Shrikant M Mane

List of Publications by Year
in descending order

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79 papers	12,885 citations	109264 35 h-index	66879 78 g-index
82 all docs	82 docs citations	82 times ranked	25664 citing authors

#	Article	IF	Citations
1	The contribution of de novo coding mutations to autism spectrum disorder. Nature, 2014, 515, 216-221.	13.7	2,188
2	Spatio-temporal transcriptome of the human brain. Nature, 2011, 478, 483-489.	13.7	1,753
3	Insights into Autism Spectrum Disorder Genomic Architecture and Biology from 71 Risk Loci. Neuron, 2015, 87, 1215-1233.	3.8	1,219
4	Analysis of shared heritability in common disorders of the brain. Science, 2018, 360, .	6.0	1,085
5	Neuroinvasion of SARS-CoV-2 in human and mouse brain. Journal of Experimental Medicine, 2021, 218, .	4.2	677
6	The Genetic Basis of Mendelian Phenotypes: Discoveries, Challenges, and Opportunities. American Journal of Human Genetics, 2015, 97, 199-215.	2.6	574
7	Exome sequencing links mutations in PARN and RTEL1 with familial pulmonary fibrosis and telomere shortening. Nature Genetics, 2015, 47, 512-517.	9.4	385
8	Autoantibodies neutralizing type I IFNs are present in ~4% of uninfected individuals over 70 years old and account for ~20% of COVID-19 deaths. Science Immunology, 2021, 6, .	5.6	357
9	Exome sequencing identifies recurrent mutations in NF1 and RASopathy genes in sun-exposed melanomas. Nature Genetics, 2015, 47, 996-1002.	9.4	348
10	Early Assessment of Lung Cancer Immunotherapy Response via Circulating Tumor DNA. Clinical Cancer Research, 2018, 24, 1872-1880.	3.2	319
11	X-linked recessive TLR7 deficiency in ~1% of men under 60 years old with life-threatening COVID-19. Science Immunology, 2021, 6, .	5.6	267
12	Molecular and cellular reorganization of neural circuits in the human lineage. Science, 2017, 358, 1027-1032.	6.0	192
13	Mutational landscape of uterine and ovarian carcinosarcomas implicates histone genes in epithelialâ€“mesenchymal transition. Proceedings of the National Academy of Sciences of the United States of America, 2016, 113, 12238-12243.	3.3	181
14	Monogenic causes of chronic kidney disease in adults. Kidney International, 2019, 95, 914-928.	2.6	174
15	Whole Exome Sequencing of Patients with Steroid-Resistant Nephrotic Syndrome. Clinical Journal of the American Society of Nephrology: CJASN, 2018, 13, 53-62.	2.2	170
16	Two locus inheritance of non-syndromic midline craniosynostosis via rare SMAD6 and common BMP2 alleles. ELife, 2016, 5, .	2.8	168
17	Mutations in KEOPS-complex genes cause nephrotic syndrome with primary microcephaly. Nature Genetics, 2017, 49, 1529-1538.	9.4	164
18	Insights into genetics, human biology and disease gleaned from family based genomic studies. Genetics in Medicine, 2019, 21, 798-812.	1.1	161

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19	De Novo Insertions and Deletions of Predominantly Paternal Origin Are Associated with Autism Spectrum Disorder. Cell Reports, 2014, 9, 16-23.	2.9	151
20	Whole-Exome Sequencing Identifies Causative Mutations in Families with Congenital Anomalies of the Kidney and Urinary Tract. Journal of the American Society of Nephrology: JASN, 2018, 29, 2348-2361.	3.0	147
21	Whole exome sequencing frequently detects a monogenic cause in early onset nephrolithiasis andÂnephrocalcinosis. Kidney International, 2018, 93, 204-213.	2.6	133
22	A Form of the Metabolic Syndrome Associated with Mutations inDYRK1B. New England Journal of Medicine, 2014, 370, 1909-1919.	13.9	116
23	Whole-Exome Sequencing Enables a Precision Medicine Approach for Kidney Transplant Recipients. Journal of the American Society of Nephrology: JASN, 2019, 30, 201-215.	3.0	110
24	The risk of COVID-19 death is much greater and age dependent with type I IFN autoantibodies. Proceedings of the National Academy of Sciences of the United States of America, 2022, 119, e2200413119.	3.3	110
25	Mutations in KATNB1 Cause Complex Cerebral Malformations by Disrupting Asymmetrically Dividing Neural Progenitors. Neuron, 2014, 84, 1226-1239.	3.8	95
26	ALG9 Mutation Carriers Develop Kidney and Liver Cysts. Journal of the American Society of Nephrology: JASN, 2019, 30, 2091-2102.	3.0	91
27	Mutations in six nephrosis genes delineate a pathogenic pathway amenable to treatment. Nature Communications, 2018, 9, 1960.	5.8	90
28	Mutations in multiple components of the nuclear pore complex cause nephrotic syndrome. Journal of Clinical Investigation, 2018, 128, 4313-4328.	3.9	89
29	Exome sequencing implicates genetic disruption of prenatal neuro-gliogenesis in sporadic congenital hydrocephalus. Nature Medicine, 2020, 26, 1754-1765.	15.2	84
30	Whole-Exome Sequencing Reveals Somatic Mutations in HRAS and KRAS , which Cause Nevus Sebaceus. Journal of Investigative Dermatology, 2013, 133, 827-830.	0.3	79
31	Exome-wide Association Study Identifies GREB1L Mutations in Congenital Kidney Malformations. American Journal of Human Genetics, 2017, 101, 789-802.	2.6	63
32	Neomorphic effects of recurrent somatic mutations in Yin Yang 1 in insulin-producing adenomas. Proceedings of the National Academy of Sciences of the United States of America, 2015, 112, 4062-4067.	3.3	59
33	Homozygous loss of DIAPH1 is a novel cause of microcephaly in humans. European Journal of Human Genetics, 2015, 23, 165-172.	1.4	57
34	Genome-Wide Association Studies of Schizophrenia and Bipolar Disorder in a Diverse Cohort of US Veterans. Schizophrenia Bulletin, 2021, 47, 517-529.	2.3	48
35	Individual exome analysis in diagnosis and management of paediatric liver failure of indeterminate aetiology. Journal of Hepatology, 2014, 61, 1056-1063.	1.8	46
36	Centers for Mendelian Genomics: A decade of facilitating gene discovery. Genetics in Medicine, 2022, 24, 784-797.	1.1	44

