Stefan KÃ¶lker

List of Publications by Year in descending order

194 papers	8,366 citations	41323 49 h-index	62565 80 g-index
207 all docs	207 docs citations	207 times ranked	9592 citing authors

#	Article	IF	Citations
1	Diagnosis and management of glutaric aciduria type I â€“ revised recommendations. Journal of Inherited Metabolic Disease, 2011, 34, 677-694.	1.7	327
2	The incidence of urea cycle disorders. Molecular Genetics and Metabolism, 2013, 110, 179-180.	0.5	232
3	Recessive mutations in EPG5 cause Vici syndrome, a multisystem disorder with defective autophagy. Nature Genetics, 2013, 45, 83-87.	9.4	231
4	Natural History, Outcome, and Treatment Efficacy in Children and Adults with Glutaryl-CoA Dehydrogenase Deficiency. Pediatric Research, 2006, 59, 840-847.	1.1	224
5	Long-Term Outcome in Methylmalonic Acidurias Is Influenced by the Underlying Defect (mut0, mutâ˜,) Tj ETQq1 1 0.784314 rgBT /Over	1.1	210
6	The phenotypic spectrum of organic acidurias and urea cycle disorders. Part 1: the initial presentation. Journal of Inherited Metabolic Disease, 2015, 38, 1041-1057.	1.7	186
7	The phenotypic spectrum of organic acidurias and urea cycle disorders. Part 2: the evolving clinical phenotype. Journal of Inherited Metabolic Disease, 2015, 38, 1059-1074.	1.7	175
8	Proposed recommendations for diagnosing and managing individuals with glutaric aciduria type I: second revision. Journal of Inherited Metabolic Disease, 2017, 40, 75-101.	1.7	173
9	Secondary mitochondrial dysfunction in propionic aciduria: a pathogenic role for endogenous mitochondrial toxins. Biochemical Journal, 2006, 398, 107-112.	1.7	163
10	Dynamic changes of striatal and extrastriatal abnormalities in glutaric aciduria type I. Brain, 2009, 132, 1764-1782.	3.7	160
11	Neurodegeneration in Methylmalonic Aciduria Involves Inhibition of Complex II and the Tricarboxylic Acid Cycle, and Synergistically Acting Excitotoxicity. Journal of Biological Chemistry, 2002, 277, 14674-14680.	1.6	153
12	Intracerebral accumulation of glutaric and 3-hydroxyglutaric acids secondary to limited flux across the blood-brain barrier constitute a biochemical risk factor for neurodegeneration in glutaryl-CoA dehydrogenase deficiency. Journal of Neurochemistry, 2006, 97, 899-910.	2.1	147
13	Use of guidelines improves the neurological outcome in glutaric aciduria type I. Annals of Neurology, 2010, 68, 743-752.	2.8	147
14	An international classification of inherited metabolic disorders (<scp>ICIMD</scp>). Journal of Inherited Metabolic Disease, 2021, 44, 164-177.	1.7	146
15	Neuropathological, biochemical and molecular findings in a glutaric acidemia type 1 cohort. Brain, 2005, 128, 711-722.	3.7	137
16	Impaired Mitochondrial Dynamics and Mitophagy in Neuronal Models of Tuberous Sclerosis Complex. Cell Reports, 2016, 17, 1053-1070.	2.9	125
17	NMDA receptor activation and respiratory chain complex V inhibition contribute to neurodegeneration ind-2-hydroxyglutaric aciduria. European Journal of Neuroscience, 2002, 16, 21-28.	1.2	115
18	Bioenergetics in Glutaryl-Coenzyme A Dehydrogenase Deficiency. Journal of Biological Chemistry, 2005, 280, 21830-21836.	1.6	111

#	Article	IF	Citations
19	Iron Regulatory Proteins Secure Mitochondrial Iron Sufficiency and Function. Cell Metabolism, 2010, 12, 194-201.	7.2	110
20	Biallelic Mutations in NBAS Cause Recurrent Acute Liver Failure with Onset in Infancy. American Journal of Human Genetics, 2015, 97, 163-169.	2.6	110
21	Pathomechanisms of neurodegeneration in glutaryl-CoA dehydrogenase deficiency. Annals of Neurology, 2004, 55, 7-12.	2.8	104
22	Decline of Acute Encephalopathic Crises in Children with Glutaryl-CoA Dehydrogenase Deficiency Identified by Newborn Screening in Germany. Pediatric Research, 2007, 62, 357-363.	1.1	102
23	Therapeutic modulation of cerebral l-lysine metabolism in a mouse model for glutaric aciduria type I. Brain, 2011, 134, 157-170.	3.7	102
24	EPG5-related Vici syndrome: a paradigm of neurodevelopmental disorders with defective autophagy. Brain, 2016, 139, 765-781.	3.7	99
25	DHTKD1 Mutations Cause 2-Aminoadipic and 2-Oxoadipic Aciduria. American Journal of Human Genetics, 2012, 91, 1082-1087.	2.6	94
