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Titolo	Nutrition Management of Inherited Metabolic Diseases : Lessons from Metabolic University / / edited by Laurie E. Bernstein, Fran Rohr, Sandy van Calcar
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Nota di bibliografia	Includes bibliographical references and index.
Nota di contenuto	Background: Introduction to Genetics -- Expanded Newborn Screening for Inherited Metabolic Diseases -- Nutrition Education -- Pathophysiology of Inherited Metabolic Disease -- Metabolic Intoxication Syndrome in a Newborn -- Anabolism: Practical Strategies -- Protein Requirements in Inherited Metabolic Diseases -- Laboratory Evaluations in Inherited Metabolic Diseases. Aminoacidopathies: Phenyketonuria: Phenylalanine Neurotoxicity -- Phenylketonuria: The Diet Basics -- Understanding Large Neutral Amino Acids and the Blood Brain Barrier -- Tetrahydrobiopterin Therapy in Phenylketonuria -- Maternal Phenylketonuria -- Homocystinuria: Diagnosis and Management -- Nutrition Management of Urea Cycle Disorders -- Nutrition Management of Maple Syrup Urine Disease. Organic Acidemias: Organic Acidemias -- Glutaric Acidemia Type 1: Diagnosis and Management -- Nutrition Management of Glutaric Acidemia Type 1 -- Nutrition Management of Propionic Acidemia and Methylmalonic Acidemia -- NutritionManagement during Pregnancy: Maple Syrup

Urine Disease, Propionic Acidemia and Urea Cycle Disorders. Fatty Acid Oxidation Disorders: Fatty Acid Oxidation Disorders -- Nutrition Studies in Long Chain Fatty Acid Oxidation Disorders: Diet Composition and Monitoring -- Nutrition Management of Fatty Acid Oxidation Disorders. Disorders of Carbohydrate Metabolism: Nutrition Management of Galactosemia -- Glycogen Storage Disease -- Nutrition Management of Glycogen Storage Disease Type 1.

Sommario/riassunto

This text presents a compilation of topics that have been taught at Metabolic University (MU), an interactive, didactic educational program that has trained over 600 metabolic dietitians/nutritionists, physicians, nurses and genetic counselors. This book was created in 2014 for the metabolic community. The 1st edition contains only subject matter covered at Metabolic University; therefore, it is not a comprehensive treatise on Inherited Metabolic Disorders (IMD) but rather a text on the most frequently encountered challenges in IMD nutrition. Each chapter in the book highlights principles of nutrition management, how to initiate a diet, and biomarkers to monitor the diet. Recognizing that there are variations in practice, this book addresses that the key to management lies in the understanding how the inactivity of an enzyme in a metabolic pathway determines which components of the diet must be restricted and which must be supplemented as well as the monitoring of appropriate biomarkers to make diet adjustments and ensure the goals of therapy are met The 2nd edition is an updated and more extensive version covering the nutrition management of IMD, and covers a wide range of these disorders, including phenylketonuria and other aminoacidopathies, organic acidemias, urea cycle disorders, fatty acid oxidation disorders, galactosemia and glycogen storage diseases. Guidance is also provided on laboratory evaluations and biochemical testing and monitoring. Topics such as newborn screening for IMD, as well as nutrition management during pregnancy and transplantation, are also addressed. In addition, current medical management therapies is included.