Priyanka Upadhyai

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/4463926/publications.pdf

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19	227	1306789	1058022
papers	citations	7	14
		h-index	g-index

26	26	26	311
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Homozygous p.(Glu87Lys) variant in ISCA1 is associated with a multiple mitochondrial dysfunctions syndrome. Journal of Human Genetics, 2017, 62, 723-727.	1.1	53
2	Modulation of the Promoter Activation Rate Dictates the Transcriptional Response to Graded BMP Signaling Levels in the Drosophila Embryo. Developmental Cell, 2020, 54, 727-741.e7.	3.1	47
3	Autosomal recessive spinocerebellar ataxia 20: Report of a new patient and review of literature. European Journal of Medical Genetics, 2017, 60, 118-123.	0.7	29
4	Biâ€allelic missense variant, p.<scp>Ser35Leu</scp> in <scp>EXOSC1</scp> is associated with pontocerebellar hypoplasia. Clinical Genetics, 2021, 99, 594-600.	1.0	16
5	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. BMC Genetics, 2017, 18, 109.	2.7	10
6	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
7	Phenotypic diversity and genetic complexity of PAX3 â€related Waardenburg syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 2951-2958.	0.7	8
8	Characterization of primary cilia features reveal cell-type specific variability in in vitro models of osteogenic and chondrogenic differentiation. PeerJ, 2020, 8, e9799.	0.9	8
9	Brinker possesses multiple mechanisms for repression because its primary co-repressor, Groucho, may be unavailable in some cell types. Development (Cambridge), 2013, 140, 4256-4265.	1.2	6
10	An Ancestry Informative Marker Set Which Recapitulates the Known Fine Structure of Populations in South Asia. Genome Biology and Evolution, 2018, 10, 2408-2416.	1.1	5
11	Application of the geographic population structure (GPS) algorithm for biogeographical analyses of wild and captive gorillas. BMC Bioinformatics, 2019, 20, 35.	1.2	5
12	Recurrent 1q21.1 deletion syndrome: report on variable expression, nonpenetrance and review of literature. Clinical Dysmorphology, 2020, 29, 127-131.	0.1	5
13	The story of the lost twins: decoding the genetic identities of the Kumhar and Kurcha populations from the Indian subcontinent. BMC Genetics, 2020, 21, 117.	2.7	5
14	Genomic and Ancestral Variation Underlies the Severity of COVID-19 Clinical Manifestation in Individuals of European Descent. Life, 2021, 11, 921.	1.1	4
15	Investigating the West Eurasian ancestry of Pakistani Hazaras. Journal of Genetics, 2019, 98, 1.	0.4	3
16	Biallelic deep intronic variant c.5457+81T>A in TRIP11 causes loss of function and results in achondrogenesis 1A. Human Mutation, 2021, 42, 1005-1014.	1.1	3
17	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 Drosophila. Experimental Cell Research, 2022, 418, 113273.	1.2	3
18	Unraveling the Population History of Indian Siddis. Genome Biology and Evolution, 2017, 9, 1385-1392.	1.1	2

#	Article	IF	Citations
19	Novel splice site and nonsense variants in INVS cause infantile nephronophthisis. Gene, 2020, 729, 144229.	1.0	2