Sheng Chih Jin

List of Publications by Year
in descending order

70	5,286	218677	98798
papers	citations	26 h-index	67 g-index

83	83	83	9536
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Familial and syndromic forms of arachnoid cyst implicate genetic factors in disease pathogenesis. Cerebral Cortex, 2023, 33, 3012-3025.	2.9	6
2	Biallelic <scp>AOPEP</scp> Lossâ€ofâ€Function Variants Cause Progressive Dystonia with Prominent Limb Involvement. Movement Disorders, 2022, 37, 137-147.	3.9	14
3	The phenotypic spectrum of PCDH12 associated disorders - Five new cases and review of the literature. European Journal of Paediatric Neurology, 2022, 36, 7-13.	1.6	4
4	Brain ventricles as windows into brain development and disease. Neuron, 2022, 110, 12-15.	8.1	23
5	Computational Genomics in the Era of Precision Medicine: Applications to Variant Analysis and Gene Therapy. Journal of Personalized Medicine, 2022, 12, 175.	2.5	4
6	Variants in Mitochondrial <scp>ATP</scp> Synthase Cause Variable Neurologic Phenotypes. Annals of Neurology, 2022, 91, 225-237.	5.3	12
7	Sequencing of a Chinese tetralogy of Fallot cohort reveals clustering mutations in myogenic heart progenitors. JCI Insight, 2022, 7, .	5.0	9
8	Genomic approaches to improve the clinical diagnosis and management of patients with congenital hydrocephalus. Journal of Neurosurgery: Pediatrics, 2022, 29, 168-177.	1.3	6
9	Impaired neurogenesis alters brain biomechanics in a neuroprogenitor-based genetic subtype of congenital hydrocephalus. Nature Neuroscience, 2022, 25, 458-473.	14.8	46
10	Whole-exome sequencing reveals damaging gene variants associated with hypoalphalipoproteinemia. Journal of Lipid Research, 2022, 63, 100209.	4.2	2
11	Biallelic Variants in the Ectonucleotidase <scp>ENTPD1</scp> Cause a Complex Neurodevelopmental Disorder with Intellectual Disability, Distinct White Matter Abnormalities, and Spastic Paraplegia. Annals of Neurology, 2022, 92, 304-321.	5.3	2
12	Mutation spectrum of congenital heart disease in a consanguineous Turkish population. Molecular Genetics & Genomic Medicine, 2022, 10, e1944.	1.2	4
13	Network assisted analysis of de novo variants using protein-protein interaction information identified 46 candidate genes for congenital heart disease. PLoS Genetics, 2022, 18, e1010252.	3.5	3
14	Association of Damaging Variants in Genes With Increased Cancer Risk Among Patients With Congenital Heart Disease. JAMA Cardiology, 2021, 6, 457.	6.1	34
15	Variant recurrence confirms the existence of a FBXO31â€related spasticâ€dystonic cerebral palsy syndrome. Annals of Clinical and Translational Neurology, 2021, 8, 951-955.	3.7	6
16	Analysis workflow to assess de novo genetic variants from human whole-exome sequencing. STAR Protocols, 2021, 2, 100383.	1.2	7
17	Exome Sequencing as a Potential Diagnostic Adjunct in Sporadic Congenital Hydrocephalus. JAMA Pediatrics, 2021, 175, 310.	6.2	10
18	Recessive COL4A2 Mutation Leads to Intellectual Disability, Epilepsy, and Spastic Cerebral Palsy. Neurology: Genetics, 2021, 7, e583.	1.9	3

