Priyanka Srivastava

List of Publications by Year
in descending order

68	626	623734	677142
papers	citations	14 h-index	22 g-index

68	68	68	966
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Characterization of the CYP21A2 Gene Mutations in Children with Classic Congenital Adrenal Hyperplasia. Indian Journal of Pediatrics, 2024, 91, 137-142.	0.8	2
2	Gas Chromatography Mass Spectrometry Aided Diagnosis of Glutathione Synthetase Deficiency. Laboratory Medicine, 2022, 53, e59-e61.	1.2	1
3	Molecular analysis of severe hemophilia B in Indian families: Identification of mutational hotspot and novel variants. International Journal of Laboratory Hematology, 2022, 44, 186-192.	1.3	1
4	Whole-exome sequencing and variant spectrum in children with suspected inherited renal tubular disorder: the East India Tubulopathy Gene Study. Pediatric Nephrology, 2022, 37, 1811-1836.	1.7	5
5	A novel c.1937T>C (p.Leu646Pro) missense mutation in a patient with Leber congenital amaurosis. Journal of AAPOS, 2022, , .	0.3	0
6	Role of miRNA polymorphism in recurrent pregnancy loss: aÂsystematic review and meta-analysis. Biomarkers in Medicine, 2022, 16, 101-115.	1.4	3
7	Genotype-phenotype spectrum of 130 unrelated Indian families with Mucopolysaccharidosis type II. European Journal of Medical Genetics, 2022, 65, 104447.	1.3	3
8	How Experts Make a Call: Copy Number Variation Analysis in Unusual/Rare Case Scenarios. Neurology India, 2022, 70, 148.	0.4	1
9	Exploration of Potential Biomarker Genes and Pathways in Kawasaki Disease: An Integrated in-Silico Approach. Frontiers in Genetics, 2022, 13, .	2.3	6
10	Association of SNP (rs1042579) in thrombomodulin gene and plasma thrombomodulin level in North Indian children with Kawasaki disease. Molecular Biology Reports, 2022, 49, 7399-7407.	2.3	2
11	Delineating the epilepsy phenotype of NRROS-related microgliopathy: A case report and literature review. Seizure: the Journal of the British Epilepsy Association, 2022, 100, 15-20.	2.0	3
12	Clinical and molecular characterization of four patients with Robinow syndrome from different families. American Journal of Medical Genetics, Part A, 2021, 185, 1105-1112.	1.2	4
13	Deciphering the Pathogenic Nature of Two de novo Sequence Variations in a Patient with Shprintzen-Goldberg Syndrome. Molecular Syndromology, 2021, 12, 141-147.	0.8	1
14	Homozygosity stretches around homozygous mutations in autosomal recessive disorders: patients from nonconsanguineous Indian families. Journal of Genetics, 2021, 100, 1.	0.7	8
15	Novel variation in <scp>ANTXR2</scp> gene causing hyaline fibromatosis syndrome: A report from India. Congenital Anomalies (discontinued), 2021, 61, 140-141.	0.6	1
16	Partial Trisomy of Chromosome 8q and Partial Monosomy of Chromosome 6p with Robinow Syndrome-Like Phenotype. Indian Journal of Pediatrics, 2021, 88, 813-818.	0.8	0
17	Genetic heterogeneity of disorders with overgrowth and intellectual disability: Experience from a center in North India. American Journal of Medical Genetics, Part A, 2021, 185, 2345-2355.	1.2	6
18	Monozygotic twins with development delay and a characteristic electroencephalographic pattern: Just look at the face. Journal of Paediatrics and Child Health, 2021, , .	0.8	0