26	Recurrent acute liver failure due to NBAS deficiency: phenotypic spectrum, disease mechanisms, and therapeutic concepts. Journal of Inherited Metabolic Disease, 2016, 39, 3-16.	1.7	92
27	Deficiency of <scp>ECHS</scp>1 causes mitochondrial encephalopathy with cardiac involvement. Annals of Clinical and Translational Neurology, 2015, 2, 492-509.	1.7	90
28	Safety and efficacy of mTOR inhibitor treatment in patients with tuberous sclerosis complex under 2â€‰years of age â€“ a multicenter retrospective study. Orphanet Journal of Rare Diseases, 2019, 14, 96.	1.2	90
29	A nonâ€enzymatic function of 17Î²â€hydroxysteroid dehydrogenase type 10 is required for mitochondrial integrity and cell survival. EMBO Molecular Medicine, 2010, 2, 51-62.	3.3	89
30	Deorphanization of GPR109B as a Receptor for the Î²-Oxidation Intermediate 3-OH-octanoic Acid and Its Role in the Regulation of Lipolysis. Journal of Biological Chemistry, 2009, 284, 21928-21933.	1.6	78
31	Methylmalonic Acid, a Biochemical Hallmark of Methylmalonic Acidurias but No Inhibitor of Mitochondrial Respiratory Chain. Journal of Biological Chemistry, 2003, 278, 47388-47393.	1.6	77
32	Mutations in PCYT2 disrupt etherlipid biosynthesis and cause a complex hereditary spastic paraplegia. Brain, 2019, 142, 3382-3397.	3.7	76
33	Contribution of Reactive Oxygen Species to 3-Hydroxyglutarate Neurotoxicity in Primary Neuronal Cultures from Chick Embryo Telencephalons. Pediatric Research, 2001, 50, 76-82.	1.1	74
34	Biallelic IARS Mutations Cause Growth Retardation with Prenatal Onset, Intellectual Disability, Muscular Hypotonia, and Infantile Hepatopathy. American Journal of Human Genetics, 2016, 99, 414-422.	2.6	73
35	Newborn screening: A diseaseâ€changing intervention for glutaric aciduria type 1. Annals of Neurology, 2018, 83, 970-979.	2.8	65
36	Impaired mitophagy links mitochondrial disease to epithelial stress in methylmalonyl-CoA mutase deficiency. Nature Communications, 2020, 11, 970.	5.8	65

#	Article	IF	Citations
37	Glutaric aciduria type I and methylmalonic aciduria: Simulation of cerebral import and export of accumulating neurotoxic dicarboxylic acids in in vitro models of the bloodâ€“brain barrier and the choroid plexus. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2010, 1802, 552-560.	1.8	64
38	Crossâ€sectional observational study of 208 patients with nonâ€classical urea cycle disorders. Journal of Inherited Metabolic Disease, 2014, 37, 21-30.	1.7	62
39	Impact of age at onset and newborn screening on outcome in organic acidurias. Journal of Inherited Metabolic Disease, 2016, 39, 341-353.	1.7	60
40	Phenylalanine Reduces Synaptic Density in Mixed Cortical Cultures from Mice. Pediatric Research, 2006, 59, 544-548.	1.1	59
41	Ca2+ and Na+ Dependence of 3-Hydroxyglutarate-Induced Excitotoxicity in Primary Neuronal Cultures from Chick Embryo Telencephalons. Pediatric Research, 2002, 52, 199-206.	1.1	56
42	¹Hâ€MRS in glutaric aciduria type 1: impact of biochemical phenotype and age on the cerebral accumulation of neurotoxic metabolites. Journal of Inherited Metabolic Disease, 2015, 38, 829-838.	1.7	56
43	Adenosine kinase deficiency: expanding the clinical spectrum and evaluating therapeutic options. Journal of Inherited Metabolic Disease, 2016, 39, 273-283.	1.7	55
44	Biallelic Mutations in LIPT2 Cause a Mitochondrial Lipoylation Defect Associated with Severe Neonatal Encephalopathy. American Journal of Human Genetics, 2017, 101, 283-290.	2.6	55
45	Maturation-Dependent Neurotoxicity of 3-Hydroxyglutaric and Glutaric Acids In Vitro : A New Pathophysiologic Approach to Glutaryl-CoA Dehydrogenase Deficiency. Pediatric Research, 2000, 47, 495-503.	1.1	54
46	Potentiation of 3-hydroxyglutarate neurotoxicity following induction of astrocytic iNOS in neonatal rat hippocampal cultures. European Journal of Neuroscience, 2001, 13, 2115-2122.	1.2	53
47	Chronic treatment with glutaric acid induces partial tolerance to excitotoxicity in neuronal cultures from chick embryo telencephalons. Journal of Neuroscience Research, 2002, 68, 424-431.	1.3	53
