Joseph T Shieh

List of Publications by Year
in descending order

48	1,169	394421	434195
papers	citations	19 h-index	31 g-index

51	51	51	2336
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Somatic mosaicism in the MAPK pathway in sporadic brain arteriovenous malformation and association with phenotype. Journal of Neurosurgery, 2022, 136, 148-155.	1.6	12
2	Platelet VPS16B is dependent on VPS33B expression, as determined in two siblings with arthrogryposis, renal dysfunction and cholestasis (ARC) syndrome. Journal of Thrombosis and Haemostasis, 2022, , .	3.8	1
3	Tumor and Constitutional Sequencing for Neurofibromatosis Type 1. JCO Precision Oncology, 2022, 6, e2100540.	3.0	4
4	Common genetic variation associated with Mendelian disease severity revealed through cryptic phenotype analysis. Nature Communications, 2022, 13, .	12.8	5
5	De novo heterozygous variants in <scp>SLC30A7</scp> are a candidate cause for Joubert syndrome. American Journal of Medical Genetics, Part A, 2022, 188, 2360-2366.	1.2	3
6	Endovascular Biopsy for Detection of Somatic Mosaicism in Human Fusiform Cerebral Aneurysms. , 2022, 2, .		0
7	De novo variants in SNAP25 cause an early-onset developmental and epileptic encephalopathy. Genetics in Medicine, 2021, 23, 653-660.	2.4	20
8	A dyadic approach to the delineation of diagnostic entities in clinical genomics. American Journal of Human Genetics, 2021, 108, 8-15.	6.2	71
9	DLG4-related synaptopathy: a new rare brain disorder. Genetics in Medicine, 2021, 23, 888-899.	2.4	16
10	Expanding the genotypic and phenotypic spectrum in a diverse cohort of 104 individuals with Wiedemannâ€Steiner syndrome. American Journal of Medical Genetics, Part A, 2021, 185, 1649-1665.	1.2	34
11	Segmental overgrowth and aneurysms due to mosaic PDGFRB p.(Tyr562Cys). American Journal of Medical Genetics, Part A, 2021, 185, 1430-1436.	1.2	7
12	Hereditary Hemorrhagic Telangiectasia: The Convergence of Genotype, Phenotype, and Imaging in Modern Diagnosis and Management of a Multisystem Disease. Radiology, 2021, 300, 17-30.	7.3	22
13	Pathogenic SPTBN1 variants cause an autosomal dominant neurodevelopmental syndrome. Nature Genetics, 2021, 53, 1006-1021.	21.4	44
14	Application of full-genome analysis to diagnose rare monogenic disorders. Npj Genomic Medicine, 2021, 6, 77.	3.8	22
15	Response to Hamosh etÂal.. American Journal of Human Genetics, 2021, 108, 1809-1810.	6.2	0
16	Genetics workforce: distribution of genetics services and challenges to health care in California. Genetics in Medicine, 2020, 22, 227-231.	2.4	47
17	Case Report of Floating-Harbor Syndrome With Bilateral Cleft Lip. Cleft Palate-Craniofacial Journal, 2020, 57, 132-136.	0.9	3
18	Functional characterization of a novel PBX1 de novo missense variant identified in a patient with syndromic congenital heart disease. Human Molecular Genetics, 2020, 29, 1068-1082.	2.9	26

