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	De novo mutations in congenital heart disease with neurodevelopmental and other congenital anomalies. Science, 2015, 350, 1262-1266.	12.6	646
2	Contribution of rare inherited and de novo variants in 2,871 congenital heart disease probands. Nature Genetics, 2017, 49, 1593-1601.	21.4	624
3	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
4	Rare coding variants in the phospholipase D3 gene confer risk for Alzheimerâ€™s disease. Nature, 2014, 505, 550-554.	27.8	425
5	GWAS of Cerebrospinal Fluid Tau Levels Identifies Risk Variants for Alzheimerâ€™s Disease. Neuron, 2013, 78, 256-268.	8.1	344
6	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
7	Coding variants in TREM2 increase risk for Alzheimer's disease. Human Molecular Genetics, 2014, 23, 5838-5846.	2.9	263
8	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
9	CLCN2 chloride channel mutations in familial hyperaldosteronism type II. Nature Genetics, 2018, 50, 349-354.	21.4	188
10	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
11	TREM2 is associated with increased risk for Alzheimerâ€™s disease in African Americans. Molecular Neurodegeneration, 2015, 10, 19.	10.8	130
12	De Novo Mutation in Genes Regulating Neural Stem Cell Fate in Human Congenital Hydrocephalus. Neuron, 2018, 99, 302-314.e4.	8.1	112
13	Missense variant in TREML2 protects against Alzheimer's disease. Neurobiology of Aging, 2014, 35, 1510.e19-1510.e26.	3.1	110
14	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
15	Mutations disrupting neuritogenesis genes confer risk for cerebral palsy. Nature Genetics, 2020, 52, 1046-1056.	21.4	96
16	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
17	Exome sequencing implicates genetic disruption of prenatal neuro-gliogenesis in sporadic congenital hydrocephalus. Nature Medicine, 2020, 26, 1754-1765.	30.7	84
18	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

#	Article	IF	Citations
19	Mutations in Chromatin Modifier and Ephrin Signaling Genes in Vein of Galen Malformation. Neuron, 2019, 101, 429-443.e4.	8.1	56
20	The PSEN1, p.E318G Variant Increases the Risk of Alzheimer's Disease in APOE-Îµ4 Carriers. PLoS Genetics, 2013, 9, e1003685.	3.5	55
21	Molecular Genetics and Complex Inheritance of Congenital Heart Disease. Genes, 2021, 12, 1020.	2.4	46
22	Impaired neurogenesis alters brain biomechanics in a neuroprogenitor-based genetic subtype of congenital hydrocephalus. Nature Neuroscience, 2022, 25, 458-473.	14.8	46
23	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
24	EphrinB2-EphB4-RASA1 Signaling in Human Cerebrovascular Development and Disease. Trends in Molecular Medicine, 2019, 25, 265-286.	6.7	39
25	Association of Damaging Variants in Genes With Increased Cancer Risk Among Patients With Congenital Heart Disease. JAMA Cardiology, 2021, 6, 457.	6.1	34
26	Unique features in the intracellular transport of typhoid toxin revealed by a genome-wide screen. PLoS Pathogens, 2019, 15, e1007704.	4.7	33
27	DIAPH1 Variants in Nonâ€“East Asian Patients With Sporadic Moyamoya Disease. JAMA Neurology, 2021, 78, 993.	9.0	33
28	Exome Sequencing Implicates Impaired GABA Signaling and Neuronal Ion Transport in Trigeminal Neuralgia. IScience, 2020, 23, 101552.	4.1	32
29	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
30	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
31	Brain ventricles as windows into brain development and disease. Neuron, 2022, 110, 12-15.	8.1	23
32	PPIL4 is essential for brain angiogenesis and implicated in intracranial aneurysms in humans. Nature Medicine, 2021, 27, 2165-2175.	30.7	23
33	SLC12A ion transporter mutations in sporadic and familial human congenital hydrocephalus. Molecular Genetics & Genomic Medicine, 2019, 7, e892.	1.2	22
34	Biallelic variants in HPDL cause pure and complicated hereditary spastic paraplegia. Brain, 2021, 144, 1422-1434.	7.6	22
35	PTEN mutations in autism spectrum disorder and congenital hydrocephalus: developmental pleiotropy and therapeutic targets. Trends in Neurosciences, 2021, 44, 961-976.	8.6	19
36	Insights From Genetic Studies of Cerebral Palsy. Frontiers in Neurology, 2020, 11, 625428.	2.4	18

