third most prevalent muscular dystrophy of men,  
12 women and children. It is inherited genetically and  
13 has an estimated incidence of 1 in 20,000 persons.  
14 Many leading FSHD scientists note that the preva-  
15 lence may be 3 times higher due to undiagnosed and  
16 misdiagnosed cases. FSHD, affecting between  
17 15,000 to 40,000 persons, causes a lifelong progres-  
18 sive and severe loss of all skeletal muscles gradually  
19 bringing weakness and reduced mobility. It is geneti-  
20 cally transmitted to children, can occur spontane-  
21 ously, and may affect entire families. Persons with  
22 FSHD may also experience hearing loss, vision prob-  
23 lems, and respiratory insufficiency; some may be-  
24 come severely physically disabled and spend decades  
25 in a wheelchair and on a ventilator. FSHD is caused

1 by a novel epigenetic phenomenon not found in other  
2 forms of muscular dystrophy and is caused by a con-  
3 traction of repetitive DNA previously thought to be  
4 “junk DNA”. The unique epigenetic structure of  
5 FSHD is unprecedented in other muscular dys-  
6 trophies and genetic disorders and demands novel  
7 approaches and new research groups. Understanding  
8 this mechanism will have great benefit to other areas  
9 of biomedical research including cancer and other  
10 disease of epigenetic origin.

11 (5) Congenital muscular dystrophies represent a  
12 group of distinct diseases, which begin at birth, with  
13 varying severity and involvement of both muscle  
14 strength and brain. These diseases often lead to pre-  
15 mature infant death, or severely disabled young chil-  
16 dren who require 24-hour care given their develop-  
17 mental delay compounded by muscle weakness.  
18 Other children live to young adulthood and typically  
19 require the use of a wheelchair for mobility.

20 (6) Forms of muscular dystrophy affecting chil-  
21 dren and adults include Becker, congenital, distal,  
22 Duchenne, Emery-Dreifuss, facioscapulohumeral,  
23 limb-girdle, myotonic, and oculopharyngeal muscular  
24 dystrophies. The limb-girdle muscular dystrophies  
25 are of 15 known different types.

19 (9) Investments made by the Federal Govern-  
20 ment as a result of the MD-CARE Act include the  
21 creation of the MD Coordinating Committee  
22 (MDCC), the development of the MDCC Action  
23 Plan, expansion of the National Institutes of Health  
24 (referred to in this section as the “NIH”) research  
25 portfolios, establishment of 6 Paul D. Wellstone

1       Muscular Dystrophy Cooperative Research Centers,  
2       funding of a \$15,400,000 National Institutes of  
3       Health U54 grant and others focused on DMD, de-  
4       velopment of the Muscular Dystrophy Surveillance,  
5       Tracking and Research Network (MD STARnet),  
6       and the launch of a comprehensive education and  
7       outreach initiative.

8                   (10) In the past few years, the NIH program  
9       in translational research in muscular dystrophy has  
10       grown significantly and funded a number of large-  
11       scale projects to further the development of thera-  
12       pies for muscular dystrophy. As part of this pro-  
13       gram, the National Institute of Neurological Dis-  
14       orders and Stroke (NINDS) and the National Insti-  
15       tute of Arthritis and Musculoskeletal and Skin Dis-  
16       eases (NIAMS) of the National Institutes of Health  
17       (NIH) awarded a \$15,400,000, 5-year cooperative  
18       agreement to develop new small molecule drugs for  
19       the treatment of DMD and potentially other forms  
20       of muscular dystrophy as well. The project is a  
21       unique research collaboration between private, pub-  
22       lic, and nonprofit partners to build upon previous re-  
23       search and discovery work originally initiated by  
24       non-profit partners to identify new treatments for  
25       muscular dystrophy. Also through the translational

1 program, 3 other major cooperative agreements have  
2 been awarded for highly targeted therapy develop-  
3 ment projects in the muscular dystrophies.

