

Yufeng Shen

List of Publications by Year in descending order

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Version: 2024-02-01

95
papers

9,290
citations

53794

45
h-index

48315

88
g-index

112
all docs

112
docs citations

112
times ranked

18369
citing authors

#	ARTICLE	IF	CITATIONS
1	Imputing cognitive impairment in <sc>SPARK</sc>, a large autism cohort. Autism Research, 2022, 15, 156-170.	3.8	12
2	The genetic architecture of pediatric cardiomyopathy. American Journal of Human Genetics, 2022, 109, 282-298.	6.2	21
3	Genome-Wide De Novo Variants in Congenital Heart Disease Are Not Associated With Maternal Diabetes or Obesity. Circulation Genomic and Precision Medicine, 2022, 15, CIRCGEN121003500.	3.6	8
4	Neither cardiac mitochondrial DNA variation nor copy number contribute to congenital heart disease risk. American Journal of Human Genetics, 2022, 109, 961-966.	6.2	5
5	Identification and validation of candidate risk genes in endocytic vesicular trafficking associated with esophageal atresia and tracheoesophageal fistulas. Human Genetics and Genomics Advances, 2022, 3, 100107.	1.7	2
6	Discovering the Developmental Basis of Trachea–Esophageal Birth Defects: Evidence for Endosome–opathies. FASEB Journal, 2022, 36, .	0.5	0
7	Clinical and genetic characterization of <sc><i>CACNA1A</i></sc>-related disease. Clinical Genetics, 2022, 102, 288-295.	2.0	9
8	Newborn screening for neurodevelopmental diseases: Are we there yet?. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2022, 190, 222-230.	1.6	10
9	Bayesian Inference Associates Rare <i>KDR</i> Variants With Specific Phenotypes in Pulmonary Arterial Hypertension. Circulation Genomic and Precision Medicine, 2021, 14, .	3.6	29
10	Novel candidate genes in esophageal atresia/tracheoesophageal fistula identified by exome sequencing. European Journal of Human Genetics, 2021, 29, 122-130.	2.8	17
11	Association of Damaging Variants in Genes With Increased Cancer Risk Among Patients With Congenital Heart Disease. JAMA Cardiology, 2021, 6, 457.	6.1	34
12	Beta-lactam-induced immediate hypersensitivity reactions: A–genome-wide association study of a deeply phenotyped cohort. Journal of Allergy and Clinical Immunology, 2021, 147, 1830-1837.e15.	2.9	26
13	MVP predicts the–pathogenicity of missense variants by deep learning. Nature Communications, 2021, 12, 510.	12.8	85
14	Functional interrogation of DNA damage response variants with base editing screens. Cell, 2021, 184, 1081-1097.e19.	28.9	145
15	Human plasmacytoid dendritic cells mount a distinct antiviral response to virus-infected cells. Science Immunology, 2021, 6, .	11.9	28
16	Lymphohematopoietic graft-versus-host responses promote mixed chimerism in patients receiving intestinal transplantation. Journal of Clinical Investigation, 2021, 131, .	8.2	31
17	Genotype and defects in microtubule-based motility correlate with clinical severity in KIF1A-associated neurological disorder. Human Genetics and Genomics Advances, 2021, 2, 100026.	1.7	34
18	Penetrance of Breast Cancer Susceptibility Genes from the eMERGE III Network. JNCI Cancer Spectrum, 2021, 5, pkab044.	2.9	14

