

Gandham Srilakshmi Bhavani

List of Publications by Year in descending order

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Version: 2024-02-01

25
papers

443
citations

932766

10
h-index

794141

19
g-index

25
all docs

25
docs citations

25
times ranked

1310
citing authors

#	ARTICLE	IF	CITATIONS
1	The promise of discovering population-specific disease-associated genes in South Asia. <i>Nature Genetics</i> , 2017, 49, 1403-1407.	9.4	129
2	The homozygous variant c.797G>A/p.(Cys266Tyr) in <i>PISD</i> is associated with a Spondyloepimetaphyseal dysplasia with large epiphyses and disturbed mitochondrial function. <i>Human Mutation</i> , 2019, 40, 299-309.	1.1	54
3	Diagnostic strategies and genotype-phenotype correlation in a large Indian cohort of osteogenesis imperfecta. <i>Bone</i> , 2018, 110, 368-377.	1.4	38
4	SCUBE3 loss-of-function causes a recognizable recessive developmental disorder due to defective bone morphogenetic protein signaling. <i>American Journal of Human Genetics</i> , 2021, 108, 115-133.	2.6	37
5	Clinical and mutation profile of multicentric osteolysis nodulosis and arthropathy. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 410-417.	0.7	31
6	Novel subtype of mucopolysaccharidosis caused by arylsulfatase K (ARSK) deficiency. <i>Journal of Medical Genetics</i> , 2022, 59, 957-964.	1.5	29
7	Novel and recurrent mutations in <i>WISP3</i> and an atypical phenotype. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 2481-2484.	0.7	21
8	Additional three patients with Smith-McCort dysplasia due to novel <i>RAB33B</i> mutations. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 588-595.	0.7	17
9	A novel multiple joint dislocation syndrome associated with a homozygous nonsense variant in the <i>EXOC6B</i> gene. <i>European Journal of Human Genetics</i> , 2016, 24, 1206-1210.	1.4	16
10	Spectrum of <i>SMPD1</i> mutations in Asian-Indian patients with acid sphingomyelinase (ASM)-deficient Niemann-Pick disease. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 2719-2730.	0.7	15
11	Phenotyping and genotyping of skeletal dysplasias: Evolution of a center and a decade of experience in India. <i>Bone</i> , 2019, 120, 204-211.	1.4	10
12	Homozygous variant, p.(Arg643Trp) in <i>VAC14</i> causes striatonigral degeneration: report of a novel variant and review of <i>VAC14</i> -related disorders. <i>Journal of Human Genetics</i> , 2019, 64, 1237-1242.	1.1	8
13	Deficiency of <i>TMEM53</i> causes a previously unknown sclerosing bone disorder by dysregulation of BMP-SMAD signaling. <i>Nature Communications</i> , 2021, 12, 2046.	5.8	7
14	A syndrome of facial dysmorphism, cubital pterygium, short distal phalanges, swan neck deformity of fingers, and scoliosis. <i>American Journal of Medical Genetics, Part A</i> , 2014, 164, 1035-1040.	0.7	5
15	Burn-McKeown syndrome with biallelic promoter type 2 deletion in <i>TXNL4A</i> in two siblings. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 1313-1315.	0.7	5
16	Clinical and Genetic Spectrum of Inborn Errors of Immunity in a Tertiary Care Center in Southern India. <i>Indian Journal of Pediatrics</i> , 2022, 89, 233-242.	0.3	4
17	Novel Mutation in an Indian Patient with Transcobalamin II Deficiency. <i>Indian Journal of Pediatrics</i> , 2015, 82, 1073-1074.	0.3	3
18	Metatropic dysplasia with a novel mutation in <i>TRPV4</i> . <i>Indian Pediatrics</i> , 2016, 53, 735-737.	0.2	3

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19	Bosleyâ€“Salihâ€“Alorainy syndrome in patients from India. American Journal of Medical Genetics, Part A, 2020, 182, 2699-2703.	0.7	3
20	A homozygous hypomorphic <i>BNIP1</i> variant causes an increase in autophagosomes and reduced autophagic flux and results in a spondyloâ€“epiphyseal dysplasia. Human Mutation, 2022, 43, 625-642.	1.1	3
21	<i>Ectodysplasin</i> pathogenic variants affecting the furinâ€“cleavage site and unusual clinical features define Xâ€“linked hypohidrotic ectodermal dysplasia in India. American Journal of Medical Genetics, Part A, 2022, 188, 788-805.	0.7	3
22	Pseudoachondroplasia: Phenotype and genotype in 11 Indian patients. American Journal of Medical Genetics, Part A, 2022, 188, 751-759.	0.7	1
23	Steel syndrome: Report of three patients, including monozygotic twins and review of clinical and mutation profiles. European Journal of Medical Genetics, 2022, 65, 104521.	0.7	1
24	Cover Image, Volume 173A, Number 3, March 2017. American Journal of Medical Genetics, Part A, 2017, 173, i.	0.7	0
25	Clinical, radiological and molecular studies in 24 individuals with Dyggve-Melchior-Clausen dysplasia and Smith-McCort dysplasia from India. Journal of Medical Genetics, 2023, 60, 204-211.	1.5	0