4 (11) Due to the initiatives made possible  
5 through the MD-CARE Act, national nonprofit orga-  
6 nizations have joined in model strategic collabora-  
7 tions with academic research institutions, public  
8 funders of muscular dystrophy research, and indus-  
9 try to expand investments in muscular dystrophy re-  
10 search activities and to create new platforms for  
11 translational research. These have led to the devel-  
12 opment of the first potential therapies for DMD,  
13 myotonic, facioscapulohumeral, limb-girdle, and  
14 other conditions that are proceeding through clinical  
15 trials.

16 (12) Advancements in care have helped prolong  
17 life and quality of life for patients with muscular  
18 dystrophy.

19 (13) Notwithstanding these promising develop-  
20 ments, the majority of the directions envisioned by  
21 the Action Plan for the Muscular Dystrophies, devel-  
22 oped pursuant to the MD-CARE Act, have not been  
23 realized. Where recent momentum has been  
24 achieved, its sustainability is fragile and directly de-  
25 pending upon continued Federal support for the

1       early phase planning and programs created through  
2       the MD-CARE Act.

3               (14) There remains a shortage of qualified re-  
4       searchers in the field of muscular dystrophy re-  
5       search. Many family physicians and health care pro-  
6       fessionals still lack the knowledge and resources to  
7       detect and properly diagnose muscular dystrophy as  
8       early as possible, thus delaying management of  
9       symptoms in cases that go undetected or  
10      misdiagnosed.

11               (15) As new understandings of the genetic basis  
12      for disease and potential treatment has emerged, the  
13      public and health care communities are in urgent  
14      need of education and outreach to ensure competent,  
15      informed engagement in genetic testing and coun-  
16      seling and appropriate patient characterization so  
17      that patients are able to participate in new avenues  
18      of research and clinical trials.

19               (16) As basic research into the muscular dys-  
20      trophies points the way to new therapeutic targets,  
21      there is an urgent need to support the clinical re-  
22      search infrastructure necessary to bring these thera-  
23      peutic leads to human trials; these infrastructure  
24      needs include validated endpoints, current natural

1 history studies, biomarkers, clinical research net-  
2 works, patient registries, and databases.

3 (17) In order to improve lives and develop effec-  
4 tive treatments for individuals with muscular dys-  
5 trophy, there must be improved communications and  
6 partnerships between patients, patient advocacy, re-  
7 searchers, and clinical care providers. To that end,  
8 renewed effort to work together by all parties is a  
9 critical element for successful outcomes in the years  
10 to come.

11 (18) Continued focus and investment are re-  
12 quired to build on the current momentum, respond  
13 to public need, and ensure that federally funded re-  
14 search and other innovation is translated to thera-  
15 peutic targets as quickly as possible.

16 **SEC. 3. EXPANSION, INTENSIFICATION, AND COORDINA-**  
17 **TION OF ACTIVITIES OF NATIONAL INSTI-**  
18 **TUTES OF HEALTH WITH RESPECT TO RE-**  
19 **SEARCH ON MUSCULAR DYSTROPHY.**

20 Section 404E of the Public Health Service Act (42  
21 U.S.C. 283g) is amended—

22 (1) in subsection (a)(1), by inserting “the Na-  
23 tional Heart, Lung, and Blood Institute,” after  
24 “Child Health and Human Development,”;

5 (3) by adding at the end the following:

6       “(h) CLINICAL RESEARCH.—The Coordinating Com-  
7       mittee shall give special consideration to the urgent need  
8       to enhance the clinical research infrastructure required to  
9       test emerging therapies for the various forms of muscular  
10      dystrophy by prioritizing the achievement of those goals  
11      in the plan related to this topic.

12       “(i) AUTHORIZATION OF APPROPRIATIONS.—There is  
13       authorized to be appropriated to carry out this section,  
14       such sums as may be necessary for each of fiscal years  
15       2008 through 2012.”.

