1. Record Nr. UNINA9910298428603321

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| Titolo | JIMD Reports, Volume 40 [[electronic resource] /] / edited by Eva Morava, Matthias Baumgartner, Marc Patterson, Shamima Rahman, Johannes Zschocke, Verena Peters |
| Pubbl/distr/stampa | Berlin, Heidelberg : , : Springer Berlin Heidelberg : , : Imprint : Springer, , 2018 |
| ISBN | 3-662-57880-8 |
| Edizione | [1st ed. 2018.] |
| Descrizione fisica | 1 online resource (VI, 103 p. 20 illus., 13 illus. in color.) |
| Collana | JIMD Reports, , 2192-8304 ; ; 40 |
| Disciplina | 611.01816<br>599.935 |
| Soggetti | Human genetics<br>Metabolic diseases<br>Pediatrics<br>Molecular biology<br>Human Genetics<br>Metabolic Diseases<br>Molecular Medicine |
| Lingua di pubblicazione | Inglese |
| Formato | Materiale a stampa |
| Livello bibliografico | Monografia |
| Nota di contenuto | Natural History of Aromatic L-Amino Acid Decarboxylase Deficiency in Taiwan -- Nitisinone-Induced Keratopathy in Alkaptonuria: A Challenging Diagnosis Despite Clinical Suspicion -- ALG13-CDG with Infantile Spasms in a Male Patient Due to a De Novo ALG13 Gene Mutation -- Liver Failure as the Presentation of Ornithine Transcarbamylase Deficiency in a 13-Month-Old Female -- The Use of d2 and Benton Tests for Assessment of Attention Deficits and Visual Memory in Teenagers with Phenylketonuria -- Asymptomatic Corneal Keratopathy Secondary to Hypertyrosinaemia Following Low Dose Nitisinone and a Literature Review of Tyrosine Keratopathy in Alkaptonuria -- Hyperphenylalaninaemias in Estonia: Genotype–Phenotype Correlation and Comparative Overview of the Patient Cohort Before and After Nation-Wide Neonatal Screening -- Clinical, Biochemical, and Molecular Features in 37 Saudi Patients with Very |

Long Chain Acyl CoA Dehydrogenase Deficiency -- Novel Missense LCAT Gene Mutation Associated with an Atypical Phenotype of Familial LCAT Deficiency in Two Portuguese Brothers -- Mitochondrial 3-Hydroxy-3-Methylglutaryl-CoA Synthase Deficiency: Unique Presenting Laboratory Values and a Review of Biochemical and Clinical Features -- Extended Experience of Lower Dose Sapropterin in Irish Adults with Mild Phenylketonuria -- Fumarase Deficiency: A Safe and Potentially Disease Modifying Effect of High Fat/Low Carbohydrate Diet -- Early Diagnosed and Treated Glutaric Acidemia Type 1 Female Presenting with Subependymal Nodules in Adulthood -- Mitochondrial Trifunctional Protein Deficiency: Severe Cardiomyopathy and Cardiac Transplantation -- Three Cases of Hereditary Tyrosinaemia Type 1: Neuropsychiatric Outcomes and Brain Imaging Following Treatment with NTBC.

Sommario/riassunto

JIMD Reports publishes case and short research reports in the area of inherited metabolic disorders. Case reports highlight some unusual or previously unrecorded feature relevant to the disorder, or serve as an important reminder of clinical or biochemical features of a Mendelian disorder.

2. Record Nr. UNINA9910707318203321

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| Autore | Lewis Richard Quintin <1919-> |
| Titolo | Geology and uranium deposits of Monument Valley, San Juan County, Utah / / by Richard Q. Lewis, Sr., and Donald E. Trimble |
| Pubbl/distr/stampa | Washington, D.C. : , : United States Department of the Interior, Geological Survey, , 1959<br>Washington, D.C. : , : Government Printing Office |
| Descrizione fisica | 1 online resource (iv, 105-131 pages, 2 pages of plates) : illustrations, maps |
| Collana | Contributions to the geology of uranium<br>Geological Survey bulletin ; ; 1087-D |
| Soggetti | Geology - Utah - San Juan County<br>Uranium ores - Utah - San Juan County<br>Geology<br>Uranium ores<br>Utah San Juan County |
| Lingua di pubblicazione | Inglese |
| Formato | Materiale a stampa |
| Livello bibliografico | Monografia |
| Note generali | "This report concerns work done on behalf of the U.S. Atomic Energy Commission and is published with the permission of the Commission."<br>Title from title screen (viewed July 23, 2014). |
| Nota di bibliografia | Includes bibliographical references (pages 130-131). |