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	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
2	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
3	Rare Disease Registries Are Key to Evidence-Based Personalized Medicine: Highlighting the European Experience. Frontiers in Endocrinology, 2022, 13, 832063.	1.5	28
4	Opportunities and challenges in machine learningâ€based newborn screeningâ€”A systematic literature review. JIMD Reports, 2022, 63, 250-261.	0.7	13
5	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
6	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
7	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
8	How longitudinal observational studies can guide screening strategy for rare diseases. Journal of Inherited Metabolic Disease, 2022, 45, 889-901.	1.7	5
9	Towards Achieving Equity and Innovation in Newborn Screening across Europe. International Journal of Neonatal Screening, 2022, 8, 31.	1.2	14
10	Unmet Needs of Parents of Children with Urea Cycle Disorders. Children, 2022, 9, 712.	0.6	4
11	Organic acidurias: Major gaps, new challenges, and a yet unfulfilled promise. Journal of Inherited Metabolic Disease, 2021, 44, 9-21.	1.7	28
12	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
13	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
14	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
15	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
16	Cross-sectional quantitative analysis of the natural history of TUBA1A and TUBB2B tubulinopathies. Genetics in Medicine, 2021, 23, 516-523.	1.1	8
17	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
18	An international classification of inherited metabolic disorders (<scp>ICIMD</scp>). Journal of Inherited Metabolic Disease, 2021, 44, 164-177.	1.7	146

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19	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
20	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
21	Quantitative retrospective natural history modeling for orphan drug development. Journal of Inherited Metabolic Disease, 2021, 44, 99-109.	1.7	16
22	U-IMD: the first Unified European registry for inherited metabolic diseases. Orphanet Journal of Rare Diseases, 2021, 16, 95.	1.2	15
23	Newborn screening and disease variants predict neurological outcome in isovaleric aciduria. Journal of Inherited Metabolic Disease, 2021, 44, 857-870.	1.7	18
24	Genomic newborn screening: Proposal of a <scp>twoâ€stage</scp> approach. Journal of Inherited Metabolic Disease, 2021, 44, 518-520.	1.7	6
25	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
26	Further evidence for de novo variants in SYNCRIP as the cause of a neurodevelopmental disorder. Human Mutation, 2021, 42, 1094-1100.	1.1	9
27	NBAS Variants Are Associated with Quantitative and Qualitative NK and B Cell Deficiency. Journal of Clinical Immunology, 2021, 41, 1781-1793.	2.0	10
28	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
29	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
30	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
31	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
32	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
33	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
34	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
35	Defining clinical subgroups and genotypeâ€“phenotype correlations in NBAS-associated disease across 110 patients. Genetics in Medicine, 2020, 22, 610-621.	1.1	46
36	Defining the clinical, molecular and imaging spectrum of adaptor protein complex 4-associated hereditary spastic paraplegia. Brain, 2020, 143, 2929-2944.	3.7	29

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37	Long-term Outcomes of Individuals With Metabolic Diseases Identified Through Newborn Screening. Pediatrics, 2020, 146, .	1.0	37
38	Long-term effects of medical management on growth and weight in individuals with urea cycle disorders. Scientific Reports, 2020, 10, 11948.	1.6	11
39	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
40	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
41	ADP-dependent glucokinase as a novel onco-target for haematological malignancies. Scientific Reports, 2020, 10, 13584.	1.6	4
42	Axenfeld-Rieger Anomaly and Neuropsychiatric Problemsâ€"More than Meets the Eye. Neuropediatrics, 2020, 51, 192-197.	0.3	5
43	Urinary NMR Profiling in Pediatric Acute Kidney Injuryâ€"A Pilot Study. International Journal of Molecular Sciences, 2020, 21, 1187.	1.8	12
44	Impaired mitophagy links mitochondrial disease to epithelial stress in methylmalonyl-CoA mutase deficiency. Nature Communications, 2020, 11, 970.	5.8	65
45	From genotype to phenotype: Early prediction of disease severity in argininosuccinic aciduria. Human Mutation, 2020, 41, 946-960.	1.1	14
46	Chronic hyperammonemia causes a hypoglutamatergic and hyperGABAergic metabolic state associated with neurobehavioral abnormalities in zebrafish larvae. Experimental Neurology, 2020, 331, 113330.	2.0	12
47	FDA orphan drug designations for lysosomal storage disorders â€“ a cross-sectional analysis. PLoS ONE, 2020, 15, e0230898.	1.1	17
48	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
49	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
50	Organoazidurien. Springer Reference Medizin, 2020, , 689-704.	0.0	0
51	FDA orphan drug designations for lysosomal storage disorders â€“ a cross-sectional analysis. , 2020, 15, e0230898.		0
52	FDA orphan drug designations for lysosomal storage disorders â€“ a cross-sectional analysis. , 2020, 15, e0230898.		0
53	FDA orphan drug designations for lysosomal storage disorders â€“ a cross-sectional analysis. , 2020, 15, e0230898.		0
54	FDA orphan drug designations for lysosomal storage disorders â€“ a cross-sectional analysis. , 2020, 15, e0230898.		0

