

**Calendar No. 298**

107<sup>TH</sup> CONGRESS  
1<sup>ST</sup> SESSION

**S. 1379**

**[Report No. 107-129]**

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

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IN THE SENATE OF THE UNITED STATES

AUGUST 3, 2001

Mr. KENNEDY (for himself, Mr. HATCH, Mr. HOLLINGS, Mr. BINGAMAN, Mr. DURBIN, Mr. JEFFORDS, Mrs. CLINTON, Mr. SMITH of Oregon, Ms. COLLINS, and Mr. KERRY) introduced the following bill; which was read twice and referred to the Committee on Health, Education, Labor, and Pensions

DECEMBER 18, 2001

Reported by Mr. KENNEDY, with an amendment

[Strike out all after the enacting clause and insert the part printed in *italic*]

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**A BILL**

To amend the Public Health Service Act to establish an Office of Rare Diseases 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,*

1 **SECTION 1. SHORT TITLE.**

2 This Act may be cited as the “Rare Diseases Act of  
3 2001”.

4 **SEC. 2. FINDINGS AND PURPOSES.**

5 (a) FINDINGS.—Congress makes the following find-  
6 ings:

7 (1) Rare diseases and disorders are those which  
8 affect small patient populations, typically popu-  
9 lations smaller than 200,000 individuals in the  
10 United States. Such diseases and conditions include  
11 Huntington’s disease, amyotrophic lateral sclerosis  
12 (Lou Gehrig’s disease), Tourette syndrome, Crohn’s  
13 disease, cystic fibrosis, cystinosis, and Duchenne  
14 muscular dystrophy.

15 (2) For many years, the 25,000,000 Americans  
16 suffering from the over 6,000 rare diseases and dis-  
17 orders were denied access to effective medicines be-  
18 cause prescription drug manufacturers could rarely  
19 make a profit from marketing drugs for such small  
20 groups of patients. The prescription drug industry  
21 did not adequately fund research into such treat-  
22 ments. Despite the urgent health need for these  
23 medicines, they came to be known as “orphan  
24 drugs” because no companies would commercialize  
25 them.

1           (3) During the 1970s, an organization called  
2           the National Organization for Rare Disorders  
3           (NORD) was founded to provide services and to  
4           lobby on behalf of patients with rare diseases and  
5           disorders. NORD was instrumental in pressing Con-  
6           gress for legislation to encourage the development of  
7           orphan drugs.

8           (4) The Orphan Drug Act created financial in-  
9           centives for the research and production of such or-  
10          phan drugs. New federal programs at the National  
11          Institutes of Health and the Food and Drug Admin-  
12          istration encouraged clinical research and commer-  
13          cial product development for products that target  
14          rare diseases. An Orphan Products Board was estab-  
15          lished to promote the development of drugs and de-  
16          vices for rare diseases or disorders.

17          (5) Before 1983, some 38 orphan drugs had  
18          been developed. Since the enactment of the Orphan  
19          Drug Act, more than 220 new orphan drugs have  
20          been approved and marketed in the United States  
21          and more than 800 additional drugs are in the re-  
22          search pipeline.

23          (6) Despite the tremendous success of the Or-  
24          phan Drug Act, rare diseases and disorders deserve  
25          greater emphasis in the national biomedical research

1 enterprise. The Office of Rare Diseases at the Na-  
2 tional Institutes of Health was created in 1993, but  
3 lacks a statutory authorization.

4 (7) The National Institutes of Health has re-  
5 ceived a substantial increase in research funding  
6 from Congress for the purpose of expanding the na-  
7 tional investment of the United States in behavioral  
8 and biomedical research.

9 (8) Notwithstanding such increases, funding for  
10 rare diseases and disorders at the National Insti-  
11 tutes of Health has not increased appreciably.

12 (9) To redress this oversight, the Department  
13 of Health and Human Services has proposed the es-  
14 tablishment of a network of regional centers of excel-  
15 lence for research on rare diseases.

