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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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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.