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19	Rare variant analysis of 4241 pulmonary arterial hypertension cases from an international consortium implicates FBLN2, PDGFD, and rare de novo variants in PAH. <i>Genome Medicine</i> , 2021, 13, 80.	8.2	43
20	Medical Records-Based Genetic Studies of the Complement System. <i>Journal of the American Society of Nephrology: JASN</i> , 2021, 32, 2031-2047.	6.1	10
21	Recent ultra-rare inherited variants implicate new autism candidate risk genes. <i>Nature Genetics</i> , 2021, 53, 1125-1134.	21.4	68
22	Gene expression atlas of energy balance brain regions. <i>JCI Insight</i> , 2021, 6, .	5.0	6
23	Developmental basis of trachea-esophageal birth defects. <i>Developmental Biology</i> , 2021, 477, 85-97.	2.0	21
24	Rare and de novo variants in 827 congenital diaphragmatic hernia probands implicate LONP1 as candidate risk gene. <i>American Journal of Human Genetics</i> , 2021, 108, 1964-1980.	6.2	22
25	Integrated analysis toolset for defining and tracking alloreactive T-cell clones after human solid organ and hematopoietic stem cell transplantation. <i>Software Impacts</i> , 2021, 10, 100142.	1.4	11
26	A disorder-related variant (E420K) of a PP2A-regulatory subunit (PPP2R5D) causes constitutively active AKT-mTOR signaling and uncoordinated cell growth. <i>Journal of Biological Chemistry</i> , 2021, 296, 100313.	3.4	18
27	Heterogeneity of human anti-viral immunity shaped by virus, tissue, age, and sex. <i>Cell Reports</i> , 2021, 37, 110071.	6.4	34
28	Deletion of donor-reactive T cell clones after human liver transplant. <i>American Journal of Transplantation</i> , 2020, 20, 538-545.	4.7	31
29	Comprehensive analyses of B-cell compartments across the human body reveal novel subsets and a gut-resident memory phenotype. <i>Blood</i> , 2020, 136, 2774-2785.	1.4	74
30	Likely damaging de novo variants in congenital diaphragmatic hernia patients are associated with worse clinical outcomes. <i>Genetics in Medicine</i> , 2020, 22, 2020-2028.	2.4	21
31	Deep whole-genome sequencing of multiple proband tissues and parental blood reveals the complex genetic etiology of congenital diaphragmatic hernias. <i>Human Genetics and Genomics Advances</i> , 2020, 1, 100008.	1.7	5
32	Novel Mutations and Decreased Expression of the Epigenetic Regulator <i>TET2</i> in Pulmonary Arterial Hypertension. <i>Circulation</i> , 2020, 141, 1986-2000.	1.6	75
33	Genomic analyses implicate noncoding de novo variants in congenital heart disease. <i>Nature Genetics</i> , 2020, 52, 769-777.	21.4	97
34	Tissue Determinants of Human NK Cell Development, Function, and Residence. <i>Cell</i> , 2020, 180, 749-763.e13.	28.9	242
35	Beliefs in vaccine as causes of autism among SPARK cohort caregivers. <i>Vaccine</i> , 2020, 38, 1794-1803.	3.8	12
36	EM-mosaic detects mosaic point mutations that contribute to congenital heart disease. <i>Genome Medicine</i> , 2020, 12, 42.	8.2	17

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37	Large-scale targeted sequencing identifies risk genes for neurodevelopmental disorders. <i>Nature Communications</i> , 2020, 11, 4932.	12.8	105
38	Template-based prediction of protein structure with deep learning. <i>BMC Genomics</i> , 2020, 21, 878.	2.8	15
39	Exome sequencing of 457 autism families recruited online provides evidence for autism risk genes. <i>Npj Genomic Medicine</i> , 2019, 4, 19.	3.8	163
40	Single cell RNA-Seq reveals pre-cDCs fate determined by transcription factor combinatorial dose. <i>BMC Molecular and Cell Biology</i> , 2019, 20, 20.	2.0	18
41	De novo and recessive forms of congenital heart disease have distinct genetic and phenotypic landscapes. <i>Nature Communications</i> , 2019, 10, 4722.	12.8	58
42	Shared Genetic Risk Factors Across Carbamazepine-Induced Hypersensitivity Reactions. <i>Clinical Pharmacology and Therapeutics</i> , 2019, 106, 1028-1036.	4.7	52
43	Novel risk genes and mechanisms implicated by exome sequencing of 2572 individuals with pulmonary arterial hypertension. <i>Genome Medicine</i> , 2019, 11, 69.	8.2	86
44	Recessive Rare Variants in Deoxyhypusine Synthase, an Enzyme Involved in the Synthesis of Hypusine, Are Associated with a Neurodevelopmental Disorder. <i>American Journal of Human Genetics</i> , 2019, 104, 287-298.	6.2	38
45	Human Intestinal Allografts Contain Functional Hematopoietic Stem and Progenitor Cells that Are Maintained by a Circulating Pool. <i>Cell Stem Cell</i> , 2019, 24, 227-239.e8.	11.1	43
46	Cross-reactive public TCR sequences undergo positive selection in the human thymic repertoire. <i>Journal of Clinical Investigation</i> , 2019, 129, 2446-2462.	8.2	55
47	Exome Sequencing in Children With Pulmonary Arterial Hypertension Demonstrates Differences Compared With Adults. <i>Circulation Genomic and Precision Medicine</i> , 2018, 11, e001887.	3.6	104
48	SPARK: A US Cohort of 50,000 Families to Accelerate Autism Research. <i>Neuron</i> , 2018, 97, 488-493.	8.1	265
49	Robust identification of mosaic variants in congenital heart disease. <i>Human Genetics</i> , 2018, 137, 183-193.	3.8	43
50	Early expansion of donor-specific Tregs in tolerant kidney transplant recipients. <i>JCI Insight</i> , 2018, 3, .	5.0	54
51	De novo variants in congenital diaphragmatic hernia identify MYRF as a new syndrome and reveal genetic overlaps with other developmental disorders. <i>PLoS Genetics</i> , 2018, 14, e1007822.	3.5	79
52	Quantifying size and diversity of the human T cell alloresponse. <i>JCI Insight</i> , 2018, 3, .	5.0	69
53	Distinct epigenomic patterns are associated with haploinsufficiency and predict risk genes of developmental disorders. <i>Nature Communications</i> , 2018, 9, 2138.	12.8	28
54	Rare variants in SOX17 are associated with pulmonary arterial hypertension with congenital heart disease. <i>Genome Medicine</i> , 2018, 10, 56.	8.2	112