16 (10) The Food and Drug Administration sup-  
17 ports small clinical trials through Orphan Products  
18 Research Grants. Such grants embody successful  
19 partnerships of government and industry, and have  
20 led to the development of at least 23 drugs and four  
21 medical devices for rare diseases and disorders. Yet  
22 the appropriations in Fiscal Year 2001 for such  
23 grants were less than in Fiscal Year 1995.

24 (b) PURPOSES.—The purposes of this Act are to—

1           (1) amend the Public Health Service Act to es-  
 2           tablish an Office of Rare Diseases at the National  
 3           Institutes of Health; and

4           (2) increase the national investment in the de-  
 5           velopment of diagnostics and treatments for patients  
 6           with rare diseases and disorders.

## 7   **TITLE I—NATIONAL INSTITUTES** 8           **OF HEALTH**

### 9   **SEC. 101. NIH OFFICE OF RARE DISEASES.**

10          Title IV of the Public Health Service Act (42 U.S.C.  
 11   281 et seq.) is amended by inserting after section 404D  
 12   the following:

13                           “OFFICE OF RARE DISEASES

14          “SEC. 404E. (a) ESTABLISHMENT.—There is estab-  
 15   lished within the Office of the Director of NIH an office  
 16   to be known as the Office of Rare Diseases (in this section  
 17   referred to as the ‘Office’), which shall be headed by a  
 18   Director (in this section referred to as the ‘Director’), ap-  
 19   pointed by the Director of NIH.

20                           “(b) DUTIES.—

21                           “(1) IN GENERAL.—The Director of the Office  
 22   shall carry out the following:

23                           “(A) The Director shall recommend an  
 24                           agenda for conducting and supporting research  
 25                           on rare diseases through the national research  
 26                           institutes and centers. The agenda shall provide

1 for a broad range of research and education ac-  
2 tivities, including scientific workshops and  
3 symposia to identify research opportunities for  
4 rare diseases.

5 “(B) The Director shall, with respect to  
6 rare diseases, promote coordination and co-  
7 operation among the national research insti-  
8 tutes and centers and entities whose research is  
9 supported by such institutes.

10 “(C) The Director shall enter into coopera-  
11 tive agreements with and make grants for re-  
12 gional centers of excellence on rare diseases in  
13 accordance with section 404F.

14 “(D) The Director shall promote the suffi-  
15 cient allocation of the resources of the National  
16 Institutes of Health for conducting and sup-  
17 porting research on rare diseases.

18 “(E) The Director shall promote and en-  
19 courage the establishment of a centralized  
20 clearinghouse for rare and genetic disease infor-  
21 mation that will provide understandable infor-  
22 mation about these diseases to the public, med-  
23 ical professionals, patients and families.

24 “(F) The Director shall biennially prepare  
25 a report that describes the research and edu-

1           cation activities on rare diseases being con-  
 2           ducted or supported through the national re-  
 3           search institutes and centers, and that identi-  
 4           fies particular projects or types of projects that  
 5           should in the future be conducted or supported  
 6           by the national research institutes and centers  
 7           or other entities in the field of research on rare  
 8           diseases.

9           “(G) The Director shall prepare the NIH  
 10          Director’s annual report to Congress on rare  
 11          disease research conducted by or supported  
 12          through the national research institutes and  
 13          centers.

14          “(2) PRINCIPAL ADVISOR REGARDING ORPHAN  
 15          DISEASES.—With respect to rare diseases, the Direc-  
 16          tor shall serve as the principal advisor to the Direc-  
 17          tor of NIH and shall provide advice to other relevant  
 18          agencies. The Director shall provide liaison with na-  
 19          tional and international patient, health and scientific  
 20          organizations concerned with rare diseases.

21          “(e) DEFINITION.—For purposes of this section, the  
 22          term ‘rare disease’ means any disease or condition that  
 23          affects less than 200,000 persons in the United States.

24          “(d) AUTHORIZATION OF APPROPRIATIONS.—For the  
 25          purpose of carrying out this section, there are authorized

1 to be appropriated \$4,000,000 for fiscal year 2002, and  
 2 such sums as may be necessary for each subsequent fiscal  
 3 year.”.

