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The impact of physician education and public awareness on early diagnosis of primary immunodeficiencies

Robert A. Good Immunology Symposium

Vicki Modell

Thank you Hans for that kind and gracious introduction. We are deeply honored to have been asked by Bibi Day to speak at the opening session of the first Robert A. Good Immunology Symposium. It is with deep humility that we come before such an illustrious faculty and assembled delegates. There are no words to describe how moved we are that this session is dedicated to our son, Jeffrey. What a great honor.

We first met Dr. Good in 1975 when our son Jeffrey was 5 years old. He had already been diagnosed with Hypogammaglobulinenemia.

He was treated superbly by his doctors, many of whom are here with us today. We asked Jeffrey's doctors to reach out to the greatest experts, anywhere in the world, on this disease; especially experts on the specific B cell complexities that affected our son.

In our quest for answers, we eventually reached Dr. Joshua Lederberg who, at the time, was President of Rockefeller University. To our surprise, he told us that the greatest expert, anywhere in the world on Primary Immunodeficiencies, was not half way around the globe but, in fact, was only two city blocks from our home. Of course he was talking about Dr. Robert A. Good, President of the Sloan Kettering Cancer Institute.

Dr. Good took our call, met with Jeffrey, and monitored Jeffrey's progress over many years. Doctor and patient got along famously. Both of them were constantly filled with energy and enthusiasm. Dr. Good gave Fred and I the confidence to share a normal life with our son. Jeffrey did well in school, played sports, traveled, enjoyed summer camp, and even had a girlfriend. But repeated episodes of high fever, multiple hospitalizations, and eventually serious bouts of pneumonia were too much for Jeffrey to overcome and he lost his courageous battle at the age of 15.

Throughout those years, we never met another family and Jeffrey never met another child facing this condition. We were sure that he was the only one. A few months after we lost Jeffrey, with the urging of family and friends to do something to remember his life, we

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established the Jeffrey Modell Foundation. Almost immediately, we began to hear from families across the nation and even from other parts of the world that were going through some of the same things we had experienced. It was at a time where there was very little research; virtually no funding and few treatments were available. There were no cures and little hope.

We set out on an uncertain course, a passionate journey committed to changing that landscape by building greater awareness, finding more meaningful treatments and ultimately, by finding cures for PI. We became close friends with Bob and Bibi, often sharing wonderful, very stimulating evenings in New York. They both helped us to eventually structure the JMF mission to include:

- Basic and clinical research
- Physician education
- Patient support
- Public awareness and
- Government advocacy

We learned, soon enough, that the kind of immunodeficiency that took Jeffrey's life needed to be addressed at the highest levels. We reached out to government, visited every Senate Office, called on most of the Congress, NIH Directors, the CDC, and other healthcare agencies, to generate attention about these diseases.

Together with Dr. Anthony Fauci, at NIAID, we created a working partnership and developed program announcements and RFPs that were subjected to the rigorous NIH peer review processes. We helped to fund an unique, for its time, multi-million dollar research program, which ran for 5 years. Since that time, we have established 25 Jeffrey Modell Diagnostic and Research Centers in the United States, Canada, Europe, Asia and the Middle East. There is also a JM network of more than 118 Referral Centers worldwide.

Soon, we will begin construction of the Jeffrey Modell Immunology Center at Harvard Medical School, the only one of its kind in the world today. The Center will be home to the Robert A. Good Library which will help 'bring to life' Bob Good's tremendous achievements and accomplishments.

Our Foundation has partnered with the CDC to create a successful and well received Public Service Advertising Campaign, broadly reaching out to the public and important, underserved, minorities. We have organized more than 30 continuing medical education symposia, conferences and lectures in the United States and Europe reaching out to primary care physicians and sub-specialists to help ensure the earliest possible precise diagnoses of PI. Since 1994, we have organized the biannual WHO/IUIS Expert meeting on Primary Immunodeficiencies to be held this June 6–10th in Jackson Hole, Wyoming.

The JMF, in consultation with our Board of Medical Advisors, creates and distributes educational materials including the 10 Warning Signs and 4 Stage Physician Algorithm which has been translated into 20 languages and distributed in 25 countries. We developed a colorful and educational poster about the immune system and how it works. JMF publications reach primary care physicians, sub-specialists, nurses, daycare centers and others in the health care industry. The Foundation has organized more than 30 KIDS Days in communities across the United States and in China. These fun-filled events bring families and professionals alike together to share their experiences, ask questions and get answers and to meet new friends.

As a result of these programs, our Centers report, during a 12-month period; a 98% increase in diagnosed patients, a 77% increase in patients receiving appropriate treatment, a 79% increase in patient referrals and an impressive 492% increase in the number of diagnostic test performed. Equally impressive is the fact that during that same period of time the JMF website INFO4PI received six million hits and the Hotline received 10,000 calls [1].

