

Patient & Family Handbook

For Primary
Immunodeficiency
Diseases

 IMMUNE
DEFICIENCY
FOUNDATION



This book contains general medical information which cannot be applied safely to any individual case. Medical knowledge and practice can change rapidly. Therefore, this book should not be used as a substitute for professional medical advice.

FOURTH EDITION
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For Primary Immunodeficiency Diseases

4th Edition

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Preface

The first edition of the *Patient & Family Handbook* was written nearly two decades ago in response to requests from patients with primary immunodeficiency diseases, their families and their physicians. We hoped that it would help patients and their families to learn more about the immune system, the primary immunodeficiency diseases, currently available therapies and possible future treatments. Since then, tens of thousands of copies have been distributed to patients and their families!

This fourth edition, like those editions that have come before, has been inspired by the many new and exciting advances in the diagnosis and therapy of the primary immunodeficiencies. Many chapters in this edition are new and all of the existing chapters have been revised to include important new information. We hope that the first chapter will be useful to all who read this book. It explains how the immune system works and how the failure of the immune system leads to primary immunodeficiency diseases. Individual chapters on most of the specific primary immunodeficiencies follow. We have included three new chapters on disorders that were not covered in previous editions and updated the existing chapters to include information on new diagnostic tools, more precise clinical information, and new therapies. The *General Care* chapter has been updated to reflect new nutritional guidance and conveys commonsense guidelines that the patient and family may find useful. The chapter on *Specific Medical Therapy* has been revised to reflect recent advances in treatments available to the primary immunodeficient individual, including newer methods of treatment with immunoglobulin. A new chapter dedicated to the adolescent with a primary immunodeficiency has been added, making three chapters that deal with issues relating to patients from infancy through adult life. The *Health Insurance* chapter has been expanded and updated to reflect changes in reimbursement and insurance; it should be a good place to start when trying to understand this complex and important area. *Resources* includes additional information available both in printed form and on the Internet. Finally, the *Glossary* offers definitions of the more common, and possibly confusing, medical terms.

A few words on how to use this book: this book is not a substitute for a dialogue between the patient, his/her family, their physician, and other members of the healthcare team. Rather, it is intended to provide the patient and family with tools to enhance the communication process and to understand the information they receive from the healthcare team. Most importantly, this book is not intended to suggest diagnostic approaches or to recommend specific therapy for any patient. Each patient's condition and treatment is unique and the management of their illness should be customized to their individual medical needs.

We thank all those individuals who contributed to this book: those who wrote specific chapters, the members of the Medical Advisory Committee who reviewed the chapters, the Board of Trustees and the staff of the Immune Deficiency Foundation who made this book a high priority, and finally, the patients and their families whose suggestions will make this edition even better than the last three!

The Editors
Baltimore 2007

The Immune Deficiency Foundation

The Immune Deficiency Foundation (IDF) was founded in 1980 by parents of children with primary immunodeficiency diseases and their physicians. At that time, there were no educational materials or programs for patients and no public advocacy initiatives. One of the greatest challenges faced by people who find themselves or their children diagnosed with a primary immunodeficiency is getting the right information at the right time. To fill this void, one of the most important publications developed by IDF is the *Patient & Family Handbook* and we are proud to offer this fourth edition. We know that this book has served as the basis of understanding primary immunodeficiency diseases for over two decades and we are pleased to present this updated version.

A few years ago, Kinsey Moore, then an eighth grader, had the assignment of writing an essay on what book she would choose to be. Kinsey wrote:

“The book I would want to be turned into is the Immune Deficiency Foundation (IDF) Family Handbook. When I was born, I was very ill and almost died several times. In all of my baby pictures, I have cords and wires connected to me. One time when I was in the hospital and I had a life-threatening infection for the third time that month, my mom walked into the library and saw a little blue and white book poking off the shelf. It was the IDF’s family handbook. Everything in that book applied to me. When my mom told the doctors, they did not believe her. After two years of going to different doctors, I was finally diagnosed when I was four. This book saved my life. I would want to become this book so I could save more people in my situation. My family knows what it is like to feel lost and not know whether I would live till tomorrow, but this book gave us hope.”

We hope this handbook gives you hope, knowledge and empowerment to help cope with the challenges of living with a primary immunodeficiency disease. As a patient-focused organization dedicated to our community, we encourage you to contact IDF to help meet your needs. Whether you want to talk to a peer support volunteer, need assistance from a patient advocate or want to attend an educational meeting, know that IDF is the place to turn for help and information.

Marcia Boyle
President & Founder
Immune Deficiency Foundation

About the Immune Deficiency Foundation

The Immune Deficiency Foundation, founded in 1980, is the national non-profit patient organization dedicated to improving the diagnosis and treatment of patients with primary immunodeficiency diseases through research, education and advocacy.

Educational Publications

- Patient & Family Handbook for Primary Immunodeficiency Diseases
- Our Immune System
- A Guide for School Personnel on Primary Immune Deficiency Diseases
- Diagnostic and Clinical Care Guidelines for Primary Immunodeficiency Diseases
- IDF Guide for Nurses on Immunoglobulin Therapy for Primary Immunodeficiency Diseases
- IDF Advocate—newsletter
- Primary Immune Tribune—e-newsletter

Services for Patients and Families

- Patient Advocacy—inquiries related to diagnosis, treatment, health insurance, peer support and literature requests
- IDF Educational Meetings—local and regional patient meetings, national conference
- IDF Volunteer Network—Peer Support, Grassroots Advocacy and Fundraising
- Student Scholarships—post-secondary education

Services for Medical Professionals

- Consulting Immunologist Program (877-666-0866) provides physicians with a free consult or second opinion on patients with primary immunodeficiency diseases
- LeBien Visiting Professor Program offers Grand Rounds and clinical presentations at medical institutions throughout North America
- United States Immunodeficiency Network (USIDNET). IDF administers this National Institute of Health contract for research and mentoring for primary immunodeficiency diseases
- National Registries of Primary Immunodeficiency Diseases

Public Policy Initiatives

- Advocacy efforts on public policy issues at national and state levels by monitoring issues that are critical to patients
- IDF Grassroots Advocacy Program mobilizes the primary immunodeficiency community to contact their government representatives to promote healthcare legislation that will positively affect the community.
- Advocacy for increased funding for research on primary immunodeficiency diseases
- Work with other organizations on quality of care initiatives for users of plasma products

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