

Peter Burgard

List of Publications by Year in descending order

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Version: 2024-02-01

43
papers

1,911
citations

279798

23
h-index

254184

43
g-index

51
all docs

51
docs citations

51
times ranked

2098
citing authors

#	ARTICLE	IF	CITATIONS
1	The phenotypic spectrum of organic acidurias and urea cycle disorders. Part 1: the initial presentation. <i>Journal of Inherited Metabolic Disease</i> , 2015, 38, 1041-1057.	3.6	186
2	The phenotypic spectrum of organic acidurias and urea cycle disorders. Part 2: the evolving clinical phenotype. <i>Journal of Inherited Metabolic Disease</i> , 2015, 38, 1059-1074.	3.6	175
3	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
4	Use of guidelines improves the neurological outcome in glutaric aciduria type I. <i>Annals of Neurology</i> , 2010, 68, 743-752.	5.3	147
5	Efficacy and outcome of expanded newborn screening for metabolic diseases - Report of 10 years from South-West Germany *. <i>Orphanet Journal of Rare Diseases</i> , 2011, 6, 44.	2.7	128
6	Biallelic Mutations in NBAS Cause Recurrent Acute Liver Failure with Onset in Infancy. <i>American Journal of Human Genetics</i> , 2015, 97, 163-169.	6.2	110
7	Recurrent acute liver failure due to NBAS deficiency: phenotypic spectrum, disease mechanisms, and therapeutic concepts. <i>Journal of Inherited Metabolic Disease</i> , 2016, 39, 3-16.	3.6	92
8	Newborn screening programmes in Europe; arguments and efforts regarding harmonization. Part 2 – From screening laboratory results to treatment, follow-up and quality assurance. <i>Journal of Inherited Metabolic Disease</i> , 2012, 35, 613-625.	3.6	88
9	Cross-sectional observational study of 208 patients with non-classical urea cycle disorders. <i>Journal of Inherited Metabolic Disease</i> , 2014, 37, 21-30.	3.6	62
10	Impact of age at onset and newborn screening on outcome in organic acidurias. <i>Journal of Inherited Metabolic Disease</i> , 2016, 39, 341-353.	3.6	60
11	Age at disease onset and peak ammonium level rather than interventional variables predict the neurological outcome in urea cycle disorders. <i>Journal of Inherited Metabolic Disease</i> , 2016, 39, 661-672.	3.6	52
12	Neonatal mortality and outcome at the end of the first year of life in early onset urea cycle disorders – review and meta-analysis of observational studies published over more than 35 years. <i>Journal of Inherited Metabolic Disease</i> , 2016, 39, 219-229.	3.6	50
13	Allelic phenotype values: a model for genotype-based phenotype prediction in phenylketonuria. <i>Genetics in Medicine</i> , 2019, 21, 580-590.	2.4	48
14	Impact of Diagnosis and Therapy on Cognitive Function in Urea Cycle Disorders. <i>Annals of Neurology</i> , 2019, 86, 116-128.	5.3	42
15	Long-term Outcomes of Individuals With Metabolic Diseases Identified Through Newborn Screening. <i>Pediatrics</i> , 2020, 146, .	2.1	37
16	Transatlantic combined and comparative data analysis of 1095 patients with urea cycle disorders – A successful strategy for clinical research of rare diseases. <i>Journal of Inherited Metabolic Disease</i> , 2019, 42, 93-106.	3.6	35
17	Living with an inborn error of metabolism detected by newborn screening – Parents’ perspectives on child development and impact on family life. <i>Journal of Inherited Metabolic Disease</i> , 2014, 37, 189-195.	3.6	34
18	Biochemical markers and neuropsychological functioning in distal urea cycle disorders. <i>Journal of Inherited Metabolic Disease</i> , 2018, 41, 657-667.	3.6	31

