Bwee Tien Poll-The

List of Publications by Year
in descending order

108 papers	5,718 citations	101384 36 h-index	82410 72 g-index
111 all docs	111 docs citations	111 times ranked	6637 citing authors

#	Article	IF	Citations
1	Mutations in MVK, encoding mevalonate kinase, cause hyperimmunoglobulinaemia D and periodic fever syndrome. Nature Genetics, 1999, 22, 175-177.	9.4	480
2	X-linked adrenoleukodystrophy (X-ALD): clinical presentation and guidelines for diagnosis, follow-up and management. Orphanet Journal of Rare Diseases, 2012, 7, 51.	1.2	403
3	Zellweger spectrum disorders: clinical overview and management approach. Orphanet Journal of Rare Diseases, 2015, 10, 151.	1.2	286
4	Smith-Lemli-Opitz Syndrome Is Caused by Mutations in the 7-Dehydrocholesterol Reductase Gene. American Journal of Human Genetics, 1998, 63, 329-338.	2.6	271
5	tRNA splicing endonuclease mutations cause pontocerebellar hypoplasia. Nature Genetics, 2008, 40, 1113-1118.	9.4	217
6	Clinical, neuroradiological and genetic findings in pontocerebellar hypoplasia. Brain, 2011, 134, 143-156.	3.7	200
7	Clinical and biochemical spectrum of D-bifunctional protein deficiency. Annals of Neurology, 2006, 59, 92-104.	2.8	175
8	Classification, diagnosis and potential mechanisms in Pontocerebellar Hypoplasia. Orphanet Journal of Rare Diseases, 2011, 6, 50.	1.2	149
9	Pyridoxine-Dependent Epilepsy: An Expanding Clinical Spectrum. Pediatric Neurology, 2016, 59, 6-12.	1.0	136
10	The enigmatic role of tafazzin in cardiolipin metabolism. Biochimica Et Biophysica Acta - Biomembranes, 2009, 1788, 2003-2014.	1.4	135
11	Pristanic acid and phytanic acid: naturally occurring ligands for the nuclear receptor peroxisome proliferator-activated receptor Î±. Journal of Lipid Research, 2000, 41, 1801-1807.	2.0	133
12	X-Linked Adrenoleukodystrophy: Pathogenesis and Treatment. Current Neurology and Neuroscience Reports, 2014, 14, 486.	2.0	127
13	Sepiapterin reductase deficiency: A Treatable Mimic of Cerebral Palsy. Annals of Neurology, 2012, 71, 520-530.	2.8	125
14	Cardiolipin and monolysocardiolipin analysis in fibroblasts, lymphocytes, and tissues using high-performance liquid chromatographyâ€“mass spectrometry as a diagnostic test for Barth syndrome. Analytical Biochemistry, 2009, 387, 230-237.	1.1	120
15	2-Methyl-3-Hydroxybutyryl-CoA Dehydrogenase Deficiency Is Caused by Mutations in the HADH2 Gene. American Journal of Human Genetics, 2003, 72, 1300-1307.	2.6	116
16	Ataxia with loss of Purkinje cells in a mouse model for Refsum disease. Proceedings of the National Academy of Sciences of the United States of America, 2008, 105, 17712-17717.	3.3	108
17	Molecular Characterization of 3-Phosphoglycerate Dehydrogenase Deficiencyâ€”a Neurometabolic Disorder Associated with Reduced L-Serine Biosynthesis. American Journal of Human Genetics, 2000, 67, 1389-1399.	2.6	104
18	Pontine tegmental cap dysplasia: a novel brain malformation with a defect in axonal guidance. Brain, 2007, 130, 2258-2266.	3.7	104

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19	Whatâ€™s new in pontocerebellar hypoplasia? An update on genes and subtypes. Orphanet Journal of Rare Diseases, 2018, 13, 92.	1.2	101
