Shane McKee

List of Publications by Year
in descending order

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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	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
2	SPECC1L Mutations Are Not Common in Sporadic Cases of Opitz G/BBB Syndrome. Genes, 2022, 13, 252.	2.4	2
3	Rare deleterious mutations of HNRNP genes result in shared neurodevelopmental disorders. Genome Medicine, 2021, 13, 63.	8.2	50
4	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
5	Making sense of missense variants in TTN-related congenital myopathies. Acta Neuropathologica, 2021, 141, 431-453.	7.7	34
6	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
7	Delineation of phenotypes and genotypes related to cohesin structural protein RAD21. Human Genetics, 2020, 139, 575-592.	3.8	24
8	A scoping review and proposed workflow for multi-omic rare disease research. Orphanet Journal of Rare Diseases, 2020, 15, 107.	2.7	24
9	Finding Diagnostically Useful Patterns in Quantitative Phenotypic Data. American Journal of Human Genetics, 2019, 105, 933-946.	6.2	8
10	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
11	The clinical presentation caused by truncating CHD8 variants. Clinical Genetics, 2019, 96, 72-84.	2.0	32
12	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
13	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
14	Delineation of dominant and recessive forms of LZTR1â€associated Noonan syndrome. Clinical Genetics, 2019, 95, 693-703.	2.0	35
15	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
16	The ARID1B spectrum in 143 patients: from nonsyndromic intellectual disability to Coffinâ€“Siris syndrome. Genetics in Medicine, 2019, 21, 1295-1307.	2.4	80
17	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
18	Histone Lysine Methylases and Demethylases in the Landscape of Human Developmental Disorders. American Journal of Human Genetics, 2018, 102, 175-187.	6.2	204

#	Article	IF	Citations
19	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
20	Elucidating the genetic architecture of Adams-Oliver syndrome in a large European cohort. Human Mutation, 2018, 39, 1246-1261.	2.5	31
21	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
22	Clinical and molecular consequences of disease-associated de novo mutations in SATB2. Genetics in Medicine, 2017, 19, 900-908.	2.4	46
23	Mutations in the histone methyltransferase gene KMT2B cause complex early-onset dystonia. Nature Genetics, 2017, 49, 223-237.	21.4	186
24	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
25	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
26	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
27	A clinical study of Aicardi syndrome in Northern Ireland: the spectrum of ophthalmic findings. Eye, 2016, 30, 1011-1016.	2.1	8
28	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
29	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
30	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
31	Coffin-Siris Syndrome and the BAF Complex: Genotype-Phenotype Study in 63 Patients. Human Mutation, 2013, 34, 1519-1528.	2.5	178
32	Heterozygous missense mutations in SMARCA2 cause Nicolaides-Baraitser syndrome. Nature Genetics, 2012, 44, 445-449.	21.4	207
33	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
34	Autosomal Dominant Early Childhood Seizures Associated with Chondrocalcinosis and a Mutation in the ANKH Gene. Epilepsia, 2004, 45, 1258-1260.	5.1	14
35	Marking of patch tests. Contact Dermatitis, 1983, 9, 240-240.	1.4	1