

110TH CONGRESS
1ST SESSION

H. RES. 524

Expressing the sense of the House of Representatives with respect to
Diamond-Blackfan Anemia.

IN THE HOUSE OF REPRESENTATIVES

JUNE 27, 2007

Mrs. MCCARTHY of New York submitted the following resolution; which was
referred to the Committee on Energy and Commerce

RESOLUTION

Expressing the sense of the House of Representatives with
respect to Diamond-Blackfan Anemia.

Whereas Diamond-Blackfan Anemia (“DBA”) is a rare genetic bone marrow failure disorder affecting children and adults, 90 percent of whom are younger than 1 year of age when they are diagnosed, and results in severe anemia due to failure to produce red blood cells;

Whereas individuals and families suffering with rare diseases such as DBA not only face the challenges of their debilitating and life-threatening diseases, but must also confront the unfair consequences of their rare disease status, which often means limited research is being done and treatment options may not be optimal;

Whereas individuals suffering from rare diseases deserve access to improved treatment options and the potential for

a cure through the advancement of specialized research initiatives despite their rare disease status;

Whereas it is recognized that although DBA is rare, the importance of the disease lies only in part in the devastating clinical consequences it may have on those affected, as research is proving that study of complex, rare diseases such as DBA are yielding tremendous advancements in other, larger disease areas that affect millions of Americans;

Whereas the children living with DBA have an increased risk of leukemia, solid tumors, and complete bone marrow failure, and 50 percent of patients with DBA are born with birth defects including abnormalities to the face, head, upper arm and hand, genitourinary, and heart with 21 percent of affected patients having more than 1 defect;

Whereas researchers believe that as a genetic disorder of red cell production and as a cancer predisposition syndrome with a high rate of congenital anomalies, the study of DBA will yield clues to several other widespread diseases providing valuable insights into the biology of blood disorders, blood cell formation (recovery from cancer chemotherapy), the true incidence of aplastic anemia, myelodysplastic syndrome, leukemia, and the predisposition to cancer in DBA, and serve as an important model for understanding the genetics of birth defects;

Whereas treatments for DBA, including the use of steroids (such as prednisone) and blood transfusions, have potential long-term side effects, including osteoporosis, impaired growth because of the steroids, diabetes, and iron overload because of the transfusions;

Whereas the only cure for DBA is a bone marrow transplant, a procedure that carries serious risks and, since most patients lack an acceptable donor, is an option available for only about 25 percent of patients;

Whereas rare diseases, such as DBA, where there are no regional or ethnic biases and a small number of patients, making progress in treatment protocols and research difficult, benefit greatly from well-established comprehensive care centers such as the DBA Comprehensive Clinical Care Center at Schneider Children's Hospital in New Hyde Park, New York, which has become the multi-dimensional hub for the care and treatment of DBA patients across the country, as well as the home of the DBA Patient Registry which has become a valuable national resource for investigators utilizing the Center to accomplish research in a multitude of areas not specific only to DBA;

Whereas the successful establishment of the DBA Comprehensive Clinical Care Center at Schneider Children's Hospital became a model for how to diagnose, treat, and improve the lives of patients with rare diseases, while learning from the disorder to yield advancements in other areas of disease research;

Whereas the success of the initial DBA Comprehensive Clinical Care Center prompted the Centers for Disease Control and Prevention's DBA Public Health Outreach and Surveillance Program to establish 3 additional DBA Centers in Texas, California, and Massachusetts to further patient access to information, treatment, and care by DBA experts, which has resulted in a doubling of patient care visits for DBA care and surveillance since their establishment;

Whereas the DBA Public Health Outreach and Surveillance Program at the Centers for Disease Control and Prevention (CDC) has resulted in the completion of the first CDC brochure for the DBA patient population, the introduction of a DBA hotline and dedicated DBA nurse, and has resulted in a 25-percent increase of enrollment of DBA patients into the DBA Patient Registry in the first 2 years of the program;

Whereas the collaboration achieved through the federally supported DBA initiatives within the National Institutes of Health and Centers for Disease Control and Prevention and their close collaboration with the Daniella Maria Arturi Foundation and the DBA Foundation have driven the many recent successes in the DBA field and serve as a model for addressing rare disease research efforts through close public and private collaboration to achieve the highest levels of success in the areas of improved patient care and disease research;

