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	Mapping Human Genetic Diversity in Asia. Science, 2009, 326, 1541-1545.	6.0	557
2	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
3	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
4	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
5	Comparative study on deletions of the dystrophin gene in three Asian populations. Journal of Human Genetics, 2002, 47, 0552-0555.	1.1	25
6	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
7	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
8	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
9	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
10	Clinical phenotypes of spinal muscular atrophy patients with hybrid SMN gene. Brain and Development, 2021, 43, 294-302.	0.6	18
11	Singapore Undiagnosed Disease Program: Genomic Analysis aids Diagnosis and Clinical Management. Archives of Disease in Childhood, 2021, 106, 31-37.	1.0	17
12	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
13	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
14	Spinal muscular atrophy carriers with two SMN1 copies. Brain and Development, 2017, 39, 851-860.	0.6	14
15	Automated DNA mutation detection using universal conditions direct sequencing: application to ten muscular dystrophy genes. BMC Genetics, 2009, 10, 66.	2.7	13
16	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
17	Specific phenotype semantics facilitate gene prioritization in clinical exome sequencing. European Journal of Human Genetics, 2019, 27, 1389-1397.	1.4	11
18	Gold nanostructures for the multiplex detection of glucose-6-phosphate dehydrogenase gene mutations. Analytical Biochemistry, 2014, 451, 56-62.	1.1	10

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19	SMA mutations in SMN Tudor and C-terminal domains destabilize the protein. Brain and Development, 2017, 39, 606-612.	0.6	10
20	Alternative splicing of a cryptic exon embedded in intron 6 of SMN1 and SMN2. Human Genome Variation, 2016, 3, 16040.	0.4	9
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	Genetic variation in the oxytocin system and its link to social motivation in human infants. Psychoneuroendocrinology, 2021, 131, 105290.	1.3	9
23	Novel Autoantibodies in Idiopathic Small Fiber Neuropathy. Annals of Neurology, 2022, 91, 66-77.	2.8	9
24	Design of Split Proximity Circuit as a Plug-and-Play Translator for Point Mutation Discrimination. Analytical Chemistry, 2020, 92, 11164-11170.	3.2	7
25	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
26	Novel variant in NSDHL gene associated with CHILD syndrome and syndactyly- a case report. BMC Medical Genetics, 2020, 21, 164.	2.1	6
27	Intron-retained transcripts of the spinal muscular atrophy genes, SMN1 and SMN2. Brain and Development, 2018, 40, 670-677.	0.6	5
28	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
29	Blending oxytocin and dopamine with everyday creativity. Scientific Reports, 2021, 11, 16185.	1.6	4
30	Germline genome modification through novel political, ethical, and social lenses. PLoS Genetics, 2021, 17, e1009741.	1.5	4
31	Effect of semaphorin 3C gene variants in multifactorial Hirschsprung disease. Journal of International Medical Research, 2021, 49, 030006052098778.	0.4	3
32	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
33	Six Novel ATM Gene Variants in Sri Lankan Patients with Ataxia Telangiectasia. Case Reports in Genetics, 2020, 2020, 1-7.	0.1	3
34	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
35	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
36	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

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37	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
38	Vulnerability and the Ethics of Human Germline Genome Editing. CRISPR Journal, 2022, 5, 358-363.	1.4	2
39	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
40	Development of clinical genetics in Asia. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2019, 181, 150-154.	0.7	1
41	OUP accepted manuscript. Human Reproduction, 2021, 36, 3018-3027.	0.4	1
42	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
43	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
44	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
45	Phosphoethanolamine Elevation in Plasma of Spinal Muscular Atrophy Type 1 Patients. Kobe Journal of Medical Sciences, 2020, 66, E1-E11.	0.2	1
46	Mutation spectrum analysis of DMD gene in Indonesian Duchenne and Becker muscular dystrophy patients. F1000Research, 0, 11, 148.	0.8	0