Diana Ballhausen

List of Publications by Year
in descending order

49 papers	2,645 citations	318942 23 h-index	242451 47 g-index
49 all docs	49 docs citations	49 times ranked	4071 citing authors

#	Article	IF	Citations
1	Postauthorization safety study of betaine anhydrous. Journal of Inherited Metabolic Disease, 2022, 45, 719-733.	1.7	5
2	Heterozygous variants in CTR9, which encodes a major component of the PAF1 complex, are associated with a neurodevelopmental disorder. Genetics in Medicine, 2022, , .	1.1	1
3	Efficacy and safety of empagliflozin in glycogen storage disease type Ib: Data from an international questionnaire. Genetics in Medicine, 2022, 24, 1781-1788.	1.1	29
4	Guidelines for the diagnosis and management of methylmalonic acidaemia and propionic acidaemia: FirstÂrevision. Journal of Inherited Metabolic Disease, 2021, 44, 566-592.	1.7	118
5	The first knock-in rat model for glutaric aciduria type I allows further insights into pathophysiology in brain and periphery. Molecular Genetics and Metabolism, 2021, 133, 157-181.	0.5	22
6	The fate of orally administered sialic acid: First insights from patients with N-acetylneuraminic acid synthase deficiency and control subjects. Molecular Genetics and Metabolism Reports, 2021, 28, 100777.	0.4	7
7	The impact of disease severity on the psychological well-being of youth affected by an inborn error of metabolism and their families: A one-year longitudinal study. Molecular Genetics and Metabolism Reports, 2021, 29, 100795.	0.4	4
8	Active site variants in STT3A cause a dominant type I congenital disorder of glycosylation with neuromusculoskeletal findings. American Journal of Human Genetics, 2021, 108, 2130-2144.	2.6	5
9	A knock-in rat model unravels acute and chronic renal toxicity in glutaric aciduria type I. Molecular Genetics and Metabolism, 2021, 134, 287-300.	0.5	17
10	The use of Ga-EDTA PET allows detecting progressive decline of renal function in rats.. American Journal of Nuclear Medicine and Molecular Imaging, 2021, 11, 519-528.	1.0	1
11	Severe Distal Motor Involvement in a Non-compliant Adult With Biotinidase Deficiency: The Necessity of Life-Long Biotin Therapy. Frontiers in Neurology, 2020, 11, 516799.	1.1	2
12	Hepatosplenomegaly, pneumopathy, bone changes and fronto-temporal dementia: Niemannâ€“Pick type B and SQSTM1-associated Pagetâ€™s disease in the same individual. Journal of Bone and Mineral Metabolism, 2019, 37, 378-383.	1.3	1
13	Ammonium accumulation and chemokine decrease in culture media of Gcdhâ^'/â^' 3D reaggregated brain cell cultures. Molecular Genetics and Metabolism, 2019, 126, 416-428.	0.5	6
14	Phenotype, treatment practice and outcome in the cobalaminâ€dependent remethylation disorders and MTHFR deficiency: Data from the Eâ€HOD registry. Journal of Inherited Metabolic Disease, 2019, 42, 333-352.	1.7	53
15	Treatment outcome of twenty-two patients with guanidinoacetate methyltransferase deficiency: An international retrospective cohort study. European Journal of Paediatric Neurology, 2018, 22, 369-379.	0.7	24
16	New in vitro model derived from brain-specific Mut-/- mice confirms cerebral ammonium accumulation in methylmalonic aciduria. Molecular Genetics and Metabolism, 2018, 124, 266-277.	0.5	6
17	Immunolocalization of glutaryl-CoA dehydrogenase (GCDH) in adult and embryonic rat brain and peripheral tissues. Neuroscience, 2017, 343, 355-363.	1.1	12
18	Epidemiology of mucopolysaccharidoses. Molecular Genetics and Metabolism, 2017, 121, 227-240.	0.5	290