4 **SEC. 102. RARE DISEASE REGIONAL CENTERS OF EXCEL-**  
 5 **LENCE.**

6 Title IV of the Public Health Service Act (42 U.S.C.  
 7 281 et seq.), as amended by section 101, is further amend-  
 8 ed by inserting after section 404E the following:

9 “RARE DISEASE REGIONAL CENTERS OF EXCELLENCE  
 10 “SEC. 404F. (a) COOPERATIVE AGREEMENTS AND  
 11 GRANTS.—

12 “(1) IN GENERAL.—The Director of the Office  
 13 of Rare Diseases (in this section referred to as the  
 14 ‘Director’) shall enter into cooperative agreements  
 15 with and make grants to public or private nonprofit  
 16 entities to pay all or part of the cost of planning, es-  
 17 tablishing, or strengthening, and providing basic op-  
 18 erating support for regional centers of excellence for  
 19 clinical research into, training in, and demonstration  
 20 of diagnostic, prevention, control, and treatment  
 21 methods for rare diseases.

22 “(2) POLICIES.—A cooperative agreement or  
 23 grant under paragraph (1) shall be entered into in  
 24 accordance with policies established by the Director  
 25 of NIH.



1       “(b) COORDINATION WITH OTHER INSTITUTES.—

2   The Director shall coordinate the activities under this sec-  
3   tion with similar activities conducted by other national re-  
4   search institutes, centers and agencies of the National In-  
5   stitutes of Health and by the Food and Drug Administra-  
6   tion to the extent that such institutes, centers and agen-  
7   cies have responsibilities that are related to rare diseases.

8       “(c) USES FOR FEDERAL PAYMENTS UNDER COOP-

9   ERATIVE AGREEMENTS OR GRANTS.—Federal payments  
10   made under a cooperative agreement or grant under sub-  
11   section (a) may be used for—

12           “(1) staffing, administrative, and other basic  
13           operating costs, including such patient care costs as  
14           are required for research;

15           “(2) clinical training, including training for al-  
16           lied health professionals, continuing education for  
17           health professionals and allied health professions  
18           personnel, and information programs for the public  
19           with respect to rare diseases; and

20           “(3) clinical research and demonstration pro-  
21           grams.

22       “(d) PERIOD OF SUPPORT; ADDITIONAL PERIODS.—

23   Support of a center under subsection (a) may be for a  
24   period of not to exceed 5 years. Such period may be ex-  
25   tended by the Director for additional periods of not more

1 than 5 years if the operations of such center have been  
 2 reviewed by an appropriate technical and scientific peer  
 3 review group established by the Director and if such group  
 4 has recommended to the Director that such period should  
 5 be extended.

6 “(e) AUTHORIZATION OF APPROPRIATIONS.—For the  
 7 purpose of carrying out this section, there are authorized  
 8 to be appropriated \$20,000,000 for fiscal year 2002, and  
 9 such sums as may be necessary for each subsequent fiscal  
 10 year.”.

## 11 **TITLE II—FOOD AND DRUG** 12 **ADMINISTRATION**

### 13 **SEC. 201. GRANTS AND CONTRACTS FOR THE DEVELOP-** 14 **MENT OF ORPHAN DRUGS.**

15 Subsection (e) of section 5 of the Orphan Drug Act  
 16 (21 U.S.C. 360ee(e)) is amended to read as follows:

17 “(e) For grants and contracts under subsection (a)  
 18 there are authorized to be appropriated \$25,000,000 for  
 19 fiscal year 2002, and such sums as may be necessary for  
 20 each subsequent fiscal year.”.

### 21 **SEC. 202. TECHNICAL AMENDMENT.**

22 Section 527(a) of the Federal Food, Drug, and Cos-  
 23 metic Act (21 U.S.C.360ee(a)) is amended in the matter  
 24 following paragraph (2)—

25 (1) by striking “, of such certification,”; and

1           (2) by striking “, the issuance of the certifi-  
2           cation,”.

