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Note generali	Description based upon print version of record.
Nota di bibliografia	Includes bibliographical references at the end of each chapters.
Nota di contenuto	Contents; Severe Neonatal Holocarboxylase Synthetase Deficiency in West African Siblings; Abstract; Introduction; Subjects, Methods, and Results; Case 1; Case 2; Discussion; Synopsis; Compliance with Ethics Guidelines; Conflict of Interest Statements; Details of the Contributions of Individual Authors; References; Expanding Our Understanding of Lower Urinary Tract Symptoms and Incontinence in Adults with Pompe Disease; Abstract; Introduction; Methods; Patient Population; Questionnaire Details; Statistical Analysis; Results; Discussion; Conclusion; Synopsis; Compliance with Ethics Guidelines

Conflict of InterestReferences; Carnitine-Acylcarnitine Translocase Deficiency: Experience with Four Cases in Spain and Review of the Literature; Abstract; Introduction; Case Reports; Results and Discussion; Clinical and Laboratory Presenting Findings; Biochemical Findings; Mutational Findings; Long-Term Prognosis; Dietary Treatment and Monitoring; Final Considerations; Take-Home Message; Compliance with Ethics Guidelines; Conflict of Interest; Informed Consent; Details of the Contributions of Individual Authors; References
Lack of Glibenclamide Response in a Case of Permanent Neonatal Diabetes Caused by Incomplete Inactivation of GlucokinaseAbstract; Introduction; Case History and Glibenclamide Treatment; Methods; Genetic Studies; Enzymatic Analysis of Wild-Type and Mutant GCK; Statistical Analysis; Results; Identification of GCK Mutations; Functional Characterisation of p.Ile19Asn Mutation; Glibenclamide Treatment; Discussion; Synopsis; Compliance with Ethics Guidelines; Conflict of Interest; Informed Consent; Animal Rights; Contributions of Individual Authors; References
Morphology and Function of Cerebral Arteries in Adults with Pompe DiseaseAbstract; Introduction; Patients and Methods; Patients; Gd-MRA; Sonography; Statistics; Informed Consent and Ethics; Results; Discussion; Compliance with Ethics Guidelines; Conflict of Interest; Informed Consent; Individual Contributions; References; Parkinsonism in Phenylketonuria: A Consequence of Dopamine Depletion?; Abstract; Introduction; Patients and Methods; Discussion; Synopsis; Compliance with Ethics Guidelines; Conflict of Interest; Informed Consent; Contribution of the Authors; References
Characterization of Variegate Porphyria Mutations Using a Minigene ApproachAbstract; Introduction; Materials and Methods; Patients; Constructs; Minigenes Expression Assays; Statistical Analysis; Bioinformatic Tools; Results; Discussion; Synopsis; Details of Contribution of Individual Authors; Conflict of Interest; Details of Funding; Details of Ethic Approval; References; Homozygous Truncating Intragenic Duplication in TUSC3 Responsible for Rare Autosomal Recessive Nonsyndromic Intellectual Disab...; Abstract; Introduction; Clinical Report; Patient 1; Patient 2; Methods; Array-CGH
Molecular Investigations

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.