Stacey Kiat-Hong Tay

List of Publications by Year
in descending order

42 papers	1,651 citations	304368 22 h-index	315357 38 g-index
43 all docs	43 docs citations	43 times ranked	2694 citing authors

#	Article	IF	Citations
1	Clinical and biochemical features of aromatic L-amino acid decarboxylase deficiency. Neurology, 2010, 75, 64-71.	1.5	198
2	Fatal infantile neuromuscular presentation of glycogen storage disease type IV. Neuromuscular Disorders, 2004, 14, 253-260.	0.3	185
3	Nonconvulsive Status Epilepticus in Children: Clinical and EEG Characteristics. Epilepsia, 2006, 47, 1504-1509.	2.6	149
4	CISHand Susceptibility to Infectious Diseases. New England Journal of Medicine, 2010, 362, 2092-2101.	13.9	129
5	The Cerebral Vasculopathy of PHACES Syndrome. Stroke, 2008, 39, 308-316.	1.0	108
6	Spinal Muscular Atrophy: From Gene Discovery to Clinical Trials. Annals of Human Genetics, 2013, 77, 435-463.	0.3	84
7	SIL1 mutations and clinical spectrum in patients with Marinesco-SjÃ¶gren syndrome. Brain, 2013, 136, 3634-3644.	3.7	65
8	Unusually mild phenotype of AADC deficiency in 2 siblings. Molecular Genetics and Metabolism, 2007, 91, 374-378.	0.5	50
9	Mitochondrial Encephalomyopathies: Diagnostic Approach. Annals of the New York Academy of Sciences, 2004, 1011, 217-231.	1.8	49
10	Changes in PDE4D Isoforms in the Hippocampus of a Patient With Advanced Alzheimer Disease. Archives of Neurology, 2007, 64, 456.	4.9	45
11	Impaired Development Of Neural-Crest Cell Derived Organs and Intellectual Disability Caused ByMED13LHaploinsufficiency. Human Mutation, 2014, 35, n/a-n/a.	1.1	43
12	Simplified Molecular Diagnosis of Fragile X Syndrome by Fluorescent Methylation-Specific PCR and GeneScan Analysis. Clinical Chemistry, 2006, 52, 1492-1500.	1.5	42
13	Detection of Chromosomal Breakpoints in Patients with Developmental Delay and Speech Disorders. PLoS ONE, 2014, 9, e90852.	1.1	41
14	Identification of the first COGâ€“CDG patient of Indian origin. Molecular Genetics and Metabolism, 2011, 102, 364-367.	0.5	39
15	Association of Mutations in SCO2, a Cytochrome c Oxidase Assembly Gene, With Early Fetal Lethality. Archives of Neurology, 2004, 61, 950.	4.9	38
16	Generation of a novel mouse model that recapitulates early and adult onset glycogenosis type IV. Human Molecular Genetics, 2011, 20, 4430-4439.	1.4	38
17	Identification and characterization of novel mouse PDE4D isoforms: Molecular cloning, subcellular distribution and detection of isoform-specific intracellular localization signals. Cellular Signalling, 2008, 20, 139-153.	1.7	35
18	Unusual Clinical Presentations in Four Cases of Leigh Disease, Cytochrome C Oxidase Deficiency, and SURF1 Gene Mutations. Journal of Child Neurology, 2005, 20, 670-674.	0.7	32

