Sarju G Mehta

List of Publications by Year
in descending order

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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	Leukoencephalopathy with calcifications and cysts: Genetic and phenotypic spectrum. American Journal of Medical Genetics, Part A, 2021, 185, 15-25.	1.2	15
2	The broad phenotypic spectrum of PPP2R1A-related neurodevelopmental disorders correlates with the degree of biochemical dysfunction. Genetics in Medicine, 2021, 23, 352-362.	2.4	23
3	Evidence for 28 genetic disorders discovered by combining healthcare and research data. Nature, 2020, 586, 757-762.	27.8	343
4	PAPSS2â€related brachyolmia: Clinical and radiological phenotype in 18 new cases. American Journal of Medical Genetics, Part A, 2019, 179, 1884-1894.	1.2	9
5	Mutations in ACTL6B Cause Neurodevelopmental Deficits and Epilepsy and Lead to Loss of Dendrites in Human Neurons. American Journal of Human Genetics, 2019, 104, 815-834.	6.2	59
6	Delineation of dominant and recessive forms of LZTR1â€associated Noonan syndrome. Clinical Genetics, 2019, 95, 693-703.	2.0	35
7	Extending the clinical and genetic spectrum of ARID2 related intellectual disability. A case series of 7 patients. European Journal of Medical Genetics, 2019, 62, 27-34.	1.3	13
8	A mutation update on the LDS-associated genesTGFB2/3andSMAD2/3. Human Mutation, 2018, 39, 621-634.	2.5	116
9	Histone Lysine Methylases and Demethylases in the Landscape of Human Developmental Disorders. American Journal of Human Genetics, 2018, 102, 175-187.	6.2	204
10	Maternal variants in NLRP and other maternal effect proteins are associated with multilocus imprinting disturbance in offspring. Journal of Medical Genetics, 2018, 55, 497-504.	3.2	126
11	Quantifying the contribution of recessive coding variation to developmental disorders. Science, 2018, 362, 1161-1164.	12.6	158
12	Clinical and molecular consequences of disease-associated de novo mutations in SATB2. Genetics in Medicine, 2017, 19, 900-908.	2.4	46
13	Haploinsufficiency of the Chromatin Remodeler BPTF Causes Syndromic Developmental and Speech Delay, Postnatal Microcephaly, and Dysmorphic Features. American Journal of Human Genetics, 2017, 101, 503-515.	6.2	61
14	Mutations of AKT3 are associated with a wide spectrum of developmental disorders including extreme megalencephaly. Brain, 2017, 140, 2610-2622.	7.6	102
15	Detection of structural mosaicism from targeted and whole-genome sequencing data. Genome Research, 2017, 27, 1704-1714.	5.5	44
16	A missense TGFB2 variant p.(Arg320Cys) causes a paradoxical and striking increase in aortic TGFB1/2 expression. European Journal of Human Genetics, 2017, 25, 157-160.	2.8	0
17	Cervical artery dissection and iliac artery aneurysm in an SMAD-4 mutation carrier. Neurology: Genetics, 2017, 3, e191.	1.9	1
18	Mutations in SNORD118 cause the cerebral microangiopathy leukoencephalopathy with calcifications and cysts. Nature Genetics, 2016, 48, 1185-1192.	21.4	114

#	Article	IF	Citations
19	136â€...Beyond the Aorta: Experiences of Neurovascular Imaging in Loeys-dietz and Vascular Ehlers Danlos Syndrome. Heart, 2016, 102, A97.1-A97.	2.9	0
20	HeterozygousKIDINS220/ARMSnonsense variants cause spastic paraplegia, intellectual disability, nystagmus, and obesity. Human Molecular Genetics, 2016, 25, 2158-2167.	2.9	37
21	Mutations in DDX3X Are a Common Cause of Unexplained Intellectual Disability with Gender-Specific Effects on Wnt Signaling. American Journal of Human Genetics, 2015, 97, 343-352.	6.2	230
22	Genetic heterogeneity in Cornelia de Lange syndrome (CdLS) and CdLS-like phenotypes with observed and predicted levels of mosaicism. Journal of Medical Genetics, 2014, 51, 659-668.	3.2	141
23	Expanding the phenotypic spectrum of ARID1B-mediated disorders and identification of altered cell-cycle dynamics due to ARID1B haploinsufficiency. Orphanet Journal of Rare Diseases, 2014, 9, 43.	2.7	16
24	Coffin-Siris Syndrome and the BAF Complex: Genotype-Phenotype Study in 63 Patients. Human Mutation, 2013, 34, 1519-1528.	2.5	178
25	Heterozygous missense mutations in SMARCA2 cause Nicolaides-Baraitser syndrome. Nature Genetics, 2012, 44, 445-449.	21.4	207