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Preface

The Mission Statement of the Immune Deficiency Foundation (IDF) pledges that, as the national patient organization for primary immunodeficiency, it is dedicated to improving the diagnosis, treatment and quality of life of persons with primary immunodeficiency diseases through advocacy, education and research. Since its founding in 1980, one of the ways IDF has tried to achieve this goal is by the publication of pamphlets, booklets and more extensive publications such as this IDF Patient and Family Handbook to provide a reliable source of information about these often unfamiliar diseases to patients, their families and their healthcare providers. The first edition of the Handbook was published in 1987, and since then tens of thousands of copies have been distributed and it has been translated in part or in whole into at least seven different languages.

The first edition was composed of nine chapters covering five primary immunodeficiency diseases. By the time the 4th edition was published in 2007, it contained 22 chapters and covered about 60 disorders. This new 5th edition expands the content of the Handbook by another 50% to 33 chapters with descriptions of nearly 100 different primary immunodeficiencies. The Handbook includes an overview of The Immune System and Primary Immunodeficiency Diseases to provide a basic description of the components of the immune system and how its defects lead to disease. There are 18 chapters covering the specific details of many of the individual primary immunodeficiency diseases themselves. There are additional chapters with general information relevant to the inheritance, laboratory diagnosis, general care and specific medical treatments of primary immunodeficiencies as well as chapters on life management issues for patients of different ages.

Important new chapters have been added on subjects such as autoimmunity, allergies and infections-topics of critical interest to many in our community. There are also new chapters on Stem Cell and Gene Therapy, Innate Immune Defects and an enlarged section on phagocytic cell disorders within the Chronic Granulomatous Disease chapter. In addition to the new chapters, all of the existing chapters have been revised and updated with new information and many have been completely rewritten.

The authors and editors have tried to condense the often highly technical information available into a form that is informative yet still understandable to a reader not trained in medicine or immunology. We hope that you find the Handbook to be a useful source of information about primary immunodeficiency diseases and would appreciate your feedback so that we can continue to make improvements in future editions. In addition, we plan to develop an online supplement that will provide more detailed reports about some of these disorders as well as breaking research news of new insights or treatment successes. This will keep our community informed of advances between publication of new print editions of the Handbook.

It is important to recognize that a regular dialogue between the patient, the family and the healthcare provider team is essential to facilitate the highest quality care. This Handbook is not intended to be a substitute for those critical interactions, but it should be used as a tool for patients and their families. Our goal is to help them understand the information that they receive from their providers and arm them with background information so that they can better communicate with their healthcare team. Each patient’s situation is unique and the management of illness and its treatment must be customized to meet their individual needs. The development of a partnership between the patient and family and healthcare providers is critically important for success in the management of life-long challenges like those presented by primary immunodeficiency diseases.

The Editors
Baltimore 2013
Letter from the President & Founder

Years ago, my son’s immunologist told me about a saying that many doctors are taught in medical school, “when you hear hoof beats, think horses, not zebras.” However, immunologists are taught to look for zebras, not horses. Rather than focus on the likeliest possibilities when making a diagnosis, they look for the unusual ones.

At IDF, we believe that patients with primary immunodeficiency diseases are the zebras of the medical world, and that more physicians need to think about the unique diagnosis, not the horse, but a zebra. Zebras in a herd might all look alike, but their stripe patterns are as distinctive as fingerprints—no two are the same. And no two members of our patient community are the same either!

Since IDF was founded in 1980, we have strived to develop and provide innovative resources to meet the individual needs of patients and families living with primary immunodeficiency diseases. Our first edition of the *IDF Patient & Family Handbook* represented the first information in the world on primary immunodeficiency developed specifically for patients. We continue that effort as we introduce this fifth edition of the *IDF Patient & Family Handbook for Primary Immunodeficiency Diseases*. We are incredibly proud of this publication! Written and edited by leading immunologists, nurses and life management specialists, it focuses on what is important to our community—sharing information to advance diagnosis and treatment, as well as quality healthcare and life management skills that can make a positive difference in daily life.

We hope that you will benefit from this expanded and updated version of the Handbook and that you will continue to connect with IDF’s vital network of resources. Our programs offer a wealth of valuable education, information and support when you need it. We welcome you to stay in touch, online or by phone. Rest assured you can rely on IDF!

As the national patient organization dedicated to persons living with primary immunodeficiency diseases, IDF encourages you to THINK ZEBRA! and live a fulfilling life with your primary immunodeficiency!

Marcia Boyle  
*President & Founder*  
*Immune Deficiency Foundation*

Efforts have been made to ensure that the material presented in this Handbook is accurate, reliable and in accordance with current standards at the time of publication. However, as research and experience expands, recommendations for care, treatment and management may change. Please visit [www.primaryimmune.org](http://www.primaryimmune.org) for updated information.
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Please use this search list to locate the chapter that contains information on the particular disease in which you are interested. With more than 185 primary immunodeficiency diseases recognized by the World Health Organization, it was not possible to include them all in this Handbook. However, there is information about many of the disorders, including those that are less common.

Over the years, the names of some disorders have changed. Those alternative names are listed. Sometimes a chapter discusses several different primary immunodeficiency diseases. In that case this list directs you to the chapter subsection where you can find information about that disease.

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