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19	Issues with European guidelines for phenylketonuria. Lancet Diabetes and Endocrinology,the, 2017, 5, 681-683.	5.5	26
20	(5aR)-5a-C-Pentyl-4-epi-isofagomine: A powerful inhibitor of lysosomal Î²-galactosidase and a remarkable chaperone for mutations associated with GM1-gangliosidosis and Morquio disease type B. European Journal of Medicinal Chemistry, 2017, 126, 160-170.	2.6	32
21	Ammonium accumulation is a primary effect of 2-methylcitrate exposure in an in vitro model for brain damage in methylmalonic aciduria. Molecular Genetics and Metabolism, 2016, 119, 57-67.	0.5	13
22	Adenosine kinase deficiency: expanding the clinical spectrum and evaluating therapeutic options. Journal of Inherited Metabolic Disease, 2016, 39, 273-283.	1.7	55
23	NBAS mutations cause a multisystem disorder involving bone, connective tissue, liver, immune system, and retina. American Journal of Medical Genetics, Part A, 2015, 167, 2902-2912.	0.7	66
24	Early co-occurrence of a neurologic-psychiatric disease pattern in Niemann-Pick type C disease: a retrospective Swiss cohort study. Orphanet Journal of Rare Diseases, 2014, 9, 176.	1.2	14
25	Proposed guidelines for the diagnosis and management of methylmalonic and propionic acidemia. Orphanet Journal of Rare Diseases, 2014, 9, 130.	1.2	482
26	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
27	Clinical presentation and outcome in a series of 88 patients with the cblC defect. Journal of Inherited Metabolic Disease, 2014, 37, 831-840.	1.7	133
28	Brain damage in methylmalonic aciduria: 2-methylcitrate induces cerebral ammonium accumulation and apoptosis in 3D organotypic brain cell cultures. Orphanet Journal of Rare Diseases, 2013, 8, 4.	1.2	23
29	Ammonium Accumulation and Cell Death in a Rat 3D Brain Cell Model of Glutaric Aciduria Type I. PLoS ONE, 2013, 8, e53735.	1.1	23
30	Recurrent Dominant Mutations Affecting Two Adjacent Residues in the Motor Domain of the Monomeric Kinesin KIF22 Result in Skeletal Dysplasia and Joint Laxity. American Journal of Human Genetics, 2012, 90, 170.	2.6	0
31	The unsolved puzzle of neuropathogenesis in glutaric aciduria type I. Molecular Genetics and Metabolism, 2011, 104, 425-437.	0.5	53
32	Recurrent Dominant Mutations Affecting Two Adjacent Residues in the Motor Domain of the Monomeric Kinesin KIF22 Result in Skeletal Dysplasia and Joint Laxity. American Journal of Human Genetics, 2011, 89, 767-772.	2.6	31
33	Born at 27 weeks of Gestation with Classical PKU: Challenges of Dietetic Management in a very Preterm Infant. Mental Illness, 2011, 3, e26.	0.8	6
34	Recurrent postpartum cerebral sinus vein thrombosis as a presentation of cystathionine-Î²-synthase deficiency. Thrombosis and Haemostasis, 2010, 103, 871-873.	1.8	14
35	Glucose transporter-1 deficiency syndrome: the expanding clinical and genetic spectrum of a treatable disorder. Brain, 2010, 133, 655-670.	3.7	356
36	Mitochondrial tRNA^{Leu(UUR)} mutation m.3302Aâ€‰>â€‰G presenting as childhoodâ€onset severe myopathy: threshold determination through segregation study. Journal of Inherited Metabolic Disease, 2010, 33, 219-226.	1.7	10

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37	Early neurological impairment and severe anemia in a newborn with Pearson syndrome. European Journal of Pediatrics, 2009, 168, 311-315.	1.3	14
38	Sudden unexpected death in an infant with L-2-hydroxyglutaric aciduria. European Journal of Pediatrics, 2009, 168, 957-962.	1.3	13
39	Mutations in the Heparan-Sulfate Proteoglycan Glypican 6 (GPC6) Impair Endochondral Ossification and Cause Recessive Omodysplasia. American Journal of Human Genetics, 2009, 84, 760-770.	2.6	106
40	Glutâ€1 deficiency syndrome masqueradingâ€¨ as idiopathic generalized epilepsy. Epilepsia, 2008, 49, 1955-1958.	2.6	41
41	Outcome and long-term follow-up of 36 patients with tetrahydrobiopterin deficiency. Molecular Genetics and Metabolism, 2008, 93, 295-305.	0.5	73
42	Clinical, biochemical, and molecular findings in three patients with 3-hydroxyisobutyric aciduria. Molecular Genetics and Metabolism, 2006, 87, 243-248.	0.5	24
43	Spondyloenchondrodysplasia with spasticity, cerebral calcifications, and immune dysregulation: Clinical and radiographic delineation of a pleiotropic disorder. American Journal of Medical Genetics, Part A, 2006, 140A, 541-550.	0.7	58
44	Familial X-linked cardiomyopathy (Danon disease): diagnostic confirmation by mutation analysis of the LAMP2gene. European Journal of Pediatrics, 2005, 164, 509-514.	1.3	43
45	Extended tetrahydrobiopterin loading test in the diagnosis of cofactor-responsive phenylketonuria: A pilot study. Molecular Genetics and Metabolism, 2005, 86, 91-95.	0.5	75
46	Transport, enzymatic activity, and stability of mutant sulfamidase (SGSH) identified in patients with mucopolysaccharidosis type III A. Human Mutation, 2004, 23, 559-566.	1.1	26
47	Plasma tetrahydrobiopterin and its pharmacokinetic following oral administration. Molecular Genetics and Metabolism, 2004, 81, 45-51.	0.5	114
48	Mass Spectrometric Analysis of Human Transferrin in Different Body Fluids. Clinical Chemistry and Laboratory Medicine, 2003, 41, 1580-8.	1.4	35
49	Hydroxylated residues influence desensitization behaviour of recombinant Î±3 glycine receptor channels. Journal of Neurochemistry, 2002, 83, 30-36.	2.1	21