Nirmala D Sirisena

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/7657406/publications.pdf

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52	725	840728	580810
papers	citations	11 h-index	25 g-index

53	53	53	1154
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	22q11.2 deletion syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2017, 173, 879-888.	1.2	103
2	Genetic determinants of inherited susceptibility to hypercholesterolemia â€“ a comprehensive literature review. Lipids in Health and Disease, 2017, 16, 103.	3.0	89
3	Down syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2017, 173, 42-53.	1.2	75
4	Noonan syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2017, 173, 2323-2334.	1.2	68
5	Williamsâ€“Beuren syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2018, 176, 1128-1136.	1.2	55
6	Cornelia de Lange syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2019, 179, 150-158.	1.2	40
7	Candidate gene study of genetic thrombophilic polymorphisms in preâ€eclampsia and recurrent pregnancy loss in Sinhalese women. Journal of Obstetrics and Gynaecology Research, 2012, 38, 1168-1176.	1.3	26
8	Susceptible and Prognostic Genetic Factors Associated with Diabetic Peripheral Neuropathy: A Comprehensive Literature Review. International Journal of Endocrinology, 2018, 2018, 1-9.	1.5	25
9	Dyskeratosis congenita with a novel genetic variant in the DKC1 gene: a case report. BMC Medical Genetics, 2018, 19, 85.	2.1	16
10	Genetic Variants Associated with Clinicopathological Profiles in Sporadic Breast Cancer in Sri Lankan Women. Journal of Breast Cancer, 2018, 21, 165.	1.9	16
11	Turner syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2020, 182, 303-313.	1.2	15
12	A Case Series of Five Sri Lankan Patients with Ovotesticular Disorder of Sex Development. Clinical Pediatric Endocrinology, 2012, 21, 69-73.	0.8	14
13	Novel mutation in the SLC12A3 gene in a Sri Lankan family with Gitelman syndrome & coexistent diabetes: a case report. BMC Nephrology, 2017, 18, 140.	1.8	13
14	Cytogenetic analysis of chromosomal abnormalities in Sri Lankan children. World Journal of Pediatrics, 2015, 11, 374-379.	1.8	12
15	Implementation of genomic medicine in Sri Lanka: Initial experience and challenges. Applied & Translational Genomics, 2016, 9, 33-36.	2.1	11
16	Child with Deletion 9p Syndrome Presenting with Craniofacial Dysmorphism, Developmental Delay, and Multiple Congenital Malformations. Case Reports in Genetics, 2013, 2013, 1-4.	0.2	10
17	A Child with a Novel de novo Mutation in the Aristaless Domain of the Aristaless-Related Homeobox <i>(ARX)</i> Gene Presenting with Ambiguous Genitalia and Psychomotor Delay. Sexual Development, 2014, 8, 156-159.	2.0	10
18	Focusing attention on ancestral diversity within genomics research: a potential means for promoting equity in the provision of genomics based healthcare services in developing countries. Journal of Community Genetics, 2017, 8, 275-281.	1.2	10

