

# Bwee Tien Poll-The

## List of Publications by Year in descending order

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108  
papers

5,718  
citations

101543

36  
h-index

82547

72  
g-index

111  
all docs

111  
docs citations

111  
times ranked

6637  
citing authors

#	ARTICLE	IF	CITATIONS
1	Endocrine and Growth Abnormalities in 4H Leukodystrophy Caused by Variants in <i>POLR3A</i> , <i>POLR3B</i> , and <i>POLR1C</i> . <i>Journal of Clinical Endocrinology and Metabolism</i> , 2021, 106, e660-e674.	3.6	26
2	Clinical, biochemical, and molecular characterization of mild (nonclassic) rhizomelic chondrodysplasia punctata. <i>Journal of Inherited Metabolic Disease</i> , 2021, 44, 1021-1038.	3.6	19
3	Postnatal Brain Growth Patterns in Pontocerebellar Hypoplasia. <i>Neuropediatrics</i> , 2021, 52, 163-169.	0.6	5
4	Optical coherence tomography shows neuroretinal thinning in myelopathy of adrenoleukodystrophy. <i>Journal of Neurology</i> , 2020, 267, 679-687.	3.6	6
5	Rhizomelic chondrodysplasia punctata morbidity and mortality, an update. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 579-583.	1.2	10
6	Hyperinsulinism in a patient with a Zellweger Spectrum Disorder and a 16p11.2 deletion syndrome. <i>Molecular Genetics and Metabolism Reports</i> , 2020, 23, 100590.	1.1	4
7	Liver disease predominates in a mouse model for mild human Zellweger spectrum disorder. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2019, 1865, 2774-2787.	3.8	12
8	Hepatic symptoms and histology in 13 patients with a Zellweger spectrum disorder. <i>Journal of Inherited Metabolic Disease</i> , 2019, 42, 955-965.	3.6	9
9	The cholic acid extension study in Zellweger spectrum disorders: Results and implications for therapy. <i>Journal of Inherited Metabolic Disease</i> , 2019, 42, 303-312.	3.6	18
10	Disease progression in women with X-linked adrenoleukodystrophy is slow. <i>Orphanet Journal of Rare Diseases</i> , 2019, 14, 30.	2.7	58
11	A SEPSECS mutation in a 23-year-old woman with microcephaly and progressive cerebellar ataxia. <i>Journal of Inherited Metabolic Disease</i> , 2018, 41, 897-898.	3.6	15
12	Clinical, Biomarker, and Molecular Delineations and Genotype-Phenotype Correlations of Ataxia With Oculomotor Apraxia Type 1. <i>JAMA Neurology</i> , 2018, 75, 495.	9.0	28
13	Relevance of neuroimaging for neurocognitive and behavioral outcome after pediatric traumatic brain injury. <i>Brain Imaging and Behavior</i> , 2018, 12, 29-43.	2.1	38
14	Oral Cholic Acid in Zellweger Spectrum Disorders. <i>Journal of Pediatric Gastroenterology and Nutrition</i> , 2018, 66, e57.	1.8	3
15	Development and validation of a severity scoring system for Zellweger spectrum disorders. <i>Clinical Genetics</i> , 2018, 93, 613-621.	2.0	11
16	Coagulopathy in Zellweger spectrum disorders: a role for vitamin K. <i>Journal of Inherited Metabolic Disease</i> , 2018, 41, 249-255.	3.6	7
17	Brain atrophy following hemiplegic migraine attacks. <i>Cephalalgia</i> , 2018, 38, 1199-1202.	3.9	19
18	The cholic acid extension study in Zellweger spectrum disorders: results and implications for therapy. <i>Journal of Inherited Metabolic Disease</i> , 2018, , .	3.6	2

