Gandham Srilakshmi Bhavani

List of Publications by Year
in descending order

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	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
2	Pseudoachondroplasia: Phenotype and genotype in 11 Indian patients. American Journal of Medical Genetics, Part A, 2022, 188, 751-759.	0.7	1
3	Clinical and Genetic Spectrum of Inborn Errors of Immunity in a Tertiary Care Center in Southern India. Indian Journal of Pediatrics, 2022, 89, 233-242.	0.3	4
4	A homozygous hypomorphicBNIP1variant 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
5	Novel subtype of mucopolysaccharidosis caused by arylsulfatase K (ARSK) deficiency. Journal of Medical Genetics, 2022, 59, 957-964.	1.5	29
6	Ectodysplasin 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
7	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
8	SCUBE3 loss-of-function causes a recognizable recessive developmental disorder due to defective bone morphogenetic protein signaling. American Journal of Human Genetics, 2021, 108, 115-133.	2.6	37
9	Deficiency of TMEM53 causes a previously unknown sclerosing bone disorder by dysregulation of BMP-SMAD signaling. Nature Communications, 2021, 12, 2046.	5.8	7
10	Bosleyâ€“Salihâ€“Alorainy syndrome in patients from India. American Journal of Medical Genetics, Part A, 2020, 182, 2699-2703.	0.7	3
11	Burnâ€McKeown syndrome with biallelic promoter type 2 deletion in <scp>TXNL4A</scp> in two siblings. American Journal of Medical Genetics, Part A, 2020, 182, 1313-1315.	0.7	5
12	Homozygous variant, p.(Arg643Trp) in VAC14 causes striatonigral degeneration: report of a novel variant and review of VAC14-related disorders. Journal of Human Genetics, 2019, 64, 1237-1242.	1.1	8
13	Phenotyping and genotyping of skeletal dysplasias: Evolution of a center and a decade of experience in India. Bone, 2019, 120, 204-211.	1.4	10
14	The homozygous variant c.797G>A/p.(Cys266Tyr) inPISDis associated with a Spondyloepimetaphyseal dysplasia with large epiphyses and disturbed mitochondrial function. Human Mutation, 2019, 40, 299-309.	1.1	54
15	Diagnostic strategies and genotype-phenotype correlation in a large Indian cohort of osteogenesis imperfecta. Bone, 2018, 110, 368-377.	1.4	38
16	Additional three patients with Smithâ€McCort dysplasia due to novel RAB33B mutations. American Journal of Medical Genetics, Part A, 2017, 173, 588-595.	0.7	17
17	Cover Image, Volume 173A, Number 3, March 2017. American Journal of Medical Genetics, Part A, 2017, 173, i.	0.7	0
18	The promise of discovering population-specific disease-associated genes in South Asia. Nature Genetics, 2017, 49, 1403-1407.	9.4	129

#	Article	IF	Citations
19	Clinical and mutation profile of multicentric osteolysis nodulosis and arthropathy. American Journal of Medical Genetics, Part A, 2016, 170, 410-417.	0.7	31
20	Metatropic dysplasia with a novel mutation in TRPV4. Indian Pediatrics, 2016, 53, 735-737.	0.2	3
21	Spectrum of SMPD1 mutations in Asianâ€Indian patients with acid sphingomyelinase (ASM)â€deficient Niemannâ€“Pick disease. American Journal of Medical Genetics, Part A, 2016, 170, 2719-2730.	0.7	15
22	A novel multiple joint dislocation syndrome associated with a homozygous nonsense variant in the EXOC6B gene. European Journal of Human Genetics, 2016, 24, 1206-1210.	1.4	16
23	Novel and recurrent mutations in WISP3 and an atypical phenotype. American Journal of Medical Genetics, Part A, 2015, 167, 2481-2484.	0.7	21
24	Novel Mutation in an Indian Patient with Transcobalamin II Deficiency. Indian Journal of Pediatrics, 2015, 82, 1073-1074.	0.3	3
25	A syndrome of facial dysmorphism, cubital pterygium, short distal phalanges, swan neck deformity of fingers, and scoliosis. American Journal of Medical Genetics, Part A, 2014, 164, 1035-1040.	0.7	5