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| Nota di contenuto | First Successful Conception Induced by a Male Cystinosis Patient -- Glutaric Acidemia Type 1: A Case of Infantile Stroke -- Treatment of Depression in Adults with Fabry Disease -- Mutations in GMPPB Presenting with Pseudometabolic Myopathy -- Heterogeneous Phenotypes in Lipid Storage Myopathy Due to ETFDH Gene Mutations -- Successful Management of Pregnancies in Patients with Inherited Disorders of Ketone Body Metabolism -- Improvement of Fabry Disease-Related Gastrointestinal Symptoms in a Significant Proportion of Female Patients Treated with Agalsidase Beta: Data from the Fabry Registry -- Ketone Bodies as a Possible Adjuvant to Ketogenic Diet in PDHc Deficiency but Not in GLUT1 Deficiency -- GM2 Activator Deficiency Caused by a Homozygous Exon 2 Deletion in GM2A -- Effect of Lorenzo's Oil on Hepatic Gene Expression and the Serum Fatty Acid Level in abcd1-Deficient Mice -- Introduction of a Simple Second Tier Screening Test for C5 Isobars in Dried Blood Spots: Reducing the False |

Positive Rate for Isovaleric Acidaemia in Expanded Newborn Screening -- Open-Label Single-Sequence Crossover Study Evaluating Pharmacokinetics, Efficacy, and Safety of Once-Daily Dosing of Nitisinone in Patients with Hereditary Tyrosinemia Type 1 -- A Rapid Two-Step Iduronate-2-Sulfatase Enzymatic Activity Assay for MPSII Pharmacokinetic Assessment -- An Unexplained Congenital Disorder of Glycosylation-II in a Child with Neurohepatic Involvement, Hypercholesterolemia and Hypoceruloplasminemia -- Peripheral Neuropathy, Episodic Rhabdomyolysis, and Hypoparathyroidism in a Patient with Mitochondrial Trifunctional Protein Deficiency.

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. The chapter 'Open-Label Single-Sequence Crossover Study Evaluating Pharmacokinetics, Efficacy, and Safety of Once-Daily Dosing of Nitisinone in Patients with Hereditary Tyrosinemia Type 1 (HT-1)' is open access under a CC BY 4.0 license via link.springer.com.