Whereas the interagency collaboration achieved within the National Institutes of Health between the National Heart, Lung, and Blood Institute, the National Institute of Diabetes and Digestive and Kidney Diseases, the National Cancer Institute, and the Office of Rare Diseases to advance the research and understanding of DBA has resulted in significant advancements not only in the DBA scientific arena, but in understanding its many links to more prevalent disorders; and

Whereas the DBA research initiatives have already yielded tremendous success including the discovery of 2 ribosomal protein (“RP”) genes and the identification that DBA is the first human disease linked to a ribosomal protein problem which, as a fundamental unit

of cellular function, has been implicated in a wide range of human disorders including cancer, making this discovery a profound example of the additional benefits that may result from the study of DBA: Now, therefore, be it

1 *Resolved*, That—

2 (1) the House of Representatives—

3 (A) recognizes that the identification of Di-
4 amond-Blackfan Anemia (in this resolution re-
5 ferred to as “DBA”) as the first human dis-
6 order with a ribosomal deficiency is a primary
7 example of the importance of the Federal Gov-
8 ernment’s continued support of DBA research,
9 which may advance the understanding of the
10 basic mechanisms that affect red cell produc-
11 tion, identify connections of ribosomal function
12 and cell cycle production, identify implications
13 of cancer predisposition, and serve as an impor-
14 tant model for understanding human develop-
15 ment and the molecular basis for certain birth
16 defects;

17 (B) recognizes that Federal support of
18 comprehensive centers for rare disease patients
19 enhances the ability for experienced doctors to
20 provide the most complete care for each patient,
21 leading to an increase in correct and early diag-

1 nosis and the most appropriate treatment for
2 each patient;

3 (C) commends Schneider Children’s Hos-
4 pital for providing the first DBA Comprehen-
5 sive Clinical Care Center for patients across the
6 country, for developing the DBA Patient Reg-
7 istry which has proven a robust surveillance tool
8 to understand the epidemiology, biology, and
9 treatment of DBA, and for proving a valuable
10 resource for investigators at a national level,
11 working to understand DBA’s link to more
12 prevalent disorders facing Americans; and

13 (D) commends the Daniella Maria Arturi
14 Foundation and the Diamond-Blackfan Anemia
15 Foundation for their efforts to facilitate the
16 successful collaboration among the National In-
17 stitutes of Health and the Centers of Disease
18 Control and Prevention to achieve a successful
19 multidisciplinary approach between clinical and
20 scientific DBA efforts with the goal of short-
21 ening the life cycle of success realized between
22 the laboratory and applied patient care; and

23 (2) it is the sense of the House of Representa-
24 tives that the Federal Government has a responsi-
25 bility to—

1 (A) encourage further efforts to clarify the
2 natural history of DBA to—

3 (i) advance hematopoietic research in
4 the areas of bone marrow failure disorders;

5 (ii) develop a well-characterized data-
6 base of patients linked to a cell and DNA
7 repository to facilitate gene discovery;

8 (iii) understand the cellular and mo-
9 lecular biology of DBA;

10 (iv) understand the links to cancer
11 and birth defects; and

12 (v) provide models for preclinical gene
13 therapy trials;

14 (B) continue efforts to raise awareness and
15 ease access to information about DBA among
16 patient groups and the medical community to
17 improve accuracy of diagnosis and identification
18 of appropriate treatment options available;

19 (C) encourage research efforts that will ad-
20 vance the treatment options available to pa-
21 tients with DBA and seek a cure;

22 (D) encourage the National Institutes of
23 Health to develop a cross-institutional research
24 initiative to study ribosomal protein deficiencies
25 in rare inherited disease, including DBA,

1 among the relevant institute stakeholders inter-
2 ested in ribosome synthesis including—

3 (i) the National Heart, Lung, and
4 Blood Institute;

5 (ii) the National Institute of Diabetes
6 and Digestive and Kidney Diseases; and

7 (iii) the National Cancer Institute;
8 and

9 (E) encourage the continued Federal sup-
10 port of the DBA Comprehensive Clinical Care
11 Centers to further provide a definitive charac-
12 terization of the patients with DBA, which will
13 expand research and clinical care in order to
14 help manage this rare illness, while also ena-
15 bling hematologists, cancer researchers, geneti-
16 cists, basic scientists, and others to continue to
17 utilize the Center to enhance the study of this
18 disease to better understand its links to many
19 other problems facing Americans relating to
20 blood cell formation, cancer predisposition,
21 birth defects, and more.

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