1. Record Nr. UNINA9910298416303321

Titolo	JIMD Reports, Volume 41 [[electronic resource]] : Focus Issue: Adults and Metabolism / / 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	3-662-58081-0
Edizione	[1st ed. 2018.]
Descrizione fisica	1 online resource (128 pages)
Collana	JIMD Reports, , 2192-8304 ; ; 41
Disciplina	616.39
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	Assessment of the Effect of Once Daily Nitisinone Therapy on 24-h Urinary Metadrenalines and 5-Hydroxyindole Acetic Acid Excretion in Patients with Alkaptonuria After 4 Weeks of Treatment -- Severe Hyperammonemic Encephalopathy Requiring Dialysis Aggravated by Prolonged Fasting and Intermittent High Fat Load in a Ramadan Fasting Month in a Patient with CPTII Homozygous Mutation -- Haematopoietic Stem Cell Transplantation Arrests the Progression of Neurodegenerative Disease in Late-Onset Tay-Sachs Disease -- Expert Opinion vs Patient Perspective in Treatment of Rare Disorders: Tooth Removal in Lesch-Nyhan Disease as an Example -- Two Uneventful Pregnancies in a Woman with Glutaric Aciduria Type 1 -- The Influence of Patient-Reported Joint Manifestations on Quality of Life in Fabry Patients -- Probable Diagnosis of a Patient with Niemann–Pick Disease Type C: Managing Pitfalls of Exome Sequencing -- Alkaptonuria Severity Score Index Revisited: Analysing the AKUSSI and Its Subcomponent Features

-- Reduced Muscle Strength in Barth Syndrome May Be Improved by Resistance Exercise Training: A Pilot Study -- Cognitive Impairments and Subjective Cognitive Complaints in Fabry Disease: A Nationwide Study and Review of the Literature -- Effectiveness of Early Hematopoietic Stem Cell Transplantation in Preventing Neurocognitive Decline in Mucopolysaccharidosis Type II: A Case Series -- Parenting a Child with Phenylketonuria: An Investigation into the Factors That Contribute to Parental Distress -- P-Tau and Subunit c Mitochondrial ATP Synthase Accumulation in the Central Nervous System of a Woman with Hurler–Scheie Syndrome Treated with Enzyme Replacement Therapy for 12 Years -- Serum Amino Acid Profiling in Patients with Alkaptonuria Before and After Treatment with Nitisinone -- Burden of Illness in Acid Sphingomyelinase Deficiency: A Retrospective Chart Review of 100 Patients.

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.