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19	What's new in pontocerebellar hypoplasia? An update on genes and subtypes. Orphanet Journal of Rare Diseases, 2018, 13, 92.	2.7	101
20	Biallelic loss of function variants in COASY cause prenatal onset pontocerebellar hypoplasia, microcephaly, and arthrogryposis. European Journal of Human Genetics, 2018, 26, 1752-1758.	2.8	32
21	Role of peroxisomes in human lipid metabolism and its importance for neurological development. Neuroscience Letters, 2017, 637, 11-17.	2.1	34
22	Reply: Low bone mineral density is a common feature of Zellweger spectrum disorders. Molecular Genetics and Metabolism Reports, 2017, 11, 94.	1.1	1
23	The structural connectome of children with traumatic brain injury. Human Brain Mapping, 2017, 38, 3603-3614.	3.6	30
24	Clinical and Laboratory Diagnosis of Peroxisomal Disorders. Methods in Molecular Biology, 2017, 1595, 329-342.	0.9	24
25	Non-motor symptoms and quality of life in dopa-responsive dystonia patients. Parkinsonism and Related Disorders, 2017, 45, 57-62.	2.2	19
26	Mitochondrial Encephalopathy and Transient 3-Methylglutaconic Aciduria in ECHS1 Deficiency: Long-Term Follow-Up. JIMD Reports, 2017, 39, 83-87.	1.5	23
27	Evaluation of C26:0 lysophosphatidylcholine and C26:0 carnitine as diagnostic markers for Zellweger spectrum disorders. Journal of Inherited Metabolic Disease, 2017, 40, 875-881.	3.6	37
28	A de novo missense mutation in the inositol 1,4,5-triphosphate receptor type 1 gene causing severe pontine and cerebellar hypoplasia: Expanding the phenotype of <i>ITPR1</i> -related spinocerebellar ataxia's. American Journal of Medical Genetics, Part A, 2017, 173, 207-212.	1.2	32
29	Pediatric traumatic brain injury affects multisensory integration.. Neuropsychology, 2017, 31, 137-148.	1.3	12
30	Clinical and Biochemical Pitfalls in the Diagnosis of Peroxisomal Disorders. Neuropediatrics, 2016, 47, 205-220.	0.6	41
31	RARS2 Mutations: Is Pontocerebellar Hypoplasia Type 6 a Mitochondrial Encephalopathy?. JIMD Reports, 2016, 33, 87-92.	1.5	14
32	Cholic acid therapy in Zellweger spectrum disorders. Journal of Inherited Metabolic Disease, 2016, 39, 859-868.	3.6	37
33	Position statement on the role of healthcare professionals, patient organizations and industry in European Reference Networks. Orphanet Journal of Rare Diseases, 2016, 11, 7.	2.7	14
34	Pyridoxine-Dependent Epilepsy: An Expanding Clinical Spectrum. Pediatric Neurology, 2016, 59, 6-12.	2.1	136
35	Zellweger spectrum disorders: clinical manifestations in patients surviving into adulthood. Journal of Inherited Metabolic Disease, 2016, 39, 93-106.	3.6	73
36	Inborn Errors of Non-Mitochondrial Fatty Acid Metabolism Including Peroxisomal Disorders. , 2016, , 591-606.		1

