

1. Record Nr.	UNINA9910300209903321
Titolo	JIMD Reports, Volume 15 [[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, , 2015
ISBN	3-662-43751-1
Edizione	[1st ed. 2015.]
Descrizione fisica	1 online resource (133 p.)
Collana	JIMD Reports, , 2192-8304 ; ; 15
Disciplina	616.39042
Soggetti	Human genetics Metabolic diseases Pediatrics Human physiology Human Genetics Metabolic Diseases Human Physiology Genètica humana Trastorns del metabolisme Pediatría Fisiologia humana Llibres electrònics
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""; ""4-Hydroxyglutamate Is a Biomarker for Primary Hyperoxaluria Type 3""; ""Abstract""; ""Introduction""; ""Materials and Methods""; ""Results""; ""Assay Performance""; ""4OHGlu Levels in Controls and PH3 Families""; ""Discussion""; ""Compliance with Ethics Guidelines""; ""Author Contributions""; ""Conflicts of Interest""; ""Ethical Approval""; ""References""; ""Excellent Response to a Ketogenic Diet in a Patient with Alternating Hemiplegia of Childhood""; ""Abstract""; ""Introduction""; ""Patient and Methods""; ""Procedure""; ""Results""; ""Discussion""; ""Synopsis"" ""Compliance with Ethics and Guidelines""""Conflict of Interest"";

""Informed Consent""; ""Animal Rights""; ""Details of the Contributions of Individual Authors""; ""References""; ""Thiamine-Responsive and Non-responsive Patients with PDHC-E1 Deficiency: A Retrospective Assessment""; ""Abstract""; ""Introduction""; ""Methods""; ""Results""; ""Discussion""; ""Compliance with Ethics Guidelines""; ""Conflict of Interest""; ""Details of the Contributions of Individual Authors""; ""References""

""Diagnostic Exome Sequencing and Tailored Bioinformatics of the Parents of a Deceased Child with Cobalamin Deficiency Suggests ..."" Abstract""; ""Introduction""; ""Subjects and Methods""; ""Results""; ""Discussion""; ""Compliance with Ethics Guidelines""; ""Synopsis""; ""Conflict of Interest""; ""Informed Consent""; ""Animal Rights""; ""Details of the Contributions of Individual Authors""; ""Electronic-Database Information""; ""References""; ""Report of Two Never Treated Adult Sisters with Aromatic I-Amino Acid Decarboxylase Deficiency: A Portrait of the Natural Histo...""; ""Abstract""

""Introduction""""Patients""; ""Patient 1 (Fig.1; II-3)""; ""Patient 2 (Fig.1; II-4)""; ""Biochemical Phenotype and Genotype""; ""Treatments""; ""Discussion""; ""Compliance with Ethics Guidelines""; ""Synopsis""; ""Conflict of Interest""; ""Ethics Approval""; ""PatientA?s Informed Consent""; ""Animal Rights""; ""AuthorsA? Contribution""; ""References""; ""Lysine-Restricted Diet as Adjunct Therapy for Pyridoxine-Dependent Epilepsy: The PDE Consortium Consensus Recommendations""; ""Abstract""; ""Introduction""; ""Methods""; ""Results""; ""Literature Search""

""Pharmacotherapy: Recommendation""""Add-on Dietary Treatment: General Recommendations and Rationale""; ""Recommendations (Fig. 2)""; ""Rationale""; ""Diet Prescriptions (See Online Supplement for More Details)""; ""Nutritional Aims""; ""Diet Management in (Breastfed) Infants""; ""Assessment of Lysine Content""; ""Lysine-Free Amino Acid Formulas""; ""Monitoring""; ""Clinical Monitoring""; ""Biochemical and Routine Laboratory Monitoring""; ""Nutritional Markers""; ""Neurological Monitoring""; ""Neurodevelopmental Monitoring""; ""Neuro-Radiological Monitoring""; ""Emergency Treatment"" ""Discussion""

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.