#	Article	IF	Citations
19	Myotoxicity of Lipid-Lowering Agents in a Teenager With MELAS Mutation. Pediatric Neurology, 2008, 39, 426-428.	1.0	30
20	Mitochondrial D-loop Polymorphisms and Mitochondrial DNA Content in Childhood Acute Lymphoblastic Leukemia. Journal of Pediatric Hematology/Oncology, 2011, 33, e239-e244.	0.3	24
21	Clinical and Biochemical Characterization of a Patient with Congenital Disorder of Glycosylation (CDG) IIx. Journal of Pediatrics, 2005, 147, 851-853.	0.9	23
22	Headache diagnosis, disability and co-morbidities in a multi-ethnic, heterogeneous paediatric Asian population. Cephalalgia, 2010, 30, 953-961.	1.8	23
23	A Novel Splice-Site Mutation in ALS2 Establishes the Diagnosis of Juvenile Amyotrophic Lateral Sclerosis in a Family with Early Onset Anarthria and Generalized Dystonias. PLoS ONE, 2014, 9, e113258.	1.1	22
24	Responsiveness of sphingosine phosphate lyase insufficiency syndrome to vitamin <scp>B6</scp> cofactor supplementation. Journal of Inherited Metabolic Disease, 2020, 43, 1131-1142.	1.7	21
25	Congenital cardiomyopathy and pulmonary hypertension: Another fatal variant of cytochrome-c oxidase deficiency. Journal of Inherited Metabolic Disease, 2004, 27, 735-739.	1.7	20
26	Is Trunk Posture in Walking a Better Marker than Gait Speed in Predicting Decline in Function and Subsequent Frailty?. Journal of the American Medical Directors Association, 2016, 17, 65-70.	1.2	20
27	Congenital or late-onset myopathy in patients with the T14709C mtDNA mutation. Journal of the Neurological Sciences, 2005, 228, 93-97.	0.3	14
28	Identification and molecular characterization of a novel PDE4D11 cAMP-specific phosphodiesterase isoform. Cellular Signalling, 2008, 20, 2247-2255.	1.7	14
29	Enterovirus infections in Singaporean children: an assessment of neurological manifestations and clinical outcomes. Singapore Medical Journal, 2017, 58, 189-195.	0.3	12
30	Novel LRPPRC Mutation in a Boy With Mild Leigh Syndrome, Frenchâ€“Canadian Type Outside of QuÃ©bec. Child Neurology Open, 2017, 4, 2329048X1773763.	0.5	9
31	Acute starvation in C57BL/6J mice increases myocardial UCP2 and UCP3 protein expression levels and decreases mitochondrial bio-energetic function. Stress, 2011, 14, 66-72.	0.8	8
32	Studies of COX16, COX19, and PET191 in Human Cytochrome-c Oxidase Deficiency. Archives of Neurology, 2004, 61, 1935-7.	4.9	7
33	Elevated Thyroid Peroxidase Antibodies with Encephalopathy in MELAS Syndrome. Pediatric Neurology, 2007, 36, 414-417.	1.0	6
34	Managing tuberous sclerosis in the Asia-Pacific region: Refining practice and the role of targeted therapy. Journal of Clinical Neuroscience, 2014, 21, 1180-1187.	0.8	6
35	Atherothrombotic events and clopidogrel therapy. Cmaj, 2007, 176, 349-349.	0.9	5
36	Detection of hemi/homozygotes through heteroduplex formation in high-resolution melting analysis. Analytical Biochemistry, 2011, 410, 158-160.	1.1	4

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37	Displacement of the Beating Heart Induces an Immediate and Sustained Increase in Myocardial Reactive Oxygen Species. Circulation Journal, 2006, 70, 1226-1228.	0.7	3
38	Chronic Phenobarbital-Induced Mitochondrial Pleomorphism in the Rat Liver. Toxicologic Pathology, 2007, 35, 831-833.	0.9	3
39	Simultaneous Screening of the FRAXA and FRAXE Loci for Rapid Detection of FMR1 CGG and/or AFF2 CCG Repeat Expansions by Triplet-Primed PCR. Journal of Molecular Diagnostics, 2021, 23, 941-951.	1.2	3
40	Alternating hemiplegia of childhood: successful treatment with topiramate and flunarizine, a case report. Annals of Tropical Paediatrics, 2011, 31, 149-152.	1.0	2
41	Mitochondrial Changes in Aging Orbicularis Oculi Muscles. Asia-Pacific Journal of Ophthalmology, 2012, 1, 180-184.	1.3	1
42	First pediatric organ donation after circulatory determination of death in Singapore: Facing challenges in the absence of a local practice guideline. Pediatric Transplantation, 2020, 24, e13740.	0.5	1