

1. Record Nr.	UNINA9910300350803321
Titolo	JIMD Reports, Volume 17 [[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, , 2014
ISBN	3-662-44578-6
Edizione	[1st ed. 2014.]
Descrizione fisica	1 online resource (95 p.)
Collana	JIMD Reports, , 2192-8304 ; ; 17
Disciplina	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	""Contents""; ""Reversible Keratopathy Due to Hypertyrosinaemia Following Intermittent Low-Dose Nitisinone in Alkaptonuria: A Case Report""; ""Abstract""; ""Introduction""; ""Case Report""; ""Discussion""; ""Compliance with Ethics Guidelines""; ""Contribution from Authors""; ""References""; ""Sports in LCHAD Deficiency: Maximal Incremental and Endurance Exercise Tests in a 13-Year-Old Patient with Long-Chain 3-Hydrox...""; ""Abstract""; ""Introduction""; ""Case Report""; ""Applied Tests (Methods)""; ""Results""; ""Standard Incremental Cycle Test""; ""Standard Endurance Cycle Test"" ""Anaerobic Threshold, Maximal Lactate Steady State (MLSS)"" ""Metabolic Laboratory Tests""; ""Discussion""; ""Compliance with Ethical Standards""; ""Conflict of Interest""; ""Informed Consent""; ""References""; ""A Hunter Patient with a Severe Phenotype Reveals Two Large Deletions and Two Duplications Extending 1.2Mb Distally to IDS Locus""; ""Abstract""; ""Introduction""; ""Materials and Methods""; ""Results""; ""Clinical Description""; ""Array CGH Analysis"";

""Discussion""; ""Conclusion""; ""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""; ""References""; ""Widening Phenotypic Spectrum of AADC Deficiency, a Disorder of Dopamine and Serotonin Synthesis""; ""Abstract""; ""Introduction""; ""Case Reports""; ""Discussion""; ""Synopsis""; ""Compliance with Ethics Guidelines""; ""References""; ""Antiepileptic Medications Increase Osteoporosis Risk in Male Fabry Patients: Bone Mineral Density in an Australian Cohort""; ""Abstract""; ""Introduction""; ""Methods""
""Study Population""""Dual X-Ray Absorptiometry""; ""Biochemistry""; ""Statistical Analysis""; ""Results""; ""Study Population""; ""Dual X-Ray Absorptiometry""; ""Biochemistry""; ""Patient Outcomes""; ""Discussion""; ""Take-Home Message""; ""Compliance with Ethics Guidelines""; ""Details of Contributions of Individual Authors""; ""Conflict of Interest""; ""Informed Consent""; ""References""; ""The Complexity of Newborn Screening Follow-Up in Phenylketonuria""; ""Abstract""; ""Introduction""; ""Clinical Report""; ""Discussion""; ""Synopsis""; ""Compliance with Ethics Guidelines""
""Conflict of Interest""""Informed Consent and Animal Rights""; ""References""; ""Revised Proposal for the Prevention of Low Bone Mass in Patients with Classic Galactosemia""; ""Abstract""; ""Introduction""; ""Diagnostic Procedures""; ""Dietary Evaluation and Bone Metabolism Markers""; ""Estrogen Supplementation""; ""Physical Activity Evaluation""; ""Assessment of Spinal Deformities""; ""Dual-Energy X-Ray Absorptiometry""; ""Follow-Up and Therapeutic Interventions""; ""Dietary Evaluation and Bone Metabolism Markers""; ""Estrogen Supplementation""; ""Physical Activity Evaluation""
""Assessment of Spinal Deformities""

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.