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19	Newborn screening by tandem mass spectrometry for glutaric aciduria type 1: a cost-effectiveness analysis. <i>Orphanet Journal of Rare Diseases</i> , 2013, 8, 167.	2.7	30
20	Evaluation of dietary treatment and amino acid supplementation in organic acidurias and urea cycle disorders: On the basis of information from a European multicenter registry. <i>Journal of Inherited Metabolic Disease</i> , 2019, 42, 1162-1175.	3.6	30
21	Behavioural and emotional problems, intellectual impairment and health-related 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
22	Networking Across Borders for Individuals with Organic Acidurias and Urea Cycle Disorders: The E-IMD Consortium. <i>JIMD Reports</i> , 2015, 22, 29-38.	1.5	26
23	Issues with European guidelines for phenylketonuria. <i>Lancet Diabetes and Endocrinology</i> , 2017, 5, 681-683.	11.4	26
24	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. <i>Molecular Genetics and Metabolism</i> , 2019, 126, 397-405.	1.1	26
25	Multifactorial modulation of susceptibility to l-lysine in an animal model of glutaric aciduria type I. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2015, 1852, 768-777.	3.8	21
26	Newborn Screening for Vitamin B6 Non-responsive Classical Homocystinuria: Systematical Evaluation of a Two-Tier Strategy. <i>JIMD Reports</i> , 2016, 32, 87-94.	1.5	19
27	Newborn Screening Programmes in Europe, Arguments and Efforts Regarding Harmonisation: Focus on Organic Acidurias. <i>JIMD Reports</i> , 2016, 32, 105-115.	1.5	17
28	Diagnosis and therapeutic monitoring of inborn errors of creatine metabolism and transport using liquid chromatography-tandem mass spectrometry in urine, plasma and CSF. <i>Gene</i> , 2014, 538, 188-194.	2.2	16
29	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
30	Impairment of cognitive function in ornithine transcarbamylase deficiency is global rather than domain-specific and is associated with disease onset, sex, maximum ammonium, and number of hyperammonemic events. <i>Journal of Inherited Metabolic Disease</i> , 2019, 42, 243-253.	3.6	15
31	Determinants of Plasma Docosahexaenoic Acid Levels and Their Relationship to Neurological and Cognitive Functions in PKU Patients: A Double Blind Randomized Supplementation Study. <i>Nutrients</i> , 2018, 10, 1944.	4.1	12
32	Impact of glycogen storage disease type I on adult daily life: a survey. <i>Orphanet Journal of Rare Diseases</i> , 2021, 16, 371.	2.7	12
33	A cross-sectional controlled developmental study of neuropsychological functions in patients with glutaric aciduria type I. <i>Orphanet Journal of Rare Diseases</i> , 2015, 10, 163.	2.7	10
34	An Integrated clinical pathway for diagnosis, treatment and care of rare diseases: model, operating procedures, and results of the project TRANSLATE-NAMSE funded by the German Federal Joint Committee. <i>Orphanet Journal of Rare Diseases</i> , 2021, 16, 474.	2.7	7
35	On being an editor, reviewer, and author—different sides of the same coin. <i>Journal of Inherited Metabolic Disease</i> , 2019, 42, 1-2.	3.6	6
36	In vivo monitoring of urea cycle activity with ¹³ C-acetate as a tracer of ureagenesis. <i>Molecular Genetics and Metabolism</i> , 2016, 117, 19-26.	1.1	5

#	ARTICLE	IF	CITATIONS
37	High blood pressure, a red flag for the neonatal manifestation of urea cycle disorders. Orphanet Journal of Rare Diseases, 2019, 14, 80.	2.7	4
38	Transatlantic combined and comparative data analysis of 1095 patients with urea cycle disorders—a successful strategy for clinical research of rare diseases. Journal of Inherited Metabolic Disease, 2019, 42, 93.	3.6	4
39	Differences of Phenylalanine Concentrations in Dried Blood Spots and in Plasma: Erythrocytes as a Neglected Component for This Observation. Metabolites, 2021, 11, 680.	2.9	3
40	A holistic approach to the patients/ Families with inborn errors of metabolism. Medycyna Wieku Rozwojowego, 2020, 24, 65-72.	0.2	3
41	Response. Pediatric Research, 2007, 61, 134-135.	2.3	0
42	Urea Cycle Disorders in the US and Europe — Evidence-based Clinical Outcomes Derived from Two Decades of Experience with Prospective Registry Studies. , 2019, 50, .		0
43	William L.Nyhan, Georg F.Hoffmann. Atlas of inherited metabolic diseases (Hardcover and eBook)4th ed. Boca Raton: CRC Press, Taylor & Francis Group, 2020, 855p., \$175; £120, -124. ISBN: 9781138196599. (With contributions from Aida I. Al-Aqeel and Bruce A. Barshop).. Journal of Inherited Metabolic Disease, 2021, 44, 284-284.	3.6	0