

TESTIMONY

OF

RICHARD BARR, MD

MEMBER, BOARD OF TRUSTEES

IMMUNE DEFICIENCY FOUNDATION

BEFORE THE

HOUSE APPROPRIATIONS SUBCOMMITTEE ON LABOR, HEALTH
AND HUMAN SERVICES, EDUCATION AND RELATED AGENCIES

MARCH 13, 2001 - 10AM

Chairman Regula and members of the subcommittee, thank you for the opportunity to testify today on Primary Immune Deficiency and the need for continued research and education on these diseases. .

Primary immune deficiency diseases are inherited disorders in which parts of the body's immune system are missing or do not function properly. The World Health Organization has identified more than 70 different primary immunodeficiency diseases. These disorders are very underdiagnosed, but we believe that more than 45,000 Americans, of all races, ages, and gender. Fortunately, most primary immune deficient patients, once diagnosed, are able to maintain their health through infusions of a pooled plasma derivative known as intravenous immunoglobulin (IGIV) every three to four weeks for the rest of their life. However, if primary immunodeficiency diseases are not properly diagnosed and treated, they can lead to serious illness and early death.

The Immune Deficiency Foundation (IDF) is the national non-profit, charitable organization dedicated to improving the health of primary immune deficient patients through research and education. Headquartered in Towson, Maryland, IDF was founded in 1980 by a group of parents of primary immune deficient children who wanted to focus attention on the needs of primary immune deficient patients, physicians, and researchers.

IDF provides a wide variety of patient and family services, medical research and education, and advocacy for issues related to these diseases. Specifically, the Foundation acts as an information clearinghouse for newly diagnosed patients and provides these individuals with an opportunity to interact with other primary immune deficient patients and families. Oftentimes, the most reassuring call a parent of a newly-diagnosed child will make is not to a doctor or hospital, but to one of our local patient representatives with his or her own children playing loudly in the backyard. This opportunity to speak directly and frankly to another parent in a similar situation often is the first chance to seek support and results in a level of comfort that with proper treatment their child can grow up with a near-normal life.

The foundation is also active in medical research to try to better define and diagnose these diseases. While we search for these longer-term answers, we are also looking to improve current treatment options for patients as well as improve understanding of these diseases

within the medical community. Because primary immune deficiency is a rare disease, we focus on providing educational opportunities such as visiting professorships and grand rounds for physicians and medical students who might otherwise not be exposed to this knowledge. The foundation is also working through an NIH grant that I will discuss in more detail later to help better identify the range and occurrence of these diseases.

Finally, IDF has a very active public policy program that focus on blood safety issues, patient reimbursement for treatment, and advancing scientific knowledge regarding primary immune deficiency diseases.

Mr. Chairman, I am here today to speak as a patient, a physician, and the newly elected Chairman of the IDF Board of Trustees. My case is representative of a typical immune deficient patient. I was diagnosed with Common Variable Immunodeficiency 10 years ago, following years of repeated infections, which were unresponsive to antibiotics, and undiagnosed by numerous physicians who happened to be colleagues of mine. This led to numerous unsuccessful surgeries resulting in permanent lung and sinus damage. Prior to my diagnosis, a day was considered successful if I had enough energy to get out of bed. Following appropriate diagnosis and treatment with IGIV, I was able to return to my medical practice and developed a new lease on life.

In my testimony today, I would like to highlight three areas of importance to the IDF and the primary immune deficiency community we represent:

- 1) National Primary Immune Deficiency Surveillance Program.
- 2) Primary Immune Deficiency Research at the National Institutes of Health
- 3) Primary Immune Deficiency Registries at the National Institute of Allergy and Infectious Diseases

1) NATIONAL PRIMARY IMMUNE DEFICIENCY SURVEILLANCE PROGRAM

Mr. Chairman, because primary immune deficient patients are the only patient population that require life-long infusions of IGIV to maintain their health, the Immune Deficiency Foundation has been working to establish a national surveillance study of this group to evaluate the short and long term effects of IGIV use. The establishment of this surveillance initiative is vitally important because although primary immune patients have been treated with IGIV for over 20 years, a prospective study on adverse events associated with its use has not been performed.

