Simone Sanna-Cherchi

List of Publications by Year
in descending order

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Version: 2024-02-01

63 papers	5,593 citations	101543 36 h-index	98798 67 g-index
75 all docs	75 docs citations	75 times ranked	6840 citing authors

#	Article	IF	Citations
1	Genome-wide association study identifies susceptibility loci for IgA nephropathy. Nature Genetics, 2011, 43, 321-327.	21.4	528
2	Discovery of new risk loci for IgA nephropathy implicates genes involved in immunity against intestinal pathogens. Nature Genetics, 2014, 46, 1187-1196.	21.4	505
3	Diagnostic Utility of Exome Sequencing for Kidney Disease. New England Journal of Medicine, 2019, 380, 142-151.	27.0	456
4	Renal outcome in patients with congenital anomalies of the kidney and urinary tract. Kidney International, 2009, 76, 528-533.	5.2	309
5	Geographic Differences in Genetic Susceptibility to IgA Nephropathy: GWAS Replication Study and Geospatial Risk Analysis. PLoS Genetics, 2012, 8, e1002765.	3.5	301
6	Scara5 Is a Ferritin Receptor Mediating Non-Transferrin Iron Delivery. Developmental Cell, 2009, 16, 35-46.	7.0	264
7	Copy-Number Disorders Are a Common Cause of Congenital Kidney Malformations. American Journal of Human Genetics, 2012, 91, 987-997.	6.2	201
8	Whole-Exome Sequencing in Adults With Chronic Kidney Disease. Annals of Internal Medicine, 2018, 168, 100.	3.9	154
9	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.	6.1	147
10	Recurrence of focal segmental glomerulosclerosis after renal transplantation in patients with mutations of podocin. American Journal of Kidney Diseases, 2003, 41, 1314-1321.	1.9	144
11	The copy number variation landscape of congenital anomalies of the kidney and urinary tract. Nature Genetics, 2019, 51, 117-127.	21.4	144
12	Isolated polycystic liver disease genes define effectors of polycystin-1 function. Journal of Clinical Investigation, 2017, 127, 1772-1785.	8.2	137
13	Î±â€“Intercalated cells defend the urinary system from bacterial infection. Journal of Clinical Investigation, 2014, 124, 2963-2976.	8.2	127
14	Genetic Drivers of Kidney Defects in the DiGeorge Syndrome. New England Journal of Medicine, 2017, 376, 742-754.	27.0	120
15	The genetic architecture of membranous nephropathy and its potential to improve non-invasive diagnosis. Nature Communications, 2020, 11, 1600.	12.8	120
16	Mutations in DSTYK and Dominant Urinary Tract Malformations. New England Journal of Medicine, 2013, 369, 621-629.	27.0	119
17	HNF1B and PAX2 mutations are a common cause of renal hypodysplasia in the CKiD cohort. Pediatric Nephrology, 2011, 26, 897-903.	1.7	114
18	Genetic approaches to human renal agenesis/hypoplasia and dysplasia. Pediatric Nephrology, 2007, 22, 1675-1684.	1.7	99

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19	Genetic basis of human congenital anomalies of the kidney and urinary tract. Journal of Clinical Investigation, 2018, 128, 4-15.	8.2	91
20	Clinical Implications of the Solitary Functioning Kidney. Clinical Journal of the American Society of Nephrology: CJASN, 2014, 9, 978-986.	4.5	81
21	Mutations in TBX18 Cause Dominant Urinary Tract Malformations via Transcriptional Dysregulation of Ureter Development. American Journal of Human Genetics, 2015, 97, 291-301.	6.2	72
22	Exome sequencing identified MYO1E and NEIL1 as candidate genes for human autosomal recessive steroid-resistant nephrotic syndrome. Kidney International, 2011, 80, 389-396.	5.2	69
23	Genomic imbalances in pediatric patients with chronic kidney disease. Journal of Clinical Investigation, 2015, 125, 2171-2178.	8.2	68
24	CureGN Study Rationale, Design, and Methods: Establishing a Large Prospective Observational Study of Glomerular Disease. American Journal of Kidney Diseases, 2019, 73, 218-229.	1.9	68
25	Copy number variation analysis identifies novel CAKUT candidate genes in children with a solitary functioning kidney. Kidney International, 2015, 88, 1402-1410.	5.2	65
26	Exome-wide Association Study Identifies GREB1L Mutations in Congenital Kidney Malformations. American Journal of Human Genetics, 2017, 101, 789-802.	6.2	63
27	The Burden of Candidate Pathogenic Variants for Kidney and Genitourinary Disorders Emerging From Exome Sequencing. Annals of Internal Medicine, 2019, 170, 11.	3.9	60
28	Phenotypic Expansion of DGKE-Associated Diseases. Journal of the American Society of Nephrology: JASN, 2014, 25, 1408-1414.	6.1	59
29	Familial Vesicoureteral Reflux: Testing Replication of Linkage in Seven New Multigenerational Kindreds. Journal of the American Society of Nephrology: JASN, 2005, 16, 1781-1787.	6.1	56
30	Cyclosporine in patients with steroid-resistant nephrotic syndrome: an open-label, nonrandomized, retrospective study. Clinical Therapeutics, 2004, 26, 1411-1418.	2.5	48
31	Urinary NGAL Marks Cystic Disease in HIV-Associated Nephropathy. Journal of the American Society of Nephrology: JASN, 2009, 20, 1687-1692.	6.1	47
32	Genetics in chronic kidney disease: conclusions from a Kidney Disease: Improving Global Outcomes (KDIGO) Controversies Conference. Kidney International, 2022, 101, 1126-1141.	5.2	46
33	A Recessive Gene for Primary Vesicoureteral Reflux Maps to Chromosome 12p11-q13. Journal of the American Society of Nephrology: JASN, 2009, 20, 1633-1640.	6.1	42
34	Familial forms of nephrotic syndrome. Pediatric Nephrology, 2010, 25, 241-252.	1.7	41
35	A Dominant Mutation in Nuclear Receptor Interacting Protein 1 Causes Urinary Tract Malformations via Dysregulation of Retinoic Acid Signaling. Journal of the American Society of Nephrology: JASN, 2017, 28, 2364-2376.	6.1	40
36	Exome-Based Rare-Variant Analyses in CKD. Journal of the American Society of Nephrology: JASN, 2019, 30, 1109-1122.	6.1	40

