

110TH CONGRESS
2D SESSION

H. R. 5265

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 HOUSE OF REPRESENTATIVES

FEBRUARY 7, 2008

Mr. ENGEL (for himself and Mr. BURGESS) introduced the following bill;
which was referred to the Committee on Energy and Commerce

A BILL

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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 of 2008”.

1 **SEC. 2. FINDINGS.**

2 The Congress finds as follows:

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) DMD is the most common lethal genetic
9 disorder of childhood worldwide, affecting approxi-
10 mately 1 in every 3,500 boys born each year around
11 the globe. It is characterized by a rapidly progressive
12 muscle weakness that almost always results in death
13 from respiratory or cardiac failure, typically in the
14 late teens or twenties.

15 (3) Myotonic muscular dystrophy is the second
16 most prominent form of muscular dystrophy and the
17 type most commonly found in adults affecting an es-
18 timated 1 in 8,000 people. However, it can affect
19 people of any age—from birth to old age. Described
20 as the most variable disease known in medicine, it
21 is multi-systemic and can cause not only muscle at-
22 rophy and myotonia, but also serious cardiac, res-
23 piratory, endocrine, gastrointestinal, skeletal and
24 central nervous system complications, as well as
25 problems with the eyes, teeth and hair. As it passes
26 from one generation to the next, it generally worsens

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

7 (4) Facioscapulohumeral muscular dystrophy
8 (referred to in this section as “FSHD”) is the sec-
9 ond most prevalent adult muscular dystrophy and
10 the third most prevalent muscular dystrophy of men,
11 women and children. It is inherited genetically and
12 has an estimated incidence of 1 in 20,000 persons.
13 Many leading FSHD scientists note that the preva-
14 lence may be three times higher due to undiagnosed
15 and misdiagnosed cases. FSHD, affecting between
16 15,000 to 40,000 persons, causes a lifelong progres-
17 sive and severe loss of all skeletal muscles gradually
18 bringing weakness and reduced mobility. It is geneti-
19 cally transmitted to children, can occur spontane-
20 ously, and may affect entire families. Persons with
21 FSHD may also experience hearing loss, vision prob-
22 lems and respiratory insufficiency; some may become
23 severely physically disabled and spend decades in a
24 wheelchair and on a ventilator. FSHD is caused by
25 a novel epigenetic phenomenon not found in other

1 forms of muscular dystrophy and is caused by a con-
2 traction of repetitive DNA previously thought to be
3 “junk DNA”. The unique epigenetic structure of
4 FSHD is unprecedented in other muscular dys-
5 trophies and genetic disorders and demands novel
6 approaches and new research groups. Understanding
7 this mechanism will have great benefit to other areas
8 of biomedical research including cancer and other
9 disease of epigenetic origin.

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

19 (6) Forms of muscular dystrophy affecting chil-
20 dren and adults include Becker, congenital, distal,
21 Duchenne, Emery-Dreifuss, facioscapulohumeral,
22 limb-girdle, myotonic, and oculopharyngeal muscular
23 dystrophies. The limb-girdle muscular dystrophies
24 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 (MD-CARE
15 Act) and due directly to the momentum established
16 by the MD-CARE Act, progress has been made in
17 the battle against the Muscular Dystrophies.

18 (9) Investments made by the Federal Govern-
19 ment as a result of the MD-CARE Act include the
20 creation of the MD Coordinating Committee
21 (MDCC), the development of the MDCC Action
22 Plan, expansion of the NIH research portfolios, es-
23 tablishment of 6 Paul D. Wellstone Muscular Dys-
24 trophy Cooperative Research Centers (co-funded, in
25 part, by a national non-profit health organization),

1 development of the Muscular Dystrophy Surveil-
2 lance, Tracking and Research Network (MD
3 STARnet), and the launch of a comprehensive edu-
4 cation and outreach initiative.