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55	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
56	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
57	Early prediction of phenotypic severity in Citrullinemia Type 1. Annals of Clinical and Translational Neurology, 2019, 6, 1858-1871.	1.7	26
58	Targeted Metabolic Profiling of Methionine Cycle Metabolites and Redox Thiol Pools in Mammalian Plasma, Cells and Urine. Metabolites, 2019, 9, 235.	1.3	26
59	Mutations in PCYT2 disrupt etherlipid biosynthesis and cause a complex hereditary spastic paraplegia. Brain, 2019, 142, 3382-3397.	3.7	76
60	ADP-dependent glucokinase regulates energy metabolism via ER-localized glucose sensing. Scientific Reports, 2019, 9, 14248.	1.6	15
61	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
62	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
63	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
64	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
65	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
66	Impact of Diagnosis and Therapy on Cognitive Function in Urea Cycle Disorders. Annals of Neurology, 2019, 86, 116-128.	2.8	42
67	QDPR homologues in Danio rerio regulate melanin synthesis, early gliogenesis, and glutamine homeostasis. PLoS ONE, 2019, 14, e0215162.	1.1	12
68	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
69	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
70	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
71	Bioenergetic dysfunction in a zebrafish model of acute hyperammonemic decompensation. Experimental Neurology, 2019, 314, 91-99.	2.0	16
72	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

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73	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
74	Newborn screening for homocystinurias: Recent recommendations versus current practice. Journal of Inherited Metabolic Disease, 2019, 42, 128-139.	1.7	37
75	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
76	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
77	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
78	AB1029â€…FIBRODYSPLASIA OSSIFICANS PROGRESSIVA: A CHALLENGE TO DIAGNOSE AND TO TREAT. , 2019, , .		0
79	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
80	Organoazidurien. Springer Reference Medizin, 2019, , 1-17.	0.0	0
81	Newborn screening: A diseaseâ€changing intervention for glutaric aciduria type 1. Annals of Neurology, 2018, 83, 970-979.	2.8	65
82	Organic acidurias in adults: late complications and management. Journal of Inherited Metabolic Disease, 2018, 41, 765-776.	1.7	27
83	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
84	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
85	Impairment of astrocytic glutaminolysis in glutaric aciduria type I. Journal of Inherited Metabolic Disease, 2018, 41, 91-99.	1.7	9
86	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
87	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
88	Pharmacologic rescue of hyperammonemia-induced toxicity in zebrafish by inhibition of ornithine aminotransferase. PLoS ONE, 2018, 13, e0203707.	1.1	19
89	Arachnoid Cysts in Glutaric Aciduria Type I (GA-I). , 2018, , 39-55.		0
90	Metabolism of amino acid neurotransmitters: the synaptic disorder underlying inherited metabolic diseases. Journal of Inherited Metabolic Disease, 2018, 41, 1055-1063.	1.7	26

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91	Age-Related Changes and Reference Values of Bicaudate Ratio and Sagittal Brainstem Diameters on MRI. Neuropediatrics, 2018, 49, 269-275.	0.3	14
92	FV 754. Newborn Screening, a Disease-Modifying Intervention for Glutaric Aciduria Type 1. , 2018, 49, .		0
93	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
94	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
95	Linking mitochondrial dysfunction to neurodegeneration in lysosomal storage diseases. Journal of Inherited Metabolic Disease, 2017, 40, 631-640.	1.7	46
96	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
97	Quantitative clinical characteristics of 53 patients with MPS VII: a cross-sectional analysis. Genetics in Medicine, 2017, 19, 983-988.	1.1	42
98	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
99	Issues with European guidelines for phenylketonuria. Lancet Diabetes and Endocrinology,the, 2017, 5, 681-683.	5.5	26
100	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
101	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
102	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
103	Impact of clinical exomes in neurodevelopmental and neurometabolic disorders. Molecular Genetics and Metabolism, 2017, 121, 297-307.	0.5	50
104	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
105	Emergency Treatment of Inherited Metabolic Diseases. , 2017, , 125-131.		0
106	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
107	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
108	Genetic cause and prevalence of hydroxyprolinemia. Journal of Inherited Metabolic Disease, 2016, 39, 625-632.	1.7	17

