Shane McKee

List of Publications by Year
in descending order

Source: https://exaly.com/author-pdf/3243868/publications.pdf

Version: 2024-02-01

35	1,638	361413 20	361022 35
papers	citations	h-index	g-index

38	38	38	3656
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Heterozygous missense mutations in SMARCA2 cause Nicolaides-Baraitser syndrome. Nature Genetics, 2012, 44, 445-449.	21.4	207
2	Histone Lysine Methylases and Demethylases in the Landscape of Human Developmental Disorders. American Journal of Human Genetics, 2018, 102, 175-187.	6.2	204
3	Mutations in the histone methyltransferase gene KMT2B cause complex early-onset dystonia. Nature Genetics, 2017, 49, 223-237.	21.4	186
4	Coffin-Siris Syndrome and the BAF Complex: Genotype-Phenotype Study in 63 Patients. Human Mutation, 2013, 34, 1519-1528.	2.5	178
5	A Restricted Repertoire of De Novo Mutations in ITPR1 Cause Gillespie Syndrome with Evidence for Dominant-Negative Effect. American Journal of Human Genetics, 2016, 98, 981-992.	6.2	81
6	The ARID1B spectrum in 143 patients: from nonsyndromic intellectual disability to Coffinâ€“Siris syndrome. Genetics in Medicine, 2019, 21, 1295-1307.	2.4	80
7	Biallelic Variants in UBA5 Link Dysfunctional UFM1ÂUbiquitin-like Modifier Pathway to Severe Infantile-Onset Encephalopathy. American Journal of Human Genetics, 2016, 99, 683-694.	6.2	72
8	PURA syndrome: clinical delineation and genotype-phenotype study in 32 individuals with review of published literature. Journal of Medical Genetics, 2018, 55, 104-113.	3.2	59
9	Rare deleterious mutations of HNRNP genes result in shared neurodevelopmental disorders. Genome Medicine, 2021, 13, 63.	8.2	50
10	Clinical and molecular consequences of disease-associated de novo mutations in SATB2. Genetics in Medicine, 2017, 19, 900-908.	2.4	46
11	Recurrent heterozygous PAX6 missense variants cause severe bilateral microphthalmia via predictable effects on DNAâ€“protein interaction. Genetics in Medicine, 2020, 22, 598-609.	2.4	43
12	Delineation of dominant and recessive forms of LZTR1â€associated Noonan syndrome. Clinical Genetics, 2019, 95, 693-703.	2.0	35
13	Variants in members of the cadherinâ€“catenin complex, CDH1 and CTNND1, cause blepharocheilodontic syndrome. European Journal of Human Genetics, 2018, 26, 210-219.	2.8	34
14	Making sense of missense variants in TTN-related congenital myopathies. Acta Neuropathologica, 2021, 141, 431-453.	7.7	34
15	The CHD8 overgrowth syndrome: A detailed evaluation of an emerging overgrowth phenotype in 27 patients. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2019, 181, 557-564.	1.6	33
16	The clinical presentation caused by truncating CHD8 variants. Clinical Genetics, 2019, 96, 72-84.	2.0	32
17	A Syndromic Neurodevelopmental Disorder Caused by Mutations in SMARCD1, a Core SWI/SNF Subunit Needed for Context-Dependent Neuronal Gene Regulation in Flies. American Journal of Human Genetics, 2019, 104, 596-610.	6.2	32
18	Elucidating the genetic architecture of Adams-Oliver syndrome in a large European cohort. Human Mutation, 2018, 39, 1246-1261.	2.5	31

#	Article	IF	Citations
19	Delineation of phenotypes and genotypes related to cohesin structural protein RAD21. Human Genetics, 2020, 139, 575-592.	3.8	24
20	A scoping review and proposed workflow for multi-omic rare disease research. Orphanet Journal of Rare Diseases, 2020, 15, 107.	2.7	24
21	De novo and inherited TCF20 pathogenic variants are associated with intellectual disability, dysmorphic features, hypotonia, and neurological impairments with similarities to Smithâ€“Magenis syndrome. Genome Medicine, 2019, 11, 12.	8.2	23
22	Histiocytoid cardiomyopathy and microphthalmia with linear skin defects syndrome: phenotypes linked by truncating variants in NDUFB11. Journal of Physical Education and Sports Management, 2017, 3, a001271.	1.2	19
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	Atypical COL3A1 variants (glutamic acid to lysine) cause vascular Ehlersâ€“Danlos syndrome with a consistent phenotype of tissue fragility and skin hyperextensibility. Genetics in Medicine, 2019, 21, 2081-2091.	2.4	16
25	Expanding the clinical spectrum of recessive truncating mutations ofKLHL7to a Bohring-Opitz-like phenotype. Journal of Medical Genetics, 2017, 54, 830-835.	3.2	15
26	Autosomal Dominant Early Childhood Seizures Associated with Chondrocalcinosis and a Mutation in the ANKH Gene. Epilepsia, 2004, 45, 1258-1260.	5.1	14
27	Genetic Analysis of a Cohort of 275 Patients with Hyper-IgE Syndromes and/or Chronic Mucocutaneous Candidiasis. Journal of Clinical Immunology, 2021, 41, 1804-1838.	3.8	12
28	A clinical study of Aicardi syndrome in Northern Ireland: the spectrum of ophthalmic findings. Eye, 2016, 30, 1011-1016.	2.1	8
29	Finding Diagnostically Useful Patterns in Quantitative Phenotypic Data. American Journal of Human Genetics, 2019, 105, 933-946.	6.2	8
30	Whole-genome analysis as a diagnostic tool for patients referred for diagnosis of Silver-Russell syndrome: a real-world study. Journal of Medical Genetics, 2022, 59, 613-622.	3.2	8
31	Bilateral microtia with severe cardiac defect: a new syndrome, or a severe manifestation of the oculoauriculovertebral spectrum?. Clinical Dysmorphology, 2006, 15, 121-122.	0.3	2
32	SPECC1L Mutations Are Not Common in Sporadic Cases of Opitz G/BBB Syndrome. Genes, 2022, 13, 252.	2.4	2
33	Marking of patch tests. Contact Dermatitis, 1983, 9, 240-240.	1.4	1
34	CADASIL IN NORTHERN IRELAND: A RETROSPECTIVE CLINICAL, NEUROIMAGING AND GENETIC STUDY. Journal of Neurology, Neurosurgery and Psychiatry, 2015, 86, e4.101-e4.	1.9	0
35	P35â€…Histiocytoid cardiomyopathy and microphthalmia with linear skin defects syndrome: Phenotypes linked by truncating variants inNDUFB11. Heart, 2016, 102, A18.2-A18.	2.9	0