48	Current concepts in organic acidurias: understanding intraâ€and extracerebral disease manifestation. Journal of Inherited Metabolic Disease, 2013, 36, 635-644.	1.7	52
49	Age at disease onset and peak ammonium level rather than interventional variables predict the neurological outcome in urea cycle disorders. Journal of Inherited Metabolic Disease, 2016, 39, 661-672.	1.7	52
50	Neonatal mortality and outcome at the end of the first year of life in early onset urea cycle disordersâ€”review and metaâ€analysis of observational studies published over more than 35 years. Journal of Inherited Metabolic Disease, 2016, 39, 219-229.	1.7	50
51	Impact of clinical exomes in neurodevelopmental and neurometabolic disorders. Molecular Genetics and Metabolism, 2017, 121, 297-307.	0.5	50
52	SCYL1 variants cause a syndrome with lowÎ³-glutamyl-transferase cholestasis, acute liver failure, and neurodegeneration (CALFAN). Genetics in Medicine, 2018, 20, 1255-1265.	1.1	50
53	Impact of short- and medium-chain organic acids, acylcarnitines, and acyl-CoAs onmitochondrial energy metabolism. Biochimica Et Biophysica Acta - Bioenergetics, 2008, 1777, 1276-1282.	0.5	47
54	Linking mitochondrial dysfunction to neurodegeneration in lysosomal storage diseases. Journal of Inherited Metabolic Disease, 2017, 40, 631-640.	1.7	46

#	Article	IF	Citations
55	Defining clinical subgroups and genotypeâ€“phenotype correlations in NBAS-associated disease across 110 patients. Genetics in Medicine, 2020, 22, 610-621.	1.1	46
56	Methylmalonic acid â€” an endogenous toxin?. Cellular and Molecular Life Sciences, 2005, 62, 621-624.	2.4	45
57	Complementary dietary treatment using lysine-free, arginine-fortified amino acid supplements in glutaric aciduria type I â€” A decade of experience. Molecular Genetics and Metabolism, 2012, 107, 72-80.	0.5	45
58	Extrastriatal changes in patients with late-onset glutaric aciduria type I highlight the risk of long-term neurotoxicity. Orphanet Journal of Rare Diseases, 2017, 12, 77.	1.2	45
59	Optimized Spectrophotometric Assay for the Completely Activated Pyruvate Dehydrogenase Complex in Fibroblasts. Clinical Chemistry, 2005, 51, 151-160.	1.5	44
60	Incidence, disease onset and short-term outcome in urea cycle disorders â€“cross-border surveillance in Germany, Austria and Switzerland. Orphanet Journal of Rare Diseases, 2017, 12, 111.	1.2	43
61	Simultaneous determination of 3-hydroxypropionic acid, methylmalonic acid and methylcitric acid in dried blood spots: Second-tier LC-MS/MS assay for newborn screening of propionic acidemia, methylmalonic acidemias and combined remethylation disorders. PLoS ONE, 2017, 12, e0184897.	1.1	43
62	Incidence and Short-Term Outcome of Children With Symptomatic Presentation of Organic Acid and Fatty Acid Oxidation Disorders in Germany. Pediatrics, 2002, 110, 1204-1211.	1.0	42
63	Emerging role of autophagy in pediatric neurodegenerative and neurometabolic diseases. Pediatric Research, 2014, 75, 217-226.	1.1	42
64	Quantitative clinical characteristics of 53 patients with MPS VII: a cross-sectional analysis. Genetics in Medicine, 2017, 19, 983-988.	1.1	42
65	Impact of Diagnosis and Therapy on Cognitive Function in Urea Cycle Disorders. Annals of Neurology, 2019, 86, 116-128.	2.8	42
66	Intrastriatal administration of 3-hydroxyglutaric acid induces convulsions and striatal lesions in rats. Brain Research, 2001, 916, 70-75.	1.1	41
67	Pathogenesis of CNS involvement in disorders of amino and organic acid metabolism. Journal of Inherited Metabolic Disease, 2008, 31, 194-204.	1.7	40
68	3-Ureidopropionate contributes to the neuropathology of 3-ureidopropionase deficiency and severe propionic aciduria: A hypothesis. Journal of Neuroscience Research, 2001, 66, 666-673.	1.3	39
69	Elevated glutaric acid levels in Dhtkd1-/Gcdh- double knockout mice challenge our current understanding of lysine metabolism. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2017, 1863, 2220-2228.	1.8	39
70	Cerebrospinal fluid biogenic amines depletion and brain atrophy in adult patients with phenylketonuria. Journal of Inherited Metabolic Disease, 2019, 42, 398-406.	1.7	38
