1. Record Nr. UNINA9910298425803321

Titolo	JIMD Reports, Volume 39 [[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	3-662-57577-9
Edizione	[1st ed. 2018.]
Descrizione fisica	1 online resource (VI, 116 p. 20 illus., 8 illus. in color.)
Collana	JIMD Reports, , 2192-8304 ; ; 39
Disciplina	611.01816 599.935
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 contenuto	Successful Pregnancy in a Young Woman with Multiple Acyl-CoA Dehydrogenase Deficiency -- Role of Intramuscular Levofolinate Administration in the Treatment of Hereditary Folate Malabsorption: Report of Three Cases -- The Prevalence of PMM2-CDG in Estonia Based on Population Carrier Frequencies and Diagnosed Patients -- Triheptanoin: A Rescue Therapy for Cardiogenic Shock in Carnitine-acylcarnitine Translocase Deficiency -- Glutaric Aciduria Type 1 and Acute Renal Failure: Case Report and Suggested Pathomechanisms -- Cardiovascular Histopathology of a 11-Year Old with Mucopolysaccharidosis VII Demonstrates Fibrosis, Macrophage Infiltration, and Arterial Luminal Stenosis -- Longitudinal Changes in White Matter Fractional Anisotropy in Adult-Onset Niemann-Pick Disease Type C Patients Treated with Miglustat -- Beta-Ketothiolase Deficiency Presenting with Metabolic Stroke After a Normal Newborn

Screen in Two Individuals -- Rapidly Progressive White Matter Involvement in Early Childhood: The Expanding Phenotype of Infantile Onset Pompe? -- Four Years' Experience in the Diagnosis of Very Long-Chain Acyl-CoA Dehydrogenase Deficiency in Infants Detected in Three Spanish Newborn Screening Centers -- Social Functioning and Behaviour in Mucopolysaccharidosis IH [Hurlers Syndrome] -- Mitochondrial Encephalopathy and Transient 3-Methylglutaconic Aciduria in ECHS1 Deficiency: Long-Term Follow-Up -- Glutaric Aciduria Type 3: Three Unrelated Canadian Cases, with Different Routes of Ascertainment -- High-Throughput Screen Fails to Identify Compounds That Enhance Residual Enzyme Activity of Mutant N-Acetyl--Glucosaminidase in Mucopolysaccharidosis Type IIIB -- Demographic and Psychosocial Influences on Treatment Adherence for Children and Adolescents with PKU: A Systematic Review.

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.