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19	Monozygotic twins with development delay and a characteristic electroencephalographic pattern: Just look at the face. Journal of Paediatrics and Child Health, 2021, 57, 960-961.	0.8	1
20	Epigenetics in Kawasaki Disease. Frontiers in Pediatrics, 2021, 9, 673294.	1.9	10
21	Phenotypic and genotypic spectrum of CTSK variants in a cohort of twenty-five Indian patients with pycnodysostosis. European Journal of Medical Genetics, 2021, 64, 104235.	1.3	6
22	Ayme gripp syndrome in an Indian patient. American Journal of Medical Genetics, Part A, 2021, 185, 1312-1316.	1.2	1
23	Achondroplasiaâ€"First Report from India of a Rare FGFR3 Gene Variant. Laboratory Medicine, 2021, 52, 499-502.	1.2	2
24	Kallmann Syndrome and X-linked Ichthyosis Caused by Translocation Between Chromosomes X and Y: A Case Report. Journal of Reproduction and Infertility, 2021, 22, 302-306.	1.0	2
25	Desbuquois dysplasia Kim variant: a rare case report syndrome. Clinical Dysmorphology, 2021, 30, 62-65.	0.3	0
26	A mild phenotype of LGI4-Related arthrogryposis multiplex congenita with intrafamilial variability. European Journal of Medical Genetics, 2020, 63, 103756.	1.3	5
27	Hypotonic infant with Pallisterâ€"Killian syndrome diagnosed by cytogenetic microarray, without pigmentary skin changes and malformations. Journal of Genetics, 2020, 99, 1.	0.7	0
28	Sequence variations in TENM3 gene causing eye anomalies with intellectual disability: Expanding the phenotypic spectrum. European Journal of Medical Genetics, 2019, 62, 61-64.	1.3	11
29	Cytogenetic microarray in structurally normal and abnormal foetuses: a fiveÂyears experience elucidating increasing acceptance and clinical utility. Journal of Genetics, 2019, 98, 1.	0.7	4
30	Deletion 7q21.2-q22.1 in a case with split hand-split foot malformation, sensorineural hearing loss and intellectual disability: Phenotype subtypes and the correlation with genotypes. European Journal of Medical Genetics, 2019, 62, 103597.	1.3	3
31	Connexin 26 (GJB2) Mutations Associated with Non-Syndromic Hearing Loss (NSHL). Indian Journal of Pediatrics, 2018, 85, 1061-1066.	0.8	22
32	Phenotypic characterization of derivative 22 syndrome: case series and review. Journal of Genetics, 2018, 97, 205-211.	0.7	5
33	A large interstitial 11q deletion with isolated mild intellectual disability: review of the literature for genotypeâ€"phenotype correlation. Clinical Dysmorphology, 2018, 27, 142-144.	0.3	1
34	Molecular Testing of MECP2 Gene in Rett Syndrome Phenotypes in Indian Girls. Indian Pediatrics, 2018, 55, 474-477.	0.4	3
35	Molecular Testing of MECP2 Gene in Rett Syndrome Phenotypes in Indian Girls. Indian Pediatrics, 2018, 55, 474-477.	0.4	1
36	Spondyloepiphyseal dysplasia Omani type: CHST3 mutation spectrum and phenotypes in three Indian families. American Journal of Medical Genetics, Part A, 2017, 173, 163-168.	1.2	15

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37	Pyruvate Carboxylase Deficiency Mimicking Diabetic Ketoacidosis. Indian Journal of Pediatrics, 2017, 84, 959-960.	0.8	3
38	KBG syndrome: 16q24.3 microdeletion in an Indian patient. Clinical Dysmorphology, 2017, 26, 161-166.	0.3	2
39	Expansion of the phenotypic spectrum in three families of methyl CpG-binding protein 2 duplication syndrome. Clinical Dysmorphology, 2017, 26, 73-77.	0.3	2
40	Fanconi-Bickel Syndrome: Another Novel Mutation in SLC2A2. Indian Journal of Pediatrics, 2017, 84, 236-237.	0.8	5
41	Pycnodysostosis: mutation spectrum in five unrelated Indian children. Clinical Dysmorphology, 2016, 25, 113-120.	0.3	16
42	Floating Harbor Syndrome. Indian Journal of Pediatrics, 2016, 83, 896-897.	0.8	5
43	Novel sequence variations in the thymidine phosphorylase gene causing mitochondrial neurogastrointestinal encephalopathy. Clinical Dysmorphology, 2016, 25, 156-162.	0.3	4
44	Novel mutations in the transmembrane natriuretic peptide receptor NPR-B gene in four Indian families with acromesomelic dysplasia, type Maroteaux. Journal of Genetics, 2016, 95, 905-909.	0.7	7
45	Cartilage Hair Hypoplasia: Two Unrelated Cases with g.70 AÂ>ÂG Mutation in RMRP Gene. Indian Journal of Pediatrics, 2016, 83, 1003-1005.	0.8	2
46	Hunter syndrome in northern India: Clinical features and mutation spectrum. Indian Pediatrics, 2016, 53, 134-136.	0.4	3
47	On-Demand Guided Bone Regeneration with Microbial Protection of Ornamented SPU Scaffold with Bismuth-Doped Single Crystalline Hydroxyapatite: Augmentation and Cartilage Formation. ACS Applied Materials & Interfaces, 2016, 8, 4086-4100.	8.0	35
48	Consanguinity as an Adjunct Diagnostic Tool. Indian Journal of Pediatrics, 2016, 83, 258-260.	0.8	3
49	Mutations in patients with osteogenesis imperfecta from consanguineous Indian families. European Journal of Medical Genetics, 2015, 58, 21-27.	1.3	37
50	Death receptor 4 variants enhanced prostate cancer risk in North Indian population. Tumor Biology, 2015, 36, 5655-5661.	1.8	1
51	Impact of CCL2 and Its Receptor CCR2 Gene Polymorphism in North Indian Population: A Comparative Study in Different Ethnic Groups Worldwide. Indian Journal of Clinical Biochemistry, 2013, 28, 259-264.	1.9	3
52	Association of IL-12, IL-18 variants and serum IL-18 with bladder cancer susceptibility in North Indian population. Gene, 2013, 519, 128-134.	2.2	43
53	Association of single nucleotide polymorphisms in promoter of matrix metalloproteinase-2, 8 genes with bladder cancer risk in Northern India. Urologic Oncology: Seminars and Original Investigations, 2013, 31, 247-254.	1.6	34
54	Impact of MMP-3 and TIMP-3 gene polymorphisms on prostate cancer susceptibility in North Indian cohort. Gene, 2013, 530, 273-277.	2.2	18

