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# Nutrition Management of Patients with Inherited Metabolic Disorders

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*Edited by*

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Without the many people who shared their knowledge with me over the last 50-plus years, preparation of this book would not have been possible. Three persons in particular aroused my interest in the field of inherited metabolic disorders such that they became the focus of my professional life. Willard Centerwall, MD, of the White Memorial Medical Center, Los Angeles, California, was the first person to develop an approach to screening of infants for phenylketonuria. He introduced me to the difference nutrition management could make in the long-term outcomes of these patients. Later, Richard Koch, MD, of the Los Angeles Children's Hospital, expanded my vision to the beneficial effects of nutrition management on many other patients with inherited metabolic disorders. Subsequently, Louis J. Elsas II, MD, of the Emory University School of Medicine, Atlanta, Georgia, helped broaden my knowledge of the field by requesting justification of the nutrition management prescriptions I wrote, resulting in increased professional confidence in my knowledge and expertise in the expanding field of inherited metabolic disorders.

At the same time, the several hundreds of families who trusted me to provide guidance for the nutrition management of their children with inherited metabolic disorders supported my desire to increase my knowledge in the field in order to be able to provide further help to them. Much appreciation is extended to the contributors of the chapters in this book. Without their experience, knowledge, time, and contributions, this book would not have been completed. The editors at Jones and Bartlett were so very kind in guiding this "greenhorn" through all the work in editing a book. The great help of Christine Downs, of Columbus, Ohio, was essential to the outcome and the financial support of Rick Finkel, President of Applied Nutrition Corp., Cedar Knoll, New Jersey, helped defray the cost of word processing. Thank you all very, very much.





*Nutrition Management of Patients with Inherited Metabolic Disorders* is the first book dedicated to nutrition management of patients with inherited metabolic disorders (IMDs) for which newborn screening and diagnoses are routinely practiced. The aim of this book is to supply information that will enhance the knowledge and skills needed by nutritionists, dietitians, and other healthcare professionals who provide these services to patients with IMDs. Newborn screening, followed by diagnosis, has demonstrated that about 1 of every 1000 to 3000 infants born yearly in the United States suffers from an IMD that results in mental retardation or death if untreated, and most are treatable with nutrition management.

Patients with galactosemia (galactose-1-phosphate uridyl transferase deficiency) were some of the first who were managed by diet over a century ago. Since that time, many disorders that are disastrous to patients have been diagnosed and managed by diet, improving neurological and physical outcomes. Over the years, knowledge has also accrued that has improved medical foods and nutrition management. However, nutrition problems still occur, whether due to the quality of the medical foods, inadequate prescription by healthcare providers, or poor diet adherence by the patient. It is hoped that this book will adequately describe these problems and supply appropriate knowledge to encourage medical food manufacturers, medical geneticists, nutritionists/dietitians, and other healthcare providers to search further and to conduct the research needed to find alternative forms of nutrients that would provide optimal nutrition and health for the patients. It is anticipated that nutrition management and other therapies of many disorders will continue to improve in the future. This book should be helpful in promoting nutrition management.

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