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37	GAPVD1 and ANKFY1 Mutations Implicate RAB5 Regulation in Nephrotic Syndrome. Journal of the American Society of Nephrology: JASN, 2018, 29, 2123-2138.	3.0	42
38	Contributions of Rare Gene Variants to Familial and Sporadic FSGS. Journal of the American Society of Nephrology: JASN, 2019, 30, 1625-1640.	3.0	42
39	Whole exome sequencing identified ATP6V1C2 as a novel candidate gene for recessive distal renal tubular acidosis. Kidney International, 2020, 97, 567-579.	2.6	42
40	Loss-of-Function Mutations in FRRS1L Lead to an Epileptic-Dyskinetic Encephalopathy. American Journal of Human Genetics, 2016, 98, 1249-1255.	2.6	40
41	Advillin acts upstream of phospholipase C Ïµ1 in steroid-resistant nephrotic syndrome. Journal of Clinical Investigation, 2017, 127, 4257-4269.	3.9	39
42	Clonal evolution analysis of paired anaplastic and wellâ€differentiated thyroid carcinomas reveals shared common ancestor. Genes Chromosomes and Cancer, 2018, 57, 645-652.	1.5	31
43	Dominant PAX2 mutations may cause steroid-resistant nephrotic syndrome and FSGS in children. Pediatric Nephrology, 2019, 34, 1607-1613.	0.9	31
44	Bi-allelic HPDL Variants Cause a Neurodegenerative Disease Ranging from Neonatal Encephalopathy to Adolescent-Onset Spastic Paraplegia. American Journal of Human Genetics, 2020, 107, 364-373.	2.6	30
45	Mutations in the Histone Modifier PRDM6 Are Associated with Isolated Nonsyndromic Patent Ductus Arteriosus. American Journal of Human Genetics, 2016, 98, 1082-1091.	2.6	29
46	TBC1D8B Mutations Implicate RAB11-Dependent Vesicular Trafficking in the Pathogenesis of Nephrotic Syndrome. Journal of the American Society of Nephrology: JASN, 2019, 30, 2338-2353.	3.0	25
47	Mutations of the Transcriptional Corepressor ZMYM2 Cause Syndromic Urinary Tract Malformations. American Journal of Human Genetics, 2020, 107, 727-742.	2.6	25
48	Mutations in KIRREL1, a slit diaphragm component, cause steroid-resistant nephrotic syndrome. Kidney International, 2019, 96, 883-889.	2.6	23
49	CELA2A mutations predispose to early-onset atherosclerosis and metabolic syndrome and affect plasma insulin and platelet activation. Nature Genetics, 2019, 51, 1233-1243.	9.4	23
50	Genetic variants in the LAMA5 gene in pediatric nephrotic syndrome. Nephrology Dialysis Transplantation, 2019, 34, 485-493.	0.4	22
51	A noncoding variant in GANAB explains isolated polycystic liver disease (PCLD) in a large family. Human Mutation, 2018, 39, 378-382.	1.1	21
52	Recessive NOS1AP variants impair actin remodeling and cause glomerulopathy in humans and mice. Science Advances, 2021, 7, .	4.7	21
53	CAKUT and Autonomic Dysfunction Caused by Acetylcholine Receptor Mutations. American Journal of Human Genetics, 2019, 105, 1286-1293.	2.6	18
54	Genomeâ€wide association study of cognitive performance in U.S. veterans with schizophrenia or bipolar disorder. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2020, 183, 181-194.	1.1	17

