1. Record Nr. UNINA9910253922303321

Titolo	JIMD Reports, Volume 34 / / edited by Eva Morava, Matthias Baumgartner, Marc Patterson, Shamima Rahman, Johannes Zschocke, Verena Peters
Pubbl/distr/stampa	Berlin, Heidelberg : , : Springer Berlin Heidelberg : , : Imprint : Springer, , 2017
ISBN	3-662-55586-7
Edizione	[1st ed. 2017.]
Descrizione fisica	1 online resource (VI, 115 p.)
Collana	JIMD Reports, , 2192-8304 ; ; 34
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	Diaphragmatic Eventration in Sisters with Asparagine Synthetase Deficiency: A Novel Homozygous ASNS Mutation and Expanded Phenotype -- Measurement of Elevated Concentrations of Urine Keratan Sulfate by UPLC-MSMS in Lysosomal Storage Disorders (LSDs): Comparison of Urine Keratan Sulfate Levels in MPS IVA Versus Other LSDs -- The Spectrum of PAH Mutations and Increase of Milder Forms of Phenylketonuria in Sweden During 1965–2014 -- DMP1-CDG (CDG1e) with Significant Gastrointestinal Manifestations -- Phenotype and Genotype Expansion -- Classical Galactosaemia and CDG, the N-Glycosylation Interface. A Review -- Argininosuccinic Acid Lyase Deficiency Missed by Newborn Screen -- Very Long-Chain Acyl-Coenzyme A Dehydrogenase Deficiency and Perioperative Management in Adult Patients -- Paracentric Inversion of Chromosome 21 Leading to Disruption of the HLCS Gene in a Family with Holocarboxylase Synthetase Deficiency -- Delayed Infusion Reactions to Enzyme

Replacement Therapies -- Novel PEX3 Gene Mutations Resulting in a Moderate Zellweger Spectrum Disorder -- Improved Measurement of Brain Phenylalanine and Tyrosine Related to Neuropsychological Functioning in Phenylketonuria -- Table of Phenylalanine Content of Foods: Comparative Analysis of Data Compiled in Food Composition Tables -- Inhaled Sargramostim Induces Resolution of Pulmonary Alveolar Proteinosis in Lysinuric Protein Intolerance -- COXPD9 an Evolving Multisystem Disease -- Congenital Lactic Acidosis, Sensorineural Hearing Loss, Hypertrophic Cardiomyopathy, Cirrhosis and Interstitial Nephritis -- Incidence and Geographic Distribution of Succinic Semialdehyde Dehydrogenase (SSADH) 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.