The campaign is working. All of these accomplishments are gratifying, but our journey is not finished. We are very fulfilled by what we have been able to achieve to date, but more importantly, we are filled with optimism and renewed enthusiasm for the future.

Appendix

Jeffrey Modell jm Foundation

PI Survey Reports:

Measuring the Impact of a Physician Education & Public Awareness Campaign

Comparing Undiagnosed (Dx)/Untreated (Rx)Patients with Diagnosed (Dx) /Treated (Rx) Patients

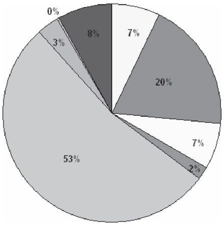
Reported by Physician/Experts from 118 Jeffrey Modell Diagnostic/Referral Centers Worldwide

Fred Modell, MA and Vicki Model
www.INFO4PI.org

June 2007

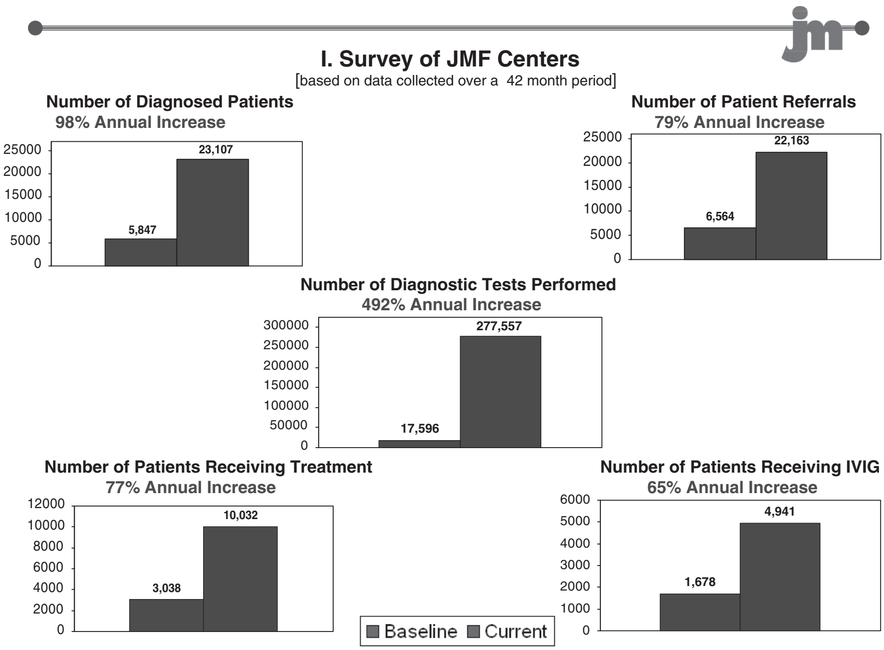
Major Immunodeficiency Groups:

□	**Predominantly Antibody Deficiencies**	**(n= 10078)**	**53%**
■	**Cellular Immunodeficiencies**	**(n= 3713)**	**20%**
□	**Combined Immunodeficiencies**	**(n= 1364)**	**7%**
□	**Defects of Phagocytic Function**	**(n= 1254)**	**7%**
▨	**Disease of Immune Dysregulation**	**(n= 606)**	**3%**
▨	**Complement Deficiencies**	**(n= 356)**	**2%**
▨	**Defects in Innate Immunity**	**(n= 57)**	**0%**
■	**Other Immunodeficiencies**	**(n= 1545)**	**8%**



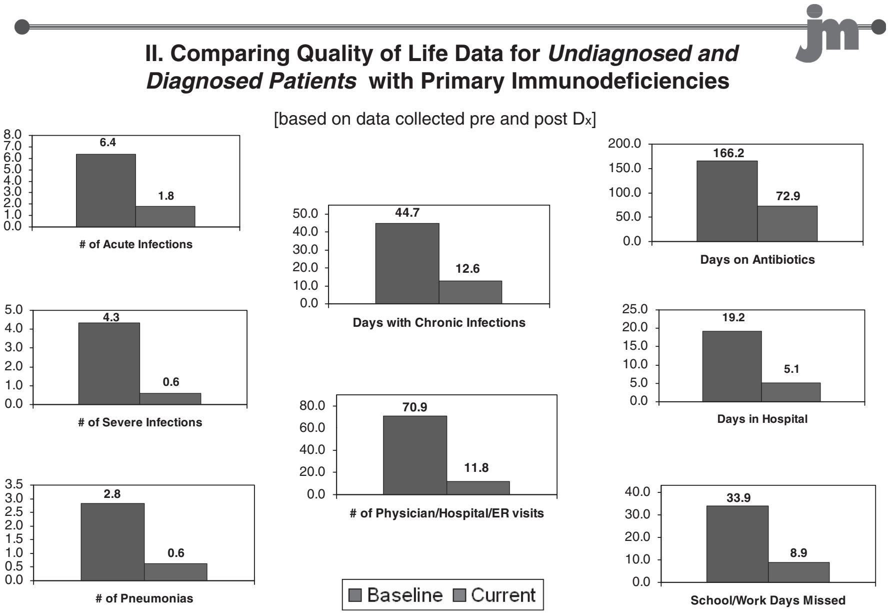
Classification of Immunodeficiency Diseases:

