

111TH CONGRESS  
1ST SESSION

# S. 2858

To amend the Public Health Service Act to establish an Office of Mitochondrial Disease at the National Institutes of Health, and for other purposes.

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## IN THE SENATE OF THE UNITED STATES

DECEMBER 9, 2009

Mrs. BOXER (for herself, Mr. DURBIN, Mr. KERRY, and Mr. CASEY) 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 establish an Office of Mitochondrial Disease at the National Institutes of Health, and for other purposes.

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 “Brittany Wilkinson  
5 Mitochondrial Disease Research and Treatment Enhance-  
6 ment Act”.

7 **SEC. 2. FINDINGS AND PURPOSE.**

8 (a) FINDINGS.—Congress finds the following:

1           (1) Mitochondrial disease results when there is  
2           a defect that reduces the ability of the mitochondria  
3           in a cell to produce energy. As mitochondria fail to  
4           produce enough energy, the cells will cease to func-  
5           tion properly and will eventually die. Organ systems  
6           will begin to fail, and the life of the individual is  
7           compromised or ended.

8           (2) There are more than 40 mitochondrial dis-  
9           eases.

10          (3) Mitochondrial diseases are a relatively newly  
11          diagnosed group of diseases, first recognized in the  
12          late 1960s. Diagnosis of these diseases is extremely  
13          difficult.

14          (4) Mitochondrial diseases can present them-  
15          selves at any age, with associated mortality rates  
16          that vary depending upon the particular disease. The  
17          most severe diseases result in progressive loss of  
18          neurological and liver function, and death within  
19          several years.

20          (5) According to the National Institute of Envi-  
21          ronmental Health Sciences, half of those affected by  
22          mitochondrial disease are children, who show symp-  
23          toms before age five and approximately 80 percent  
24          of whom will not survive beyond the age of 20.

1           (6) Mitochondrial dysfunction is also associated  
2 with numerous other disorders, including many neu-  
3 rological diseases (such as Parkinson's, Alzheimer's,  
4 ALS, and autism), and other diseases associated  
5 with aging, diabetes, and cancer.

6           (7) Mitochondrial diseases are most commonly  
7 the result of genetic mutation, either in the nuclear  
8 DNA or in the mitochondrial DNA. Some  
9 mitochondrial diseases have been attributable to en-  
10 vironmental factors that interfere with mitochondrial  
11 function.

12           (8) Researchers estimate that one in 4,000 chil-  
13 dren will develop a mitochondrial disease related to  
14 an inherited mutation by the age of 10 years, and  
15 that 1,000–2,000 children are born each year in the  
16 United States who will develop mitochondrial disease  
17 in their lifetimes. However, studies of umbilical cord  
18 blood samples show that one in 200 children are  
19 born with both normal and mutant mitochondrial  
20 DNA, and the number of children with these  
21 mutations who actually develop a disease is un-  
22 known.

23           (9) There are no cures for any of the specifi-  
24 cally identified mitochondrial diseases, nor is there a  
25 specific treatment for any of these diseases.



1 **“SEC. 404H. OFFICE OF MITOCHONDRIAL DISEASE.**

2       “(a) ESTABLISHMENT.—There is established within  
3 the Office of the Director of NIH at the Division of Pro-  
4 gram Coordination, Planning and Strategic Initiatives, an  
5 office to be known as the Office of Mitochondrial Disease  
6 (in this section referred to as the ‘Office’), which shall be  
7 headed by a Director (in this section referred to as the  
8 ‘Director’), appointed by the Director of NIH.

9       “(b) MITOCHONDRIAL DISEASE RESEARCH PLAN.—

10           “(1) IN GENERAL.—The Director shall develop,  
11 make publicly available, and implement a written  
12 plan to facilitate and coordinate research into  
13 mitochondrial disease.

14           “(2) CONTENTS.—The plan required under  
15 paragraph (1) shall include the following objectives:

16           “(A) Improving coordination of research  
17 related to mitochondrial disease among the na-  
18 tional research institutes and between the Na-  
19 tional Institutes of Health and outside research-  
20 ers.

21           “(B) Providing training to research sci-  
22 entists and clinical researchers engaged in re-  
23 search related to mitochondrial disease.