71	Newborn screening for homocystinurias: Recent recommendations versus current practice. Journal of Inherited Metabolic Disease, 2019, 42, 128-139.	1.7	37
72	Long-term Outcomes of Individuals With Metabolic Diseases Identified Through Newborn Screening. Pediatrics, 2020, 146, .	1.0	37

#	Article	IF	Citations
73	Metabolic decompensation in methylmalonic aciduria: which biochemical parameters are discriminative?. Journal of Inherited Metabolic Disease, 2012, 35, 797-806.	1.7	35
74	Novel Mouse Models of Methylmalonic Aciduria Recapitulate Phenotypic Traits with a Genetic Dosage Effect. Journal of Biological Chemistry, 2016, 291, 20563-20573.	1.6	35
75	Transatlantic combined and comparative data analysis of 1095 patients with urea cycle disordersâ€”A successful strategy for clinical research of rare diseases. Journal of Inherited Metabolic Disease, 2019, 42, 93-106.	1.7	35
76	Increase in glutamate-induced neurotoxicity by activated astrocytes involves stimulation of protein kinase C. Journal of Neurochemistry, 2002, 82, 504-515.	2.1	34
77	Patterns, evolution, and severity of striatal injury in insidiousâ€vs acuteâ€onset glutaric aciduria type 1. Journal of Inherited Metabolic Disease, 2019, 42, 117-127.	1.7	34
78	Liver and/or kidney transplantation in amino and organic acidâ€related inborn errors of metabolism: An overview on European data. Journal of Inherited Metabolic Disease, 2021, 44, 593-605.	1.7	34
79	Clinical characteristics of 248 patients with Krabbe disease: quantitative natural history modeling based on published cases. Genetics in Medicine, 2019, 21, 2208-2215.	1.1	33
80	Diagnosis of glutaric aciduria type 1 by measuring 3â€hydroxyglutaric acid in dried urine spots by liquid chromatography tandem mass spectrometry. Journal of Inherited Metabolic Disease, 2011, 34, 173-180.	1.7	32
81	Understanding cerebral Lâ€lysine metabolism: the role of Lâ€pipecolate metabolism in Gcdhâ€deficient mice as a model for glutaric aciduria type I. Journal of Inherited Metabolic Disease, 2015, 38, 265-272.	1.7	31
82	Newborn screening by tandem mass spectrometry for glutaric aciduria type 1: a cost-effectiveness analysis. Orphanet Journal of Rare Diseases, 2013, 8, 167.	1.2	30
83	Unravelling the complex MRI pattern in glutaric aciduria type I using statistical modelsâ€”a cohort study in 180 patients. Journal of Inherited Metabolic Disease, 2014, 37, 763-773.	1.7	30
84	Novel challenges in spinal muscular atrophy â€“ How to screen and whom to treat?. Annals of Clinical and Translational Neurology, 2019, 6, 197-205.	1.7	30
85	Evaluation of dietary treatment and amino acid supplementation in organic acidurias and ureaâ€cycle disorders: On the basis of information from a European multicenter registry. Journal of Inherited Metabolic Disease, 2019, 42, 1162-1175.	1.7	30
86	Impact of newborn screening and quality of therapy on the neurological outcome in glutaric aciduria type 1: a meta-analysis. Genetics in Medicine, 2021, 23, 13-21.	1.1	30
87	Transport and distribution of 3-hydroxyglutaric acid before and during induced encephalopathic crises in a mouse model of glutaric aciduria type 1. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2008, 1782, 385-390.	1.8	29
88	Low lysine diet in glutaric aciduria type I â€“ effect on anthropometric and biochemical followâ€up parameters. Journal of Inherited Metabolic Disease, 2013, 36, 525-533.	1.7	29
89	Behavioural and emotional problems, intellectual impairment and healthâ€related quality of life in patients with organic acidurias and urea cycle disorders. Journal of Inherited Metabolic Disease, 2016, 39, 231-241.	1.7	29
90	Defining the clinical, molecular and imaging spectrum of adaptor protein complex 4-associated hereditary spastic paraplegia. Brain, 2020, 143, 2929-2944.	3.7	29

#	Article	IF	Citations
91	Organic acidurias: Major gaps, new challenges, and a yet unfulfilled promise. Journal of Inherited Metabolic Disease, 2021, 44, 9-21.	1.7	28
92	Rare Disease Registries Are Key to Evidence-Based Personalized Medicine: Highlighting the European Experience. Frontiers in Endocrinology, 2022, 13, 832063.	1.5	28
93	Organic acidurias in adults: late complications and management. Journal of Inherited Metabolic Disease, 2018, 41, 765-776.	1.7	27
