Poh-San Lai

List of Publications by Year
in descending order

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46	1,039	686830	433756
papers	citations	13 h-index	31 g-index

46	46	46	2108
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Novel Autoantibodies in Idiopathic Small Fiber Neuropathy. Annals of Neurology, 2022, 91, 66-77.	2.8	9
2	Novel frameshift variant in the PCNT gene associated with Microcephalic Osteodysplastic Primordial Dwarfism (MOPD) Type II and small kidneys. BMC Medical Genomics, 2022, 15, 82.	0.7	2
3	Vulnerability and the Ethics of Human Germline Genome Editing. CRISPR Journal, 2022, 5, 358-363.	1.4	2
4	Clinical phenotypes of spinal muscular atrophy patients with hybrid SMN gene. Brain and Development, 2021, 43, 294-302.	0.6	18
5	The orphan nuclear receptor NR0B2 could be a novel susceptibility locus associated with microsatelliteâ€stable, APC mutationâ€negative earlyâ€onset colorectal carcinomas with metabolic manifestation. Genes Chromosomes and Cancer, 2021, 60, 61-72.	1.5	5
6	Singapore Undiagnosed Disease Program: Genomic Analysis aids Diagnosis and Clinical Management. Archives of Disease in Childhood, 2021, 106, 31-37.	1.0	17
7	OUP accepted manuscript. Human Reproduction, 2021, 36, 3018-3027.	0.4	1
8	Effect of semaphorin 3C gene variants in multifactorial Hirschsprung disease. Journal of International Medical Research, 2021, 49, 030006052098778.	0.4	3
9	Dried Blood Spot Screening System for Spinal Muscular Atrophy with Allele-Specific Polymerase Chain Reaction and Melting Peak Analysis. Genetic Testing and Molecular Biomarkers, 2021, 25, 293-301.	0.3	3
10	Blending oxytocin and dopamine with everyday creativity. Scientific Reports, 2021, 11, 16185.	1.6	4
11	Germline genome modification through novel political, ethical, and social lenses. PLoS Genetics, 2021, 17, e1009741.	1.5	4
12	Genetic variation in the oxytocin system and its link to social motivation in human infants. Psychoneuroendocrinology, 2021, 131, 105290.	1.3	9
13	Design of Split Proximity Circuit as a Plug-and-Play Translator for Point Mutation Discrimination. Analytical Chemistry, 2020, 92, 11164-11170.	3.2	7
14	Novel variant in NSDHL gene associated with CHILD syndrome and syndactyly- a case report. BMC Medical Genetics, 2020, 21, 164.	2.1	6
15	Six Novel ATM Gene Variants in Sri Lankan Patients with Ataxia Telangiectasia. Case Reports in Genetics, 2020, 2020, 1-7.	0.1	3
16	Phosphoethanolamine Elevation in Plasma of Spinal Muscular Atrophy Type 1 Patients. Kobe Journal of Medical Sciences, 2020, 66, E1-E11.	0.2	1
17	Mutational spectrum of dystrophinopathies in Singapore: Insights for genetic diagnosis and precision therapy. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2019, 181, 230-244.	0.7	12
18	Specific phenotype semantics facilitate gene prioritization in clinical exome sequencing. European Journal of Human Genetics, 2019, 27, 1389-1397.	1.4	11