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37	Characterization of a large Lebanese family segregating IgA nephropathy. Nephrology Dialysis Transplantation, 2007, 22, 772-777.	0.7	39
38	Clinical Characteristics and Treatment Patterns of Children and Adults With IgA Nephropathy or IgA Vasculitis: Findings From the CureGN Study. Kidney International Reports, 2018, 3, 1373-1384.	0.8	39
39	Health-related quality of life in glomerular disease. Kidney International, 2019, 95, 1209-1224.	5.2	38
40	Infantile Encephaloneuromyopathy and Defective Mitochondrial Translation Are Due to a Homozygous RMND1 Mutation. American Journal of Human Genetics, 2012, 91, 729-736.	6.2	35
41	Localization of a Gene for Nonsyndromic Renal Hypodysplasia to Chromosome 1p32-33. American Journal of Human Genetics, 2007, 80, 539-549.	6.2	33
42	Pilot Study of Return of Genetic Results to Patients in Adult Nephrology. Clinical Journal of the American Society of Nephrology: CJASN, 2020, 15, 651-664.	4.5	28
43	Type IV Collagen Mutations in Familial IgA Nephropathy. Kidney International Reports, 2020, 5, 1075-1078.	0.8	26
44	Mutations of the Transcriptional Corepressor ZMYM2 Cause Syndromic Urinary Tract Malformations. American Journal of Human Genetics, 2020, 107, 727-742.	6.2	25
45	Apolipoprotein E in idiopathic nephrotic syndrome and focal segmental glomerulosclerosis. Kidney International, 2003, 63, 686-695.	5.2	23
46	'Congenital solitary functioning kidneys: which ones warrant follow-up into adult life?'. Nephrology Dialysis Transplantation, 2011, 26, 1458-1460.	0.7	23
47	A noncoding variant in GANAB explains isolated polycystic liver disease (PCLD) in a large family. Human Mutation, 2018, 39, 378-382.	2.5	21
48	Integrative analysis of rare copy number variants and gene expression data in alopecia areata implicates an aetiological role for autophagy. Experimental Dermatology, 2020, 29, 243-253.	2.9	21
49	Longitudinal Changes in Health-Related Quality of Life in Primary Glomerular Disease: Results From the CureGN Study. Kidney International Reports, 2020, 5, 1679-1689.	0.8	17
50	Copy Number Variant Analysis and Genome-wide Association Study Identify Loci with Large Effect for Vesicoureteral Reflux. Journal of the American Society of Nephrology: JASN, 2021, 32, 805-820.	6.1	17
51	Human and mouse studies establish TBX6 in Mendelian CAKUT and as a potential driver of kidney defects associated with the 16p11.2 microdeletion syndrome. Kidney International, 2020, 98, 1020-1030.	5.2	17
52	Multidisciplinary approaches for elucidating genetics and molecular pathogenesis of urinary tract malformations. Kidney International, 2022, 101, 473-484.	5.2	16
53	The emerging role of genomics in the diagnosis and workup of congenital urinary tract defects: a novel deletion syndrome on chromosome 3q13.31-22.1. Pediatric Nephrology, 2014, 29, 257-267.	1.7	15
54	Targeted sequencing of 96 renal developmental microRNAs in 1213 individuals from 980 families with congenital anomalies of the kidney and urinary tract. Nephrology Dialysis Transplantation, 2016, 31, 1280-1283.	0.7	15

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55	A Novel SUCLA2 Mutation Presenting as a Complex Childhood Movement Disorder. Journal of Child Neurology, 2017, 32, 246-250.	1.4	15
56	Immunohistochemical expression pattern of RIP5, FGFR1, FGFR2 and HIP2 in the normal human kidney development. Acta Histochemica, 2019, 121, 531-538.	1.8	14
57	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.	6.2	14
58	Alterations of Type IV Collagen Î± Chains in Patients with Chronic Acquired Glomerulopathies: mRNA Levels, Protein Expression and Urinary Loss. American Journal of Nephrology, 2007, 27, 129-137.	3.1	7
59	Phenocopies, Phenotypic Expansion, and Coincidental Diagnoses: Time to Abandon Targeted Gene Panels?. American Journal of Kidney Diseases, 2020, 76, 451-453.	1.9	7
60	Recessive mutations in CAKUT and VACTERL association. Kidney International, 2014, 85, 1253-1255.	5.2	4
61	GWAS in Mice Maps Susceptibility to HIV-Associated Nephropathy to the Ssbp2 Locus. Journal of the American Society of Nephrology: JASN, 2022, 33, 108-120.	6.1	3
62	tarSVM: Improving the accuracy of variant calls derived from microfluidic PCR-based targeted next generation sequencing using a support vector machine. BMC Bioinformatics, 2016, 17, 233.	2.6	2
63	Persistent Disease Activity in Patients With Long-Standing Glomerular Disease. Kidney International Reports, 2020, 5, 860-871.	0.8	2