# 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 | 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 |
| 2 | 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 |
| 3 | Reverse phenotyping facilitates disease allele calling in exome sequencing of patients with CAKUT. Genetics in Medicine, 2022, 24, 307-318. | 1.1 | 13 |
| 4 | 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 |
| 5 | Centers for Mendelian Genomics: A decade of facilitating gene discovery. Genetics in Medicine, 2022, 24, 784-797. | 1.1 | 44 |
| 6 | 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 |
| 7 | Mutation spectrum of congenital heart disease in a consanguineous Turkish population. Molecular Genetics & Genomic Medicine, 2022, 10, e1944. | 0.6 | 4 |
| 8 | 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 |
| 9 | 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 |
| 10 | Recessive Mutations in SYNPO2 as a Candidate of Monogenic Nephrotic Syndrome. Kidney International Reports, 2021, 6, 472-483. | 0.4 | 7 |
| 11 | Generation of Monogenic Candidate Genes for Human Nephrotic Syndrome Using 3 Independent Approaches. Kidney International Reports, 2021, 6, 460-471. | 0.4 | 2 |
| 12 | Neuroinvasion of SARS-CoV-2 in human and mouse brain. Journal of Experimental Medicine, 2021, 218, . | 4.2 | 677 |
| 13 | Recessive NOS1AP variants impair actin remodeling and cause glomerulopathy in humans and mice. Science Advances, 2021, 7, . | 4.7 | 21 |
| 14 | 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 |
| 15 | 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 |
| 16 | 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 |
| 17 | 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 |
| 18 | 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 |

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|---|---|---|---|
| 19 | Cystin genetic variants cause autosomal recessive polycystic kidney disease associated with altered Myc expression. Scientific Reports, 2021, 11, 18274. | 1.6 | 13 |
| 20 | 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 |
| 21 | 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 |
| 22 | 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 |
| 23 | 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 |
| 24 | Exome sequencing implicates genetic disruption of prenatal neuro-gliogenesis in sporadic congenital hydrocephalus. Nature Medicine, 2020, 26, 1754-1765. | 15.2 | 84 |
| 25 | DAAM2 Variants Cause Nephrotic Syndrome via Actin Dysregulation. American Journal of Human Genetics, 2020, 107, 1113-1128. | 2.6 | 12 |
| 26 | 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 |
| 27 | 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 |
| 28 | Mutations of the Transcriptional Corepressor ZMYM2 Cause Syndromic Urinary Tract Malformations. American Journal of Human Genetics, 2020, 107, 727-742. | 2.6 | 25 |
| 29 | ALG9 Mutation Carriers Develop Kidney and Liver Cysts. Journal of the American Society of Nephrology: JASN, 2019, 30, 2091-2102. | 3.0 | 91 |
| 30 | 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 |
| 31 | Mutations in KIRREL1, a slit diaphragm component, cause steroid-resistant nephrotic syndrome. Kidney International, 2019, 96, 883-889. | 2.6 | 23 |
| 32 | 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 |
| 33 | 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 |
| 34 | COL4A1 mutations as a potential novel cause of autosomal dominant CAKUT in humans. Human Genetics, 2019, 138, 1105-1115. | 1.8 | 13 |
| 35 | Dominant PAX2 mutations may cause steroid-resistant nephrotic syndrome and FSGS in children. Pediatric Nephrology, 2019, 34, 1607-1613. | 0.9 | 31 |
| 36 | Monogenic causes of chronic kidney disease in adults. Kidney International, 2019, 95, 914-928. | 2.6 | 174 |

