1. Record Nr. UNINA9910159538303321

Autore	Bezenac Agnes de
Titolo	El Padre nuestro : padre nuestro que estas en los cielos / / Creado por Agnes y Salem de Bezenac ; Ilustrado por Agnes de Bezenac ; Coloreado por Sabine Rich
Pubbl/distr/stampa	[Paris, France] : , : [iCharacter.org], , 2012 ©2012
ISBN	1-62387-117-4
Descrizione fisica	1 online resource (32 p.)
Disciplina	226.9606
Soggetti	Lord's prayer Spiritual life - Christianity
Lingua di pubblicazione	Spagnolo
Formato	Materiale a stampa
Livello bibliografico	Monografia
Note generali	Description based upon print version of record.
Sommario/riassunto	The ""Lord's Prayer"" is offered alongside an easy-to-understand application for children. Also includes 10 written prayers.

2. Record Nr. UNINA9910300078203321

Titolo	JIMD Reports - Volume 12 / / edited by Johannes Zschocke, K Michael Gibson, Garry Brown, Eva Morava, Verena Peters
Pubbl/distr/stampa	Cham : , : Springer International Publishing : , : Imprint : Springer, , 2014
ISBN	3-319-03461-8
Edizione	[1st ed. 2014.]
Descrizione fisica	1 online resource (130 p.)
Collana	JIMD Reports, , 2192-8304 ; ; 12
Disciplina	616.39042
Soggetti	Human genetics Metabolism - Disorders Pediatrics Human physiology Human Genetics Metabolic Diseases Human Physiology Genètica humana Fisiologia humana Trastorns del metabolisme Pediatria 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 at the end of each chapters.
Nota di contenuto	""Contents""; ""Propionic Acidemia and Optic Neuropathy: A Report of Two Cases""; ""Abstract""; ""Introduction""; ""Case Reports""; ""Discussion""; ""Synopsis""; ""References""; ""Chronic Kidney Disease in an Adult with Propionic Acidemia""; ""Abstract""; ""Introduction""; ""Methods and Results""; ""Clinical Summary""; ""Renal Evaluation""; ""Summary and Discussion""; ""Synopsis""; ""References""; ""Transient Massive Trimethylaminuria Associated with Food Protein-Induced Enterocolitis Syndrome""; ""Abstract""; ""Introduction""; ""Case Presentation""; ""Discussion""; ""Synopsis"" ""References""""""Increased Prevalence of Hypertension in Young Adults

with High Heteroplasmy Levels of the MELAS m.3243A>G Mutation""; ""Abstract""; ""Introduction""; ""Methods""; ""Study Design and Population""; ""Statistical Analysis""; ""Results""; ""Discussion""; ""Study Limitations""; ""Conclusion""; ""Contribution""; ""Disclosures""; ""References""; ""Niemann-Pick Disease Type C: New Aspects in a Long Published Family - Partial Manifestations in Heterozygotes""; ""Abstract""; ""Introduction""; ""Case Report""; ""Genetic Testing""; ""Results and Discussion""; ""Synopsis""
""Compliance with Ethics Guidelines""""References""; ""Chiari 1 Malformation and Holocord Syringomyelia in Hunter Syndrome""; ""Abstract""; ""Introduction""; ""Clinical Description""; ""Discussion""; ""Take-Home Messages""; ""References""; ""A Patient with Complex I Deficiency Caused by a Novel ACAD9 Mutation Not Responding to Riboflavin Treatment""; ""Abstract""; ""Introduction""; ""Materials and Methods""; ""Clinical Data""; ""Enzyme Measurements""; ""SNP Array Analysis and Homozygosity Mapping""; ""Mutation Analysis""; ""Blue Native, SDS-PAGE, and In-Gel Activity Assays""
""Antibodies and ECL Detection""""Lentiviral Complementation of Patient Fibroblasts""; ""Riboflavin Treatment of Patient Cells""; ""Modeling""; ""Results""; ""Enzyme Measurements and Gel Analyses""; ""Gene and Conservation Analyses""; ""Modeling of the p.Ala220Val Mutation""; ""Functional Complementation""; ""Riboflavin Treatment of Fibroblasts""; ""Discussion""; ""Take-Home Message""; ""References""; ""Pulmonary Manifestations in a Patient with Transaldolase Deficiency""; ""Abstract""; ""Introduction""; ""Case Report""; ""Discussion""; ""Conclusion""; ""Conflict of Interest""
""AuthorsA? Contributions""""Take-Home Message""; ""References""; ""Burden of Lysosomal Storage Disorders in India: Experience of 387 Affected Children from a Single Diagnostic Facility""; ""Abstract""; ""Introduction""; ""Material and Methods""; ""Results""; ""Discussion""; ""Synopsis""; ""Conflict of Interest""; ""Informed Consent""; ""Details of the Contributions of Individual Authors""; ""References""; ""A Japanese Adult Case of Guanidinoacetate Methyltransferase Deficiency""; ""Abstract""; ""Introduction""; ""Case Report""; ""Discussion""; ""Take-Home Message""
""Compliance with Ethics Guidelines""

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.