1. Record Nr. UNINA9910253898003321

Titolo	JIMD Reports, Volume 28 / / edited by Eva Morava, Matthias Baumgartner, Marc Patterson, Shamima Rahman, Johannes Zschocke, Verena Peters
Pubbl/distr/stampa	Berlin, Heidelberg : , : Springer Berlin Heidelberg : , : Imprint : Springer, , 2016
ISBN	3-662-52847-9
Edizione	[1st ed. 2016.]
Descrizione fisica	1 online resource (VI, 135 p.)
Collana	JIMD Reports, , 2192-8304 ; ; 28
Disciplina	616.39042
Soggetti	Human genetics Metabolic diseases Pediatrics Molecular biology Human Genetics Metabolic Diseases Molecular Medicine
Lingua di pubblicazione	Inglese
Formato	Materiale a stampa
Livello bibliografico	Monografia
Nota di bibliografia	Includes bibliographical references at the end of each chapters.
Nota di contenuto	Lethal Neonatal Progression of Fetal Cardiomegaly Associated to ACAD9 Deficiency -- Novel Direct Assay for Acetyl-CoA:-Glucosaminide N-Acetyltransferase Using BODIPY-Glucosamine as a Substrate -- Electrical Changes in Resting, Exercise, and Holter Electrocardiography in Fabry Cardiomyopathy -- The Nutritional Intake of Patients with Organic Acidaemias on Enteral Tube Feeding: Can We Do Better? -- Normoglycemic Ketonemia as Biochemical Presentation in Ketotic Glycogen Storage Disease -- LARS2 Variants Associated with Hydrops, Lactic Acidosis, Sideroblastic Anemia, and Multisystem Failure -- Lower Urinary Tract Symptoms and Incontinence in Children with Pompe Disease -- Cerebral Lipid Accumulation Detected by MRS in a Child with Carnitine Palmitoyltransferase 2 Deficiency: A Case Report and Review of the Literature on Genetic Etiologies of Lipid Peaks on MRS -- Neuropsychological Development in Patients with Long-Chain 3-Hydroxyacyl-CoA Dehydrogenase (LCHAD) Deficiency --

Enhancement by Uridine Diphosphate of Macrophage Inflammatory Protein-1 Alpha Production in Microglia Derived from Sandhoff Disease Model Mice -- In Patients with an -Galactosidase A Variant, Small Nerve Fibre Assessment Cannot Confirm a Diagnosis of Fabry Disease -- In Utero Diagnosis of Niemann–Pick Type C in the Absence of Family History -- Multiple, Successful Pregnancies in Pompe Disease -- Heterologous Expression in Yeast of Human Ornithine Carriers ORNT1 and ORNT2 and of ORNT1 Alleles Implicated in HHH Syndrome in Humans -- Inborn Errors of Metabolism in the United Arab Emirates: Disorders Detected by Newborn Screening (2011–2014).

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.