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Nota di contenuto	Contents; Molecular Analysis of Turkish Maroteaux-Lamy Patients and Identification of One Novel Mutation in the Arylsulfatase B (ARSB) G...; Abstract; Introduction; Materials and Methods; Patients; ARSB Mutation Analysis; Sequence Variations Nomenclature; Analysis of the New Missense Sequence Variation; Results; Genotype-Phenotype

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ConclusionConflict of Interest; Informed Consent; Details of the Contributions of Individual Authors; References; Abnormalities in Glycogen Metabolism in a Patient with Alpers´ Syndrome Presenting with Hypoglycemia; Abstract; Introduction; Case Report; Discussion; Competing Interest; Synopsis; Compliance with Ethics Guidelines; Conflict of Interest; Informed Consent; Animal Rights; Contributions of Individual Authors; References; Assessment of Basal Metabolic Rate and Nutritional Status in Patients with Gaucher Disease Type III; Abstract; Introduction; Methods; Results; Basal Metabolic Rate
Nutritional StatusDiscussion; Conclusion; Summary; References; Mutations in the Complex III Assembly Factor Tetratricopeptide 19 Gene TTC19 Are a Rare Cause of Leigh Syndrome; Abstract; Introduction; Conclusions; Compliance with Ethics Guidelines; Informed Consent; References; Quality of Life in Adult Patients with Glycogen Storage Disease Type I: Results of a Multicenter Italian Study; Abstract; Introduction; Methods; Statistical Analysis; Results; Discussion; Conclusions; Compliance with Ethics Guidelines; References
The Molecular Bases of Phenylketonuria (PKU) in New South Wales, Australia: Mutation Profile and Correlation with Tetrahydrobi...Abstract; Introduction; Materials and Methods; Results; Discussion; Conflict of Interest; Synopsis; Compliance with Ethics Guidelines; Details of Contributions of Individual Authors; References; Extraosseous Extension Caused by Epidural Hematoma in Gaucher Disease Mimicking Malignant Bone Tumor; Abstract; Introduction; Case Report; Discussion; Conflict of Interest; Informed Consent; Contributions of Individual Authors; References
Thirteen Patients with MAT1A Mutations Detected Through Newborn Screening: 13 Years´ Experience

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.