Déborah Mathis

List of Publications by Year in descending order

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| # | Article | IF | CITATIONS |
|----|---|---|-----------|
| 1 | Decrease of diseaseâ€related metabolites upon fasting in a hemizygous knockâ€in mouse model (<scp><i>Mut</i></scp> â€ko/ki) of methylmalonic aciduria. JIMD Reports, 2021, 58, 44-51. | 1.5 | 2 |
| 2 | In-depth phenotyping reveals common and novel disease symptoms in a hemizygous knock-in mouse model (Mut-ko/ki) of mut-type methylmalonic aciduria. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2020, 1866, 165622. | 3.8 | 12 |
| 3 | Condensation of deltaâ€lâ€piperideineâ€6â€carboxylate with orthoâ€aminobenzaldehyde allows its simple, fast, and inexpensive quantification in the urine of patients with antiquitin deficiency. Journal of Inherited Metabolic Disease, 2020, 43, 891-900. | 3.6 | 6 |
| 4 | LCâ€MS / MS method for the differential diagnosis of treatable early onset inherited metabolic epilepsies. Journal of Inherited Metabolic Disease, 2020, 43, 1102-1111. | 3.6 | 8 |
| 5 | Validation of a Food Frequency Questionnaire to Assess Intake of n-3 Polyunsaturated Fatty Acids in Switzerland. Nutrients, 2019, 11, 1863. | 4.1 | 18 |
| 6 | Protein profile of dairy products: Simultaneous quantification of twenty bovine milk proteins. International Dairy Journal, 2019, 97, 167-175. | 3.0 | 30 |
| 7 | New insights into human lysine degradation pathways with relevance to pyridoxineâ€dependent epilepsy due to antiquitin deficiency. Journal of Inherited Metabolic Disease, 2019, 42, 620-628. | 3.6 | 45 |
| 8 | Antiquitin Deficiency with Adolescent Onset Epilepsy: Molecular Diagnosis in a Mother of Affected Offsprings. Neuropediatrics, 2018, 49, 154-157. | 0.6 | 15 |
| 9 | Confirmation of mutations in <i>PROSC</i> as a novel cause of vitamin B _₆ -dependent epilepsy. Journal of Medical Genetics, 2017, 54, 809-814. | 3.2 | 66 |
| 10 | A simple dried blood spot-method for in vivo measurement of ureagenesis by gas chromatography–mass spectrometry using stable isotopes. Clinica Chimica Acta, 2017, 464, 236-243. | 1.1 | 17 |
| 11 | Early biomarker response and patient preferences to oral and intramuscular vitamin B12 substitution in primary care: a randomised parallel-group trial. Swiss Medical Weekly, 2017, 147, w14421. | 1.6 | 10 |
| 12 | Novel Mouse Models of Methylmalonic Aciduria Recapitulate Phenotypic Traits with a Genetic Dosage Effect. Journal of Biological Chemistry, 2016, 291, 20563-20573. | 3.4 | 35 |
| 13 | The value of plasma vitamin B ₆ profiles in early onset epileptic encephalopathies. Journal of Inherited Metabolic Disease, 2016, 39, 733-741. | 3.6 | 19 |
| 14 | N ⁸ â€acetylspermidine as a potential plasma biomarker for Snyderâ€Robinson syndrome identified by clinical metabolomics. Journal of Inherited Metabolic Disease, 2016, 39, 131-137. | 3.6 | 33 |
| 15 | LC-MS/MS based assay and reference intervals in children and adolescents for oxysterols elevated in Niemann–Pick diseases. Clinical Biochemistry, 2015, 48, 596-602. | 1.9 | 50 |
| 16 | Controlled Ethylene Polymerization Catalyzed by Cp\$m{{_{2}^{ast}}}\$ZrBu ₂ /[Ph ₃ C] [B(C ₆ F ₅) _{4above Room Temperature. Helvetica Chimica Acta, 2010, 93, 212-219.} | s ube]/ <sup< td=""><td>ɔ≉iBu</td></sup<> | ɔ≉iBu |
| 17 | Narrowly Distributed Polyethylene via Reversible Chain Transfer to Aluminum by a Sterically Hindered Zirconocene/MAO. Organometallics, 2010, 29, 294-302. | 2.3 | 38 |

18Artificial Metalloenzyme for Enantioselective Sulfoxidation Based on Vanadyl-Loaded Streptavidin.
Journal of the American Chemical Society, 2008, 130, 8085-8088.13.7145