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| Soggetti                | Human genetics<br>Metabolism - Disorders<br>Pediatrics<br>Human physiology<br>Human Genetics<br>Metabolic Diseases<br>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; Leucine Loading Test is Only Discriminative for 3-Methylglutaconic Aciduria Due to AUH Defect; Abstract; Introduction; Material and Methods; Patients; Leucine Loading Tests; Urinary Organic Acid Analysis; Leucine Loading Tests in Patients Described in the Literature; Results; Discussion; Disclosures; Compliance with Ethics Guidelines; Conflict of Interest; Informed Consent; Details of the Contributions of Individual Authors; References; Fabry Disease: Multidisciplinary Evaluation After 10 Years of Treatment with Agalsidase Beta; Abstract; Introduction; Objective Materials and MethodsStatistical Analysis; Results; Discussion; |

Conclusions; Take-Home Message; Compliance with Ethics Guidelines; Conflict of Interest; Informed Consent; Details of the Contributions of Individual Authors; References; Chondroitin 6-Sulfate as a Novel Biomarker for Mucopolysaccharidosis IVA and VII; Abstract; Introduction; Materials and Method; Subjects; Chemicals and Materials; Sample Preparation; Apparatus; Method Validation; Statistical Analysis; Results; LC-MS/MS Conditions; Calibration Curves; Precision and Accuracy; Chondroitin 6-Sulfate (C6S) Levels  
Keratan Sulfate (KS) LevelCorrelation Between KS and C6S Levels; Discussion; Compliance with Ethics Guidelines; Conflict of Interest; Informed Consent; Animal Rights; Contributions to the Project; References; Carnitine Profile and Effect of Suppletion in Children with Renal Fanconi Syndrome due to Cystinosis; Abstract; Background; Patients and Methods; Results; Discussion; Synopsis; Compliance with Ethics Guidelines; Conflict of Interest; Informed Consent; Details of the Contributions of Individual Authors; References  
Cognitive and Antipsychotic Medication Use in Monoallelic GBA-Related Parkinson DiseaseAbstract; Introduction; Methods; Data Collection; Statistical Analysis; Results; Discussion; Sources of Support; Synopsis Sentence; Compliance with Ethics Guidelines; Conflict of Interest; Other Disclosures; Informed Consent; Animal Rights; Individual Author Contributions; Guarantor; References; Laronidase Replacement Therapy and Left Ventricular Function in Mucopolysaccharidosis I; Abstract; Introduction; Case Report; Methods; Conventional, Pulse Doppler, and Speckle Tracking Echocardiograms  
Statistical AnalysisResults; Clinical Findings After LRT; Echocardiographic Findings After LRT; Relation of LV Weight to Urinary Uronic Acid Concentration, Diastolic Function, and Contraction Synchrony; Discussion; Limitations; Funding; Disclosure; Synopsis; Compliance with Ethics Guidelines; Conflict of Interest; Informed Consent; Animal Rights; Author Contributions; Competing Interests; Funding; References; Fibrolamellar Hepatocellular Carcinoma Mimicking Ornithine Transcarbamylase Deficiency; Abstract; Introduction; Case Report; Discussion; Synopsis  
Details of the Contributions of Individual Authors

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#### 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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