

1. Record Nr.	UNINA9910253932403321
Titolo	JIMD Reports, Volume 33 // 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	3-662-55012-1
Edizione	[1st ed. 2017.]
Descrizione fisica	1 online resource (VI, 110 p. 26 illus., 15 illus. in color.)
Collana	JIMD Reports, , 2192-8304 ; ; 33
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	Difficulties in Daily Life and Associated Factors, and QoL of Children with Inherited Metabolic Disease and Their Parents in Japan: A Literature Review -- Swallow Prognosis and Follow-Up Protocol in Infantile Onset Pompe Disease -- Clinical and Genetic Characteristics of Romanian Patients with Mucopolysaccharidosis Type II -- Gastrointestinal Health in Classic Galactosemia -- Management of Life-Threatening Tracheal Stenosis and Tracheomalacia in Patients with Mucopolysaccharidoses -- Brain White Matter Integrity Mediates the Relationship Between Phenylalanine Control and Executive Abilities in Children with Phenylketonuria -- The Challenges of a Successful Pregnancy in a Patient with Adult Refsum's Disease due to Phytanoyl-CoA Hydroxylase Deficiency -- Novel Homozygous Missense Mutation in SPG20 Gene Results in Troyer Syndrome Associated with Mitochondrial Cytochrome c Oxidase Deficiency -- Lethal Neonatal LTBL Associated with Biallelic EARS2 Variants: Case Report and Review

of the Reported Neuroradiological Features -- Leukoencephalopathy due to Complex II Deficiency and Bi-Allelic SDHB Mutations: Further Cases and Implications for Genetic Counselling -- Peak Jump Power Reflects the Degree of Ambulatory Ability in Patients with Mitochondrial and Other Rare Diseases -- RARS2 Mutations: Is Pontocerebellar Hypoplasia Type 6 a Mitochondrial Encephalopathy? -- Missed Newborn Screening Case of Carnitine Palmitoyltransferase-II Deficiency -- Leigh-Like Syndrome Due to Homoplasmic m.8993T>G Variant with Hypocitrullinemia and Unusual Biochemical Features Suggestive of Multiple Carboxylase Deficiency (MCD) -- Erratum to: Missed Newborn Screening Case of Carnitine Palmitoyltransferase-II Deficiency -- Erratum to: Leigh-Like Syndrome Due to Homoplasmic m.8993T>G Variant with Hypocitrullinemia and Unusual Biochemical Features Suggestive of Multiple Carboxylase Deficiency (MCD).

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.