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37	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
38	Loss of TNR causes a nonprogressive neurodevelopmental disorder with spasticity and transient opisthotonus. Genetics in Medicine, 2020, 22, 1061-1068.	2.4	14
39	Biallelic <scp>AOPEP</scp> Lossâ€ofâ€Function Variants Cause Progressive Dystonia with Prominent Limb Involvement. Movement Disorders, 2022, 37, 137-147.	3.9	14
40	Human Genetics and Molecular Mechanisms of Congenital Hydrocephalus. World Neurosurgery, 2018, 119, 441-443.	1.3	12
41	Genomics of human congenital hydrocephalus. Child's Nervous System, 2021, 37, 3325-3340.	1.1	12
42	Variants in Mitochondrial <scp>ATP</scp> Synthase Cause Variable Neurologic Phenotypes. Annals of Neurology, 2022, 91, 225-237.	5.3	12
43	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
44	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
45	Exome Sequencing as a Potential Diagnostic Adjunct in Sporadic Congenital Hydrocephalus. JAMA Pediatrics, 2021, 175, 310.	6.2	10
46	De novo MYH9 mutation in congenital scalp hemangioma. Journal of Physical Education and Sports Management, 2018, 4, a002998.	1.2	9
47	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
48	Sequencing of a Chinese tetralogy of Fallot cohort reveals clustering mutations in myogenic heart progenitors. JCI Insight, 2022, 7, .	5.0	9
49	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
50	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
51	Analysis workflow to assess de novo genetic variants from human whole-exome sequencing. STAR Protocols, 2021, 2, 100383.	1.2	7
52	Mutation in ZDHHC15 Leads to Hypotonic Cerebral Palsy, Autism, Epilepsy, and Intellectual Disability. Neurology: Genetics, 2021, 7, e602.	1.9	7
53	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
54	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

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55	Familial and syndromic forms of arachnoid cyst implicate genetic factors in disease pathogenesis. Cerebral Cortex, 2023, 33, 3012-3025.	2.9	6
56	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
57	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
58	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
59	Mutation spectrum of congenital heart disease in a consanguineous Turkish population. Molecular Genetics & Genomic Medicine, 2022, 10, e1944.	1.2	4
60	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
61	Recessive COL4A2 Mutation Leads to Intellectual Disability, Epilepsy, and Spastic Cerebral Palsy. Neurology: Genetics, 2021, 7, e583.	1.9	3
62	Quantifying concordant genetic effects of de novo mutations on multiple disorders. ELife, 0, 11, .	6.0	3
63	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
64	Whole-exome sequencing reveals damaging gene variants associated with hypoalphalipoproteinemia. Journal of Lipid Research, 2022, 63, 100209.	4.2	2
65	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
66	Exome Sequencing Defines the Molecular Pathogenesis of Vein of Galen Malformation. Neurosurgery, 2019, 66, 310-341.	1.1	1
67	Protein kinase <scp>D1</scp> variant associated with human epilepsy and peripheral nerve hypermyelination. Clinical Genetics, 2021, 100, 176-186.	2.0	1
68	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
69	Exome Sequencing Implicates SWI/SNF Chromatin Remodeling Genes in Human Congenital Hydrocephalus. Neurosurgery, 2019, 66, 310-133.	1.1	0
70	The Phenotypic Spectrum of PCDH12-Associated Disorders: Five New Cases and Review of the Literature. , 2021, 52, .		0