1. Record Nr. UNINA9910739419403321

Titolo	JIMD reports : case and research reports, 2012/6 / / Johannes Zschocke, K. Michael Gibson, editors-in-chief ; Garry Brown, Eva Morava, editors ; Verena Peters, managing editor
Pubbl/distr/stampa	Heidelberg [Germany] ; ; New York, : Springer, 2013
ISBN	1-283-94503-7 3-642-35518-8
Edizione	[1st ed. 2013.]
Descrizione fisica	1 online resource (140 p.)
Collana	JIMD reports, , 2192-8304 ; ; v. 9
Altri autori (Persone)	ZschockeJohannes GibsonK. Michael BrownGarry MoravaEva PetersVerena
Disciplina	600
Soggetti	Metabolism, Inborn errors of Metabolism - Disorders
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, 2012/6; Contents; Novel Mutations in the PC Gene in Patients with Type B Pyruvate Carboxylase Deficiency; Abstract; References; Novel Mutations in the Glucocerebrosidase Gene of Brazilian Patients with Gaucher Disease; Abstract; Introduction; Materials and Methods; Patients; Sample Collection and DNA Isolation; Amplification of the Entire GBA Gene; Screening of Common Mutations; PCR Amplification and Direct DNA Sequencing; Isolation of Total RNA, cDNA Synthesis, and PCR Amplification; Evaluation of Novel Mutations; Nomenclature of Mutations; Results DiscussionSynopsis; References; Prevalence and Development of Orthopaedic Symptoms in the Dutch Hurler Patient Population after Haematopoietic Stem Cell Transplantation; Abstract; Introduction; Methods; Patients and Data Collection; Odontoid Dysplasia; Thoracolumbar Spine; Hip Dysplasia; Genu Valgum; Statistical Analysis; Results; Patients; Odontoid Hypoplasia; Thoracolumbar Kyphosis;

Scoliosis; Spinal Cord Involvement; Hip Dysplasia; Genu Valgum; Correlation Between Abnormalities; Discussion; Take-Home Message; Contribution of Individual Authors; Guarantor of the Article Competing Interests StatementDetails of Funding; References; Nutritional Changes and Micronutrient Supply in Patients with Phenylketonuria Under Therapy with Tetrahydrobiopterin (BH4); Abstract; Introduction; Study Design; Subjects and Methods; Subjects; Methods; Assessment of Plasma Phe Concentrations; Assessment of Food and Nutrient Intake; Statistical Analysis; Results; Patient Characteristics; Metabolic Control and Phe Consumption; Food Consumption of BH4-Sensitive Patients; Macronutrient Intake of BH4-Sensitive Patients; Micronutrient Intake of BH4-Sensitive Patients; Discussion
Take Home MessageReferences; Effects of Switching from Agalsidase Beta to Agalsidase Alfa in 10 Patients with Anderson-Fabry Disease; Abstract; Introduction; Materials and Methods; Study Design; Patients; Treatments; Data Collection and Study End Points; cMRI Technique and Analysis; Safety; Statistical Analysis; Results; Patients; Renal Function; Cardiac Functional Parameters; Quality of Life; Pain Symptoms; Tolerability; Discussion; Conclusions; Appendix; Details of the Contributions of Individual Authors; Guarantor; Competing Interest Statement; Provide Details of Funding
Details of Ethics Approval and Patient ConsentSynopsis; References; Molecular Genetics and Genotype-Based Estimation of BH4-Responsiveness in Serbian PKU Patients: Spotlight on Phenotypic Implications of p.L48S; Abstract; Introduction; Subjects and Methods; Patients and Phenotypic Classification; Molecular Genetic Analysis; Results; Genotyping; Phenotypic Characterization; Genotype-Phenotype Correlation Study; BH4 Responsiveness; Discussion; Genotype-Phenotype Inconsistencies; BH4 Responsiveness in Serbia; Synopsis; References
Subjective and Objective Assessment of Hand Function in Mucopolysaccharidosis IVa Patients

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.