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Altri autori (Persone)	ZschockeJohannes GibsonK. Michael BrownGarry MoravaEva PetersVerena
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Nota di contenuto	Contents; Prevalence and Novel Mutations of Lysosomal Storage Disorders in United Arab Emirates; Abstract; Introduction; Materials and Methods; Patients; Birth Prevalence; Mutation Analysis; Results; Discussion; Disclosure Statement; References; Chitotriosidase Deficiency: A Mutation Update in an African Population; Abstract; Introduction; Materials and Methods; Subjects; Methods; Plasma Chitotriosidase Assay; CHIT1 Mutation Detection; Population Screening for CHIT1 Mutations; Evaluation of Missense Mutations; Results; Plasma chito activities and CHIT1 genotyping; Discussion and Conclusion ReferencesNDUFS8-related Complex I Deficiency Extends Phenotype from ``PEO Plus to Leigh Syndrome; Abstract; Introduction; Case Reports; Patient 1; Patient 2; Patient 3; Results; Discussion; Synopsis; Author Contribution; Funding; Ethical Issues; Competing Interest Statement; References; Accuracy of Six Anthropometric Skinfold Formulas Versus Air Displacement Plethysmography for Estimating Percent Body Fat in Female Adolescents with Phenylketonuria; Abstract; Introduction; Subjects and Methods; Subjects; Data Collection;

Anthropometric Calculations; Statistical Analysis; Results
Demographics, BMI, and ADP Body CompositionMean Values and Correlations; Bland-Altman Agreement; Variation in Percent Error; Discussion; Agreement of Tested Equations with ADP Results; Adiposity in PKU and Clinical Relevance; Conclusion; Synopsis; Contributions of Individual Authors; Author Serving as Guarantor; Competing Interest Statement; Details of Funding; Details of Ethics Approval; Patient Consent Statement; References; Noncompaction of the Ventricular Myocardium and Hydrops Fetalis in Cobalamin C Disease; Abstract; Introduction; Case Report; Discussion; Synopsis; References
Primary Carnitine (OCTN2) Deficiency Without Neonatal Carnitine DeficiencyAbstract; Introduction; References; MNGIE Syndrome: Liver Cirrhosis Should Be Ruled Out Prior to Bone Marrow Transplantation; Abstract; Introduction; Financial Disclosure; References; Differential Phonological Awareness Skills in Children with Classic Galactosemia: A Descriptive Study of Four Cases; Abstract; Introduction; Method; Participants; Child 1; Child 2; Child 3; Child 4; Measures; Procedure; Descriptive Analyses; Results; Discussion; Clinical Implications and Conclusion; References
Dihydropteridine Reductase Deficiency and Treatment with Tetrahydrobiopterin: A Case ReportAbstract; Introduction; Case Report; Biochemical Results; Discussion; Synopsis; References to Electronic Databases; References; Stem Cell Transplantation for Adult-Onset Krabbe Disease: Report of a Case; Abstract; Introduction; Report of a Case; Disclosures and Funding; References; Detection by Urinary GAG Testing of Mucopolysaccharidosis Type II in an At-Risk Spanish Population; Abstract; Introduction; Objective; Patients and Methods; Study Population; Methods; Statistical Analysis; Results; Discussion Synopsis

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.