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Nota di contenuto	Multidisciplinary Team Approach Is Key for Managing Pregnancy and Delivery in Patient with Rare, Complex MPS I -- Clinical Evolution After Enzyme Replacement Therapy in Twins with the Severe Form of Maroteaux–Lamy Syndrome -- A New Approach for Fast Metabolic Diagnostics in CMAMMA -- Pilot Experience with an External Quality Assurance Scheme for Acylcarnitines in Plasma/Serum -- ECHS1 Deficiency as a Cause of Severe Neonatal Lactic Acidosis -- Chronic Oral l-Carnitine Supplementation Drives Marked Plasma TMAO Elevations in Patients with Organic Acidemias Despite Dietary Meat Restrictions -- A Founder Effect for the HGD G360R Mutation in Italy: Implications for a Regional Screening of Alkaptonuria -- Missed Newborn Screening Case of Carnitine Palmitoyltransferase-II Deficiency -- Acute Metabolic Crises in Maple Syrup Urine Disease After Liver Transplantation from a Related Heterozygous Living Donor -- Identification of Cryptic Novel -Galactosidase A Gene Mutations:

Abnormal mRNA Splicing and Large Deletions -- Severe Neonatal Presentation of Mitochondrial Citrate Carrier (SLC25A1) Deficiency -- Biomarkers in a Taurine Trial for Succinic Semialdehyde Dehydrogenase Deficiency -- A Modified Enzymatic Method for Measurement of Glycogen Content in Glycogen Storage Disease Type IV -- The Effect of Multiple Sulfatase Deficiency (MSD) on Dental Development: Can We Use the Teeth as an Early Diagnostic Tool? -- Novel Report of Phosphoserine Phosphatase Deficiency in an Adult with Myeloneuropathy and Limb Contractures -- Erratum to: Novel Report of Phosphoserine Phosphatase Deficiency in an Adult with Myeloneuropathy and Limb Contractures.

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.