

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.

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

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.

8 (2) Duchenne muscular dystrophy (referred to
9 in this section as “DMD”) is the most common le-
10 thal genetic disorder of childhood worldwide, affect-
11 ing approximately 1 in every 3,500 boys born each
12 year around the globe. It is characterized by a rap-
13 idly progressive muscle weakness that almost always
14 results in death from respiratory or cardiac failure,
15 typically in the late teens or twenties.

16 (3) Myotonic muscular dystrophy is the second
17 most prominent form of muscular dystrophy and the
18 type most commonly found in adults, affecting an
19 estimated 1 in 8,000 people. However, it can affect
20 people of any age, from birth to old age. Described
21 as the most variable disease known in medicine, it
22 is multisystemic and can cause not only muscle atro-
23 phy and myotonia, but also serious cardiac, res-
24 piratory, endocrine, gastrointestinal, skeletal and
25 central nervous system complications, as well as
26 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 mus-
4 cular 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.

1 (7) Each of the muscular dystrophies, though
2 distinct in progressivity and severity of symptoms,
3 has a devastating impact on hundreds of thousands
4 of children and adults throughout the United States
5 and worldwide, as well as imposes severe physical
6 and economic burdens on those affected. In many of
7 the muscular dystrophies, there are associated med-
8 ical problems arising from pulmonary issues, res-
9 piratory insufficiency, cardiomyopathy, which in
10 many cases is the cause of death for persons with
11 muscular dystrophy.

12 (8) In the 5 years since enactment of the Mus-
13 cular Dystrophy Community Assistance, Research
14 and Education Amendments of 2001 (the MD-
15 CARE Act (Public Law 107–84)), and due directly
16 to the momentum established by such Act, progress
17 has been made in the battle against the muscular
18 dystrophies.

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 pendent 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,”;

1 (2) in subsection (b)(1), by adding at the end
 2 the following: “Such centers of excellence shall be
 3 known as the ‘Paul D. Wellstone Muscular Dys-
 4 trophy Cooperative Research Centers’.”; and

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.”.

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