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55	Human Lymph Nodes Maintain TCF-1hi Memory T Cells with High Functional Potential and Clonal Diversity throughout Life. <i>Journal of Immunology</i> , 2018, 201, 2132-2140.	0.8	63
56	High Dimensional Functionomic Analysis of Human Hematopoietic Stem and Progenitor Cells at a Single Cell Level. <i>Bio-protocol</i> , 2018, 8, e2851.	0.4	0
57	De novo missense variants in <i>HECW2</i> are associated with neurodevelopmental delay and hypotonia. <i>Journal of Medical Genetics</i> , 2017, 54, 84-86.	3.2	46
58	Genome-wide enrichment of damaging de novo variants in patients with isolated and complex congenital diaphragmatic hernia. <i>Human Genetics</i> , 2017, 136, 679-691.	3.8	53
59	Association of Liver Injury From Specific Drugs, or Groups of Drugs, With Polymorphisms in HLA and Other Genes in a Genome-Wide Association Study. <i>Gastroenterology</i> , 2017, 152, 1078-1089.	1.3	174
60	Contribution of rare inherited and de novo variants in 2,871 congenital heart disease probands. <i>Nature Genetics</i> , 2017, 49, 1593-1601.	21.4	624
61	Human Tissue-Resident Memory T Cells Are Defined by Core Transcriptional and Functional Signatures in Lymphoid and Mucosal Sites. <i>Cell Reports</i> , 2017, 20, 2921-2934.	6.4	792
62	Contrasting Determinants of Mutation Rates in Germline and Soma. <i>Genetics</i> , 2017, 207, 255-267.	2.9	24
63	Congenital diaphragmatic hernias: from genes to mechanisms to therapies. <i>DMM Disease Models and Mechanisms</i> , 2017, 10, 955-970.	2.4	143
64	Lineage specification of human dendritic cells is marked by IRF8 expression in hematopoietic stem cells and multipotent progenitors. <i>Nature Immunology</i> , 2017, 18, 877-888.	14.5	101
65	A Cell Type-Specific Expression Signature Predicts Haploinsufficient Autism-Susceptibility Genes. <i>Human Mutation</i> , 2017, 38, 204-215.	2.5	38
66	Long-term maintenance of human naïve T cells through in situ homeostasis in lymphoid tissue sites. <i>Science Immunology</i> , 2016, 1, .	11.9	127
67	A recurrent de novo CTBP1 mutation is associated with developmental delay, hypotonia, ataxia, and tooth enamel defects. <i>Neurogenetics</i> , 2016, 17, 173-178.	1.4	32
68	Deep Genetic Connection Between Cancer and Developmental Disorders. <i>Human Mutation</i> , 2016, 37, 1042-1050.	2.5	24
69	Variants in HNRNPH2 on the X Chromosome Are Associated with a Neurodevelopmental Disorder in Females. <i>American Journal of Human Genetics</i> , 2016, 99, 728-734.	6.2	75
70	Bidirectional intragraft alloreactivity drives the repopulation of human intestinal allografts and correlates with clinical outcome. <i>Science Immunology</i> , 2016, 1, .	11.9	98
71	Long-read sequencing and de novo assembly of a Chinese genome. <i>Nature Communications</i> , 2016, 7, 12065.	12.8	242
72	Loss of RNA expression and allele-specific expression associated with congenital heart disease. <i>Nature Communications</i> , 2016, 7, 12824.	12.8	51