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|---|---|---|---|
| 37 | 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 |
| 38 | CAKUT and Autonomic Dysfunction Caused by Acetylcholine Receptor Mutations. American Journal of Human Genetics, 2019, 105, 1286-1293. | 2.6 | 18 |
| 39 | Insights into genetics, human biology and disease gleaned from family based genomic studies. Genetics in Medicine, 2019, 21, 798-812. | 1.1 | 161 |
| 40 | 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 |
| 41 | Genetic variants in the LAMA5 gene in pediatric nephrotic syndrome. Nephrology Dialysis Transplantation, 2019, 34, 485-493. | 0.4 | 22 |
| 42 | A noncoding variant in GANAB explains isolated polycystic liver disease (PCLD) in a large family. Human Mutation, 2018, 39, 378-382. | 1.1 | 21 |
| 43 | Early Assessment of Lung Cancer Immunotherapy Response via Circulating Tumor DNA. Clinical Cancer Research, 2018, 24, 1872-1880. | 3.2 | 319 |
| 44 | Whole exome sequencing frequently detects a monogenic cause in early onset nephrolithiasis andÂnephrocalcinosis. Kidney International, 2018, 93, 204-213. | 2.6 | 133 |
| 45 | 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 |
| 46 | Mutations in multiple components of the nuclear pore complex cause nephrotic syndrome. Journal of Clinical Investigation, 2018, 128, 4313-4328. | 3.9 | 89 |
| 47 | Mutations in six nephrosis genes delineate a pathogenic pathway amenable to treatment. Nature Communications, 2018, 9, 1960. | 5.8 | 90 |
| 48 | 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 |
| 49 | Analysis of shared heritability in common disorders of the brain. Science, 2018, 360, . | 6.0 | 1,085 |
| 50 | 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 |
| 51 | 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 |
| 52 | 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 |
| 53 | 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 |
| 54 | Exome-wide Association Study Identifies GREB1L Mutations in Congenital Kidney Malformations. American Journal of Human Genetics, 2017, 101, 789-802. | 2.6 | 63 |

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|---|---|---|---|
| 55 | Mutations in KEOPS-complex genes cause nephrotic syndrome with primary microcephaly. Nature Genetics, 2017, 49, 1529-1538. | 9.4 | 164 |
| 56 | Molecular and cellular reorganization of neural circuits in the human lineage. Science, 2017, 358, 1027-1032. | 6.0 | 192 |
| 57 | Advillin acts upstream of phospholipase C Ïµ1 in steroid-resistant nephrotic syndrome. Journal of Clinical Investigation, 2017, 127, 4257-4269. | 3.9 | 39 |
| 58 | 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 |
| 59 | Loss-of-Function Mutations in FRRS1L Lead to an Epileptic-Dyskinetic Encephalopathy. American Journal of Human Genetics, 2016, 98, 1249-1255. | 2.6 | 40 |
| 60 | 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 |
| 61 | 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 |
| 62 | Complete Genome Sequence of Enterococcus faecium ATCC 700221. Genome Announcements, 2016, 4, . | 0.8 | 9 |
| 63 | 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 |
| 64 | Two locus inheritance of non-syndromic midline craniosynostosis via rare SMAD6 and common BMP2 alleles. ELife, 2016, 5, . | 2.8 | 168 |
| 65 | 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 |
| 66 | Exome sequencing identifies recurrent mutations in NF1 and RASopathy genes in sun-exposed melanomas. Nature Genetics, 2015, 47, 996-1002. | 9.4 | 348 |
| 67 | The Genetic Basis of Mendelian Phenotypes: Discoveries, Challenges, and Opportunities. American Journal of Human Genetics, 2015, 97, 199-215. | 2.6 | 574 |
| 68 | 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 |
| 69 | Exome sequencing links mutations in PARN and RTEL1 with familial pulmonary fibrosis and telomere shortening. Nature Genetics, 2015, 47, 512-517. | 9.4 | 385 |
| 70 | Insights into Autism Spectrum Disorder Genomic Architecture and Biology from 71 Risk Loci. Neuron, 2015, 87, 1215-1233. | 3.8 | 1,219 |
| 71 | Homozygous loss of DIAPH1 is a novel cause of microcephaly in humans. European Journal of Human Genetics, 2015, 23, 165-172. | 1.4 | 57 |
| 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 | Mutations in KATNB1 Cause Complex Cerebral Malformations by Disrupting Asymmetrically Dividing Neural Progenitors. Neuron, 2014, 84, 1226-1239. | 3.8 | 95 |
| 74 | De Novo Insertions and Deletions of Predominantly Paternal Origin Are Associated with Autism Spectrum Disorder. Cell Reports, 2014, 9, 16-23. | 2.9 | 151 |
| 75 | A Form of the Metabolic Syndrome Associated with Mutations inDYRK1B. New England Journal of Medicine, 2014, 370, 1909-1919. | 13.9 | 116 |
| 76 | The contribution of de novo coding mutations to autism spectrum disorder. Nature, 2014, 515, 216-221. | 13.7 | 2,188 |
| 77 | Individual exome analysis in diagnosis and management of paediatric liver failure of indeterminate aetiology. Journal of Hepatology, 2014, 61, 1056-1063. | 1.8 | 46 |
| 78 | 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 |
| 79 | Spatio-temporal transcriptome of the human brain. Nature, 2011, 478, 483-489. | 13.7 | 1,753 |