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19	Biallelic variants in HPDL cause pure and complicated hereditary spastic paraplegia. Brain, 2021, 144, 1422-1434.	7.6	22
20	Molecular Genetics and Complex Inheritance of Congenital Heart Disease. Genes, 2021, 12, 1020.	2.4	46
21	Protein kinase <scp>D1</scp> variant associated with human epilepsy and peripheral nerve hypermyelination. Clinical Genetics, 2021, 100, 176-186.	2.0	1
22	Mutation in ZDHHC15 Leads to Hypotonic Cerebral Palsy, Autism, Epilepsy, and Intellectual Disability. Neurology: Genetics, 2021, 7, e602.	1.9	7
23	Genomics of human congenital hydrocephalus. Child's Nervous System, 2021, 37, 3325-3340.	1.1	12
24	Biallelic loss-of-function variants in the splicing regulator NSRP1 cause a severe neurodevelopmental disorder with spastic cerebral palsy and epilepsy. Genetics in Medicine, 2021, 23, 2455-2460.	2.4	9
25	DIAPH1 Variants in Nonâ€“East Asian Patients With Sporadic Moyamoya Disease. JAMA Neurology, 2021, 78, 993.	9.0	33
26	Integrative modeling of transmitted and de novo variants identifies novel risk genes for congenital heart disease. Quantitative Biology, 2021, 9, 216-227.	0.5	4
27	Bi-allelic variants in SPATA5L1 lead to intellectual disability, spastic-dystonic cerebral palsy, epilepsy, and hearing loss. American Journal of Human Genetics, 2021, 108, 2006-2016.	6.2	11
28	PTEN mutations in autism spectrum disorder and congenital hydrocephalus: developmental pleiotropy and therapeutic targets. Trends in Neurosciences, 2021, 44, 961-976.	8.6	19
29	The Phenotypic Spectrum of PCDH12-Associated Disorders: Five New Cases and Review of the Literature. , 2021, 52, .		0
30	PPIL4 is essential for brain angiogenesis and implicated in intracranial aneurysms in humans. Nature Medicine, 2021, 27, 2165-2175.	30.7	23
31	Exome sequencing implicates genetic disruption of prenatal neuro-gliogenesis in sporadic congenital hydrocephalus. Nature Medicine, 2020, 26, 1754-1765.	30.7	84
32	Mutations disrupting neuritogenesis genes confer risk for cerebral palsy. Nature Genetics, 2020, 52, 1046-1056.	21.4	96
33	Exome Sequencing Implicates Impaired GABA Signaling and Neuronal Ion Transport in Trigeminal Neuralgia. IScience, 2020, 23, 101552.	4.1	32
34	Loss of TNR causes a nonprogressive neurodevelopmental disorder with spasticity and transient opisthotonus. Genetics in Medicine, 2020, 22, 1061-1068.	2.4	14
35	Insights From Genetic Studies of Cerebral Palsy. Frontiers in Neurology, 2020, 11, 625428.	2.4	18
36	Recessive Inheritance of Congenital Hydrocephalus With Other Structural Brain Abnormalities Caused by Compound Heterozygous Mutations in ATP1A3. Frontiers in Cellular Neuroscience, 2019, 13, 425.	3.7	14

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37	Exome Sequencing Implicates SWI/SNF Chromatin Remodeling Genes in Human Congenital Hydrocephalus. Neurosurgery, 2019, 66, 310-133.	1.1	0
38	Exome Sequencing Defines the Molecular Pathogenesis of Vein of Galen Malformation. Neurosurgery, 2019, 66, 310-341.	1.1	1
39	SLC12A ion transporter mutations in sporadic and familial human congenital hydrocephalus. Molecular Genetics & Genomic Medicine, 2019, 7, e892.	1.2	22
40	Histone H2B monoubiquitination regulates heart development via epigenetic control of cilia motility. Proceedings of the National Academy of Sciences of the United States of America, 2019, 116, 14049-14054.	7.1	30
41	Unique features in the intracellular transport of typhoid toxin revealed by a genome-wide screen. PLoS Pathogens, 2019, 15, e1007704.	4.7	33
42	EphrinB2-EphB4-RASA1 Signaling in Human Cerebrovascular Development and Disease. Trends in Molecular Medicine, 2019, 25, 265-286.	6.7	39
43	Mutations in TFAP2B and previously unimplicated genes of the BMP, Wnt, and Hedgehog pathways in syndromic craniosynostosis. Proceedings of the National Academy of Sciences of the United States of America, 2019, 116, 15116-15121.	7.1	24
44	Mutations in Chromatin Modifier and Ephrin Signaling Genes in Vein of Galen Malformation. Neuron, 2019, 101, 429-443.e4.	8.1	56
45	CLCN2 chloride channel mutations in familial hyperaldosteronism type II. Nature Genetics, 2018, 50, 349-354.	21.4	188
46	Human Genetics and Molecular Mechanisms of Congenital Hydrocephalus. World Neurosurgery, 2018, 119, 441-443.	1.3	12
47	De Novo Pathogenic Variants in CACNA1E Cause Developmental and Epileptic Encephalopathy with Contractures, Macrocephaly, and Dyskinesias. American Journal of Human Genetics, 2018, 103, 666-678.	6.2	87
48	A novel association of campomelic dysplasia and hydrocephalus with an unbalanced chromosomal translocation upstream of SOX9. Journal of Physical Education and Sports Management, 2018, 4, a002766.	1.2	8
49	De Novo Mutation in Genes Regulating Neural Stem Cell Fate in Human Congenital Hydrocephalus. Neuron, 2018, 99, 302-314.e4.	8.1	112
50	De novo MYH9 mutation in congenital scalp hemangioma. Journal of Physical Education and Sports Management, 2018, 4, a002998.	1.2	9
51	A common haplotype lowers PU.1 expression in myeloid cells and delays onset of Alzheimer's disease. Nature Neuroscience, 2017, 20, 1052-1061.	14.8	330
52	Contribution of rare inherited and de novo variants in 2,871 congenital heart disease probands. Nature Genetics, 2017, 49, 1593-1601.	21.4	624
53	Alzheimer's diseaseâ€associated TREM2 variants exhibit either decreased or increased ligandâ€dependent activation. Alzheimer's and Dementia, 2017, 13, 381-387.	0.8	192
54	Digenic mutations of human OCRL paralogs in Dentâ€™s disease type 2 associated with Chiari I malformation. Human Genome Variation, 2016, 3, 16042.	0.7	8

