

1. Record Nr.	UNINA9910787948803321
Titolo	Inborn errors of metabolism : from neonatal screening to metabolic pathways // edited by Brendan H. Lee and Fernando Scaglia
Pubbl/distr/stampa	Oxford, [England] ; ; New York, New York : , : Oxford University Press, , 2015 ©2015
ISBN	0-19-939850-X 0-19-979768-4
Descrizione fisica	1 online resource (393 p.)
Collana	Oxford Monographs on Medical Genetics
Disciplina	616.3/9042
Soggetti	Metabolism, Inborn errors of
Lingua di pubblicazione	Inglese
Formato	Materiale a stampa
Livello bibliografico	Monografia
Note generali	Description based upon print version of record.
Nota di bibliografia	Includes bibliographical references and index.
Nota di contenuto	Cover; Series; Inborn Errors of Metabolism; Copyright; Contents; Contributors; About the Editors; Introduction; Section 1 Newborn Screening; 1 Newborn Screening for Inborn Errors of Metabolism: Introduction and Approaches for Confirmation; Section 2 Pathways; 2 Human Glycosylation Disorders: Many Faces, Many Pathways; 3 Gluconeogenesis; 4 Branched Chain Amino Acid Disorders; 5 Glycolysis; 6 Urea Cycle: Ureagenesis and Non-Ureagenic Functions; 7 Fatty Acid Metabolism and Defects; 8 Mitochondrial Disorders; 9 Cholesterol, Sterols, and Isoprenoids; 10 Disorders of One-Carbon Metabolism 11 Neurotransmission and Neurotoxicity (Phenylketonuria and Dopamine)Section 3 Therapeutic Approaches; 12 Cell and Organ Transplantation for Inborn Errors of Metabolism; 13 Gene Replacement Therapy for Inborn Errors of Metabolism; 14 Enzyme Replacement and Other Therapies for the Lysosomal Storage Disorders; 15 Chaperone Therapy for the Lysosomal Storage Disorders; 16 Substrate Deprivation Therapy; Index; Colour Plate
Sommario/riassunto	Texts on inborn errors of metabolism (IEMs) have traditionally focused on classical biochemistry, clinical presentation, and standard treatment approaches. Inborn Errors of Metabolism is an expansion on this model, one that establishes an innovative pathway approach and provides a new authority on this family of disease. Alongside the

standard cadre of molecular and clinical underpinnings, this volume includes coverage of newborn screenings and an overarching treatment of IEMs as complex diseases -- how basic alterations can lead to complex secondary and tertiary effects in metabolism that con
