

# Nirmala D Sirisena

## List of Publications by Year in descending order

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Version: 2024-02-01

52  
papers

725  
citations

840728

11  
h-index

580810

25  
g-index

53  
all docs

53  
docs citations

53  
times ranked

1154  
citing authors

#	ARTICLE	IF	CITATIONS
1	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
2	Frontometaphyseal dysplasia 1 in a patient from Sri Lanka. American Journal of Medical Genetics, Part A, 2021, 185, 1317-1320.	1.2	1
3	Turner syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2020, 182, 303-313.	1.2	15
4	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
5	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
6	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
7	Genotype data for single nucleotide polymorphism markers in sporadic breast cancer related genes in a Sri Lankan case-control cohort of postmenopausal women. BMC Research Notes, 2019, 12, 435.	1.4	3
8	Strategies for Genomic Medicine Education in Low- and Middle-Income Countries. Frontiers in Genetics, 2019, 10, 944.	2.3	8
9	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
10	Genetics and genomic medicine in Sri Lanka. Molecular Genetics & Genomic Medicine, 2019, 7, e744.	1.2	10
11	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
12	The Frequency and Spectrum of Chromosomal Translocations in a Cohort of Sri Lankans. BioMed Research International, 2019, 2019, 1-11.	1.9	7
13	Cornelia de Lange syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2019, 179, 150-158.	1.2	40
14	Williams-Beuren syndrome in diverse populations. American Journal of Medical Genetics, Part A, 2018, 176, 1128-1136.	1.2	55
15	Genetic determinants of sporadic breast cancer in Sri Lankan women. BMC Cancer, 2018, 18, 180.	2.6	10
16	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
17	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
18	Dyskeratosis congenita with a novel genetic variant in the DKC1 gene: a case report. BMC Medical Genetics, 2018, 19, 85.	2.1	16

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19	Genetic Variants Associated with Clinicopathological Profiles in Sporadic Breast Cancer in Sri Lankan Women. <i>Journal of Breast Cancer</i> , 2018, 21, 165.	1.9	16
20	Taking Genomics From the Bench to the Bedside in Developing Countries. , 2018, , 13-26.		3
21	Partial trisomy 16q21 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. <i>BMC Pediatrics</i> , 2018, 18, 4.	1.7	7
22	A child with multiple congenital anomalies due to partial trisomy 7q22.1 due to a maternally inherited balanced translocation: a case report and review of literature. <i>BMC Medical Genomics</i> , 2018, 11, 44.	1.5	5
23	22q11.2 deletion syndrome in diverse populations. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 879-888.	1.2	103
24	Cover Image, Volume 173A, Number 4, April 2017. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, i.	1.2	0
25	Down syndrome in diverse populations. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 42-53.	1.2	75
26	Noonan syndrome in diverse populations. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 2323-2334.	1.2	68
27	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. <i>Journal of Community Genetics</i> , 2017, 8, 275-281.	1.2	10
28	Novel mutation in the SLC12A3 gene in a Sri Lankan family with Gitelman syndrome & coexistent diabetes: a case report. <i>BMC Nephrology</i> , 2017, 18, 140.	1.8	13
29	Genetic determinants of inherited susceptibility to hypercholesterolemia – a comprehensive literature review. <i>Lipids in Health and Disease</i> , 2017, 16, 103.	3.0	89
30	The pattern of KRAS mutations in metastatic colorectal cancer: a retrospective audit from Sri Lanka. <i>BMC Research Notes</i> , 2017, 10, 392.	1.4	10
31	The first Sri Lankan family with Dent disease-1 due to a pathogenic variant in the CLCN5 gene: a case report. <i>BMC Research Notes</i> , 2017, 10, 539.	1.4	0
32	A case series of three Sri Lankan families with hereditary breast and ovarian cancer syndrome due to pathogenic germline mutations in the BRCA1 gene. <i>Ceylon Medical Journal</i> , 2017, 62, 65.	0.2	1
33	The Provision of Medical and Health Genetics and Genomics in the Developing World. , 2016, , 285-294.		4
34	Ring Chromosome 4 in a Child with Multiple Congenital Abnormalities: A Case Report and Review of the Literature. <i>Case Reports in Genetics</i> , 2016, 2016, 1-7.	0.2	4
35	Implementation of genomic medicine in Sri Lanka: Initial experience and challenges. <i>Applied &amp; Translational Genomics</i> , 2016, 9, 33-36.	2.1	11
36	Novel <i>AGXT</i> gene mutation in a Sri Lankan family with primary <i>Hyperoxaluria</i> type 1. <i>Nephrology</i> , 2016, 21, 76-77.	1.6	0

#	ARTICLE	IF	CITATIONS
37	Correspondence. World Journal of Pediatrics, 2016, 12, 374-375.	1.8	0
38	Establishing a relationship between clinical features and one specific type of chromosome abnormality. World Journal of Pediatrics, 2016, 12, 374-374.	1.8	0
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	HLA-B27 allele frequency in Sri Lankan patients with spondyloarthritides. Ceylon Medical Journal, 2016, 61, 71.	0.2	2
41	Authors reply re: HLA-B27 allele frequency in Sri Lankan patients with spondyloarthritides. Ceylon Medical Journal, 2016, 61, 203.	0.2	0
42	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
43	Novel COL4A3 gene mutations in a consanguineous family with autosomal recessive Alport syndrome. Nephrology, 2015, 20, 580-580.	1.6	2
44	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
45	Cytogenetic analysis of chromosomal abnormalities in Sri Lankan children. World Journal of Pediatrics, 2015, 11, 374-379.	1.8	12
46	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
47	A Child with a Novel de novo Mutation in the Aristaless Domain of the Aristaless-Related Homeobox &lt;b>&lt;i>(ARX)&lt;/i>&lt;/b> Gene Presenting with Ambiguous Genitalia and Psychomotor Delay. Sexual Development, 2014, 8, 156-159.	2.0	10
48	Cancer genetics and the surgeon â€œ new frontiers. The Sri Lanka Journal of Surgery, 2014, 32, 12.	0.0	2
49	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
50	Prevalence of chromosomal abnormalities in Sri Lankan women with primary amenorrhea. Journal of Obstetrics and Gynaecology Research, 2013, 39, 991-997.	1.3	5
51	Candidate gene study of genetic thrombophilic polymorphisms in pre-eclampsia and recurrent pregnancy loss in Sinhalese women. Journal of Obstetrics and Gynaecology Research, 2012, 38, 1168-1176.	1.3	26
52	A Case Series of Five Sri Lankan Patients with Ovotesticular Disorder of Sex Development. Clinical Pediatric Endocrinology, 2012, 21, 69-73.	0.8	14