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37	Zellweger spectrum disorders: clinical overview and management approach. Orphanet Journal of Rare Diseases, 2015, 10, 151.	2.7	286
38	Impaired Visual Integration in Children with Traumatic Brain Injury: An Observational Study. PLoS ONE, 2015, 10, e0144395.	2.5	4
39	Strokelike Episodes and Cutis Marmorata Telangiectatica Congenita. Journal of Child Neurology, 2015, 30, 129-132.	1.4	3
40	Hematopoietic cell transplantation does not prevent myelopathy in X-linked adrenoleukodystrophy: a retrospective study. Journal of Inherited Metabolic Disease, 2015, 38, 359-361.	3.6	54
41	Pediatric Traumatic Brain Injury and Attention Deficit. Pediatrics, 2015, 136, 534-541.	2.1	47
42	Clinical utility gene card for: Zellweger syndrome spectrum. European Journal of Human Genetics, 2015, 23, 1111-1111.	2.8	6
43	High prevalence of primary adrenal insufficiency in Zellweger spectrum disorders. Orphanet Journal of Rare Diseases, 2014, 9, 133.	2.7	33
44	Ectopic peripontine arcuate fibres, a novel finding in pontine tegmental cap dysplasia. European Journal of Paediatric Neurology, 2014, 18, 434-438.	1.6	22
45	EXOSC3 mutations in pontocerebellar hypoplasia type 1: novel mutations and genotype-phenotype correlations. Orphanet Journal of Rare Diseases, 2014, 9, 23.	2.7	75
46	X-Linked Adrenoleukodystrophy: Pathogenesis and Treatment. Current Neurology and Neuroscience Reports, 2014, 14, 486.	4.2	127
47	Infantile hypophosphatasia without bone deformities presenting with severe pyridoxine-resistant seizures. Molecular Genetics and Metabolism, 2014, 111, 404-407.	1.1	26
48	Peroxisomal Disorders. , 2014, , 375-397.		1
49	Genetic basis of hyperlysinemia. Orphanet Journal of Rare Diseases, 2013, 8, 57.	2.7	41
50	Chemical chaperones improve peroxisomal biogenesis in fibroblasts of mild Zellweger syndrome spectrum patients. Tijdschrift Voor Kindergeneeskunde, 2013, 81, 21-21.	0.0	0
51	S-adenosylmethionine and S-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.	3.6	11
52	Arginine improves peroxisome functioning in cells from patients with a mild peroxisome biogenesis disorder. Orphanet Journal of Rare Diseases, 2013, 8, 138.	2.7	30
53	Clinical and mutational characteristics of spinal muscular atrophy with respiratory distress type 1 in the Netherlands. Neuromuscular Disorders, 2013, 23, 461-468.	0.6	15
54	Comment on the paper "Effect of statin treatment on adrenomyeloneuropathy with cerebral inflammation: A revisit". Clinical Neurology and Neurosurgery, 2013, 115, 2401-2402.	1.4	0

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55	Rhizomelic chondrodysplasia punctata and cardiac pathology. <i>Journal of Medical Genetics</i> , 2013, 50, 419-424.	3.2	20
56	The neurology of rhizomelic chondrodysplasia punctata. <i>Orphanet Journal of Rare Diseases</i> , 2013, 8, 174.	2.7	49
57	Neurologic Abnormalities in HIV-1 Infected Children in the Era of Combination Antiretroviral Therapy. <i>PLoS ONE</i> , 2013, 8, e64398.	2.5	31
58	Respiratory Disturbances in Rett Syndrome. <i>Journal of Child Neurology</i> , 2012, 27, 888-892.	1.4	38
59	Peroxisomal Leukoencephalopathy. <i>Seminars in Neurology</i> , 2012, 32, 042-050.	1.4	8
60	Bezafibrate lowers very long-chain fatty acids in X-linked adrenoleukodystrophy fibroblasts by inhibiting fatty acid elongation. <i>Journal of Inherited Metabolic Disease</i> , 2012, 35, 1137-1145.	3.6	39
61	Clinical diagnosis, biochemical findings and MRI spectrum of peroxisomal disorders. <i>Biochimica Et Biophysica Acta - Molecular Basis of Disease</i> , 2012, 1822, 1421-1429.	3.8	69
62	X-linked adrenoleukodystrophy (X-ALD): clinical presentation and guidelines for diagnosis, follow-up and management. <i>Orphanet Journal of Rare Diseases</i> , 2012, 7, 51.	2.7	403
63	Bezafibrate for X-Linked Adrenoleukodystrophy. <i>PLoS ONE</i> , 2012, 7, e41013.	2.5	26
64	Sepiapterin reductase deficiency: A Treatable Mimic of Cerebral Palsy. <i>Annals of Neurology</i> , 2012, 71, 520-530.	5.3	125
65	Peroxisomal Disorders. , 2012, , 591-605.		2
66	TSEN54 mutations cause pontocerebellar hypoplasia type 5. <i>European Journal of Human Genetics</i> , 2011, 19, 724-726.	2.8	43
67	The eye in metabolic diseases: Clues to diagnosis. <i>European Journal of Paediatric Neurology</i> , 2011, 15, 197-204.	1.6	16
68	Classification, diagnosis and potential mechanisms in Pontocerebellar Hypoplasia. <i>Orphanet Journal of Rare Diseases</i> , 2011, 6, 50.	2.7	149
69	Clinical, neuroradiological and genetic findings in pontocerebellar hypoplasia. <i>Brain</i> , 2011, 134, 143-156.	7.6	200
70	Quantification of SMN protein in leucocytes from spinal muscular atrophy patients: effects of treatment with valproic acid. <i>Journal of Neurology, Neurosurgery and Psychiatry</i> , 2011, 82, 850-852.	1.9	29
71	The EEG response to pyridoxine neither identifies nor excludes pyridoxine-dependent epilepsy. <i>Epilepsia</i> , 2010, 51, 2406-2411.	5.1	53
72	Lovastatin in X-Linked Adrenoleukodystrophy. <i>New England Journal of Medicine</i> , 2010, 362, 276-277.	27.0	58

