1. Record Nr. UNINA990006257750403321

Autore	Cerri, Augusto
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Titolo	JIMD Reports, Volume 37 / / edited by Eva Morava, Matthias Baumgartner, Marc Patterson, Shamima Rahman, Johannes Zschocke, Verena Peters
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Lingua di pubblicazione	Inglese
Formato	Materiale a stampa
Livello bibliografico	Monografia
Nota di bibliografia	Includes bibliographical references.
Nota di contenuto	Favourable Outcome in Two Pregnancies in a Patient with 3-Hydroxy-3-Methylglutaryl-CoA Lyase Deficiency -- Lysosomal Acid Lipase Deficiency in 23 Spanish Patients: High Frequency of the Novel c. 966+2T>G Mutation in Wolman Disease -- Guanidinoacetate Methyltransferase Activity in Lymphocytes, for a Fast Diagnosis -- Galactose Epimerase Deficiency: Expanding the Phenotype -- Development and Psychometric Evaluation of the MetabQoL 1.0: A Quality of Life Questionnaire for Paediatric Patients with Intoxication-Type Inborn Errors of Metabolism -- Widening the Heterogeneity of Leigh Syndrome: Clinical, Biochemical, and Neuroradiologic Features in a Patient Harboring a NDUFA10 Mutation -- Normal Neurological Development During Infancy Despite Massive Hyperammonemia in Early Treated NAGS Deficiency -- Dihydropyrimidine Dehydrogenase Deficiency: Metabolic Disease or Biochemical Phenotype? -- Potential Misdiagnosis of Hyperhomocysteinemia due to Cystathionine Beta-

	Synthase Deficiency During Pregnancy -- Clinical and Molecular Variability in Patients with PHKA2 Variants and Liver Phosphorylase b Kinase Deficiency -- Hyperphenylalaninemia Correlated with Global Decrease of Antioxidant Genes Expression in White Blood Cells of Adult Patients with Phenylketonuria -- The Impact of Fabry Disease on Reproductive Fitness -- Neonatal-Onset Hereditary Coproporphyria: A New Variant of Hereditary Coproporphyria -- Treatment Adherence and Psychological Wellbeing in Maternal Carers of Children with Phenylketonuria (PKU) -- Systematic Review and Meta-analysis of Intelligence Quotient in Early-Treated Individuals with Classical Galactosemia.
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.