IDF's surveillance program would provide valuable epidemiological data on the potential risks of IGIV therapy, and conditions which might predispose patients to adverse events. In addition, this initiative would benefit other IGIV users by serving as an early warning system should study participants be exposed to new and emerging pathogens

IDF's proposed surveillance study would focus on the following:

- * Identifying and characterizing adverse events
- * Determining their prevalence and incidence
- * Determining whether there are specific risk factors for adverse events such as:
 - a. Certain primary immunodeficient diseases (e.g., Common Variable Immunodeficiency vs. X-Linked Agammaglobulinemia)
 - b. Pre-existing medical conditions (e.g., renal and/or cardiac disease)
 - c. More common with some preparations than with others (e.g., different brands and different formulations)

Mr. Chairman, IDF has been working with the plasma fractionation industry, the Food and Drug Administration, and the Centers for Disease Control and Prevention to establish this new surveillance program, and we are grateful for the subcommittee's support of this partnership last year as we developed the details of this project. Now that the program is nearing the stages of requiring full funding, we ask that you continue to support this important public health initiative by encouraging CDC to continue working with us again in FY02 and more importantly, we ask that you encourage the National Institutes of Health to support this effort as well.

2) PRIMARY IMMUNE DEFICIENCY RESEARCH AT THE NATIONAL INSTITUTES OF HEALTH

Mr. Chairman, I would like to take this opportunity to thank the subcommittee for its longstanding support of biomedical research at the National Institutes of Health. IDF remains committed to the goal of the doubling the NIH budget by FY03. Specifically, IDF encourages the subcommittee to continue its support of primary immune deficiency research at the National Institute of Allergy and Infectious Diseases (NIAID), the National Institute of Child Health and Human Development (NICHD), and the National Cancer Institute (NCI).

In recent years, NIAID sponsored research has shed new light on the genetics of primary immunodeficiencies. NIAID investigators are using this information to develop new gene-based therapies for many primary immune disorders. This cutting-edge research has given patients hope that improved therapies, and eventually a cure, for these diseases may be on the horizon. Primary immune deficiency research also benefits people suffering from other disorders, such as autoimmune diseases and cancer, due to its acute focus on the functions of the immune system.

Recognizing the promise that biomedical research holds for improving the quality of life for primary immune deficient patients, IDF joins with the Ad Hoc Group for Medical Research in recommending a 16.5% increase for NIAID, NICHD and NCI in FY02.

3) PRIMARY IMMUNE DEFICIENCY CLINICAL REGISTRIES PROGRAM

Mr. Chairman, since 1997, IDF has contracted with NIAID to construct and maintain registries of 8 primary immunodeficiency diseases including, Chronic Granulomatous Disease, Common Variable Immunodeficiency, DiGeorge Anomaly, Hyper IgM Syndrome, Leukocyte Adhesion Defect, Severe Combined Immunodeficiency, Wiskott-Aldrich Syndrome, and X-Linked Agammaglobulinemia.

The goal of these registries is to assemble a comprehensive clinical picture of each disorder, including estimates of disease prevalence, clinical course, and complications.

This data is an invaluable resource for physicians conducting basic research on these disorders. For example, information from one registry (chronic granulomatous) is being used by four institutions to examine six different questions relating to various aspects of the disease.

Further expansion of these registries is essential if we are to increase our understanding of additional primary immune deficiency disorders. IDF appreciates the subcommittee's longstanding support of the NIAID/IDF clinical registries partnership and encourages you to continue to support these important programs in FY02.

Mr. Chairman, thank you once again for the opportunity to present the views of the Immune Deficiency Foundation. We look forward to working with you and your staff. I would be happy to answer any questions that you or your colleagues may have.