20	Organization of the mevalonate kinase (MVK) gene and identification of novel mutations causing mevalonic aciduria and hyperimmunoglobulinaemia D and periodic fever syndrome. European Journal of Human Genetics, 2001, 9, 253-259.	1.4	85
21	Beneficial effects ofL-serine and glycine in the management of seizures in 3-phosphoglycerate dehydrogenase deficiency. Annals of Neurology, 1998, 44, 261-265.	2.8	84
22	Peroxisome biogenesis disorders with prolonged survival: Phenotypic expression in a cohort of 31 patients. American Journal of Medical Genetics Part A, 2004, 126A, 333-338.	2.4	77
23	EXOSC3 mutations in pontocerebellar hypoplasia type 1: novel mutations and genotype-phenotype correlations. Orphanet Journal of Rare Diseases, 2014, 9, 23.	1.2	75
24	Zellweger spectrum disorders: clinical manifestations in patients surviving into adulthood. Journal of Inherited Metabolic Disease, 2016, 39, 93-106.	1.7	73
25	Clinical diagnosis, biochemical findings and MRI spectrum of peroxisomal disorders. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2012, 1822, 1421-1429.	1.8	69
26	Sepiapterin reductase deficiency an autosomal recessive DOPA-responsive dystonia. Molecular Genetics and Metabolism, 2006, 89, 116-120.	0.5	61
27	Lovastatin in X-Linked Adrenoleukodystrophy. New England Journal of Medicine, 2010, 362, 276-277.	13.9	58
28	Disease progression in women with X-linked adrenoleukodystrophy is slow. Orphanet Journal of Rare Diseases, 2019, 14, 30.	1.2	58
29	Hematopoietic cell transplantation does not prevent myelopathy in Xâ€linked adrenoleukodystrophy: a retrospective study. Journal of Inherited Metabolic Disease, 2015, 38, 359-361.	1.7	54
30	The EEG response to pyridoxineâ€IV neither identifies nor excludes pyridoxineâ€dependent epilepsy. Epilepsia, 2010, 51, 2406-2411.	2.6	53
31	The neurology of rhizomelic chondrodysplasia punctata. Orphanet Journal of Rare Diseases, 2013, 8, 174.	1.2	49
32	Pediatric Traumatic Brain Injury and Attention Deficit. Pediatrics, 2015, 136, 534-541.	1.0	47
33	Congenital heart defects in spinal muscular atrophy type I: A clinical report of two siblings and a review of the literature. American Journal of Medical Genetics, Part A, 2008, 146A, 740-744.	0.7	43
34	TSEN54 mutations cause pontocerebellar hypoplasia type 5. European Journal of Human Genetics, 2011, 19, 724-726.	1.4	43
35	Genetic basis of hyperlysinemia. Orphanet Journal of Rare Diseases, 2013, 8, 57.	1.2	41
36	Clinical and Biochemical Pitfalls in the Diagnosis of Peroxisomal Disorders. Neuropediatrics, 2016, 47, 205-220.	0.3	41

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37	Bezafibrate lowers very longâ€chain fatty acids in Xâ€linked adrenoleukodystrophy fibroblasts by inhibiting fatty acid elongation. Journal of Inherited Metabolic Disease, 2012, 35, 1137-1145.	1.7	39
38	Respiratory Disturbances in Rett Syndrome. Journal of Child Neurology, 2012, 27, 888-892.	0.7	38
39	Relevance of neuroimaging for neurocognitive and behavioral outcome after pediatric traumatic brain injury. Brain Imaging and Behavior, 2018, 12, 29-43.	1.1	38
40	Carnitine-acylcarnitine translocase deficiency: phenotype, residual enzyme activity and outcome. European Journal of Pediatrics, 2001, 160, 101-104.	1.3	37
41	Cholic acid therapy in Zellweger spectrum disorders. Journal of Inherited Metabolic Disease, 2016, 39, 859-868.	1.7	37
