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Nota di contenuto	LC-MS/MS Analysis of Cerebrospinal Fluid Metabolites in the Pterin Biosynthetic Pathway -- Renal Involvement in a French Paediatric Cohort of Patients with Lysinuric Protein Intolerance -- A Highly Diverse Portrait: Heterogeneity of Neuropsychological Profiles in cblC Defect -- Heterozygous Monocarboxylate Transporter 1 (MCT1, SLC16A1) Deficiency as a Cause of Recurrent Ketoacidosis -- Spectrum of Mutations in 60 Saudi Patients with Mut Methylmalonic Acidemia -- CoQ10 Deficiency Is Not a Common Finding in GLUT1 Deficiency Syndrome -- Correlation Between Flexible Fiberoptic Laryngoscopic and Polysomnographic Findings in Patients with Mucopolysaccharidosis Type VI -- Continual Low-Dose Infusion of Sulfamidase Is Superior to Intermittent High-Dose Delivery in Ameliorating Neuropathology in the MPS IIIA Mouse Brain -- A Short Synthetic Peptide Mimetic of Apolipoprotein A1 Mediates Cholesterol and Globotriaosylceramide Efflux from Fabry Fibroblasts -- Development of Metabolic Phenotype

in Phenylketonuria: Evaluation of the Blaskovics Protein Loading Test at 5 Years of Age -- The Lactose and Galactose Content of Cheese Suitable for Galactosaemia: New Analysis -- Atypical Clinical Presentations of TAZ Mutations: An Underdiagnosed Cause of Growth Retardation? -- Long-Term Cognitive and Functional Outcomes in Children with Mucopolysaccharidosis (MPS)-IH (Hurler Syndrome) Treated with Hematopoietic Cell Transplantation -- Treatment with Mefolinate (5-Methyltetrahydrofolate), but Not Folic Acid or Folinic Acid, Leads to Measurable 5-Methyltetrahydrofolate in Cerebrospinal Fluid in Methylenetetrahydrofolate Reductase Deficiency -- Abnormal Glycosylation Profile and High Alpha-Fetoprotein in a Patient with Twinkle Variants -- Erratum to: LC-MS/MS Analysis of Cerebrospinal Fluid Metabolites in the Pterin Biosynthetic Pathway -- Erratum to: Treatment with Mefolinate (5-Methyltetrahydrofolate), but Not Folic Acid or Folinic Acid, Leads to Measurable 5-Methyltetrahydrofolate in Cerebrospinal Fluid in Methylenetetrahydrofolate Reductase 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.