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55	No Association of Matrix Metalloproteinase [MMP]-2 (â^'735CÂ>ÂT) and Tissue Inhibitor of Metalloproteinase [TIMP]-2 (â^'418GÂ>ÂC) Gene Polymorphisms with Cervical Cancer Susceptibility. Indian Journal of Clinical Biochemistry, 2013, 28, 13-18.	1.9	17
56	Association of inflammatory chemokine gene CCL2I/D with bladder cancer risk in North Indian population. Molecular Biology Reports, 2012, 39, 9827-9834.	2.3	13
57	Association of Promoter Polymorphisms in MMP2 and TIMP2 with Prostate Cancer Susceptibility in North India. Archives of Medical Research, 2012, 43, 117-124.	3.3	47
58	Association studies of Tollâ€like receptor gene polymorphisms with allograft survival in renal transplant recipients of North India. Clinical Transplantation, 2012, 26, 581-588.	1.6	8
59	Gene variants of XRCC4 and XRCC3 and their association with risk for urothelial bladder cancer. Molecular Biology Reports, 2012, 39, 1667-1675.	2.3	36
60	Association of Common Variants of Vascular Endothelial Growth Factor and Interleukin-18 Genes with Allograft Survival in Renal Transplant Recipients of North India. DNA and Cell Biology, 2011, 30, 309-315.	1.9	13
61	Role of p53 gene polymorphism and bladder cancer predisposition in northern India. Cancer Biomarkers, 2011, 8, 21-28.	1.7	9
62	Role of MMP-3 and MMP-9 and Their Haplotypes in Risk of Bladder Cancer in North Indian Cohort. Annals of Surgical Oncology, 2010, 17, 3068-3075.	1.5	27
63	Bladder Cancer Risk Associated with Genotypic Polymorphism of the Matrix Metalloproteinase-1 and 7 in North Indian Population. Disease Markers, 2010, 29, 37-46.	1.3	34
64	Matrix metalloproteinase (MMP-9 and MMP-2) gene polymorphisms influence allograft survival in renal transplant recipients. Nephrology Dialysis Transplantation, 2010, 25, 3393-3401.	0.7	27
65	Bladder cancer risk associated with genotypic polymorphism of the matrix metalloproteinase-1 and 7 in North Indian population. Disease Markers, 2010, 29, 37-46.	1.3	27
66	Influence of matrix metalloproteinase gene polymorphisms in healthy North Indians compared to variations in other ethnic groups worldwide. Asian Pacific Journal of Cancer Prevention, 2009, 10, 1127-30.	1.2	6
67	Case Studies of Two Classical Imprinting Growth Disorders: Silverâ€"Russell and Beckwithâ€"Wiedemann Syndromes. Journal of Pediatric Genetics, 0, , .	0.7	0
68	Pattern Recognition of Common Multiple Congenital Malformation Syndromes with Underlying Chromatinopathy. Journal of Pediatric Genetics, 0, , .	0.7	0