Yufeng Shen

List of Publications by Year in descending order

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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	The support of human genetic evidence for approved drug indications. Nature Genetics, 2015, 47, 856-860.	21.4	1,112
2	Human Tissue-Resident Memory T Cells Are Defined by Core Transcriptional and Functional Signatures in Lymphoid and Mucosal Sites. Cell Reports, 2017, 20, 2921-2934.	6.4	792
3	De novo mutations in congenital heart disease with neurodevelopmental and other congenital anomalies. Science, 2015, 350, 1262-1266.	12.6	646
4	Contribution of rare inherited and de novo variants in 2,871 congenital heart disease probands. Nature Genetics, 2017, 49, 1593-1601.	21.4	624
5	Spatial Map of Human T Cell Compartmentalization and Maintenance over Decades of Life. Cell, 2014, 159, 814-828.	28.9	476
6	Next-Generation Sequencing of Pulmonary Sarcomatoid Carcinoma Reveals High Frequency of Actionable MET Gene Mutations. Journal of Clinical Oncology, 2016, 34, 794-802.	1.6	287
7	SPARK: A US Cohort of 50,000 Families to Accelerate Autism Research. Neuron, 2018, 97, 488-493.	8.1	265
8	Long-read sequencing and de novo assembly of a Chinese genome. Nature Communications, 2016, 7, 12065.	12.8	242
9	Tissue Determinants of Human NK Cell Development, Function, and Residence. Cell, 2020, 180, 749-763.e13.	28.9	242
10	Increased Frequency of De Novo Copy Number Variants in Congenital Heart Disease by Integrative Analysis of Single Nucleotide Polymorphism Array and Exome Sequence Data. Circulation Research, 2014, 115, 884-896.	4.5	229
11	Tracking donor-reactive T cells: Evidence for clonal deletion in tolerant kidney transplant patients. Science Translational Medicine, 2015, 7, 272ra10.	12.4	191
12	Association of Liver Injury From Specific Drugs, or Groups ofÂDrugs, With Polymorphisms in HLA and Other Genes in aÂGenome-Wide Association Study. Gastroenterology, 2017, 152, 1078-1089.	1.3	174
13	Exome sequencing of 457 autism families recruited online provides evidence for autism risk genes. Npj Genomic Medicine, 2019, 4, 19.	3.8	163
14	Functional interrogation of DNA damage response variants with base editing screens. Cell, 2021, 184, 1081-1097.e19.	28.9	145
15	Congenital diaphragmatic hernias: from genes to mechanisms to therapies. DMM Disease Models and Mechanisms, 2017, 10, 955-970.	2.4	143
16	Long-term maintenance of human naÃ¯ve T cells through in situ homeostasis in lymphoid tissue sites. Science Immunology, 2016, 1, .	11.9	127
17	CANOES: detecting rare copy number variants from whole exome sequencing data. Nucleic Acids Research, 2014, 42, e97-e97.	14.5	123
18	Comparable Frequencies of Coding Mutations and Loss of Imprinting in Human Pluripotent Cells Derived by Nuclear Transfer and Defined Factors. Cell Stem Cell, 2014, 15, 634-642.	11.1	113