3 **SECTION 1. SHORT TITLE.**

4           *This Act may be cited as the “Rare Diseases Act of*  
5 *2001”.*

6 **SEC. 2. FINDINGS AND PURPOSES.**

7           (a) *FINDINGS.*—Congress makes the following findings:

8                   (1) *Rare diseases and disorders are those which*  
9 *affect small patient populations, typically popu-*  
10 *lations smaller than 200,000 individuals in the*  
11 *United States. Such diseases and conditions include*  
12 *Huntington’s disease, amyotrophic lateral sclerosis*  
13 *(Lou Gehrig’s disease), Tourette syndrome, Crohn’s*  
14 *disease, cystic fibrosis, cystinosis, and Duchenne mus-*  
15 *cular dystrophy.*

16                   (2) *For many years, the 25,000,000 Americans*  
17 *suffering from the over 6,000 rare diseases and dis-*  
18 *orders were denied access to effective medicines be-*  
19 *cause prescription drug manufacturers could rarely*  
20 *make a profit from marketing drugs for such small*  
21 *groups of patients. The prescription drug industry*  
22 *did not adequately fund research into such treat-*  
23 *ments. Despite the urgent health need for these medi-*  
24 *cines, they came to be known as “orphan drugs” be-*  
25 *cause no companies would commercialize them.*

1           (3) *During the 1970s, an organization called the*  
2           *National Organization for Rare Disorders (NORD)*  
3           *was founded to provide services and to lobby on behalf*  
4           *of patients with rare diseases and disorders. NORD*  
5           *was instrumental in pressing Congress for legislation*  
6           *to encourage the development of orphan drugs.*

7           (4) *The Orphan Drug Act created financial in-*  
8           *centives for the research and production of such or-*  
9           *phan drugs. New federal programs at the National*  
10          *Institutes of Health and the Food and Drug Adminis-*  
11          *tration encouraged clinical research and commercial*  
12          *product development for products that target rare dis-*  
13          *eases. An Orphan Products Board was established to*  
14          *promote the development of drugs and devices for rare*  
15          *diseases or disorders.*

16          (5) *Before 1983, some 38 orphan drugs had been*  
17          *developed. Since the enactment of the Orphan Drug*  
18          *Act, more than 220 new orphan drugs have been ap-*  
19          *proved and marketed in the United States and more*  
20          *than 800 additional drugs are in the research pipe-*  
21          *line.*

22          (6) *Despite the tremendous success of the Orphan*  
23          *Drug Act, rare diseases and disorders deserve greater*  
24          *emphasis in the national biomedical research enter-*  
25          *prise. The Office of Rare Diseases at the National In-*

1        *stitutes of Health was created in 1993, but lacks a*  
2        *statutory authorization.*

3            (7) *The National Institutes of Health has re-*  
4        *ceived a substantial increase in research funding from*  
5        *Congress for the purpose of expanding the national*  
6        *investment of the United States in behavioral and*  
7        *biomedical research.*

8            (8) *Notwithstanding such increases, funding for*  
9        *rare diseases and disorders at the National Institutes*  
10       *of Health has not increased appreciably.*

11           (9) *To redress this oversight, the Department of*  
12        *Health and Human Services has proposed the estab-*  
13        *lishment of a network of regional centers of excellence*  
14        *for research on rare diseases.*

15           (10) *The Food and Drug Administration sup-*  
16        *ports small clinical trials through Orphan Products*  
17        *Research Grants. Such grants embody successful part-*  
18        *nerships of government and industry, and have led to*  
19        *the development of at least 23 drugs and four medical*  
20        *devices for rare diseases and disorders. Yet the appro-*  
21        *priations in Fiscal Year 2001 for such grants were*  
22        *less than in Fiscal Year 1995.*

23        (b) *PURPOSES.—The purposes of this Act are to—*

1           (1) *amend the Public Health Service Act to es-*  
 2           *tablish an Office of Rare Diseases at the National In-*  
 3           *stitutes of Health; and*

4           (2) *increase the national investment in the devel-*  
 5           *opment of diagnostics and treatments for patients*  
 6           *with rare diseases and disorders.*