5 (10) In the past few years, the NIH program
6 in translational research in muscular dystrophy has
7 grown significantly and funded a number of large-
8 scale projects to further the development of thera-
9 pies for muscular dystrophy. As part of this pro-
10 gram, the National Institute of Neurological Dis-
11 orders and Stroke (NINDS) and the National Insti-
12 tute of Arthritis and Musculoskeletal and Skin Dis-
13 eases (NIAMS) of the National Institutes of Health
14 (NIH) awarded a \$15.4 million, five-year cooperative
15 agreement to develop new small molecule drugs for
16 the treatment of Duchenne muscular dystrophy
17 (DMD) and potentially other forms of muscular dys-
18 trophy as well. The project is a unique research col-
19 laboration between private, public, and non-profit
20 partners to build upon previous research and dis-
21 covery work originally initiated by non-profit part-
22 ners to identify new treatments for muscular dys-
23 trophy. Also through the translational program,
24 three other major cooperative agreements have been

1 awarded for highly targeted therapy development
2 projects in the muscular dystrophies.

3 (11) Due to the initiatives made possible
4 through the MD-CARE Act, national non-profit or-
5 ganizations have joined in model strategic collabora-
6 tions with academic research institutions, public
7 funders of MD research, and industry to expand in-
8 vestments in MD research activities and to create
9 new platforms for translational research. These have
10 led to the development of the first potential thera-
11 pies for DMD, myotonic, facioscapulohumeral, limb-
12 girdle and other conditions that are proceeding
13 through clinical trials.

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

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

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

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

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

1 (17) In order to improve lives and develop effective
2 treatments for individuals with muscular dystrophy,
3 there must be improved communications and partnerships
4 between patients, patient advocacy, researchers, and clinical care providers. To that end,
5 renewed effort to work together by all parties is a critical element for successful outcomes in the years
6 to come.

9 (18) Continued focus and investment are required to build on the current momentum, respond
10 to public need, and ensure that federally funded research and other innovation is translated to therapeutic
11 targets as quickly as possible.

14 **SEC. 3. EXPANSION, INTENSIFICATION, AND COORDINATION OF ACTIVITIES OF NIH WITH RESPECT**
15 **TO RESEARCH ON MUSCULAR DYSTROPHY.**

17 (a) **TECHNICAL CORRECTION.**—Section 404E of the
18 Public Health Service Act (42 U.S.C. 283g) is amended
19 by striking subsection (f) (relating to reports to Congress)
20 and redesignating subsection (g) as subsection (f).

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

23 (1) in subsection (a)(1), by inserting “the National Heart, Lung, and Blood Institute,” after “the
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1 Eunice Kennedy Shriver National Institute of Child
2 Health and Human Development,”;

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

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

8 “(g) CLINICAL RESEARCH.—The Coordinating Com-
9 mittee shall give special consideration to the urgent need
10 to enhance the clinical research infrastructure required to
11 test emerging therapies for the various forms of muscular
12 dystrophy by prioritizing the achievement of the goals re-
13 lated to this topic in the plan under subsection (e)(1).

14 “(h) AUTHORIZATION OF APPROPRIATIONS.—There
15 are authorized to be appropriated to carry out this section
16 such sums as may be necessary for each of fiscal years
17 2008 through 2012.”.

18 **SEC. 4. DEVELOPMENT AND EXPANSION OF ACTIVITIES OF**
19 **CDC WITH RESPECT TO EPIDEMIOLOGICAL**
20 **RESEARCH ON MUSCULAR DYSTROPHY.**

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

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

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

3 “(d) DATA.—In carrying out this section, the Sec-
4 retary shall ensure that any data on patients that is col-
5 lected as part of the Muscular Dystrophy STARnet (under
6 a grant under this section) is regularly updated to reflect
7 changes in patient condition over time, particularly with
8 respect to any improvements realized through patient ad-
9 herence to care considerations or utilization of a treatment
10 or therapy.

11 “(e) REPORTS AND STUDY.—

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

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

24 “(B) containing the data collected and
25 findings derived from the MD STARnet sites

1 each fiscal year (as funded under a grant under
2 this section during fiscal years 2008 through
3 2012); and

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

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

11 **SEC. 5. INFORMATION AND EDUCATION.**

12 Section 5 of the Muscular Dystrophy Community As-
13 sistance, Research and Education Amendments of 2001
14 (42 U.S.C. 247b–19) is amended—

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

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

19 “(c) REQUIREMENTS OF CDC.—In carrying out this
20 section, the Director of the Centers for Disease Control
21 and 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 as broadly

1 as possible, including through partnership opportuni-
2 ties with the muscular dystrophy patient commu-
3 nity.”.

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 methodology used to de-
18 velop the Duchenne-Becker muscular dystrophy care
19 considerations developed under paragraph (1) as a
20 model for other muscular dystrophies.”.

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