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	The role of exome sequencing in newborn screening for inborn errors of metabolism. Nature Medicine, 2020, 26, 1392-1397.	30.7	112
2	The genetic landscape of anaplastic pleomorphic xanthoastrocytoma. Brain Pathology, 2019, 29, 85-96.	4.1	88
3	Phenotype characterisation of TBX4 mutation and deletion carriers with neonatal and paediatric pulmonary hypertension. European Respiratory Journal, 2019, 54, 1801965.	6.7	77
4	A dyadic approach to the delineation of diagnostic entities in clinical genomics. American Journal of Human Genetics, 2021, 108, 8-15.	6.2	71
5	Consanguinity and the risk of congenital heart disease. American Journal of Medical Genetics, Part A, 2012, 158A, 1236-1241.	1.2	66
6	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
7	Genetics workforce: distribution of genetics services and challenges to health care in California. Genetics in Medicine, 2020, 22, 227-231.	2.4	47
8	Automated syndrome diagnosis by three-dimensional facial imaging. Genetics in Medicine, 2020, 22, 1682-1693.	2.4	47
9	Pathogenic SPTBN1 variants cause an autosomal dominant neurodevelopmental syndrome. Nature Genetics, 2021, 53, 1006-1021.	21.4	44
10	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
11	GBM heterogeneity as a function of variable epidermal growth factor receptor variant III activity. Oncotarget, 2016, 7, 79101-79116.	1.8	39
12	MN1 C-terminal truncation syndrome is a novel neurodevelopmental and craniofacial disorder with partial rhombencephalosynapsis. Brain, 2020, 143, 55-68.	7.6	38
13	Disruptive variants of CSDE1 associate with autism and interfere with neuronal development and synaptic transmission. Science Advances, 2019, 5, eaax2166.	10.3	35
14	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
15	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
16	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
17	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
18	Copy Number Variation Analysis in 98 Individuals with PHACE Syndrome. Journal of Investigative Dermatology, 2013, 133, 677-684.	0.7	25

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19	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
20	Application of full-genome analysis to diagnose rare monogenic disorders. Npj Genomic Medicine, 2021, 6, 77.	3.8	22
21	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
22	De novo variants in SNAP25 cause an early-onset developmental and epileptic encephalopathy. Genetics in Medicine, 2021, 23, 653-660.	2.4	20
23	Prioritizing genes for X-linked diseases using population exome data. Human Molecular Genetics, 2015, 24, 599-608.	2.9	18
24	DLG4-related synaptopathy: a new rare brain disorder. Genetics in Medicine, 2021, 23, 888-899.	2.4	16
25	Genotypeâ€“phenotype correlation at codon 1740 of <scp>SETD2</scp>. American Journal of Medical Genetics, Part A, 2020, 182, 2037-2048.	1.2	14
26	Mutations in Hnrnpa1 cause congenital heart defects. JCI Insight, 2018, 3, .	5.0	13
27	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
28	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
29	Mechanisms of Resistance to EGFR Inhibition Reveal Metabolic Vulnerabilities in Human GBM. Molecular Cancer Therapeutics, 2019, 18, 1565-1576.	4.1	11
30	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
31	Uveal Ganglioneuroma due to Germline <i>PTEN</i> Mutation (Cowden) Tj ETQq1 1 0.784314 rgBT /Ov 122-128.	1.0	9
32	Potential Role of Genomic Sequencing in the Early Diagnosis of Treatable Genetic Conditions. Journal of Pediatrics, 2017, 189, 222-226.e1.	1.8	7
33	Segmental overgrowth and aneurysms due to mosaic PDGFRB p.(Tyr562Cys). American Journal of Medical Genetics, Part A, 2021, 185, 1430-1436.	1.2	7
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	Emerging RAS superfamily conditions involving GTPase function. PLoS Genetics, 2019, 15, e1007870.	3.5	5
36	Genomic Sequencing Expansion and Incomplete Penetrance. Pediatrics, 2019, 143, S22-S26.	2.1	5

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37	Common genetic variation associated with Mendelian disease severity revealed through cryptic phenotype analysis. Nature Communications, 2022, 13, .	12.8	5
38	Tumor and Constitutional Sequencing for Neurofibromatosis Type 1. JCO Precision Oncology, 2022, 6, e2100540.	3.0	4
39	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
40	Schimke immunoosseous dysplasia and management considerations for vascular risks. American Journal of Medical Genetics, Part A, 2019, 179, 1246-1252.	1.2	3
41	Case Report of Floating-Harbor Syndrome With Bilateral Cleft Lip. Cleft Palate-Craniofacial Journal, 2020, 57, 132-136.	0.9	3
42	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
43	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
44	Optimizing genetics online resources for diverse readers. Genetics in Medicine, 2020, 22, 640-645.	2.4	1
45	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
46	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
47	Response to Hamosh etÂal.. American Journal of Human Genetics, 2021, 108, 1809-1810.	6.2	0
48	Endovascular Biopsy for Detection of Somatic Mosaicism in Human Fusiform Cerebral Aneurysms. , 2022, 2, .		0