1. Record Nr. UNINA9910438005803321

Titolo	JIMD Reports - Case and Research Reports, 2012/4 [[electronic resource] /] / edited by Johannes Zschocke, K Michael Gibson, Garry Brown, Eva Morava, Verena Peters
Pubbl/distr/stampa	Berlin, Heidelberg : , : Springer Berlin Heidelberg : , : Imprint : Springer, , 2013
ISBN	1-283-63178-4 9786613944238 3-642-32442-8
Edizione	[1st ed. 2013.]
Descrizione fisica	1 online resource (131 p.)
Collana	JIMD Reports, , 2192-8304 ; ; 7
Disciplina	616.3/9042 616.39042
Soggetti	Human genetics Metabolic diseases Pediatrics Human physiology Human Genetics Metabolic Diseases Human Physiology
Lingua di pubblicazione	Inglese
Formato	Materiale a stampa
Livello bibliografico	Monografia
Note generali	Description based upon print version of record.
Nota di bibliografia	Includes bibliographical references.
Nota di contenuto	JIMD Reports -Case and Research Reports, 2012/4; Contents; Necrotizing Enterocolitis and Respiratory Distress Syndromeas First Clinical Presentation of Mitochondrial TrifunctionalProtein Deficiency; Abstract; Introduction; Materials and Methods; Case 1; Case 2; Classification; Analytical Methods; Discussion; Synopsis; Conflict of Interest; References; Temporal Intradiploic Dilative Vasculopathy: An Additional Pathogenic Factor for the Hearing Loss in Fabry Disease?; Abstract; Introduction; Case Report; Discussion; References; Hereditary Intrinsic Factor Deficiency in Chaldeans; Abstract IntroductionPatients and Methods; Patients and Samples; Methods; Results and Discussion; Declaration; Synopsis; Authors Contributions; References to Electronic Databases; References; Mutation Analysis in

Glycogen Storage DiseaseType III Patients in the Netherlands: NovelGenotype-Phenotype Relationships and FiveNovel Mutations in the AGL Gene; Abstract; Introduction; Materials and Methods; Results; Discussion; Synopsis; Conflicts of Interest; References; Cholestatic Jaundice Associated with Carnitine Palmitoyltransferase IA Deficiency; Abstract; Introduction; Case Report; Discussion
ReferencesQuality of Life of Brazilian Patients with Gaucher Disease and Fabry Disease; Abstract; Introduction; Methods; Medical Outcomes Study: 36-Item Short Form Health Survey (SF-36); Statistical Analysis; Definition of Clinically Significant Change; Results; Gaucher Disease (n=21); Fabry Disease (n=14); Comparisons Between GD and FD Patients Receiving ERT and Other Populations; Discussion; Gaucher Disease; Fabry Disease; Conclusion; Synopsis of the Article; Authors Contributions; Guarantor for the Article; Competing Interest; Funding; Ethics Approval; Patient Consent; References
Identification and Functional Characterization of GAA Mutations in Colombian Patients Affected by Pompe DiseaseAbstract; Introduction; Material and Methods; Patients; GAA Mutational Analysis; Functional Analysis of GAA Mutations; Results; GAA Mutation Detection and In Silico Analysis; Functional Characterization of GAA Mutations; Discussion; References; Successful Live Birth following Preimplantation GeneticDiagnosis for Phenylketonuria in Day 3 Embryos by SpecificMutation Analysis and Elective Single Embryo Transfer; Abstract; Introduction; Materials and Methods; Results; Discussion
SynopsisConflict of Interest; References; The Transforming Growth Factor-Beta Signaling Pathway Involvement in Cardiovascular Lesions in Mucopolysaccharidosis-I; Abstract; Introduction; Methods; Results; Discussion; Synopsis; References to Electronic Databases; References; Recommendations for Pregnancies in Patients with Crigler-Najjar Syndrome; Introduction; Case Report; Discussion; Conclusions; Conflict of Interest Statement; References; Autism Spectrum Disorder in a Child with Propionic Acidemia; Abstract; Introduction; Case Report; Psychiatric Evaluation; Discussion; Synopsis
Competing Interests

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.