

Sarju G Mehta

List of Publications by Year in descending order

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Version: 2024-02-01

25
papers

2,278
citations

430874

18
h-index

642732

23
g-index

25
all docs

25
docs citations

25
times ranked

5721
citing authors

#	ARTICLE	IF	CITATIONS
1	Evidence for 28 genetic disorders discovered by combining healthcare and research data. <i>Nature</i> , 2020, 586, 757-762.	27.8	343
2	Mutations in DDX3X Are a Common Cause of Unexplained Intellectual Disability with Gender-Specific Effects on Wnt Signaling. <i>American Journal of Human Genetics</i> , 2015, 97, 343-352.	6.2	230
3	Heterozygous missense mutations in SMARCA2 cause Nicolaides-Baraitser syndrome. <i>Nature Genetics</i> , 2012, 44, 445-449.	21.4	207
4	Histone Lysine Methylases and Demethylases in the Landscape of Human Developmental Disorders. <i>American Journal of Human Genetics</i> , 2018, 102, 175-187.	6.2	204
5	Coffin-Siris Syndrome and the BAF Complex: Genotype-Phenotype Study in 63 Patients. <i>Human Mutation</i> , 2013, 34, 1519-1528.	2.5	178
6	Quantifying the contribution of recessive coding variation to developmental disorders. <i>Science</i> , 2018, 362, 1161-1164.	12.6	158
7	Genetic heterogeneity in Cornelia de Lange syndrome (CdLS) and CdLS-like phenotypes with observed and predicted levels of mosaicism. <i>Journal of Medical Genetics</i> , 2014, 51, 659-668.	3.2	141
8	Maternal variants in <i>NLRP</i> and other maternal effect proteins are associated with multilocus imprinting disturbance in offspring. <i>Journal of Medical Genetics</i> , 2018, 55, 497-504.	3.2	126
9	A mutation update on the LDS-associated genes <i>TGFB2/3</i> and <i>SMAD2/3</i> . <i>Human Mutation</i> , 2018, 39, 621-634.	2.5	116
10	Mutations in SNORD118 cause the cerebral microangiopathy leukoencephalopathy with calcifications and cysts. <i>Nature Genetics</i> , 2016, 48, 1185-1192.	21.4	114
11	Mutations of <i>AKT3</i> are associated with a wide spectrum of developmental disorders including extreme megalencephaly. <i>Brain</i> , 2017, 140, 2610-2622.	7.6	102
12	Haploinsufficiency of the Chromatin Remodeler BPTF Causes Syndromic Developmental and Speech Delay, Postnatal Microcephaly, and Dysmorphic Features. <i>American Journal of Human Genetics</i> , 2017, 101, 503-515.	6.2	61
13	Mutations in <i>ACTL6B</i> Cause Neurodevelopmental Deficits and Epilepsy and Lead to Loss of Dendrites in Human Neurons. <i>American Journal of Human Genetics</i> , 2019, 104, 815-834.	6.2	59
14	Clinical and molecular consequences of disease-associated de novo mutations in <i>SATB2</i> . <i>Genetics in Medicine</i> , 2017, 19, 900-908.	2.4	46
15	Detection of structural mosaicism from targeted and whole-genome sequencing data. <i>Genome Research</i> , 2017, 27, 1704-1714.	5.5	44
16	Heterozygous <i>KIDINS220/ARMS</i> nonsense variants cause spastic paraplegia, intellectual disability, nystagmus, and obesity. <i>Human Molecular Genetics</i> , 2016, 25, 2158-2167.	2.9	37
17	Delineation of dominant and recessive forms of <i>LZTR1</i> associated Noonan syndrome. <i>Clinical Genetics</i> , 2019, 95, 693-703.	2.0	35
18	The broad phenotypic spectrum of <i>PPP2R1A</i> -related neurodevelopmental disorders correlates with the degree of biochemical dysfunction. <i>Genetics in Medicine</i> , 2021, 23, 352-362.	2.4	23

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19	Expanding the phenotypic spectrum of ARID1B-mediated disorders and identification of altered cell-cycle dynamics due to ARID1B haploinsufficiency. <i>Orphanet Journal of Rare Diseases</i> , 2014, 9, 43.	2.7	16
20	Leukoencephalopathy with calcifications and cysts: Genetic and phenotypic spectrum. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 15-25.	1.2	15
21	Extending the clinical and genetic spectrum of ARID2 related intellectual disability. A case series of 7 patients. <i>European Journal of Medical Genetics</i> , 2019, 62, 27-34.	1.3	13
22	<i>PAPSS2</i> -related brachyolmia: Clinical and radiological phenotype in 18 new cases. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 1884-1894.	1.2	9
23	Cervical artery dissection and iliac artery aneurysm in an SMAD-4 mutation carrier. <i>Neurology: Genetics</i> , 2017, 3, e191.	1.9	1
24	136...Beyond the Aorta: Experiences of Neurovascular Imaging in Loeys-dietz and Vascular Ehlers Danlos Syndrome. <i>Heart</i> , 2016, 102, A97.1-A97.	2.9	0
25	A missense TGFB2 variant p.(Arg320Cys) causes a paradoxical and striking increase in aortic TGFB1/2 expression. <i>European Journal of Human Genetics</i> , 2017, 25, 157-160.	2.8	0