94	Usefulness of biochemical parameters in decisionâ€making on the start of emergency treatment in patients with propionic acidemia. Journal of Inherited Metabolic Disease, 2014, 37, 31-37.	1.7	26
95	Networking Across Borders for Individuals with Organic Acidurias and Urea Cycle Disorders: The E-IMD Consortium. JIMD Reports, 2015, 22, 29-38.	0.7	26
96	Issues with European guidelines for phenylketonuria. Lancet Diabetes and Endocrinology,the, 2017, 5, 681-683.	5.5	26
97	Metabolism of amino acid neurotransmitters: the synaptic disorder underlying inherited metabolic diseases. Journal of Inherited Metabolic Disease, 2018, 41, 1055-1063.	1.7	26
98	Ultraâ€orphan lysosomal storage diseases: A crossâ€sectional quantitative analysis of the natural history of alphaâ€mannosidosis. Journal of Inherited Metabolic Disease, 2019, 42, 975-983.	1.7	26
99	Early prediction of phenotypic severity in Citrullinemia Type 1. Annals of Clinical and Translational Neurology, 2019, 6, 1858-1871.	1.7	26
100	Targeted Metabolic Profiling of Methionine Cycle Metabolites and Redox Thiol Pools in Mammalian Plasma, Cells and Urine. Metabolites, 2019, 9, 235.	1.3	26
101	Decreased plasma l-arginine levels in organic acidurias (MMA and PA) and decreased plasma branched-chain amino acid levels in urea cycle disorders as a potential cause of growth retardation: Options for treatment. Molecular Genetics and Metabolism, 2019, 126, 397-405.	0.5	26
102	High throughput newborn screening for aromatic ÊŸâ€aminoâ€acid decarboxylase deficiency by analysis of concentrations of 3â€Oâ€methyldopa from dried blood spots. Journal of Inherited Metabolic Disease, 2020, 43, 602-610.	1.7	26
103	Acute encephalopathy despite early therapy in a patient with homozygosity for E365K in the glutarylâ€“coenzyme A dehydrogenase gene. Journal of Pediatrics, 2001, 138, 277-279.	0.9	25
104	High urgency liver transplantation in ornithine transcarbamylase deficiency presenting with acute liver failure. Pediatric Transplantation, 2011, 15, E110-5.	0.5	25
105	Delineating the clinical spectrum of isolated methylmalonic acidurias: <scp>cblA</scp> and mut. Journal of Inherited Metabolic Disease, 2021, 44, 193-214.	1.7	25
106	A cross-sectional quantitative analysis of the natural history of Farber disease: an ultra-orphan condition with rheumatologic and neurological cardinal disease features. Genetics in Medicine, 2018, 20, 524-530.	1.1	24
107	Lysine intake and neurotoxicity in glutaric aciduria type I: towards a rationale for therapy?. Brain, 2006, 129, e54-e54.	3.7	23
108	Healthâ€related quality of life in paediatric patients with intoxicationâ€type inborn errors of metabolism: Analysis of an international data set. Journal of Inherited Metabolic Disease, 2021, 44, 215-225.	1.7	22

#	Article	IF	Citations
109	Defects in amino acid catabolism and the urea cycle. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2013, 113, 1755-1773.	1.0	21
110	Multifactorial modulation of susceptibility to l-lysine in an animal model of glutaric aciduria type I. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2015, 1852, 768-777.	1.8	21
111	Severe Acute Subdural Hemorrhage in a Patient With Glutaric Aciduria Type I After Minor Head Trauma. Journal of Child Neurology, 2015, 30, 1065-1069.	0.7	21
112	Quantitative natural history characterization in a cohort of 142 published cases of patients with galactosialidosisâ€"A crossâ€sectional study. Journal of Inherited Metabolic Disease, 2019, 42, 295-302.	1.7	21
113	Cystathionine Î²â€synthase deficiency in the <scp>Eâ€HOD registryâ€part</scp> I: pyridoxine responsiveness as a determinant of biochemical and clinical phenotype at diagnosis. Journal of Inherited Metabolic Disease, 2021, 44, 677-692.	1.7	20
114	Pharmacologic rescue of hyperammonemia-induced toxicity in zebrafish by inhibition of ornithine aminotransferase. PLoS ONE, 2018, 13, e0203707.	1.1	19
115	Genotypic diversity and phenotypic spectrum of infantile liver failure syndrome type 1 due to variants in LARS1. Genetics in Medicine, 2020, 22, 1863-1873.	1.1	19