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109	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
110	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
111	Newborn Screening Programmes in Europe, Arguments and Efforts Regarding Harmonisation: Focus on Organic Acidurias. JIMD Reports, 2016, 32, 105-115.	0.7	17
112	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
113	Impaired Mitochondrial Dynamics and Mitophagy in Neuronal Models of Tuberous Sclerosis Complex. Cell Reports, 2016, 17, 1053-1070.	2.9	125
114	Adenosine kinase deficiency: expanding the clinical spectrum and evaluating therapeutic options. Journal of Inherited Metabolic Disease, 2016, 39, 273-283.	1.7	55
115	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
116	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
117	EPG5-related Vici syndrome: a paradigm of neurodevelopmental disorders with defective autophagy. Brain, 2016, 139, 765-781.	3.7	99
118	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
119	Cerebral Organic Acid Disorders and Other Disorders of Lysine Catabolism. , 2016, , 333-348.		7
120	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
121	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
122	Variability of OTC deficiency in heterozygous carriers: Case report of a family. Journal of Pediatric Biochemistry, 2015, 04, 005-010.	0.2	0
123	Newborn Screening for Glutaric Aciduria Type I: Benefits and limitations. International Journal of Neonatal Screening, 2015, 1, 57-68.	1.2	8
124	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
125	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
126	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

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127	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
128	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
129	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
130	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
131	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
132	Recurrent Stroke-Like Episodes in FBXL4-Associated Early-Onset Mitochondrial Encephalomyopathy. Pediatric Neurology, 2015, 53, 549-550.	1.0	8
133	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
134	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
135	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
136	Emerging role of autophagy in pediatric neurodegenerative and neurometabolic diseases. Pediatric Research, 2014, 75, 217-226.	1.1	42
137	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
138	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
139	Cerebral Organic Acidurias. , 2014, , 143-156.		1
140	Krankheiten und StÃ¶rungen des EiweiÃŸstoffwechsel. , 2014, , 466-492.		0
141	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
142	The incidence of urea cycle disorders. Molecular Genetics and Metabolism, 2013, 110, 179-180.	0.5	232
143	Current concepts in organic acidurias: understanding intraâ€and extracerebral disease manifestation. Journal of Inherited Metabolic Disease, 2013, 36, 635-644.	1.7	52
144	Perioperative Management of a Child with Glutaric Aciduria Type I Undergoing Cardiac Surgery. A & A Case Reports, 2013, 1, 5-7.	0.7	0

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145	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
146	Recessive mutations in EPG5 cause Vici syndrome, a multisystem disorder with defective autophagy. Nature Genetics, 2013, 45, 83-87.	9.4	231
147	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
148	Metabolic decompensation in methylmalonic aciduria: which biochemical parameters are discriminative?. Journal of Inherited Metabolic Disease, 2012, 35, 797-806.	1.7	35
149	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
150	DHTKD1 Mutations Cause 2-Aminoadipic and 2-Oxoadipic Aciduria. American Journal of Human Genetics, 2012, 91, 1082-1087.	2.6	94
151	Cerebral Organic Acid Disorders and Other Disorders of Lysine Catabolism. , 2012, , 333-347.		3
152	High urgency liver transplantation in ornithine transcarbamylase deficiency presenting with acute liver failure. Pediatric Transplantation, 2011, 15, E110-5.	0.5	25
153	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
154	Diagnosis and management of glutaric aciduria type I â€" revised recommendations. Journal of Inherited Metabolic Disease, 2011, 34, 677-694.	1.7	327
155	Therapeutic modulation of cerebral l-lysine metabolism in a mouse model for glutaric aciduria type I. Brain, 2011, 134, 157-170.	3.7	102
156	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
157	Use of guidelines improves the neurological outcome in glutaric aciduria type I. Annals of Neurology, 2010, 68, 743-752.	2.8	147
158	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
159	Iron Regulatory Proteins Secure Mitochondrial Iron Sufficiency and Function. Cell Metabolism, 2010, 12, 194-201.	7.2	110
160	Protein-dependent inborn errors of metabolism. , 2010, , 1559-1595.		1
161	Dynamic changes of striatal and extrastriatal abnormalities in glutaric aciduria type I. Brain, 2009, 132, 1764-1782.	3.7	160
162	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

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