42	Evaluation of C26:0â€lysophosphatidylcholine and C26:0â€carnitine as diagnostic markers for Zellweger spectrum disorders. Journal of Inherited Metabolic Disease, 2017, 40, 875-881.	1.7	37
43	Mutation detection in the aspartoacylase gene in 17 patients with Canavan disease: four new mutations in the non-Jewish population. European Journal of Human Genetics, 2000, 8, 557-560.	1.4	34
44	â€œRole of peroxisomes in human lipid metabolism and its importance for neurological developmentâ€ Neuroscience Letters, 2017, 637, 11-17.	1.0	34
45	High prevalence of primary adrenal insufficiency in Zellweger spectrum disorders. Orphanet Journal of Rare Diseases, 2014, 9, 133.	1.2	33
46	A de novo missense mutation in the inositol 1,4,5â€triphosphate receptor type 1 gene causing severe pontine and cerebellar hypoplasia: Expanding the phenotype of ITPR1â€related spinocerebellar ataxia's. American Journal of Medical Genetics, Part A, 2017, 173, 207-212.	0.7	32
47	Biallelic loss of function variants in COASY cause prenatal onset pontocerebellar hypoplasia, microcephaly, and arthrogryposis. European Journal of Human Genetics, 2018, 26, 1752-1758.	1.4	32
48	Neurologic Abnormalities in HIV-1 Infected Children in the Era of Combination Antiretroviral Therapy. PLoS ONE, 2013, 8, e64398.	1.1	31
49	The eye as a window to inborn errors of metabolism. Journal of Inherited Metabolic Disease, 2003, 26, 229-244.	1.7	30
50	Arginine improves peroxisome functioning in cells from patients with a mild peroxisome biogenesis disorder. Orphanet Journal of Rare Diseases, 2013, 8, 138.	1.2	30
51	The structural connectome of children with traumatic brain injury. Human Brain Mapping, 2017, 38, 3603-3614.	1.9	30
52	Quantification of SMN protein in leucocytes from spinal muscular atrophy patients: effects of treatment with valproic acid. Journal of Neurology, Neurosurgery and Psychiatry, 2011, 82, 850-852.	0.9	29
53	Spastic diplegia and periventricular white matter abnormalities in 2-methyl-3-hydroxybutyryl-CoA dehydrogenase deficiency, a defect of isoleucine metabolism: differential diagnosis with hypoxicâ€“ischemic brain diseases. Molecular Genetics and Metabolism, 2004, 81, 295-299.	0.5	28
54	Renal Fanconi syndrome with ultrastructural defects in lysinuric protein intolerance. Journal of Inherited Metabolic Disease, 2007, 30, 402-403.	1.7	28

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55	Clinical, Biomarker, and Molecular Delineations and Genotype-Phenotype Correlations of Ataxia With Oculomotor Apraxia Type 1. JAMA Neurology, 2018, 75, 495.	4.5	28
56	Identification of three patients with a very mild form of Smith-Lemli-Opitz syndrome. American Journal of Medical Genetics Part A, 2003, 122A, 24-29.	2.4	27
57	Bezafibrate for X-Linked Adrenoleukodystrophy. PLoS ONE, 2012, 7, e41013.	1.1	26
58	Infantile hypophosphatasia without bone deformities presenting with severe pyridoxine-resistant seizures. Molecular Genetics and Metabolism, 2014, 111, 404-407.	0.5	26
59	Endocrine and Growth Abnormalities in 4H Leukodystrophy Caused by Variants in POLR3A, POLR3B, and POLR1C. Journal of Clinical Endocrinology and Metabolism, 2021, 106, e660-e674.	1.8	26
60	Clinical and Laboratory Diagnosis of Peroxisomal Disorders. Methods in Molecular Biology, 2017, 1595, 329-342.	0.4	24
