
Preface

Primary immunodeficiency diseases (PIDs) are a heterogeneous group of inherited disorders, characterized by different defects in the development and function of the immune system. These defects render a patient susceptible to a variety of infectious diseases. The infections in PIDs can occur repeatedly, severely, and atypically damaging the organs and reducing quality of life. PIDs also predispose affected individuals to immune dysregulation with autoimmune disease and malignancy.

About 180 PIDs have been phenotypically described so far; among them, there are over 100 primarily single-gene defects. There is no exact incidence of PIDs; however, it seems that their measured frequency may be underestimated. Early diagnosis and adequate therapy are the keys for better prognosis and quality of life of affected patients, while diagnosis delay and/or inadequate management may lead to permanent organ damage and shorten lifespan. Unfortunately, failure to recognize these conditions is still a major problem for clinicians around the world, and diagnosis of patients with PIDs is associated with a considerable delay in children and adults. Indeed, some PIDs could be undiagnosed for some years, if providers continue to treat only the individuals' complications. One major problem is that general practitioners, physicians, and pediatricians lack familiarity with PIDs and guidance regarding the appropriate treatment. This is particularly applicable in developing countries. Lack of awareness among medical communities as well as underdeveloped infrastructural diagnostic and therapeutic facilities are the main problems encountered in the management of PIDs.

This book is planned to increase the clinical awareness and knowledge of practicing clinicians about diagnosis and management of PIDs. Knowledge of normal immunity and specific warning signs and symptoms can help physicians to distinguish those children with underlying PIDs. Therefore, the ultimate orientation of the book is toward practical diagnosis and management of real cases. For this goal, this book consists of nine chapters (about 90 cases and 300 questions), resulting from valuable cooperation of more than 40 senior and junior scientists in the field of immunodeficiency. The selected case reports focus on the presenting features of patients with PIDs and further investigation and management which are needed. Although the case scenarios are tried to be according to real history, some modifications are made in some cases for educational purposes. Each of the numbered cases in the chapter is followed by questions, their answers, and completion of the statement and discussion. It should be mentioned that in some questions,

more than one lettered answers are true. Important messages in outline format are set in practical points box at the end of each case. Lastly, we would like to express our appreciation to all physicians and patients who developed the concept for this book. We hope that the book will be welcomed by clinicians, who wish to learn more about PIDs.

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A Problem-Solving Approach

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