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19	A novel truncating variant in ring finger protein 113A (RNF113A) confirms the association of this gene with Xâ€linked trichothiodystrophy. American Journal of Medical Genetics, Part A, 2020, 182, 513-520.	1.2	12
20	MN1 C-terminal truncation syndrome is a novel neurodevelopmental and craniofacial disorder with partial rhombencephalosynapsis. Brain, 2020, 143, 55-68.	7.6	38
21	Optimizing genetics online resources for diverse readers. Genetics in Medicine, 2020, 22, 640-645.	2.4	1
22	The role of exome sequencing in newborn screening for inborn errors of metabolism. Nature Medicine, 2020, 26, 1392-1397.	30.7	112
23	Genotypeâ€“phenotype correlation at codon 1740 of <scp>SETD2</scp>. American Journal of Medical Genetics, Part A, 2020, 182, 2037-2048.	1.2	14
24	Automated syndrome diagnosis by three-dimensional facial imaging. Genetics in Medicine, 2020, 22, 1682-1693.	2.4	47
25	The genetic landscape of anaplastic pleomorphic xanthoastrocytoma. Brain Pathology, 2019, 29, 85-96.	4.1	88
26	Mechanisms of Resistance to EGFR Inhibition Reveal Metabolic Vulnerabilities in Human GBM. Molecular Cancer Therapeutics, 2019, 18, 1565-1576.	4.1	11
27	Disruptive variants of CSDE1 associate with autism and interfere with neuronal development and synaptic transmission. Science Advances, 2019, 5, eaax2166.	10.3	35
28	Phenotype characterisation of TBX4 mutation and deletion carriers with neonatal and paediatric pulmonary hypertension. European Respiratory Journal, 2019, 54, 1801965.	6.7	77
29	Schimke immunoosseous dysplasia and management considerations for vascular risks. American Journal of Medical Genetics, Part A, 2019, 179, 1246-1252.	1.2	3
30	Extracutaneous manifestations in phacomatosis cesioflammea and cesiomarmorata: Case series and literature review. American Journal of Medical Genetics, Part A, 2019, 179, 966-977.	1.2	20
31	Emerging RAS superfamily conditions involving GTPase function. PLoS Genetics, 2019, 15, e1007870.	3.5	5
32	Genomic Sequencing Expansion and Incomplete Penetrance. Pediatrics, 2019, 143, S22-S26.	2.1	5
33	Xâ€linked duplication copy number variation in a familial overgrowth condition. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2019, 181, 644-649.	1.6	2
34	Using exome sequencing to decipher family history in a healthy individual: Comparison of pathogenic and population <scp>MTM</scp>1 variants. Molecular Genetics & Genomic Medicine, 2018, 6, 722-727.	1.2	5
35	Mutations in Hnrnpa1 cause congenital heart defects. JCI Insight, 2018, 3, .	5.0	13
36	De Novo Truncating Mutations in the Last and Penultimate Exons of PPM1D Cause an Intellectual Disability Syndrome. American Journal of Human Genetics, 2017, 100, 650-658.	6.2	56

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37	Potential Role of Genomic Sequencing in the Early Diagnosis of Treatable Genetic Conditions. Journal of Pediatrics, 2017, 189, 222-226.e1.	1.8	7
38	Uveal Ganglioneuroma due to Germline <i>PTEN</i> Mutation (Cowden) Tj ETQq0 0 0 rgBT /Overlock 10 T 122-128.	1.0	9
39	GBM heterogeneity as a function of variable epidermal growth factor receptor variant III activity. Oncotarget, 2016, 7, 79101-79116.	1.8	39
40	Activating NRF1-BRAF and ATG7-RAF1 fusions in anaplastic pleomorphic xanthoastrocytoma without BRAF p.V600E mutation. Acta Neuropathologica, 2016, 132, 757-760.	7.7	32
41	Missense-depleted regions in population exomes implicate ras superfamily nucleotide-binding protein alteration in patients with brain malformation. Npj Genomic Medicine, 2016, 1, .	3.8	41
42	Response to Finsterer and StÃ¶llberger â€œExplanations for discordance of noncompaction in monozygotic twinsâ€• American Journal of Medical Genetics, Part A, 2015, 167, 2495-2495.	1.2	0
43	Prioritizing genes for X-linked diseases using population exome data. Human Molecular Genetics, 2015, 24, 599-608.	2.9	18
44	Clinical and ultrastructural spectrum of diffuse lung disease associated with surfactant protein C mutations. European Journal of Human Genetics, 2015, 23, 1033-1041.	2.8	29
45	Implications of genetic testing in noncompaction/hypertrabeculation. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2013, 163, 206-211.	1.6	9
46	Copy Number Variation Analysis in 98 Individuals with PHACE Syndrome. Journal of Investigative Dermatology, 2013, 133, 677-684.	0.7	25
47	Disorders of left ventricular trabeculation/compaction or right ventricular wall formation. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2013, 163, 141-143.	1.6	3
48	Consanguinity and the risk of congenital heart disease. American Journal of Medical Genetics, Part A, 2012, 158A, 1236-1241.	1.2	66