61	Mitochondrial Encephalopathy and Transient 3-Methylglutaconic Aciduria in ECHS1 Deficiency: Long-Term Follow-Up. JIMD Reports, 2017, 39, 83-87.	0.7	23
62	Phytanoyl-CoA hydroxylase activity is induced by phytanic acid. FEBS Journal, 2000, 267, 4063-4067.	0.2	22
63	Ectopic peripontine arcuate fibres, a novel finding in pontine tegmental cap dysplasia. European Journal of Paediatric Neurology, 2014, 18, 434-438.	0.7	22
64	High incidence of hyperoxaluria in generalized peroxisomal disorders. Molecular Genetics and Metabolism, 2006, 88, 346-350.	0.5	20
65	Rhizomelic chondrodysplasia punctata and cardiac pathology. Journal of Medical Genetics, 2013, 50, 419-424.	1.5	20
66	Non-motor symptoms and quality of life in dopa-responsive dystonia patients. Parkinsonism and Related Disorders, 2017, 45, 57-62.	1.1	19
67	Brain atrophy following hemiplegic migraine attacks. Cephalalgia, 2018, 38, 1199-1202.	1.8	19
68	Clinical, biochemical, and molecular characterization of mild (nonclassic) rhizomelic chondrodysplasia punctata. Journal of Inherited Metabolic Disease, 2021, 44, 1021-1038.	1.7	19
69	The cholic acid extension study in Zellweger spectrum disorders: Results and implications for therapy. Journal of Inherited Metabolic Disease, 2019, 42, 303-312.	1.7	18
70	The eye in metabolic diseases: Clues to diagnosis. European Journal of Paediatric Neurology, 2011, 15, 197-204.	0.7	16
71	Clinical and mutational characteristics of spinal muscular atrophy with respiratory distress type 1 in the Netherlands. Neuromuscular Disorders, 2013, 23, 461-468.	0.3	15
72	A SEPSECS mutation in a 23-year-old woman with microcephaly and progressive cerebellar ataxia. Journal of Inherited Metabolic Disease, 2018, 41, 897-898.	1.7	15

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73	Prenatal diagnosis in adenylosuccinate lyase deficiency. , 2000, 20, 33-36.		14
74	RARS2 Mutations: Is Pontocerebellar Hypoplasia Type 6 a Mitochondrial Encephalopathy?. JIMD Reports, 2016, 33, 87-92.	0.7	14
75	Position statement on the role of healthcare professionals, patient organizations and industry in European Reference Networks. Orphanet Journal of Rare Diseases, 2016, 11, 7.	1.2	14
76	Glycerol kinase deficiency: residual activity explained by reduced transcription and enzyme conformation. European Journal of Human Genetics, 2004, 12, 424-432.	1.4	13
77	Skeletal Dysplasia and Myelopathy in Congenital Disorder of Glycosylation Type IA. Journal of Pediatrics, 2006, 148, 115-117.	0.9	12
78	Pediatric traumatic brain injury affects multisensory integration.. Neuropsychology, 2017, 31, 137-148.	1.0	12
79	Liver disease predominates in a mouse model for mild human Zellweger spectrum disorder. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2019, 1865, 2774-2787.	1.8	12
80	Rhabdomyolysis in pontocerebellar hypoplasia type 2 (PCH-2). Neuromuscular Disorders, 2008, 18, 52-58.	0.3	11
81	Sâ€adenosylmethionine and Sâ€adenosylhomocysteine in plasma and cerebrospinal fluid in Rett syndrome and the effect of folinic acid supplementation. Journal of Inherited Metabolic Disease, 2013, 36, 967-972.	1.7	11
82	Development and validation of a severity scoring system for Zellweger spectrum disorders. Clinical Genetics, 2018, 93, 613-621.	1.0	11
83	Rhizomelic chondrodysplasia punctata morbidity and mortality, an update. American Journal of Medical Genetics, Part A, 2020, 182, 579-583.	0.7	10