7       ***TITLE I—NATIONAL INSTITUTES***  
 8               ***OF HEALTH***

9       ***SEC. 101. NIH OFFICE OF RARE DISEASES.***

10       *Title IV of the Public Health Service Act (42 U.S.C.*  
 11       *281 et seq.) is amended by inserting after section 404D the*  
 12       *following:*

13                       ***“OFFICE OF RARE DISEASES***

14       ***“SEC. 404E. (a) ESTABLISHMENT.—****There is estab-*  
 15       *lished within the Office of the Director of NIH an office*  
 16       *to be known as the Office of Rare Diseases (in this section*  
 17       *referred to as the ‘Office’), which shall be headed by a Direc-*  
 18       *tor (in this section referred to as the ‘Director’), appointed*  
 19       *by the Director of NIH.*

20       ***“(b) DUTIES.—***

21               ***“(1) IN GENERAL.—****The Director of the Office*  
 22       *shall carry out the following:*

23                       ***“(A) The Director shall recommend an***  
 24               *agenda for conducting and supporting research*  
 25               *on rare diseases through the national research*  
 26               *institutes and centers. The agenda shall provide*

1       *for a broad range of research and education ac-*  
2       *tivities, including scientific workshops and*  
3       *symposia to identify research opportunities for*  
4       *rare diseases.*

5               *“(B) The Director shall, with respect to rare*  
6       *diseases, promote coordination and cooperation*  
7       *among the national research institutes and cen-*  
8       *ters and entities whose research is supported by*  
9       *such institutes.*

10              *“(C) The Director, in collaboration with the*  
11       *directors of the other relevant institutes and cen-*  
12       *ters of the National Institutes of Health, shall*  
13       *enter into cooperative agreements with and make*  
14       *grants for regional centers of excellence on rare*  
15       *diseases in accordance with section 404F.*

16              *“(D) The Director shall promote the suffi-*  
17       *cient allocation of the resources of the National*  
18       *Institutes of Health for conducting and sup-*  
19       *porting research on rare diseases.*

20              *“(E) The Director shall promote and en-*  
21       *courage the establishment of a centralized clear-*  
22       *inghouse for rare and genetic disease informa-*  
23       *tion that will provide understandable informa-*  
24       *tion about these diseases to the public, medical*  
25       *professionals, patients and families.*

1           “(F) *The Director shall biennially prepare*  
2           *a report that describes the research and edu-*  
3           *cation activities on rare diseases being conducted*  
4           *or supported through the national research insti-*  
5           *tutes and centers, and that identifies particular*  
6           *projects or types of projects that should in the fu-*  
7           *ture be conducted or supported by the national*  
8           *research institutes and centers or other entities*  
9           *in the field of research on rare diseases.*

10           “(G) *The Director shall prepare the NIH*  
11           *Director’s annual report to Congress on rare dis-*  
12           *ease research conducted by or supported through*  
13           *the national research institutes and centers.*

14           “(2) *PRINCIPAL ADVISOR REGARDING ORPHAN*  
15           *DISEASES.—With respect to rare diseases, the Director*  
16           *shall serve as the principal advisor to the Director of*  
17           *NIH and shall provide advice to other relevant agen-*  
18           *cies. The Director shall provide liaison with national*  
19           *and international patient, health and scientific orga-*  
20           *nizations concerned with rare diseases.*

21           “(c) *DEFINITION.—For purposes of this section, the*  
22           *term ‘rare disease’ means any disease or condition that af-*  
23           *fects less than 200,000 persons in the United States.*

24           “(d) *AUTHORIZATION OF APPROPRIATIONS.—For the*  
25           *purpose of carrying out this section, there are authorized*



1 to be appropriated \$4,000,000 for fiscal year 2002, and  
 2 such sums as may be necessary for each subsequent fiscal  
 3 year.”.

