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| 1. Record Nr.           | UNINA9910298429603321                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                        |
| Titolo                  | JIMD Reports, Volume 42 / / edited by Eva Morava, Matthias Baumgartner, Marc Patterson, Shamima Rahman, Johannes Zschocke, Verena Peters                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                     |
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| Descrizione fisica      | 1 online resource (116 pages) : illustrations                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                |
| Collana                 | JIMD Reports, , 2192-8304 ; ; 42                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                             |
| Disciplina              | 616.39042                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                    |
| Soggetti                | Human genetics<br>Metabolism - Disorders<br>Pediatrics<br>Molecular biology<br>Human Genetics<br>Metabolic Diseases<br>Molecular Medicine                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                    |
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| Livello bibliografico   | Monografia                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                                   |
| Nota di contenuto       | Acute Pancreatitis Secondary to Severe Hypertriglyceridaemia in a Patient with Type 1a Glycogen Storage Disease: Emergent Use of Plasmapheresis -- A Third Case of Glycogen Storage Disease IB and Giant Cell Tumour of the Mandible: A Disease Association or Iatrogenic Complication of Therapy -- Cardiopulmonary Exercise Testing Reflects Improved Exercise Capacity in Response to Treatment in Morquio A Patients: Results of a 52-Week Pilot Study of Two Different Doses of Elosulfase Alfa -- EPG5-Related Vici Syndrome: A Primary Defect of Autophagic Regulation with an Emerging Phenotype Overlapping with Mitochondrial Disorders -- Compound Heterozygous Inheritance of Mutations in Coenzyme Q8A Results in Autosomal Recessive Cerebellar Ataxia and Coenzyme Q10 Deficiency in a Female Sib-Pair -- The Validity of Bioelectrical Impedance Analysis to Measure Body Composition in Phenylketonuria -- Effect of Storage Conditions on Stability of Ophthalmological Compounded Cysteamine Eye Drops -- |

Leber Hereditary Optic Neuropathy and Longitudinally Extensive Transverse Myelitis -- Mitochondrial Disease in Children: The Nephrologist's Perspective -- Characterization of Phenylalanine Hydroxylase Gene Mutations in Chilean PKU Patients -- Long-Term Systematic Monitoring of Four Polish Transaldolase Deficient Patients -- Coping Strategies, Stress, and Support Needs in Caregivers of Children with Mucopolysaccharidosis -- Beneficial Effect of BH4 Treatment in a 15-Year-Old Boy with Biallelic Mutations in DNAJC12 -- Secondary Hemophagocytic Syndrome Associated with COG6 Gene Defect: Report and Review -- Mitochondrial Encephalopathy: First Portuguese Report of a VARS2 Causative Variant.

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#### 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.

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