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| Nota di contenuto | Cerebrotendinous Xanthomatosis Presenting with Infantile Spasms and Intellectual Disability -- Hyperammonemia as a Presenting Feature in Two Siblings with FBXL4 Variants -- Intracranial Hypertension in Cystinosis Is a Challenge: Experience in a Children's Hospital -- Severe Respiratory Acidosis in Status Epilepticus as a Possible Etiology of Sudden Death in Lesch–Nyhan Disease: A Case Report and Review of the Literature -- Vitamin B12 Administration by Subcutaneous Catheter Device in a Cobalamin A (cblA) Patient -- Expansion of the Phenotypic Spectrum of Propionic Acidemia with Isolated Elevated Propionylcarnitine -- Previously Unreported Biallelic Mutation in DNAJC19:Are Sensorineural Hearing Loss and Basal Ganglia Lesions Additional Features of Dilated Cardiomyopathy and Ataxia (DCMA) Syndrome? -- Lysosomal Storage Disorders in Nonimmune Hydrops Fetalis (NIHF): An Indian Experience -- The Risk of Fatty Acid Oxidation |

Disorders and Organic Acidemias in Children with Normal Newborn Screening -- Clinical and Mutational Characterizations of Ten Indian Patients with Beta-Ketothiolase Deficiency -- Atypical Presentation and Treatment Response in a Child with Familial Hypercholesterolemia Having a Novel LDLR Mutation -- Development of a Tandem Mass Spectrometry Method for Rapid Measurement of Medium- and Very-Long-Chain Acyl-CoA Dehydrogenase Activity in Fibroblasts -- Analysis of Melanin-like Pigment Synthesized from Homogentisic Acid, with or without Tyrosine, and Its Implications in Alkaptonuria -- Bone Health in Classic Galactosemia: Systematic Review and Meta-Analysis -- Cognitive Development in a Young Child with Mucolipidosis Type IV: A Case Report -- White Matter Microstructure and Subcortical Gray Matter Structure Volumes in Aspartylglucosaminuria; a 5-Year Follow-up Brain MRI Study of an Adolescent with Aspartylglucosaminuria and His Healthy Twin Brother -- Erratum to: White Matter Microstructure and Subcortical Gray Matter Structure Volumes in Aspartylglucosaminuria; a 5-Year Follow-up Brain MRI Study of an Adolescent with Aspartylglucosaminuria and His Healthy Twin Brother.

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.