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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 Correlation; Family Studies; Discussion; One-Sentence Take-Home Message; Details of the Contributions of Individual Authors; Name of One Author Who Serves as Guarantor; Details of Funding; Details of Ethics Approval; A Patient Consent Statement; Conflict of Interest ReferencesDanon Disease Due to a Novel LAMP2 Microduplication; |

Abstract; Introduction; Methods; Results; Discussion; Acknowledgments; Synopsis; Compliance with Ethics Guidelines; Author Contributions; References; Secondary Mitochondrial Respiratory Chain Defect Can Delay Accurate PFIC2 Diagnosis; Abstract; Introduction; Case Reports; Discussion; One-Sentence Take-Home Message; Contribution of Individual Authors; References; Newborn Screening for Hunter Disease: A Small-Scale Feasibility Study; Abstract; Introduction; Materials and Methods; DBS Samples; Enzyme Assay; Results and Discussion
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
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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.