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Nota di contenuto	Contents; Enzyme Replacement Therapy in a Patient with Gaucher Disease Type III: A Paradigmatic Case Showing Severe Adverse Reactions Started a Long Time After the Beginning of Treatment; Abstract; Introduction; Case Report; Discussion; Contributors; References; Expanding the Spectrum of Methylmalonic Acid-Induced Pallidal Stroke: First Reported Case of Metabolic Globus Pallidus Stroke: First Reported Case of Metabolic Globus Pallidus Stroke in Transcobalamin II Deficiency; Abstract; Introduction; Case Report; Discussion; Synopsis; Contributions; Conflict of Interest; References A Large Intragenic Deletion in the ACADM Gene Can Cause MCAD Deficiency but is not Detected on Routine SequencingAbstract; Introduction; Case Report; Discussion; Conclusion; Take-Home Message; Details of the Contributions of Individual Authors; Guarantor for the Article; Conflict of Interests Statement; Ethics Approval; Patient Consent Statement; References; Infantile Hypophosphatasia Secondary to a Novel Compound Heterozygous Mutation Presenting with Pyridoxine-Responsive Seizures; Abstract; Introduction; Patient Presentation; Discussion; Synopsis; Conflict of Interest Contributor s Statement PageReferences; Liver Transplantation Prevents Progressive Neurological Impairment in Argininemia; Introduction; Material and Methods; Case Reports; Discussion; Concise Sentence Take-Home Message; Reference to Electronic Databases; Conflict of Interest; References; Motor and Speech Disorders in Classic Galactosemia; Abstract; Methods; Statistical Analysis; Results; Speech; Strength; Coordination; Days on Milk; Discussion; Speech; Strength;

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Intracellular Metabolism Presenting as Diabetic Ketoacidosis: A Rare Manifestation; Abstract; Introduction; Case Report; Discussion; Conclusion; Conflicts of Interest; Funding; Take-Home Message; References; Cerebral Magnetic Resonance Spectroscopy Demonstrates Long-Term Effect of Bone Marrow Transplantation in -Mannosidosis; Abstract; Introduction; Patient; Methods; Results; Discussion
ConclusionReferences; Early Cardiac Changes in Children with Anderson-Fabry Disease; Abstract; Introduction; Methods; Study Population; Echocardiography; ECG Data and Holter Monitoring; Statistical Analysis; Results; Study Population Characteristics; Progression of Left Ventricular Hypertrophy; 12-Leads ECG Parameters; Holter ECG Parameters; Discussion; Left Ventricular Mass and Left Ventricular Hypertrophy; T Wave Inversion; ECG LV Mass Indexes; 24-h ECG Monitors; Conduction Intervals; N215S Mutation; Limitations; Conclusion; Synopsis; Details of the Contributions of Individual Authors Guarantor Author

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.