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19	Rare variants in SOX17 are associated with pulmonary arterial hypertension with congenital heart disease. Genome Medicine, 2018, 10, 56.	8.2	112
20	Rare variant phasing and haplotypic expression from RNA sequencing with phASER. Nature Communications, 2016, 7, 12817.	12.8	105
21	Large-scale targeted sequencing identifies risk genes for neurodevelopmental disorders. Nature Communications, 2020, 11, 4932.	12.8	105
22	Exome Sequencing in Children With Pulmonary Arterial Hypertension Demonstrates Differences Compared With Adults. Circulation Genomic and Precision Medicine, 2018, 11, e001887.	3.6	104
23	Diversity and divergence of the glioma-infiltrating T-cell receptor repertoire. Proceedings of the National Academy of Sciences of the United States of America, 2016, 113, E3529-37.	7.1	103
24	Lineage specification of human dendritic cells is marked by IRF8 expression in hematopoietic stem cells and multipotent progenitors. Nature Immunology, 2017, 18, 877-888.	14.5	101
25	Bidirectional intragraft alloreactivity drives the repopulation of human intestinal allografts and correlates with clinical outcome. Science Immunology, 2016, 1, .	11.9	98
26	Genomic analyses implicate noncoding de novo variants in congenital heart disease. Nature Genetics, 2020, 52, 769-777.	21.4	97
27	Novel risk genes and mechanisms implicated by exome sequencing of 2572 individuals with pulmonary arterial hypertension. Genome Medicine, 2019, 11, 69.	8.2	86
28	MVP predicts theÂpathogenicity of missense variants by deep learning. Nature Communications, 2021, 12, 510.	12.8	85
29	Variants in GATA4 are a rare cause of familial and sporadic congenital diaphragmatic hernia. Human Genetics, 2013, 132, 285-292.	3.8	81
30	De novo variants in congenital diaphragmatic hernia identify MYRF as a new syndrome and reveal genetic overlaps with other developmental disorders. PLoS Genetics, 2018, 14, e1007822.	3.5	79
31	Variants in HNRNPH2 on the X Chromosome Are Associated with a Neurodevelopmental Disorder in Females. American Journal of Human Genetics, 2016, 99, 728-734.	6.2	75
32	Novel Mutations and Decreased Expression of the Epigenetic Regulator TET2 in Pulmonary Arterial Hypertension. Circulation, 2020, 141, 1986-2000.	1.6	75
33	Comprehensive analyses of B-cell compartments across the human body reveal novel subsets and a gut-resident memory phenotype. Blood, 2020, 136, 2774-2785.	1.4	74
34	Quantifying size and diversity of the human T cell alloresponse. JCI Insight, 2018, 3, .	5.0	69
35	Recent ultra-rare inherited variants implicate new autism candidate risk genes. Nature Genetics, 2021, 53, 1125-1134.	21.4	68
36	Increased burden ofde novopredicted deleterious variants in complex congenital diaphragmatic hernia. Human Molecular Genetics, 2015, 24, 4764-4773.	2.9	65

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37	SeqMule: automated pipeline for analysis of human exome/genome sequencing data. Scientific Reports, 2015, 5, 14283.	3.3	63
38	Human Lymph Nodes Maintain TCF-1hi Memory T Cells with High Functional Potential and Clonal Diversity throughout Life. Journal of Immunology, 2018, 201, 2132-2140.	0.8	63
39	RNA sequencing from human neutrophils reveals distinct transcriptional differences associated with chronic inflammatory states. BMC Medical Genomics, 2015, 8, 55.	1.5	61
40	De novo and recessive forms of congenital heart disease have distinct genetic and phenotypic landscapes. Nature Communications, 2019, 10, 4722.	12.8	58
41	Whole exome sequencing identifies de novo mutations inGATA6associated with congenital diaphragmatic hernia. Journal of Medical Genetics, 2014, 51, 197-202.	3.2	55
42	Cross-reactive public TCR sequences undergo positive selection in the human thymic repertoire. Journal of Clinical Investigation, 2019, 129, 2446-2462.	8.2	55
43	Mutations in ARID2 are associated with intellectual disabilities. Neurogenetics, 2015, 16, 307-314.	1.4	54
44	Early expansion of donor-specific Tregs in tolerant kidney transplant recipients. JCI Insight, 2018, 3, .	5.0	54
45	Genome-wide enrichment of damaging de novo variants in patients with isolated and complex congenital diaphragmatic hernia. Human Genetics, 2017, 136, 679-691.	3.8	53
46	Shared Genetic Risk Factors Across Carbamazepineâ€Induced Hypersensitivity Reactions. Clinical Pharmacology and Therapeutics, 2019, 106, 1028-1036.	4.7	52
47	Genomic Signatures of Cooperation and Conflict in the Social Amoeba. Current Biology, 2015, 25, 1661-1665.	3.9	51
48	Loss of RNA expression and allele-specific expression associated with congenital heart disease. Nature Communications, 2016, 7, 12824.	12.8	51
49	De novo missense variants inHECW2are associated with neurodevelopmental delay and hypotonia. Journal of Medical Genetics, 2017, 54, 84-86.	3.2	46
50	Robust identification of mosaic variants in congenital heart disease. Human Genetics, 2018, 137, 183-193.	3.8	43
51	Human Intestinal Allografts Contain Functional Hematopoietic Stem and Progenitor Cells that Are Maintained by a Circulating Pool. Cell Stem Cell, 2019, 24, 227-239.e8.	11.1	43
52	Rare variant analysis of 4241 pulmonary arterial hypertension cases from an international consortium implicates FBLN2, PDGFD, and rare de novo variants in PAH. Genome Medicine, 2021, 13, 80.	8.2	43
53	A Cell Type-Specific Expression Signature Predicts Haploinsufficient Autism-Susceptibility Genes. Human Mutation, 2017, 38, 204-215.	2.5	38
54	Recessive Rare Variants in Deoxyhypusine Synthase, an Enzyme Involved in the Synthesis of Hypusine, Are Associated with a Neurodevelopmental Disorder. American Journal of Human Genetics, 2019, 104, 287-298.	6.2	38