84	A Rett syndrome patient with a ring X chromosome: further evidence for skewing of X inactivation and heterogeneity in the aetiology of the disease. European Journal of Human Genetics, 2001, 9, 171-177.	1.4	9
85	Hepatic symptoms and histology in 13 patients with a Zellweger spectrum disorder. Journal of Inherited Metabolic Disease, 2019, 42, 955-965.	1.7	9
86	Is hearing loss a feature of Joubert syndrome, a ciliopathy?. International Journal of Pediatric Otorhinolaryngology, 2010, 74, 1034-1038.	0.4	8
87	Peroxisomal Leukoencephalopathy. Seminars in Neurology, 2012, 32, 042-050.	0.5	8
88	Peroxisome Mosaics. Advances in Experimental Medicine and Biology, 2003, 544, 97-106.	0.8	8
89	Coagulopathy in Zellweger spectrum disorders: a role for vitamin K. Journal of Inherited Metabolic Disease, 2018, 41, 249-255.	1.7	7
90	Clinical utility gene card for: Zellweger syndrome spectrum. European Journal of Human Genetics, 2015, 23, 1111-1111.	1.4	6

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91	Optical coherence tomography shows neuroretinal thinning in myelopathy of adrenoleukodystrophy. Journal of Neurology, 2020, 267, 679-687.	1.8	6
92	Postnatal Brain Growth Patterns in Pontocerebellar Hypoplasia. Neuropediatrics, 2021, 52, 163-169.	0.3	5
93	Peroxisomal disorders. Neuroscience Research Communications, 1998, 22, 63-71.	0.2	4
94	Impaired Visual Integration in Children with Traumatic Brain Injury: An Observational Study. PLoS ONE, 2015, 10, e0144395.	1.1	4
95	Hyperinsulinism in a patient with a Zellweger Spectrum Disorder and a 16p11.2 deletion syndrome. Molecular Genetics and Metabolism Reports, 2020, 23, 100590.	0.4	4
96	Strokelike Episodes and Cutis Marmorata Telangiectatica Congenita. Journal of Child Neurology, 2015, 30, 129-132.	0.7	3
97	Oral Cholic Acid in Zellweger Spectrum Disorders. Journal of Pediatric Gastroenterology and Nutrition, 2018, 66, e57.	0.9	3
98	Phytanic and Pristanic Acid Are Naturally Occuring Ligands. Advances in Experimental Medicine and Biology, 2003, 544, 247-254.	0.8	3
99	Severe neurological complications in skeletal dysplasias: Two case reports. European Journal of Paediatric Neurology, 2006, 10, 241-243.	0.7	2
100	The cholic acid extension study in Zellweger spectrum disorders: results and implications for therapy. Journal of Inherited Metabolic Disease, 2018, , .	1.7	2
101	Biochemical Markers Predicting Survival in Peroxisome Biogenesis Disorders. Advances in Experimental Medicine and Biology, 2003, 544, 67-68.	0.8	2
102	Peroxisomal Disorders. , 2012, , 591-605.		2
103	Reply: Low bone mineral density is a common feature of Zellweger spectrum disorders. Molecular Genetics and Metabolism Reports, 2017, 11, 94.	0.4	1
104	Peroxisomal Disorders. , 2014, , 375-397.		1
105	Peroxisomal Disorders. , 2006, , 509-522.		1
106	Inborn Errors of Non-Mitochondrial Fatty Acid Metabolism Including Peroxisomal Disorders. , 2016, , 591-606.		1
107	Chemical chaperones improve peroxisomal biogenesis in fibroblasts of mild Zellweger syndrome spectrum patients. Tijdschrift Voor Kindergeneeskunde, 2013, 81, 21-21.	0.0	0
108	Comment on the paper â€œEffect of statin treatment on adrenomyeloneuropathy with cerebral inflammation: A revisitâ€• Clinical Neurology and Neurosurgery, 2013, 115, 2401-2402.	0.6	0