116	Crystal structure and interaction studies of human DHTKD1 provide insight into a mitochondrial megacomplex in lysine catabolism. IUCrJ, 2020, 7, 693-706.	1.0	19
117	Cytosine arabinofuranoside-induced activation of astrocytes increases the susceptibility of neurons to glutamate due to the release of soluble factors. Neurochemistry International, 2003, 42, 567-581.	1.9	18
118	Newborn screening and disease variants predict neurological outcome in isovaleric aciduria. Journal of Inherited Metabolic Disease, 2021, 44, 857-870.	1.7	18
119	Genetic cause and prevalence of hydroxyprolinemia. Journal of Inherited Metabolic Disease, 2016, 39, 625-632.	1.7	17
120	Newborn Screening Programmes in Europe, Arguments and Efforts Regarding Harmonisation: Focus on Organic Acidurias. JIMD Reports, 2016, 32, 105-115.	0.7	17
121	FDA orphan drug designations for lysosomal storage disorders â€“ a cross-sectional analysis. PLoS ONE, 2020, 15, e0230898.	1.1	17
122	Clinical spectrum and treatment outcome of 95 children with continuous spikes and waves during sleep (CSWS). European Journal of Paediatric Neurology, 2021, 30, 121-127.	0.7	17
123	Phenotypic diversity, disease progression, and pathogenicity of <scp>MVK</scp> missense variants in mevalonic aciduria. Journal of Inherited Metabolic Disease, 2021, 44, 1272-1287.	1.7	17
124	Health Outcomes of Infants with Vitamin B12 Deficiency Identified by Newborn Screening and Early Treated. Journal of Pediatrics, 2021, 235, 42-48.	0.9	17
125	Bioenergetic dysfunction in a zebrafish model of acute hyperammonemic decompensation. Experimental Neurology, 2019, 314, 91-99.	2.0	16
126	Quantitative retrospective natural history modeling for orphan drug development. Journal of Inherited Metabolic Disease, 2021, 44, 99-109.	1.7	16

#	Article	IF	Citations
127	Reversible end-stage renal disease in an adolescent patient with methylmalonic aciduria. Pediatric Nephrology, 2004, 19, 1182-4.	0.9	15
128	Maleic Acid â€“ but Not Structurally Related Methylmalonic Acid â€“ Interrupts Energy Metabolism by Impaired Calcium Homeostasis. PLoS ONE, 2015, 10, e0128770.	1.1	15
129	Development and Psychometric Evaluation of the MetabQoL 1.0: A Quality of Life Questionnaire for Paediatric Patients with Intoxication-Type Inborn Errors of Metabolism. JIMD Reports, 2017, 37, 27-35.	0.7	15
130	Human heterologous liver cells transiently improve hyperammonemia and ureagenesis in individuals with severe urea cycle disorders. Journal of Inherited Metabolic Disease, 2018, 41, 81-90.	1.7	15
131	ADP-dependent glucokinase regulates energy metabolism via ER-localized glucose sensing. Scientific Reports, 2019, 9, 14248.	1.6	15
132	Impairment of cognitive function in ornithine transcarbamylase deficiency is global rather than domainâ€specific and is associated with disease onset, sex, maximum ammonium, and number of hyperammonemic events. Journal of Inherited Metabolic Disease, 2019, 42, 243-253.	1.7	15
133	U-IMD: the first Unified European registry for inherited metabolic diseases. Orphanet Journal of Rare Diseases, 2021, 16, 95.	1.2	15
134	Analysis of the functional muscleâ€“bone unit of the forearm in patients with phenylketonuria by peripheral quantitative computed tomography. Journal of Inherited Metabolic Disease, 2017, 40, 219-226.	1.7	14
135	Age-Related Changes and Reference Values of Bicaudate Ratio and Sagittal Brainstem Diameters on MRI. Neuropediatrics, 2018, 49, 269-275.	0.3	14
136	A cross-sectional quantitative analysis of the natural history of free sialic acid storage diseaseâ€”an ultra-orphan multisystemic lysosomal storage disorder. Genetics in Medicine, 2019, 21, 347-352.	1.1	14
137	From genotype to phenotype: Early prediction of disease severity in argininosuccinic aciduria. Human Mutation, 2020, 41, 946-960.	1.1	14
138	Severity-adjusted evaluation of newborn screening on the metabolic disease course in individuals with cytosolic urea cycle disorders. Molecular Genetics and Metabolism, 2020, 131, 390-397.	0.5	14
139	Towards Achieving Equity and Innovation in Newborn Screening across Europe. International Journal of Neonatal Screening, 2022, 8, 31.	1.2	14