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55	Association of Damaging Variants in Genes With Increased Cancer Risk Among Patients With Congenital Heart Disease. JAMA Cardiology, 2021, 6, 457.	6.1	34
56	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
57	Heterogeneity of human anti-viral immunity shaped by virus, tissue, age, and sex. Cell Reports, 2021, 37, 110071.	6.4	34
58	A recurrent de novo CTBP1 mutation is associated with developmental delay, hypotonia, ataxia, and tooth enamel defects. Neurogenetics, 2016, 17, 173-178.	1.4	32
59	Deletion of donor-reactive T cell clones after human liver transplant. American Journal of Transplantation, 2020, 20, 538-545.	4.7	31
60	Lymphohematopoietic graft-versus-host responses promote mixed chimerism in patients receiving intestinal transplantation. Journal of Clinical Investigation, 2021, 131, .	8.2	31
61	Bayesian Inference Associates Rare KDR Variants With Specific Phenotypes in Pulmonary Arterial Hypertension. Circulation Genomic and Precision Medicine, 2021, 14, .	3.6	29
62	Distinct epigenomic patterns are associated with haploinsufficiency and predict risk genes of developmental disorders. Nature Communications, 2018, 9, 2138.	12.8	28
63	Human plasmacytoid dendritic cells mount a distinct antiviral response to virus-infected cells. Science Immunology, 2021, 6, .	11.9	28
64	Beta-lactam-induced immediate hypersensitivity reactions: AÂgenome-wide association study of a deeply phenotyped cohort. Journal of Allergy and Clinical Immunology, 2021, 147, 1830-1837.e15.	2.9	26
65	Deep Genetic Connection Between Cancer and Developmental Disorders. Human Mutation, 2016, 37, 1042-1050.	2.5	24
66	Contrasting Determinants of Mutation Rates in Germline and Soma. Genetics, 2017, 207, 255-267.	2.9	24
67	Rare and de novo variants in 827 congenital diaphragmatic hernia probands implicate LONP1 as candidate risk gene. American Journal of Human Genetics, 2021, 108, 1964-1980.	6.2	22
68	Likely damaging de novo variants in congenital diaphragmatic hernia patients are associated with worse clinical outcomes. Genetics in Medicine, 2020, 22, 2020-2028.	2.4	21
69	Developmental basis of trachea-esophageal birth defects. Developmental Biology, 2021, 477, 85-97.	2.0	21
70	The genetic architecture of pediatric cardiomyopathy. American Journal of Human Genetics, 2022, 109, 282-298.	6.2	21
71	Single cell RNA-Seq reveals pre-cDCs fate determined by transcription factor combinatorial dose. BMC Molecular and Cell Biology, 2019, 20, 20.	2.0	18
72	A disorder-related variant (E420K) of a PP2A-regulatory subunit (PPP2R5D) causes constitutively active AKT-mTOR signaling and uncoordinated cell growth. Journal of Biological Chemistry, 2021, 296, 100313.	3.4	18

