Barbara Plecko

List of Publications by Year
in descending order

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40	3,411	218677	395702
papers	citations	26 h-index	33 g-index

42	42	42	3074
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Consensus guidelines for the diagnosis and management of pyridoxineâ€dependent epilepsy due to Î±â€aminoadipic semialdehyde dehydrogenase deficiency. Journal of Inherited Metabolic Disease, 2021, 44, 178-192.	3.6	47
2	Why child neurologists should have vitamine B6 in their pockets. European Journal of Paediatric Neurology, 2021, 33, A5-A6.	1.6	0
3	Condensation of deltaâ€1â€piperideineâ€6â€carboxylate with orthoâ€aminobenzaldehyde allows its simple, fast, and inexpensive quantification in the urine of patients with antiquitin deficiency. Journal of Inherited Metabolic Disease, 2020, 43, 891-900.	3.6	6
4	LCâ€MS / MS method for the differential diagnosis of treatable early onset inherited metabolic epilepsies. Journal of Inherited Metabolic Disease, 2020, 43, 1102-1111.	3.6	8
5	Vitaminresponsive Enzephalopathien. Springer Reference Medizin, 2020, , 2539-2544.	0.0	0
6	Neonatal Seizuresâ€”Are We there Yet?. Neuropediatrics, 2019, 50, 280-293.	0.6	43
7	Disorders affecting vitamin B₆ metabolism. Journal of Inherited Metabolic Disease, 2019, 42, 629-646.	3.6	143
8	New insights into human lysine degradation pathways with relevance to pyridoxineâ€dependent epilepsy due to antiquitin deficiency. Journal of Inherited Metabolic Disease, 2019, 42, 620-628.	3.6	45
9	Vitaminresponsive Enzephalopathien bei Kindern und Jugendlichen. Springer Reference Medizin, 2019, , 1-6.	0.0	0
10	Antiquitin Deficiency with Adolescent Onset Epilepsy: Molecular Diagnosis in a Mother of Affected Offsprings. Neuropediatrics, 2018, 49, 154-157.	0.6	15
11	Confirmation of mutations inPROSCas a novel cause of vitamin B_₆-dependent epilepsy. Journal of Medical Genetics, 2017, 54, 809-814.	3.2	66
12	Disorders of Vitamin Metabolism. , 2017, , 373-382.		3
13	Disorders of Thiamine and Pyridoxine Metabolism. , 2016, , 401-412.		0
14	The value of plasma vitamin B₆ profiles in early onset epileptic encephalopathies. Journal of Inherited Metabolic Disease, 2016, 39, 733-741.	3.6	19
15	Treatable newborn and infant seizures due to inborn errors of metabolism. Epileptic Disorders, 2015, 17, 229-242.	1.3	31
16	Intragenic deletions of ALDH7A1 in pyridoxine-dependent epilepsy caused by Alu - Alu recombination. Neurology, 2015, 85, 756-762.	1.1	34
17	Positive Outcome following Early Diagnosis and Treatment of Pyridoxal-5â€²-Phosphate Oxidase Deficiency: A Case Report. Neuropediatrics, 2014, 45, 064-068.	0.6	27
18	Epilepsy due to PNPO mutations: genotype, environment and treatment affect presentation and outcome. Brain, 2014, 137, 1350-1360.	7.6	151

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19	Pyridoxine responsiveness in novel mutations of the PNPO gene. Neurology, 2014, 82, 1425-1433.	1.1	100
20	Phenotypic spectrum of eleven patients and five novel MTFMT mutations identified by exome sequencing and candidate gene screening. Molecular Genetics and Metabolism, 2014, 111, 342-352.	1.1	65
21	Lysine-Restricted Diet as Adjunct Therapy for Pyridoxine-Dependent Epilepsy: The PDE Consortium Consensus Recommendations. JIMD Reports, 2014, 15, 1-11.	1.5	37
22	Occasional seizures, epilepsy, and inborn errors of metabolism. Lancet Neurology, The, 2014, 13, 727-739.	10.2	41
23	Vitamin B6-Dependent and Responsive Disorders. , 2014, , 179-190.		2
24	Neurometabolische und neurodegenerative Erkrankungen. , 2014, , 1690-1720.		0
25	Pyridoxine and pyridoxalphosphate-dependent epilepsies. Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2013, 113, 1811-1817.	1.8	23
26	Molecular diagnosis in mitochondrial complex I deficiency using exome sequencing. Journal of Medical Genetics, 2012, 49, 277-283.	3.2	182
27	Pyridoxine dependent epilepsy and antiquitin deficiency. Molecular Genetics and Metabolism, 2011, 104, 48-60.	1.1	258
28	Status epilepticus in a neonate treated with pyridoxine because of a familial recurrence risk for antiquitin deficiency: pyridoxine toxicity?. Developmental Medicine and Child Neurology, 2011, 53, 1150-1153.	2.1	27
29	Enzyme replacement therapy for mucopolysaccharidosis VI: evaluation of longâ€term pulmonary function in patients treated with recombinant human Nâ€acetylgalactosamine 4â€sulfatase. Journal of Inherited Metabolic Disease, 2010, 33, 51-60.	3.6	80
30	Enzyme replacement therapy for mucopolysaccharidosis VI: Growth and pubertal development in patients treated with recombinant human N-acetylgalactosamine 4-sulfatase. Journal of Pediatric Rehabilitation Medicine, 2010, 3, 89-100.	0.5	58
31	Folinic acidâ€“responsive seizures are identical to pyridoxineâ€dependent epilepsy. Annals of Neurology, 2009, 65, 550-556.	5.3	170
32	Mutation detection in DNA isolated from cerebrospinal fluid and urine: Clinical utility and pitfalls of multiple displacement amplification. Molecular Genetics and Metabolism, 2009, 97, 312-314.	1.1	3
33	Long-term follow-up of endurance and safety outcomes during enzyme replacement therapy for mucopolysaccharidosis VI: Final results of three clinical studies of recombinant human N-acetylgalactosamine 4-sulfatase. Molecular Genetics and Metabolism, 2008, 94, 469-475.	1.1	198
34	Biochemical and molecular characterization of 18 patients with pyridoxine-dependent epilepsy and mutations of the antiquitin (ALDH7A1) gene. Human Mutation, 2007, 28, 19-26.	2.5	158
35	Mutations in antiquitin in individuals with pyridoxine-dependent seizures. Nature Medicine, 2006, 12, 307-309.	30.7	476
36	Enzyme replacement therapy in mucopolysaccharidosis VI (Maroteaux-Lamy syndrome). Journal of Pediatrics, 2004, 144, 574-580.	1.8	267

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37	Neutrophils in Barth syndrome (BTHS) avidly bind annexin-V in the absence of apoptosis. Blood, 2004, 103, 3915-3923.	1.4	93
38	Cardiolipin deficiency in x-linked cardioskeletal myopathy and neutropenia (barth syndrome, mim) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50	1.8	97
39	Pipecolic acid elevation in plasma and cerebrospinal fluid of two patients with pyridoxine-dependent epilepsy. Annals of Neurology, 2000, 48, 121-125.	5.3	86
40	Defective Remodeling of Cardiolipin and Phosphatidylglycerol in Barth Syndrome. Biochemical and Biophysical Research Communications, 2000, 279, 378-382.	2.1	352