140	Quantitative Acylcarnitine Profiling in Peripheral Blood Mononuclear Cells Using In Vitro Loading With Palmitic and 2-Oxoadipic Acids: Biochemical Confirmation of Fatty Acid Oxidation and Organic Acid Disorders. Pediatric Research, 2005, 58, 873-880.	1.1	13
141	Impact of interventional and nonâ€interventional variables on anthropometric longâ€term development in glutaric aciduria type 1: A national prospective multiâ€centre study. Journal of Inherited Metabolic Disease, 2021, 44, 629-638.	1.7	13
142	Opportunities and challenges in machine learningâ€based newborn screeningâ€”A systematic literature review. JIMD Reports, 2022, 63, 250-261.	0.7	13
143	On the Creation, Utility and Sustaining of Rare Diseases Research Networks: Lessons learned from the Urea Cycle Disorders Consortium, the Japanese Urea Cycle Disorders Consortium and the European Registry and Network for Intoxication Type Metabolic Diseases. Molecular Genetics and Metabolism, 2014, 113, 105-108.	0.5	12
144	QDPR homologues in Danio rerio regulate melanin synthesis, early gliogenesis, and glutamine homeostasis. PLoS ONE, 2019, 14, e0215162.	1.1	12

#	Article	IF	Citations
145	Paroxysmal and non-paroxysmal dystonia in 3 patients with biallelic ECHS1 variants: Expanding the neurological spectrum and therapeutic approaches. European Journal of Medical Genetics, 2020, 63, 104046.	0.7	12
146	Urinary NMR Profiling in Pediatric Acute Kidney Injuryâ€"A Pilot Study. International Journal of Molecular Sciences, 2020, 21, 1187.	1.8	12
147	Chronic hyperammonemia causes a hypoglutamatergic and hyperGABAergic metabolic state associated with neurobehavioral abnormalities in zebrafish larvae. Experimental Neurology, 2020, 331, 113330.	2.0	12
148	Patterns, evolution, and severity of striatal injury in insidious- versus acute-onset glutaric aciduria type 1. Journal of Inherited Metabolic Disease, 2018, , .	1.7	11
149	Long-term effects of medical management on growth and weight in individuals with urea cycle disorders. Scientific Reports, 2020, 10, 11948.	1.6	11
150	A cross-sectional controlled developmental study of neuropsychological functions in patients with glutaric aciduria type I. Orphanet Journal of Rare Diseases, 2015, 10, 163.	1.2	10
151	NBAS Variants Are Associated with Quantitative and Qualitative NK and B Cell Deficiency. Journal of Clinical Immunology, 2021, 41, 1781-1793.	2.0	10
152	Impairment of astrocytic glutaminolysis in glutaric aciduria type I. Journal of Inherited Metabolic Disease, 2018, 41, 91-99.	1.7	9
153	Further evidence for de novo variants in SYNCRIP as the cause of a neurodevelopmental disorder. Human Mutation, 2021, 42, 1094-1100.	1.1	9
154	The biochemical subtype is a predictor for cognitive function in glutaric aciduria type 1: a national prospective follow-up study. Scientific Reports, 2021, 11, 19300.	1.6	9
155	Complementing the phenotypical spectrum of TUBA1A tubulinopathy and its role in early-onset epilepsies. European Journal of Human Genetics, 2022, 30, 298-306.	1.4	9
156	Newborn Screening for Glutaric Aciduria Type I: Benefits and limitations. International Journal of Neonatal Screening, 2015, 1, 57-68.	1.2	8
157	Recurrent Stroke-Like Episodes in FBXL4-Associated Early-Onset Mitochondrial Encephalomyopathy. Pediatric Neurology, 2015, 53, 549-550.	1.0	8
158	Chiari-like displacement due to spontaneous intracranial hypotension in an adolescent: Successful treatment by epidural blood patch. European Journal of Paediatric Neurology, 2017, 21, 678-681.	0.7	8
159	Cross-sectional quantitative analysis of the natural history of TUBA1A and TUBB2B tubulinopathies. Genetics in Medicine, 2021, 23, 516-523.	1.1	8
160	1H-NMR-based metabolic profiling identifies non-invasive diagnostic and predictive urinary fingerprints in 5q spinal muscular atrophy. Orphanet Journal of Rare Diseases, 2021, 16, 441.	1.2	8
161	Generation of an induced pluripotent stem cell (iPSC) line, DHMCi005-A, from a patient with CALFAN syndrome due to mutations in SCYL1. Stem Cell Research, 2019, 37, 101428.	0.3	7
162	Cerebral Organic Acid Disorders and Other Disorders of Lysine Catabolism. , 2016, , 333-348.		7