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73	Is hearing loss a feature of Joubert syndrome, a ciliopathy?. <i>International Journal of Pediatric Otorhinolaryngology</i> , 2010, 74, 1034-1038.	1.0	8
74	Cardiolipin and monolysocardiolipin analysis in fibroblasts, lymphocytes, and tissues using high-performance liquid chromatography-mass spectrometry as a diagnostic test for Barth syndrome. <i>Analytical Biochemistry</i> , 2009, 387, 230-237.	2.4	120
75	The enigmatic role of tafazzin in cardiolipin metabolism. <i>Biochimica Et Biophysica Acta - Biomembranes</i> , 2009, 1788, 2003-2014.	2.6	135
76	Congenital heart defects in spinal muscular atrophy type I: A clinical report of two siblings and a review of the literature. <i>American Journal of Medical Genetics, Part A</i> , 2008, 146A, 740-744.	1.2	43
77	tRNA splicing endonuclease mutations cause pontocerebellar hypoplasia. <i>Nature Genetics</i> , 2008, 40, 1113-1118.	21.4	217
78	Rhabdomyolysis in pontocerebellar hypoplasia type 2 (PCH-2). <i>Neuromuscular Disorders</i> , 2008, 18, 52-58.	0.6	11
79	Ataxia with loss of Purkinje cells in a mouse model for Refsum disease. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2008, 105, 17712-17717.	7.1	108
80	Pontine tegmental cap dysplasia: a novel brain malformation with a defect in axonal guidance. <i>Brain</i> , 2007, 130, 2258-2266.	7.6	104
81	Renal Fanconi syndrome with ultrastructural defects in lysinuric protein intolerance. <i>Journal of Inherited Metabolic Disease</i> , 2007, 30, 402-403.	3.6	28
82	Skeletal Dysplasia and Myelopathy in Congenital Disorder of Glycosylation Type IA. <i>Journal of Pediatrics</i> , 2006, 148, 115-117.	1.8	12
83	High incidence of hyperoxaluria in generalized peroxisomal disorders. <i>Molecular Genetics and Metabolism</i> , 2006, 88, 346-350.	1.1	20
84	Sepiapterin reductase deficiency an autosomal recessive DOPA-responsive dystonia. <i>Molecular Genetics and Metabolism</i> , 2006, 89, 116-120.	1.1	61
85	Severe neurological complications in skeletal dysplasias: Two case reports. <i>European Journal of Paediatric Neurology</i> , 2006, 10, 241-243.	1.6	2
86	Clinical and biochemical spectrum of D-bifunctional protein deficiency. <i>Annals of Neurology</i> , 2006, 59, 92-104.	5.3	175
87	<i>Peroxisomal Disorders</i> , 2006, , 509-522.		1
88	Glycerol kinase deficiency: residual activity explained by reduced transcription and enzyme conformation. <i>European Journal of Human Genetics</i> , 2004, 12, 424-432.	2.8	13
89	Peroxisome biogenesis disorders with prolonged survival: Phenotypic expression in a cohort of 31 patients. <i>American Journal of Medical Genetics Part A</i> , 2004, 126A, 333-338.	2.4	77
90	Spastic diplegia and periventricular white matter abnormalities in 2-methyl-3-hydroxybutyryl-CoA dehydrogenase deficiency, a defect of isoleucine metabolism: differential diagnosis with hypoxic-ischemic brain diseases. <i>Molecular Genetics and Metabolism</i> , 2004, 81, 295-299.	1.1	28

