

# Chris MÃ¼hlhausen

## List of Publications by Year in descending order

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Version: 2024-02-01

20  
papers

1,917  
citations

567281

15  
h-index

752698

20  
g-index

20  
all docs

20  
docs citations

20  
times ranked

2592  
citing authors

#	ARTICLE	IF	CITATIONS
1	Managing CLN2 disease: a treatable neurodegenerative condition among other treatable early childhood epilepsies. <i>Expert Review of Neurotherapeutics</i> , 2021, 21, 1275-1282.	2.8	5
2	Mannose phosphate isomerase deficiencyâ€œcongenital disorder of glycosylation ( MPIâ€œCDG ) with cerebral venous sinus thrombosis as first and only presenting symptom: A rare but treatable cause of thrombophilia. <i>JIMD Reports</i> , 2020, 55, 38-43.	1.5	8
3	Ammonium accumulation and chemokine decrease in culture media of Gcdh <sup>+/+</sup> 3D reaggregated brain cell cultures. <i>Molecular Genetics and Metabolism</i> , 2019, 126, 416-428.	1.1	6
4	Newborn screening: A diseaseâ€œchanging intervention for glutaric aciduria type 1. <i>Annals of Neurology</i> , 2018, 83, 970-979.	5.3	65
5	Disease-Linked Glutarylation Impairs Function and Interactions of Mitochondrial Proteins and Contributes to Mitochondrial Heterogeneity. <i>Cell Reports</i> , 2018, 24, 2946-2956.	6.4	42
6	Disease-causing mutations affecting surface residues of mitochondrial glutaryl-CoA dehydrogenase impair stability, heteromeric complex formation and mitochondria architecture. <i>Human Molecular Genetics</i> , 2017, 26, ddw411.	2.9	14
7	Development and Psychometric Evaluation of the MetabQoL 1.0: A Quality of Life Questionnaire for Paediatric Patients with Intoxication-Type Inborn Errors of Metabolism. <i>JIMD Reports</i> , 2017, 37, 27-35.	1.5	15
8	Glutaric Aciduria Type 1 and Acute Renal Failure: Case Report and Suggested Pathomechanisms. <i>JIMD Reports</i> , 2017, 39, 25-30.	1.5	12
9	Proposed recommendations for diagnosing and managing individuals with glutaric aciduria type I: second revision. <i>Journal of Inherited Metabolic Disease</i> , 2017, 40, 75-101.	3.6	173
10	Living with Intoxication-Type Inborn Errors of Metabolism: A Qualitative Analysis of Interviews with Paediatric Patients and Their Parents. <i>JIMD Reports</i> , 2016, 31, 1-9.	1.5	20
11	Bi-allelic Truncating Mutations in TANGO2 Cause Infancy-Onset Recurrent Metabolic Crises with Encephalocardiomyopathy. <i>American Journal of Human Genetics</i> , 2016, 98, 358-362.	6.2	77
12	Behavioural and emotional problems, intellectual impairment and healthâ€œrelated quality of life in patients with organic acidurias and urea cycle disorders. <i>Journal of Inherited Metabolic Disease</i> , 2016, 39, 231-241.	3.6	29
13	Interaction of Glutaric Aciduria Type 1-Related glutaryl-CoA Dehydrogenase with Mitochondrial Matrix Proteins. <i>PLoS ONE</i> , 2014, 9, e87715.	2.5	20
14	Lysine Glutarylation Is a Protein Posttranslational Modification Regulated by SIRT5. <i>Cell Metabolism</i> , 2014, 19, 605-617.	16.2	647
15	Combined D2â€œL2â€œhydroxyglutaric aciduria (<i>SLC25A1</i> deficiency): clinical course and effects of citrate treatment. <i>Journal of Inherited Metabolic Disease</i> , 2014, 37, 775-781.	3.6	32
16	Diagnosis and management of glutaric aciduria type I â€œ revised recommendations. <i>Journal of Inherited Metabolic Disease</i> , 2011, 34, 677-694.	3.6	327
17	Use of guidelines improves the neurological outcome in glutaric aciduria type I. <i>Annals of Neurology</i> , 2010, 68, 743-752.	5.3	147
18	Transport and distribution of 3-hydroxyglutaric acid before and during induced encephalopathic crises in a mouse model of glutaric aciduria type 1. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2008, 1782, 385-390.	3.8	29

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19	Decline of Acute Encephalopathic Crises in Children with Glutaryl-CoA Dehydrogenase Deficiency Identified by Newborn Screening in Germany. <i>Pediatric Research</i> , 2007, 62, 357-363.	2.3	102
20	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. <i>Journal of Neurochemistry</i> , 2006, 97, 899-910.	3.9	147