#	Article	IF	Citations
163	Influence of early identification and therapy on longâ€term outcomes in earlyâ€onset <scp>MTHFR</scp> deficiency. Journal of Inherited Metabolic Disease, 2022, 45, 848-861.	1.7	7
164	Genomic newborn screening: Proposal of a <scp>twoâ€stage</scp> approach. Journal of Inherited Metabolic Disease, 2021, 44, 518-520.	1.7	6
165	Mitochondrial damage in renal epithelial cells is potentiated by protein exposure in propionic aciduria. Journal of Inherited Metabolic Disease, 2021, 44, 1330-1342.	1.7	6
166	Subdural hematoma in glutaric aciduria type 1: High excreters are prone to incidental <scp>SDH</scp> despite newborn screening. Journal of Inherited Metabolic Disease, 2021, 44, 1343-1352.	1.7	6
167	Axenfeld-Rieger Anomaly and Neuropsychiatric Problemsâ€”More than Meets the Eye. Neuropediatrics, 2020, 51, 192-197.	0.3	5
168	Quantitative retrospective natural history modeling of WDR45-related developmental and epileptic encephalopathy â€“ a systematic cross-sectional analysis of 160 published cases. Autophagy, 2022, 18, 1715-1727.	4.3	5
169	How longitudinal observational studies can guide screening strategy for rare diseases. Journal of Inherited Metabolic Disease, 2022, 45, 889-901.	1.7	5
170	High blood pressure, a red flag for the neonatal manifestation of urea cycle disorders. Orphanet Journal of Rare Diseases, 2019, 14, 80.	1.2	4
171	ADP-dependent glucokinase as a novel onco-target for haematological malignancies. Scientific Reports, 2020, 10, 13584.	1.6	4
172	Transatlantic combined and comparative data analysis of 1095 patients with urea cycle disordersâ€”a successful strategy for clinical research of rare diseases. Journal of Inherited Metabolic Disease, 2019, 42, 93.	1.7	4
173	Unmet Needs of Parents of Children with Urea Cycle Disorders. Children, 2022, 9, 712.	0.6	4
174	Cerebral Organic Acid Disorders and Other Disorders of Lysine Catabolism. , 2012, , 333-347.		3
175	Sudden neonatal death in individuals with medium-chain acyl-coenzyme A dehydrogenase deficiency: limit of newborn screening. European Journal of Pediatrics, 2022, 181, 2415-2422.	1.3	3
176	From newborn screening to genomic medicine: challenges and suggestions on how to incorporate genomic newborn screening in public health programs. Medizinische Genetik, 2022, 34, 13-20.	0.1	2
177	Generation of an iPSC line from a patient with infantile liver failure syndrome 2 due to mutations in NBAS: DHMCi004-A. Stem Cell Research, 2019, 35, 101398.	0.3	1
178	Protein-dependent inborn errors of metabolism. , 2010, , 1559-1595.		1
179	Cerebral Organic Acidurias. , 2014, , 143-156.		1
180	Response. Pediatric Research, 2007, 61, 134-135.	1.1	0

#	Article	IF	Citations
181	Perioperative Management of a Child with Glutaric Aciduria Type I Undergoing Cardiac Surgery. A & A Case Reports, 2013, 1, 5-7.	0.7	0
182	Variability of OTC deficiency in heterozygous carriers: Case report of a family. Journal of Pediatric Biochemistry, 2015, 04, 005-010.	0.2	0
183	Emergency Treatment of Inherited Metabolic Diseases. , 2017, , 125-131.		0
184	Arachnoid Cysts in Glutaric Aciduria Type I (GA-I). , 2018, , 39-55.		0
185	AB1029â€…FIBRODYSPLASIA OSSIFICANS PROGRESSIVA: A CHALLENGE TO DIAGNOSE AND TO TREAT. , 2019, , .		0
186	Krankheiten und StÃ¶rungen des EiweiÃŸstoffwechsel. , 2014, , 466-492.		0
187	FV 754. Newborn Screening, a Disease-Modifying Intervention for Glutaric Aciduria Type 1. , 2018, 49, .		0
188	Organoazidurien. Springer Reference Medizin, 2019, , 1-17.	0.0	0
189	Organoazidurien. Springer Reference Medizin, 2020, , 689-704.	0.0	0
190	The Biochemical High Excreter Phenotype Is the Major Risk Factor for Cognitive Impairment in Early Diagnosed Individuals with Glutaric Aciduria Type 1. Neuropediatrics, 2021, 52, .	0.3	0
191	FDA orphan drug designations for lysosomal storage disorders â€“ a cross-sectional analysis. , 2020, 15, e0230898.		0
192	FDA orphan drug designations for lysosomal storage disorders â€“ a cross-sectional analysis. , 2020, 15, e0230898.		0
193	FDA orphan drug designations for lysosomal storage disorders â€“ a cross-sectional analysis. , 2020, 15, e0230898.		0
194	FDA orphan drug designations for lysosomal storage disorders â€“ a cross-sectional analysis. , 2020, 15, e0230898.		0