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55	Novel homozygous ENPP1 mutation causes generalized arterial calcifications of infancy, thrombocytopenia, and cardiovascular and central nervous system syndrome. American Journal of Medical Genetics, Part A, 2019, 179, 2112-2118.	0.7	16
56	Mutations in PRDM15 Are a Novel Cause of Galloway-Mowat Syndrome. Journal of the American Society of Nephrology: JASN, 2021, 32, 580-596.	3.0	15
57	A patient with a novel homozygous missense mutation in FTO and concomitant nonsense mutation in CETP. Journal of Human Genetics, 2016, 61, 395-403.	1.1	14
58	De novo TRIM8 variants impair its protein localization to nuclear bodies and cause developmental delay, epilepsy, and focal segmental glomerulosclerosis. American Journal of Human Genetics, 2021, 108, 357-367.	2.6	14
59	Whole-exome sequencing reveals a monogenic cause in 56% of individuals with laterality disorders and associated congenital heart defects. Journal of Medical Genetics, 2022, 59, 691-696.	1.5	14
60	COL4A1 mutations as a potential novel cause of autosomal dominant CAKUT in humans. Human Genetics, 2019, 138, 1105-1115.	1.8	13
61	Beyond the tubule: pathological variants of LRP2, encoding the megalin receptor, result in glomerular loss and early progressive chronic kidney disease. American Journal of Physiology - Renal Physiology, 2020, 319, F988-F999.	1.3	13
62	Cystin genetic variants cause autosomal recessive polycystic kidney disease associated with altered Myc expression. Scientific Reports, 2021, 11, 18274.	1.6	13
63	Reverse phenotyping facilitates disease allele calling in exome sequencing of patients with CAKUT. Genetics in Medicine, 2022, 24, 307-318.	1.1	13
64	Impact of genotyping errors on statistical power of association tests in genomic analyses: A case study. Genetic Epidemiology, 2017, 41, 152-162.	0.6	12
65	DAAM2 Variants Cause Nephrotic Syndrome via Actin Dysregulation. American Journal of Human Genetics, 2020, 107, 1113-1128.	2.6	12
66	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.	2.6	11
67	Complete Genome Sequence of Enterococcus faecium ATCC 700221. Genome Announcements, 2016, 4, .	0.8	9
68	Digenic mutations of human OCRL paralogs in Dentâ€™s disease type 2 associated with Chiari I malformation. Human Genome Variation, 2016, 3, 16042.	0.4	8
69	Recessive Mutations in SYNPO2 as a Candidate of Monogenic Nephrotic Syndrome. Kidney International Reports, 2021, 6, 472-483.	0.4	7
70	Exome survey of individuals affected by VATER / VACTERL with renal phenotypes identifies phenocopies and novel candidate genes. American Journal of Medical Genetics, Part A, 2021, 185, 3784-3792.	0.7	6
71	Whole-exome sequencing identifiesFOXL2,FOXA2andFOXA3as candidate genes for monogenic congenital anomalies of the kidneys and urinary tract. Nephrology Dialysis Transplantation, 2022, 37, 1833-1843.	0.4	6
72	Mutation in GM2A Leads to a Progressive Chorea-dementia Syndrome. Tremor and Other Hyperkinetic Movements, 2015, 5, 306.	1.1	6

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73	A homozygous missense variant in VWA2, encoding an interactor of the Fraser-complex, in a patient with vesicoureteral reflux. PLoS ONE, 2018, 13, e0191224.	1.1	5
74	Noninvasive Analysis of the Sputum Transcriptome Discriminates Clinical Phenotypes of Asthma. Annals of the American Thoracic Society, 2016, 13 Suppl 1, S104-5.	1.5	5
75	Mutation spectrum of congenital heart disease in a consanguineous Turkish population. Molecular Genetics & Genomic Medicine, 2022, 10, e1944.	0.6	4
76	Generation of Monogenic Candidate Genes for Human Nephrotic Syndrome Using 3 Independent Approaches. Kidney International Reports, 2021, 6, 460-471.	0.4	2
77	Whole exome sequencing identifies potential candidate genes for spina bifida derived from mouse models. American Journal of Medical Genetics, Part A, 2022, , .	0.7	2
78	Sequencing the CaSR locus in Pakistani stone formers reveals a novel loss-of-function variant atypically associated with nephrolithiasis. BMC Medical Genomics, 2021, 14, 266.	0.7	1
79	A Novel form of Familial Vasopressin Deficient Diabetes Insipidus Transmitted in an X-linked Recessive manner. Journal of Clinical Endocrinology and Metabolism, 2022, , .	1.8	0