□	**Agammaglobulinemia, AR**	**176**
□	**Agammaglobulinemia, X-linked**	**915**
▨	**ALPS**	**322**
□	**Antibody Deficiency, Selective**	**814**
▨	**APECED**	**30**
■	**Ataxia Telangiectasia**	**906**
■	**Cartilage-hair Hypoplasia**	**38**
▨	**Chediak-Higashi Syndrome**	**61**
□	**Chronic Granulomatous Disease**	**723**
■	**Chronic Mucocutaneous Candidiasis**	**94**
□	**Common Variable**	**2,806**
▨	**Complement Deficiencies**	**356**
■	**DiGeorge Syndrome**	**2,155**
▨	**Griscelli Syndrome**	**19**
□	**Hyper IgE Syndrome**	**156**
□	**Hyper IgM, AR**	**194**
□	**Hyper IgM, X-linked (CD 40L)**	**261**
□	**Hypogammaglobulinemia of Infancy, Transient**	**471**
□	**IgA Deficiency, Selective**	**2,334**
□	**IgG Subclass Deficiency**	**2,368**
▨	**IPEX**	**78**
□	**MHC Class I Deficiency**	**0**
□	**MHC Class II Deficiency**	**67**
▨	**NEMO**	**40**
■	**Nijmegan Breakage Syndrome**	**130**
□	**Neturopenia**	**303**
□	**Leukocyte Adhesion Deficiency**	**72**
□	**Severe Combined:**	
	ADA Deficiency	**93**
	Artemis Defect	**43**
	CD3 Delta Deficiency	**6**
	CD3 Epsilon Deficiency	**3**
	CD45 Deficiency	**0**
	IL-7R Alpha Deficiency	**27**
	JAK3 Deficiency	**25**
	Omenn Syndrome	**67**
	PNP Deficiency	**2**
	RAG-1/2 Deficiency	**65**
	X-Linked Severe Combined	**239**
	ZAP-70 Deficiency	**5**
	Other SCID	**461**
▨	**WHIM Syndrome**	**17**
■	**Wiskott Aldrich**	**390**
▨	**X-Linked Lymphoproliferative**	**96**
■	**Other**	**1,545**