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19	Asia Pacific Society of Human Genetics (APSHG) from conception to 2019: 13 years of collaboration to tackle congenital malformation and genetic disorders in Asia. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2019, 181, 155-165.	0.7	1
20	Development of clinical genetics in Asia. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2019, 181, 150-154.	0.7	1
21	Training in clinical genetics and genetic counseling in Asia. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2019, 181, 177-186.	0.7	9
22	The analysis of DMD gene deletions by multiplex PCR in Indonesian DMD/BMD patients: the era of personalized medicine. BMC Research Notes, 2019, 12, 704.	0.6	15
23	Nested PCR Amplification Secures DNA Template Quality and Quantity in Real-time mCOP-PCR Screening for SMA. Kobe Journal of Medical Sciences, 2019, 65, E54-E58.	0.2	1
24	Spinal Muscular Atrophy: Advanced Version of Screening System with Real-Time mCOP-PCR and PCR-RFLP for SMN1 Deletion. Kobe Journal of Medical Sciences, 2019, 65, E49-E53.	0.2	2
25	Spinal Muscular Atrophy: New Screening System with Real-Time mCOP-PCR and PCR-RFLP for SMN1 Deletion. Kobe Journal of Medical Sciences, 2019, 65, E44-E48.	0.2	3
26	Newborn Screening for Spinal Muscular Atrophy: DNA Preparation from Dried Blood Spot and DNA Polymerase Selection in PCR. Kobe Journal of Medical Sciences, 2019, 65, E95-E99.	0.2	1
27	Intron-retained transcripts of the spinal muscular atrophy genes, SMN1 and SMN2. Brain and Development, 2018, 40, 670-677.	0.6	5
28	Genetic screening of spinal muscular atrophy using a real-time modified COP-PCR technique with dried blood-spot DNA. Brain and Development, 2017, 39, 774-782.	0.6	20
29	SMA mutations in SMN Tudor and C-terminal domains destabilize the protein. Brain and Development, 2017, 39, 606-612.	0.6	10
30	Spinal muscular atrophy carriers with two SMN1 copies. Brain and Development, 2017, 39, 851-860.	0.6	14
31	Mutation spectrum of RB1 mutations in retinoblastoma cases from Singapore with implications for genetic management and counselling. PLoS ONE, 2017, 12, e0178776.	1.1	49
32	Gender Effects on the Clinical Phenotype in Japanese Patients with Spinal Muscular Atrophy. Kobe Journal of Medical Sciences, 2017, 63, E41-E44.	0.2	2
33	SMA Diagnosis: Detection of SMN1 Deletion with Real-Time mCOP-PCR System Using Fresh Blood DNA. Kobe Journal of Medical Sciences, 2017, 63, E80-E83.	0.2	1
34	Alternative splicing of a cryptic exon embedded in intron 6 of SMN1 and SMN2. Human Genome Variation, 2016, 3, 16040.	0.4	9
35	Delay discounting, genetic sensitivity, and leukocyte telomere length. Proceedings of the National Academy of Sciences of the United States of America, 2016, 113, 2780-2785.	3.3	20
36	Genetic variation in CD38 and breastfeeding experience interact to impact infantsâ€™ attention to social eye cues. Proceedings of the National Academy of Sciences of the United States of America, 2015, 112, E5434-42.	3.3	50

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37	Intragenic mutations in SMN1 may contribute more significantly to clinical severity than SMN2 copy numbers in some spinal muscular atrophy (SMA) patients. Brain and Development, 2014, 36, 914-920.	0.6	39
38	Gold nanostructures for the multiplex detection of glucose-6-phosphate dehydrogenase gene mutations. Analytical Biochemistry, 2014, 451, 56-62.	1.1	10
39	A Prospective Study in the Rational Design of Efficient Antisense Oligonucleotides for Exon Skipping in the DMD Gene. Human Gene Therapy, 2012, 23, 781-790.	1.4	23
40	Dimeric gold nanoparticle assembly for detection and discrimination of single nucleotide mutation in Duchenne muscular dystrophy. Biosensors and Bioelectronics, 2010, 25, 2021-2025.	5.3	22
41	A Case of X-Linked Adrenal Hypoplasia Congenita, Central Precocious Puberty and Absence of the <i>DAX-1</i> Gene: Implications for Pubertal Regulation. Hormone Research in Paediatrics, 2009, 71, 298-304.	0.8	14
42	Automated DNA mutation detection using universal conditions direct sequencing: application to ten muscular dystrophy genes. BMC Genetics, 2009, 10, 66.	2.7	13
43	Mapping Human Genetic Diversity in Asia. Science, 2009, 326, 1541-1545.	6.0	557
44	Diagnostic Strategy for the Detection of Dystrophin Gene Mutations in Asian Patients and Carriers Using Immortalized Cell Lines. Journal of Child Neurology, 2006, 21, 150-155.	0.7	6
45	Comparative study on deletions of the dystrophin gene in three Asian populations. Journal of Human Genetics, 2002, 47, 0552-0555.	1.1	25
46	Mutation spectrum analysis of DMD gene in Indonesian Duchenne and Becker muscular dystrophy patients. F1000Research, 0, 11, 148.	0.8	0