16 SEC. 4. DEVELOPMENT AND EXPANSION OF ACTIVITIES OF  
17 CENTERS FOR DISEASE CONTROL AND PRE-  
18 VENTION WITH RESPECT TO EPIDEMIOLOG-  
19 ICAL RESEARCH ON MUSCULAR DYSTROPHY.

20 Section 317Q of the Public Health Service Act (42  
21 U.S.C. 247b-18) is amended—

22 (1) by redesignating subsection (d) as sub-  
23 section (f); and

24 (2) by inserting after subsection (c) the fol-  
25 lowing:

1       “(d) DATA.—In carrying out this section, the Sec-  
2  retary shall ensure that any data on patients that is col-  
3 lected as part of the Muscular Dystrophy Surveillance,  
4  Tracking and Research Network (referred to in this sec-  
5  tion as the ‘MD STARnet’) under a grant under this sec-  
6  tion is regularly updated to reflect changes in patient con-  
7  dition over time, particularly with respect to any improve-  
8  ments realized through patient adherence to care consider-  
9  ations or utilization of a treatment or therapy.

10     “(e) REPORTS AND TRACKING.—

11     “(1) ANNUAL REPORT.—Not later than 18  
12  months after the date of enactment of the Paul D.  
13  Wellstone Muscular Dystrophy Community Assist-  
14  ance, Research, and Education Amendments Act of  
15  2008, and annually thereafter, the Director of the  
16  Centers for Disease Control and Prevention shall  
17  submit to the appropriate committees of Congress a  
18  report—

19           “(A) concerning the activities carried out  
20  by MD STARnet sites funded under this sec-  
21  tion during the year for which the report is pre-  
22  pared;

23           “(B) containing the data collected and  
24  findings derived from the MD STARnet sites  
25  each fiscal year (as funded under a grant under

1           this section during the periods of fiscal years  
2           2008 through 2012); and

3           “(C) that every 2 years outlines prospec-  
4           tive data collection objectives and strategies.

5           “(2) TRACKING HEALTH OUTCOMES.—The Di-  
6           rector of the Centers for Disease Control and Pre-  
7           vention shall make publicly available prospective  
8           health outcome data on the health and survival of  
9           people with muscular dystrophy.”.

10 **SEC. 5. INFORMATION AND EDUCATION.**

11           Section 5 of the Muscular Dystrophy Community As-  
12           sistance, Research and Education Amendments of 2001  
13           (42 U.S.C. 247b–19) is amended—

14           (1) by redesignating subsection (c) as sub-  
15           section (d); and

16           (2) by inserting after subsection (b), the fol-  
17           lowing:

18           “(c) REQUIREMENTS OF THE CENTERS FOR DISEASE  
19           CONTROL AND PREVENTION.—In carrying out this sec-  
20           tion, the Director of the Centers for Disease Control and  
21           Prevention shall—

22           “(1) partner with leaders in the muscular dys-  
23           trophy patient community; and

24           “(2) widely disseminate the Duchenne-Becker  
25           Muscular Dystrophy care considerations described in

1 section 904 as broadly as possible, including through  
2 partnership opportunities with the muscular dys-  
3 trophy patient community.”.

#### 4 SEC. 6. STANDARDS OF CARE.

5       Part A of title IX of the Public Health Service Act  
6 (42 U.S.C. 299 et seq.) is amended by adding at the end  
7 the following:

## 8 "SEC. 904. STANDARDS OF CARE RELATING TO MUSCULAR

## 9 DYSTROPHY.

## 10        “The Director shall—

11                 “(1) evaluate the available scientific evidence  
12                 for the appropriate medical or patient organizations  
13                 for purposes of the development and issuance of an  
14                 initial set of care considerations for Duchenne-Beck-  
15                 er Muscular Dystrophy and provide ongoing review  
16                 and updates where appropriate; and

17           “(2) replicate the same systematic review meth-  
18       odology used to develop the Duchenne-Becker Mus-  
19       cular Dystrophy care considerations developed under  
20       paragraph (1) as a model for other muscular dys-  
21       trophies.”.