

Priyanka Upadhyai

List of Publications by Year in descending order

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Version: 2024-02-01

19
papers

227
citations

1306789

7
h-index

1058022

14
g-index

26
all docs

26
docs citations

26
times ranked

311
citing authors

#	ARTICLE	IF	CITATIONS
1	The intraflagellar transport protein IFT52 associated with short-rib thoracic dysplasia is essential for ciliary function in osteogenic differentiation in vitro and for sensory perception in <i>Drosophila</i> . <i>Experimental Cell Research</i> , 2022, 418, 113273.	1.2	3
2	Biallelic deep intronic variant c.5457+81T>A in <i>TRIP11</i> causes loss of function and results in achondrogenesis 1A. <i>Human Mutation</i> , 2021, 42, 1005-1014.	1.1	3
3	Genomic and Ancestral Variation Underlies the Severity of COVID-19 Clinical Manifestation in Individuals of European Descent. <i>Life</i> , 2021, 11, 921.	1.1	4
4	Biallelic missense variant, p.<sc>Ser35Leu</sc> in <sc><i>EXOSC1</i></sc> is associated with pontocerebellar hypoplasia. <i>Clinical Genetics</i> , 2021, 99, 594-600.	1.0	16
5	Novel splice site and nonsense variants in <i>INVS</i> cause infantile nephronophthisis. <i>Gene</i> , 2020, 729, 144229.	1.0	2
6	Modulation of the Promoter Activation Rate Dictates the Transcriptional Response to Graded BMP Signaling Levels in the <i>Drosophila</i> Embryo. <i>Developmental Cell</i> , 2020, 54, 727-741.e7.	3.1	47
7	Phenotypic diversity and genetic complexity of <i>PAX3</i> -related Waardenburg syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 2951-2958.	0.7	8
8	Recurrent 1q21.1 deletion syndrome: report on variable expression, nonpenetrance and review of literature. <i>Clinical Dysmorphology</i> , 2020, 29, 127-131.	0.1	5
9	The story of the lost twins: decoding the genetic identities of the Kumhar and Kurcha populations from the Indian subcontinent. <i>BMC Genetics</i> , 2020, 21, 117.	2.7	5
10	Characterization of primary cilia features reveal cell-type specific variability in in vitro models of osteogenic and chondrogenic differentiation. <i>PeerJ</i> , 2020, 8, e9799.	0.9	8
11	Investigating the West Eurasian ancestry of Pakistani Hazaras. <i>Journal of Genetics</i> , 2019, 98, 1.	0.4	3
12	Application of the geographic population structure (GPS) algorithm for biogeographical analyses of wild and captive gorillas. <i>BMC Bioinformatics</i> , 2019, 20, 35.	1.2	5
13	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
14	An Ancestry Informative Marker Set Which Recapitulates the Known Fine Structure of Populations in South Asia. <i>Genome Biology and Evolution</i> , 2018, 10, 2408-2416.	1.1	5
15	Autosomal recessive spinocerebellar ataxia 20: Report of a new patient and review of literature. <i>European Journal of Medical Genetics</i> , 2017, 60, 118-123.	0.7	29
16	Unraveling the Population History of Indian Siddis. <i>Genome Biology and Evolution</i> , 2017, 9, 1385-1392.	1.1	2
17	Homozygous p.(Glu87Lys) variant in <i>ISCA1</i> is associated with a multiple mitochondrial dysfunctions syndrome. <i>Journal of Human Genetics</i> , 2017, 62, 723-727.	1.1	53
18	Application of geographic population structure (GPS) algorithm for biogeographical analyses of populations with complex ancestries: a case study of South Asians from 1000 genomes project. <i>BMC Genetics</i> , 2017, 18, 109.	2.7	10

#	ARTICLE	IF	CITATIONS
19	Brinker possesses multiple mechanisms for repression because its primary co-repressor, Groucho, may be unavailable in some cell types. <i>Development (Cambridge)</i> , 2013, 140, 4256-4265.	1.2	6