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19	The pattern of KRAS mutations in metastatic colorectal cancer: a retrospective audit from Sri Lanka. BMC Research Notes, 2017, 10, 392.	1.4	10
20	Genetic determinants of sporadic breast cancer in Sri Lankan women. BMC Cancer, 2018, 18, 180.	2.6	10
21	Genetics and genomic medicine in Sri Lanka. Molecular Genetics & Genomic Medicine, 2019, 7, e744.	1.2	10
22	Split hand/foot malformation with long bone deficiency associated with BHLHA9 gene duplication: a case report and review of literature. BMC Medical Genetics, 2019, 20, 108.	2.1	9
23	Strategies for Genomic Medicine Education in Low- and Middle-Income Countries. Frontiers in Genetics, 2019, 10, 944.	2.3	8
24	Hypoparathyroidism, Sensorineural deafness and renal disease (Barakat syndrome) caused by a reduced gene dosage in GATA3: a case report and review of literature. BMC Endocrine Disorders, 2019, 19, 111.	2.2	8
25	Partial trisomy 16q21âž”qter due to an unbalanced segregation of a maternally inherited balanced translocation 46,XX,t(15;16)(p13;q21): a case report and review of literature. BMC Pediatrics, 2018, 18, 4.	1.7	7
26	The Frequency and Spectrum of Chromosomal Translocations in a Cohort of Sri Lankans. BioMed Research International, 2019, 2019, 1-11.	1.9	7
27	Prevalence of chromosomal abnormalities in Sri Lankan women with primary amenorrhea. Journal of Obstetrics and Gynaecology Research, 2013, 39, 991-997.	1.3	5
28	A child with multiple congenital anomalies due to partial trisomy 7q22.1â€‰â†’â€‰qter resulting from a maternally inherited balanced translocation: a case report and review of literature. BMC Medical Genomics, 2018, 11, 44.	1.5	5
29	Functional evaluation of five BRCA2 unclassified variants identified in a Sri Lankan cohort with inherited cancer syndromes using a mouse embryonic stem cell-based assay. Breast Cancer Research, 2020, 22, 43.	5.0	5
30	The Provision of Medical and Health Genetics and Genomics in the Developing World. , 2016, , 285-294.		4
31	Ring Chromosome 4 in a Child with Multiple Congenital Abnormalities: A Case Report and Review of the Literature. Case Reports in Genetics, 2016, 2016, 1-7.	0.2	4
32	The Prevalence of the Prothrombin (F2) 20210G>A Mutation in a Cohort of Sri Lankan Patients with Thromboembolic Disorders. Indian Journal of Hematology and Blood Transfusion, 2015, 31, 356-361.	0.6	3
33	Taking Genomics From the Bench to the Bedside in Developing Countries. , 2018, , 13-26.		3
34	Genotype data for single nucleotide polymorphism markers in sporadic breast cancer related genes in a Sri Lankan caseâ€“control cohort of postmenopausal women. BMC Research Notes, 2019, 12, 435.	1.4	3
35	NovelCOL4A3gene mutations in a consanguineous family with autosomal recessive Alport syndrome. Nephrology, 2015, 20, 580-580.	1.6	2
36	Relative normalized luciferase activity for the recombinant vector constructs carrying the ancestral and variant alleles for XRCC2:rs3218550 and PHB:rs6917. BMC Research Notes, 2018, 11, 643.	1.4	2

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37	Electrophoretic mobility shift assays implicate XRCC2:rs3218550C>T as a potential low-penetrant susceptibility allele for sporadic breast cancer. BMC Research Notes, 2019, 12, 476.	1.4	2
38	A novel variant in the COL6A1 gene causing Ullrich congenital muscular dystrophy in a consanguineous family: a case report. BMC Neurology, 2021, 21, 105.	1.8	2
39	Androgen insensitivity syndrome in a cohort of Sri Lankan children with 46, XY disorders of sex development. Ceylon Medical Journal, 2016, 60, 139.	0.2	2
40	Cancer genetics and the surgeon â€“ new frontiers. The Sri Lanka Journal of Surgery, 2014, 32, 12.	0.0	2
41	HLA-B27 allele frequency in Sri Lankan patients with spondyloarthritides. Ceylon Medical Journal, 2016, 61, 71.	0.2	2
42	A case series of three Sri Lankan families with hereditary breast and ovarian cancer syndrome due to pathogenic germline mutations in the BRCA1 gene. Ceylon Medical Journal, 2017, 62, 65.	0.2	1
43	Frontometaphyseal dysplasia 1 in a patient from Sri Lanka. American Journal of Medical Genetics, Part A, 2021, 185, 1317-1320.	1.2	1
44	Novel <scp>AGXT</scp> gene mutation in a <scp>S</scp>ri <scp>L</scp>ankan family with primary <scp>H</scp>yperoxaluria type 1. Nephrology, 2016, 21, 76-77.	1.6	0
45	Correspondence. World Journal of Pediatrics, 2016, 12, 374-375.	1.8	0
46	Establishing a relationship between clinical features and one specific type of chromosome abnormality. World Journal of Pediatrics, 2016, 12, 374-374.	1.8	0
47	Cover Image, Volume 173A, Number 4, April 2017. American Journal of Medical Genetics, Part A, 2017, 173, i.	1.2	0
48	The first Sri Lankan family with Dent disease-1 due to a pathogenic variant in the CLCN5 gene: a case report. BMC Research Notes, 2017, 10, 539.	1.4	0
49	A Sri Lankan family with cerebellar hemangioblastoma due to a heterozygous nonsense mutation in the von Hippel-Lindau tumor suppressor, E3 ubiquitin protein ligase (VHL) gene. The Sri Lanka Journal of Surgery, 2015, 33, 30.	0.0	0
50	Authors reply re: HLA-B27 allele frequency in Sri Lankan patients with spondyloarthritides. Ceylon Medical Journal, 2016, 61, 203.	0.2	0
51	A case series of 7 Sri Lankan patients with type 1 spinal muscular atrophy. Sri Lanka Journal of Child Health, 2016, 45, 247.	0.1	0
52	Hypokalaemic Paralysis - A double trouble from concurrent Thyrotoxicosis and Gitelman syndrome: A report of two cases. Sri Lanka Journal of Diabetes Endocrinology and Metabolism, 2020, 10, 26.	0.1	0