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73	EM-mosaic detects mosaic point mutations that contribute to congenital heart disease. Genome Medicine, 2020, 12, 42.	8.2	17
74	Novel candidate genes in esophageal atresia/tracheoesophageal fistula identified by exome sequencing. European Journal of Human Genetics, 2021, 29, 122-130.	2.8	17
75	Template-based prediction of protein structure with deep learning. BMC Genomics, 2020, 21, 878.	2.8	15
76	Penetrance of Breast Cancer Susceptibility Genes from the eMERGE III Network. JNCI Cancer Spectrum, 2021, 5, pkab044.	2.9	14
77	Beliefs in vaccine as causes of autism among SPARK cohort caregivers. Vaccine, 2020, 38, 1794-1803.	3.8	12
78	Imputing cognitive impairment in <scp>SPARK</scp>, a large autism cohort. Autism Research, 2022, 15, 156-170.	3.8	12
79	Integrated analysis toolset for defining and tracking alloreactive T-cell clones after human solid organ and hematopoietic stem cell transplantation. Software Impacts, 2021, 10, 100142.	1.4	11
80	Medical Records-Based Genetic Studies of the Complement System. Journal of the American Society of Nephrology: JASN, 2021, 32, 2031-2047.	6.1	10
81	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
82	ABC Transporters and the Proteasome Complex Are Implicated in Susceptibility to Stevensâ€“Johnson Syndrome and Toxic Epidermal Necrolysis across Multiple Drugs. PLoS ONE, 2015, 10, e0131038.	2.5	9
83	Clinical and genetic characterization of <scp>CACNA1A</scp>â€related disease. Clinical Genetics, 2022, 102, 288-295.	2.0	9
84	Estimating heritability of drug-induced liver injury from common variants and implications for future study designs. Scientific Reports, 2015, 4, 5762.	3.3	8
85	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
86	Gene expression atlas of energy balance brain regions. JCI Insight, 2021, 6, .	5.0	6
87	Deep whole-genome sequencing of multiple proband tissues and parental blood reveals the complex genetic etiology of congenital diaphragmatic hernias. Human Genetics and Genomics Advances, 2020, 1, 100008.	1.7	5
88	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
89	Author response to comment on â€œTracking donor-reactive T cells: Evidence for clonal deletion in tolerant kidney transplant patientsâ€• Science Translational Medicine, 2015, 7, 297lr1.	12.4	3
90	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

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91	A homozygous splice variant in ATP5PO , disrupts mitochondrial complex V function and causes Leigh syndrome in two unrelated families. Journal of Inherited Metabolic Disease, 0, , .	3.6	1
92	A parametric Bayesian method to test the association of rare variants. , 2011, , .		0
93	Exploring therapeutic targets in pulmonary sarcomatoid carcinoma by comprehensive genomic profiling.. Journal of Clinical Oncology, 2014, 32, 8073-8073.	1.6	0
94	High Dimensional Functionomic Analysis of Human Hematopoietic Stem and Progenitor Cells at a Single Cell Level. Bio-protocol, 2018, 8, e2851.	0.4	0
95	Discovering the Developmental Basis of Tracheaâ€Esophageal Birth Defects: Evidence for Endosomeâ€opathies. FASEB Journal, 2022, 36, .	0.5	0