4 **SEC. 102. RARE DISEASE REGIONAL CENTERS OF EXCEL-**  
 5 **LENCE.**

6 *Title IV of the Public Health Service Act (42 U.S.C.*  
 7 *281 et seq.), as amended by section 101, is further amended*  
 8 *by inserting after section 404E the following:*

9 “RARE DISEASE REGIONAL CENTERS OF EXCELLENCE

10 “SEC. 404F. (a) COOPERATIVE AGREEMENTS AND  
 11 GRANTS.—

12 “(1) IN GENERAL.—*The Director of the Office of*  
 13 *Rare Diseases (in this section referred to as the ‘Di-*  
 14 *rector’)* shall, in collaboration with the directors of the  
 15 *other relevant institutes and centers of the National*  
 16 *Institutes of Health, enter into cooperative agreements*  
 17 *with and make grants to public or private nonprofit*  
 18 *entities to pay all or part of the cost of planning, es-*  
 19 *tablishing, or strengthening, and providing basic op-*  
 20 *erating support for regional centers of excellence for*  
 21 *clinical research into, training in, and demonstration*  
 22 *of diagnostic, prevention, control, and treatment*  
 23 *methods for rare diseases.*

24 “(2) POLICIES.—*A cooperative agreement or*  
 25 *grant under paragraph (1) shall be entered into in*

1       *accordance with policies established by the Director of*  
2       *NIH.*

3       “(b) *COORDINATION WITH OTHER INSTITUTES.—The*  
4       *Director shall coordinate the activities under this section*  
5       *with similar activities conducted by other national research*  
6       *institutes, centers and agencies of the National Institutes*  
7       *of Health and by the Food and Drug Administration to*  
8       *the extent that such institutes, centers and agencies have*  
9       *responsibilities that are related to rare diseases.*

10       “(c) *USES FOR FEDERAL PAYMENTS UNDER COOPER-*  
11       *ATIVE AGREEMENTS OR GRANTS.—Federal payments made*  
12       *under a cooperative agreement or grant under subsection*  
13       *(a) may be used for—*

14               “(1) *staffing, administrative, and other basic op-*  
15               *erating costs, including such patient care costs as are*  
16               *required for research;*

17               “(2) *clinical training, including training for al-*  
18               *lied health professionals, continuing education for*  
19               *health professionals and allied health professions per-*  
20               *sonnel, and information programs for the public with*  
21               *respect to rare diseases; and*

22               “(3) *clinical research and demonstration pro-*  
23               *grams.*

24       “(d) *PERIOD OF SUPPORT; ADDITIONAL PERIODS.—*  
25       *Support of a center under subsection (a) may be for a pe-*

1 *riod of not to exceed 5 years. Such period may be extended*  
 2 *by the Director for additional periods of not more than 5*  
 3 *years if the operations of such center have been reviewed*  
 4 *by an appropriate technical and scientific peer review*  
 5 *group established by the Director and if such group has rec-*  
 6 *ommended to the Director that such period should be ex-*  
 7 *tended.*

8 “(e) *AUTHORIZATION OF APPROPRIATIONS.—For the*  
 9 *purpose of carrying out this section, there are authorized*  
 10 *to be appropriated \$20,000,000 for fiscal year 2002, and*  
 11 *such sums as may be necessary for each subsequent fiscal*  
 12 *year.”.*

13 ***TITLE II—FOOD AND DRUG***  
 14 ***ADMINISTRATION***

15 ***SEC. 201. GRANTS AND CONTRACTS FOR THE DEVELOP-***  
 16 ***MENT OF ORPHAN DRUGS.***

17 *Subsection (c) of section 5 of the Orphan Drug Act*  
 18 *(21 U.S.C. 360ee(c)) is amended to read as follows:*

19 “(c) *For grants and contracts under subsection (a)*  
 20 *there are authorized to be appropriated \$25,000,000 for fis-*  
 21 *cal year 2002, and such sums as may be necessary for each*  
 22 *subsequent fiscal year.”.*

1 **SEC. 202. TECHNICAL AMENDMENT.**

2        *Section 527(a) of the Federal Food, Drug, and Cos-*  
3 *metic Act (21 U.S.C.360cc(a)) is amended in the matter*  
4 *following paragraph (2)—*

5            *(1) by striking “, of such certification,”; and*

6            *(2) by striking “, the issuance of the certifi-*  
7 *cation,”.*



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**A BILL**

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

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