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91	The eye as a window to inborn errors of metabolism. <i>Journal of Inherited Metabolic Disease</i> , 2003, 26, 229-244.	3.6	30
92	Identification of three patients with a very mild form of Smith-Lemli-Opitz syndrome. <i>American Journal of Medical Genetics Part A</i> , 2003, 122A, 24-29.	2.4	27
93	2-Methyl-3-Hydroxybutyryl-CoA Dehydrogenase Deficiency Is Caused by Mutations in the HADH2 Gene. <i>American Journal of Human Genetics</i> , 2003, 72, 1300-1307.	6.2	116
94	Peroxisome Mosaics. <i>Advances in Experimental Medicine and Biology</i> , 2003, 544, 97-106.	1.6	8
95	Phytanic and Pristanic Acid Are Naturally Occuring Ligands. <i>Advances in Experimental Medicine and Biology</i> , 2003, 544, 247-254.	1.6	3
96	Biochemical Markers Predicting Survival in Peroxisome Biogenesis Disorders. <i>Advances in Experimental Medicine and Biology</i> , 2003, 544, 67-68.	1.6	2
97	Carnitine-acylcarnitine translocase deficiency: phenotype, residual enzyme activity and outcome. <i>European Journal of Pediatrics</i> , 2001, 160, 101-104.	2.7	37
98	Organization of the mevalonate kinase (MVK) gene and identification of novel mutations causing mevalonic aciduria and hyperimmunoglobulinaemia D and periodic fever syndrome. <i>European Journal of Human Genetics</i> , 2001, 9, 253-259.	2.8	85
99	A Rett syndrome patient with a ring X chromosome: further evidence for skewing of X inactivation and heterogeneity in the aetiology of the disease. <i>European Journal of Human Genetics</i> , 2001, 9, 171-177.	2.8	9
100	Prenatal diagnosis in adenylosuccinate lyase deficiency. , 2000, 20, 33-36.		14
101	Phytanoyl-CoA hydroxylase activity is induced by phytanic acid. <i>FEBS Journal</i> , 2000, 267, 4063-4067.	0.2	22
102	Mutation detection in the aspartoacylase gene in 17 patients with Canavan disease: four new mutations in the non-Jewish population. <i>European Journal of Human Genetics</i> , 2000, 8, 557-560.	2.8	34
103	Molecular Characterization of 3-Phosphoglycerate Dehydrogenase Deficiencyâ€”a Neurometabolic Disorder Associated with Reduced L-Serine Biosynthesis. <i>American Journal of Human Genetics</i> , 2000, 67, 1389-1399.	6.2	104
104	Pristanic acid and phytanic acid: naturally occurring ligands for the nuclear receptor peroxisome proliferator-activated receptor Î±. <i>Journal of Lipid Research</i> , 2000, 41, 1801-1807.	4.2	133
105	Mutations in MVK, encoding mevalonate kinase, cause hyperimmunoglobulinaemia D and periodic fever syndrome. <i>Nature Genetics</i> , 1999, 22, 175-177.	21.4	480
106	Beneficial effects of L-serine and glycine in the management of seizures in 3-phosphoglycerate dehydrogenase deficiency. <i>Annals of Neurology</i> , 1998, 44, 261-265.	5.3	84
107	Peroxisomal disorders. <i>Neuroscience Research Communications</i> , 1998, 22, 63-71.	0.2	4
108	Smith-Lemli-Opitz Syndrome Is Caused by Mutations in the 7-Dehydrocholesterol Reductase Gene. <i>American Journal of Human Genetics</i> , 1998, 63, 329-338.	6.2	271