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| 1. Record Nr.           | UNINA9910150449003321                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                        |
| Titolo                  | Inborn Metabolic Diseases : Diagnosis and Treatment / / edited by Jean-Marie Saudubray, Matthias R. Baumgartner, John H. Walter                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                              |
| Pubbl/distr/stampa      | Berlin, Heidelberg : , : Springer Berlin Heidelberg : , : Imprint : Springer, , 2016                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                         |
| ISBN                    | 3-662-49771-9                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                |
| Edizione                | [6th ed. 2016.]                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                              |
| Descrizione fisica      | 1 online resource (XXXI, 658 p.)                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                             |
| Disciplina              | 618.92                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                       |
| Soggetti                | Pediatrics<br>Human genetics<br>Endocrinology<br>Neurology<br>Human Genetics                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                 |
| Lingua di pubblicazione | Inglese                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                      |
| Formato                 | Materiale a stampa                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                           |
| Livello bibliografico   | Monografia                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                   |
| Note generali           | Includes index.<br>"With 81 figures."                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                        |
| Nota di contenuto       | Section I Diagnosis and Treatment: General Principles -- Section II Disorders of Carbohydrate Metabolism -- Section III Disorders of Mitochondrial Energy Metabolism -- Section IV Disorders of Amino Acid Metabolism and Transport -- Section V Vitamin-Responsive Disorders -- Section VI Neurotransmitter and Small Peptide Disorders -- Section VII Disorders of Lipid and Bile Acid Metabolism -- Section VIII Disorders of Nucleic and Heme Metabolism -- 35 Disorders of Purine and Pyrimidine Metabolism -- 36 Disorders of Haem Biosynthesis -- Section IX Disorders of Metal Transport -- Section X Organelle-Related Disorders: Lysosomes, Peroxisomes, and Golgi and Pre-Golgi Systems -- Section XI Appendices. |
| Sommario/riassunto      | This work is recognised as the standard textbook for professionals involved in the diagnosis and management of inborn errors of metabolism (IEM) and an essential resource in this multidisciplinary field. For the 6th edition all 43 chapters have been newly written or revised by authors with particular expertise in their subject areas. Contents A clinical and biochemical approach to the recognition and                                                                                                                                                                                                                                                                                                          |

diagnosis of IEM with algorithms to symptoms, signs, and syndromes in patients of all ages; Emergency treatments; Medications. Separate comprehensive sections on IEM of: Carbohydrates; Mitochondrial Energy; Amino and organic acids; Vitamin-responsive defects; Neurotransmitter and Small peptides, Lipid and Bile Acids; Nucleic Acid and Heme; Organelles. Disorders affecting the synthesis and remodelling of complex lipids and fatty acid homeostasis are now included. The Editors Jean-Marie Saudubray, M.D., is Emeritus Professor of Pediatrics and Expert Metabolic Consultant at the adult neuro-metabolic unit in the Hôpital La Pitié Salpêtrière from the Université Pierre et Marie Curie, Paris. Matthias R. Baumgartner, M.D., Professor of Paediatrics and Inherited Metabolic Medicine, University of Zurich, is Head of the Division of Metabolism and the Swiss Newborn Screening Programme at the University Children's Hospital, Zurich. John Walter, M.D. is Honorary Clinical Professor of Inherited Metabolic Medicine, Manchester Academic Health Science Centre, Willink Unit, Manchester.

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| 2. Record Nr.           | UNISA996640371403316                                                                                                                                                   |
| Autore                  | GUERRINI, Mauro <1953- >                                                                                                                                               |
| Titolo                  | Biblioteconomia come impegno civile : lectio magistralis a conclusione del percorso accademico : 27 ottobre 2023, Palazzo Marucelli Fenzi, Aula Magna / Mauro Guerrini |
| Pubbl/distr/stampa      | Firenze, : Università degli Studi di Firenze, Dipartimento di Storia, Archeologia, Geografia, Arte e Spettacolo, 2023                                                  |
| Descrizione fisica      | 26 p. ; 24 cm                                                                                                                                                          |
| Disciplina              | 025.3                                                                                                                                                                  |
| Soggetti                | Biblioteconomia                                                                                                                                                        |
| Collocazione            | XIV Misc..1. 66                                                                                                                                                        |
| Lingua di pubblicazione | Italiano                                                                                                                                                               |
| Formato                 | Materiale a stampa                                                                                                                                                     |
| Livello bibliografico   | Monografia                                                                                                                                                             |
| Note generali           | Edizione fuori commercio                                                                                                                                               |