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73	Rare variant phasing and haplotypic expression from RNA sequencing with phASER. <i>Nature Communications</i> , 2016, 7, 12817.	12.8	105
74	Diversity and divergence of the glioma-infiltrating T-cell receptor repertoire. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2016, 113, E3529-37.	7.1	103
75	Next-Generation Sequencing of Pulmonary Sarcomatoid Carcinoma Reveals High Frequency of Actionable <i>MET</i> Gene Mutations. <i>Journal of Clinical Oncology</i> , 2016, 34, 794-802.	1.6	287
76	Estimating heritability of drug-induced liver injury from common variants and implications for future study designs. <i>Scientific Reports</i> , 2015, 4, 5762.	3.3	8
77	SeqMule: automated pipeline for analysis of human exome/genome sequencing data. <i>Scientific Reports</i> , 2015, 5, 14283.	3.3	63
78	ABC Transporters and the Proteasome Complex Are Implicated in Susceptibility to Stevensâ€™Johnson Syndrome and Toxic Epidermal Necrolysis across Multiple Drugs. <i>PLoS ONE</i> , 2015, 10, e0131038.	2.5	9
79	Genomic Signatures of Cooperation and Conflict in the Social Amoeba. <i>Current Biology</i> , 2015, 25, 1661-1665.	3.9	51
80	RNA sequencing from human neutrophils reveals distinct transcriptional differences associated with chronic inflammatory states. <i>BMC Medical Genomics</i> , 2015, 8, 55.	1.5	61
81	Increased burden of <i>de novo</i> predicted deleterious variants in complex congenital diaphragmatic hernia. <i>Human Molecular Genetics</i> , 2015, 24, 4764-4773.	2.9	65
82	De novo mutations in congenital heart disease with neurodevelopmental and other congenital anomalies. <i>Science</i> , 2015, 350, 1262-1266.	12.6	646
83	Author response to comment on â€œTracking donor-reactive T cells: Evidence for clonal deletion in tolerant kidney transplant patientsâ€. <i>Science Translational Medicine</i> , 2015, 7, 297r1.	12.4	3
84	The support of human genetic evidence for approved drug indications. <i>Nature Genetics</i> , 2015, 47, 856-860.	21.4	1,112
85	Tracking donor-reactive T cells: Evidence for clonal deletion in tolerant kidney transplant patients. <i>Science Translational Medicine</i> , 2015, 7, 272ra10.	12.4	191
86	Mutations in <i>ARID2</i> are associated with intellectual disabilities. <i>Neurogenetics</i> , 2015, 16, 307-314.	1.4	54
87	CANOES: detecting rare copy number variants from whole exome sequencing data. <i>Nucleic Acids Research</i> , 2014, 42, e97-e97.	14.5	123
88	Whole exome sequencing identifies <i>de novo</i> mutations in <i>GATA6</i> associated with congenital diaphragmatic hernia. <i>Journal of Medical Genetics</i> , 2014, 51, 197-202.	3.2	55
89	Comparable Frequencies of Coding Mutations and Loss of Imprinting in Human Pluripotent Cells Derived by Nuclear Transfer and Defined Factors. <i>Cell Stem Cell</i> , 2014, 15, 634-642.	11.1	113
90	Spatial Map of Human T Cell Compartmentalization and Maintenance over Decades of Life. <i>Cell</i> , 2014, 159, 814-828.	28.9	476

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91	Increased Frequency of De Novo Copy Number Variants in Congenital Heart Disease by Integrative Analysis of Single Nucleotide Polymorphism Array and Exome Sequence Data. <i>Circulation Research</i> , 2014, 115, 884-896.	4.5	229
92	Exploring therapeutic targets in pulmonary sarcomatoid carcinoma by comprehensive genomic profiling.. <i>Journal of Clinical Oncology</i> , 2014, 32, 8073-8073.	1.6	0
93	Variants in GATA4 are a rare cause of familial and sporadic congenital diaphragmatic hernia. <i>Human Genetics</i> , 2013, 132, 285-292.	3.8	81
94	A parametric Bayesian method to test the association of rare variants. , 2011, , .		0
95	A homozygous splice variant in <i>ATP5PO</i> , disrupts mitochondrial complex V function and causes Leigh syndrome in two unrelated families. <i>Journal of Inherited Metabolic Disease</i> , 0, , .	3.6	1