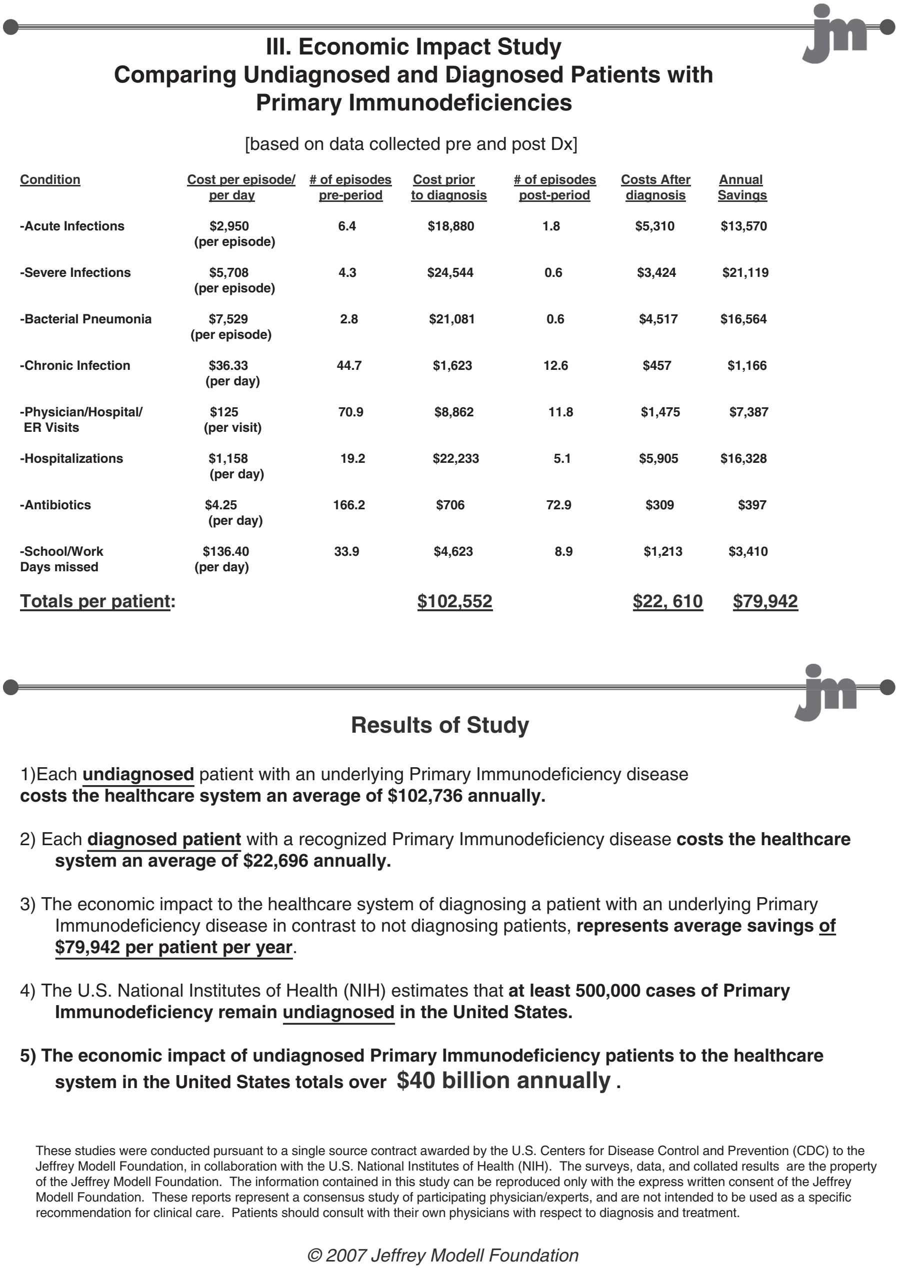
I. Survey of JMF Centers

[based on data collected over a 42 month period]



II. Comparing Quality of Life Data for *Undiagnosed and Diagnosed Patients* with Primary Immunodeficiencies

[based on data collected pre and post Dx]



III. Economic Impact Study Comparing Undiagnosed and Diagnosed Patients with Primary Immunodeficiencies

[based on data collected pre and post Dx]

Condition	Cost per episode/ per day	# of episodes pre-period	Cost prior to diagnosis	# of episodes post-period	Costs After diagnosis	Annual Savings
-Acute Infections	$2,950 (per episode)	6.4	$18,880	1.8	$5,310	$13,570
-Severe Infections	$5,708 (per episode)	4.3	$24,544	0.6	$3,424	$21,119
-Bacterial Pneumonia	$7,529 (per episode)	2.8	$21,081	0.6	$4,517	$16,564
-Chronic Infection	$36.33 (per day)	44.7	$1,623	12.6	$457	$1,166
-Physician/Hospital/ ER Visits	$125 (per visit)	70.9	$8,862	11.8	$1,475	$7,387
-Hospitalizations	$1,158 (per day)	19.2	$22,233	5.1	$5,905	$16,328
-Antibiotics	$4.25 (per day)	166.2	$706	72.9	$309	$397
-School/Work Days missed	$136.40 (per day)	33.9	$4,623	8.9	$1,213	$3,410
Totals per patient:			**$102,552**		**$22, 610**	**$79,942**

Results of Study

1)Each **undiagnosed** patient with an underlying Primary Immunodeficiency disease **costs the healthcare system an average of $102,736 annually.**

2) Each **diagnosed patient** with a recognized Primary Immunodeficiency disease **costs the healthcare system an average of $22,696 annually.**

3) The economic impact to the healthcare system of diagnosing a patient with an underlying Primary Immunodeficiency disease in contrast to not diagnosing patients, **represents average savings of $79,942 per patient per year**.

4) The U.S. National Institutes of Health (NIH) estimates that **at least 500,000 cases of Primary Immunodeficiency remain undiagnosed in the United States.**

5) The economic impact of undiagnosed Primary Immunodeficiency patients to the healthcare system in the United States totals over $40 billion annually .

These studies were conducted pursuant to a single source contract awarded by the U.S. Centers for Disease Control and Prevention (CDC) to the Jeffrey Modell Foundation, in collaboration with the U.S. National Institutes of Health (NIH). The surveys, data, and collated results are the property of the Jeffrey Modell Foundation. The information contained in this study can be reproduced only with the express written consent of the Jeffrey Modell Foundation. These reports represent a consensus study of participating physician/experts, and are not intended to be used as a specific recommendation for clinical care. Patients should consult with their own physicians with respect to diagnosis and treatment.

Reference

Modell F, Modell V (2007) Primary immunodeficiency reports: Measuring the impact of a physician education & public awareness campaign (PEPAC); comparing undiagnosed/untreated patients with diagnosed/treated patients. http://www.info4pi.org