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55	O2â€10â€06: A Common Allele in SPI1 Lowers Risk and Delays Age at Onset for Alzheimer's Disease. Alzheimer's and Dementia, 2016, 12, P253.	0.8	0
56	Resequencing analysis of five Mendelian genes andÂthe top genes from genome-wide association studies in Parkinsonâ€™s Disease. Molecular Neurodegeneration, 2016, 11, 29.	10.8	70
57	Pooled-DNA Sequencing for Elucidating New Genomic Risk Factors, Rare Variants Underlying Alzheimerâ€™s Disease. Methods in Molecular Biology, 2016, 1303, 299-314.	0.9	3
58	De novo mutations in congenital heart disease with neurodevelopmental and other congenital anomalies. Science, 2015, 350, 1262-1266.	12.6	646
59	TREM2 is associated with increased risk for Alzheimerâ€™s disease in African Americans. Molecular Neurodegeneration, 2015, 10, 19.	10.8	130
60	Coding variants in TREM2 increase risk for Alzheimer's disease. Human Molecular Genetics, 2014, 23, 5838-5846.	2.9	263
61	Rare coding variants in the phospholipase D3 gene confer risk for Alzheimerâ€™s disease. Nature, 2014, 505, 550-554.	27.8	425
62	Missense variant in TREML2 protects against Alzheimer's disease. Neurobiology of Aging, 2014, 35, 1510.e19-1510.e26.	3.1	110
63	GWAS of Cerebrospinal Fluid Tau Levels Identifies Risk Variants for Alzheimerâ€™s Disease. Neuron, 2013, 78, 256-268.	8.1	344
64	The PSEN1, p.E318G Variant Increases the Risk of Alzheimer's Disease in APOE-Îµ4 Carriers. PLoS Genetics, 2013, 9, e1003685.	3.5	55
65	Xâ€linked markers in the <scp>D</scp>uchenne muscular dystrophy gene associated with oral clefts. European Journal of Oral Sciences, 2013, 121, 63-68.	1.5	11
66	The FGF and FGFR Gene Family and Risk of Cleft Lip with or Without Cleft Palate. Cleft Palate-Craniofacial Journal, 2013, 50, 96-103.	0.9	39
67	Pooled-DNA sequencing identifies novel causative variants in PSEN1, GRN and MAPT in a clinical early-onset and familial Alzheimer's disease Ibero-American cohort. Alzheimer's Research and Therapy, 2012, 4, 34.	6.2	103
68	Evidence for gene-environment interaction in a genome wide study of nonsyndromic cleft palate. Genetic Epidemiology, 2011, 35, n/a-n/a.	1.3	145
69	A genome-wide association study of cleft lip with and without cleft palate identifies risk variants near MAFB and ABCA4. Nature Genetics, 2010, 42, 525-529.	21.4	518
70	Quantifying concordant genetic effects of de novo mutations on multiple disorders. ELife, 0, 11, .	6.0	3