24           “(C) Conducting programs to provide in-  
25 formation and continuing education to health

1 care providers regarding the diagnosis of  
2 mitochondrial disease.

3 “(D) Ensuring relevant scientific review  
4 groups contain individuals with expertise in  
5 mitochondrial disease.

6 “(3) CONSULTATION.—In developing the plan  
7 under paragraph (1), the Director shall consult  
8 with—

9 “(A) the Director of the National Cancer  
10 Institute;

11 “(B) the Director of the National Institute  
12 of Child Health and Human Development;

13 “(C) the Director of the National Institute  
14 of Environmental Health Sciences;

15 “(D) the Director of the National Heart,  
16 Lung, and Blood Institute;

17 “(E) the Director of the National Institute  
18 of Neurological Disorders and Stroke;

19 “(F) the Director of the National Institute  
20 of Diabetes and Digestive and Kidney Diseases;

21 “(G) the Director of the National Eye In-  
22 stitute;

23 “(H) the Director of the National Institute  
24 of Mental Health;

1           “(I) the Director of the National Institute  
2           of Arthritis and Musculoskeletal and Skin Dis-  
3           eases;

4           “(J) the Director of the National Human  
5           Genome Research Institute; and

6           “(K) the heads of such other institutes and  
7           offices as the Director considers appropriate.

8           “(4) UPDATES.—The Director shall update the  
9           plan required under paragraph (1) on a biennial  
10          basis.

11          “(c) RESEARCH GRANTS.—In addition to any grants  
12          otherwise awarded by the National Institutes of Health  
13          for research in mitochondrial disease, the Director may  
14          award competitive, peer-reviewed grants—

15                 “(1) for integrated, multi-project research pro-  
16                 grams related to mitochondrial disease; and

17                 “(2) for planning activities associated with inte-  
18                 grated, multi-project research programs related to  
19                 mitochondrial disease.

20          “(d) CENTERS OF EXCELLENCE.—

21                 “(1) IN GENERAL.—The Director may award  
22                 grants to institutions or consortiums of institutions  
23                 to establish Mitochondrial Disease Centers of Excel-  
24                 lence to promote interdisciplinary research and  
25                 training related to mitochondrial disease.

1           “(2) USE OF FUNDS AWARDED.—A grant  
2 awarded under paragraph (1) may be used to—

3           “(A) conduct basic and clinical research re-  
4 lated to mitochondrial disease;

5           “(B) facilitate training programs for re-  
6 search scientists and health professionals seek-  
7 ing to engage in research related to  
8 mitochondrial disease; and

9           “(C) develop and disseminate programs  
10 and materials to provide continuing education  
11 to health care professionals regarding the rec-  
12 ognition, diagnosis, and treatment of  
13 mitochondrial disease.

14           “(e) NATIONAL REGISTRY; BIOREPOSITORY.—

15           “(1) NATIONAL REGISTRY.—The Director of  
16 the Centers for Disease Control and Prevention shall  
17 establish a national registry for the maintenance and  
18 sharing for research purposes of medical information  
19 collected from patients with mitochondrial disease.

20           “(2) BIOREPOSITORY.—The Director of the  
21 Centers for Disease Control and Prevention shall es-  
22 tablish a national biorepository for the maintenance  
23 and sharing for research purposes of tissues and  
24 DNA collected from patients with mitochondrial dis-  
25 ease.

1       “(f) DEFINITION.—In this section, the term  
2 ‘mitochondrial disease’ means mitochondrial diseases,  
3 mutations, dysfunctions and functions.

4       “(g) AUTHORIZATION OF APPROPRIATIONS.—There  
5 is authorized to be appropriated, such sums as may be  
6 necessary to carry out this section.”.

7       (b) DEVELOPMENT OF MITOCHONDRIAL DISEASE  
8 RESEARCH PLAN.—The Director of the Office of  
9 Mitochondrial Disease shall develop and make publicly  
10 available the mitochondrial disease research plan required  
11 under section 404H(b)(1) of the Public Health Service  
12 Act, as added by subsection (a) of this section, not later  
13 than 180 days after the date of the enactment of this Act.

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