1. Record Nr. UNINA9910300343303321 JIMD Reports - Case and Research Reports, Volume 13 [[electronic **Titolo** resource] /] / edited by Johannes Zschocke, K. Michael Gibson, Garry Brown, Eva Morava, Verena Peters Berlin, Heidelberg:,: Springer Berlin Heidelberg:,: Imprint: Springer, Pubbl/distr/stampa **ISBN** 3-642-54149-6 Edizione [1st ed. 2014.] 1 online resource (164 p.) Descrizione fisica JIMD Reports, , 2192-8304; ; 13 Collana 616.3/9042 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. Includes bibliographical references. Nota di bibliografia Nota di contenuto ""Contents""; ""Newborn Screening for Glutaric Aciduria-II: The New England Experience""; ""Abstract""; ""Introduction""; ""Methods and Population""; ""Results""; ""Discussion""; ""Synopsis""; ""Conflict of Interest""; ""Informed Consent/Human or Animal Studies""; ""Authors Attestations""; ""References""; ""Systematic Data Collection to Inform Policy Decisions: Integration of the Region 4 Stork (R4S) Collaborative Newborn Screening...""; ""Abstract""; ""Introduction""; ""Washington Newborn Screening""; ""Analyte Selection and Review""; ""Cutoff Determinations""; ""Post-Analytical Tools"" ""Limitations""""Conclusion""; ""Synopsis""; ""Compliance with Ethics Guidelines""; ""Conflict of Interest""; ""Animal Rights and Human Subjects""; ""Details of the Contributions of Individual Authors"": ""References""; ""Application of a Second-Tier Newborn Screening Assay for C5 Isoforms""; ""Abstract""; ""Introduction""; ""Case Report"";

""Materials and Methods""; ""Results""; ""Discussion""; ""Compliance with Ethics Guidelines""; ""Details of the Contributions of Individual Authors""; ""References""; ""Cystinosis with Sclerotic Bone Lesions""; ""Abstract""; ""Case Report""; ""Discussion"" ""Conclusion""""Conflict of Interest""; ""Informed Consent""; ""Contributions of the Individual Authors""; ""References""; ""Pregnancy and Lactation Outcomes in a Turkish Patient with Lysinuric Protein Intolerance""; ""Abstract""; ""Introduction""; ""Case Report""; ""Discussion""; ""Take-Home Message""; ""Compliance with Ethical Guidelines""; ""References""; ""Adult-Onset Fatal Neurohepatopathy in a Woman Caused by MPV17 Mutation""; ""Abstract""; ""Case Report""; ""Discussion""; ""Summary""; ""Compliance with Ethics Guidelines""; ""References"" ""Multiple Acyl-CoA Dehydrogenation Deficiency (Glutaric Aciduria Type II) with a Novel Mutation of Electron Transfer Flavoprote..."""Abstract"": ""Introduction""; ""Materials and Methods""; ""Subject""; ""Diet Loading Test""; ""GC-MS Analysis""; ""LC-MS/MS Analysis""; ""Cell Culture""; ""RT-PCR""; ""PCR-Restriction Fragment Length Polymorphism (PCR-RFLP)""; ""Results""; ""Feline MADD""; ""Sequencing of Feline ETFI±, ETFbeta, and ETFDH""; ""Presence of a Novel Mutation in the Cat with MADD""; ""Discussion""; ""Synopsis""; ""Compliance with Ethics Guidelines""; ""References"" ""Biotin-Responsive Basal Ganglia Disease: A Treatable Differential Diagnosis of Leigh Syndrome"""Abstract""; ""Introduction""; ""Case Report""; ""Discussion""; ""Synopsis""; ""Compliance with Ethics Guidelines""; ""Conflict of Interest""; ""Informed Consent""; ""Animal Rights""; ""References""; ""Severe Hypertriglyceridemia in a Newborn with Monogenic Lipoprotein Lipase Deficiency: An Unconventional

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.

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""Analysis of LPL Gene""; ""Results""
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