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List of Publications by Year in descending order

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Version: 2024-02-01

50
papers

4,269
citations

126907

33
h-index

175258

52
g-index

53
all docs

53
docs citations

53
times ranked

6234
citing authors

#	ARTICLE	IF	CITATIONS
1	Recessive <i>NOS1AP</i> variants impair actin remodeling and cause glomerulopathy in humans and mice. <i>Science Advances</i> , 2021, 7, .	10.3	21
2	Mutations in <i>PRDM15</i> Are a Novel Cause of Galloway-Mowat Syndrome. <i>Journal of the American Society of Nephrology: JASN</i> , 2021, 32, 580-596.	6.1	15
3	Cystin genetic variants cause autosomal recessive polycystic kidney disease associated with altered <i>Myc</i> expression. <i>Scientific Reports</i> , 2021, 11, 18274.	3.3	13
4	<i>DAAM2</i> Variants Cause Nephrotic Syndrome via Actin Dysregulation. <i>American Journal of Human Genetics</i> , 2020, 107, 1113-1128.	6.2	12
5	Novel nephronophthisis-associated variants reveal functional importance of <i>MAPKBP1</i> dimerization for centriolar recruitment. <i>Kidney International</i> , 2020, 98, 958-969.	5.2	6
6	Panel sequencing distinguishes monogenic forms of nephritis from nephrosis in children. <i>Nephrology Dialysis Transplantation</i> , 2019, 34, 474-485.	0.7	13
7	Defects in t6A tRNA modification due to <i>GON7</i> and <i>YRDC</i> mutations lead to Galloway-Mowat syndrome. <i>Nature Communications</i> , 2019, 10, 3967.	12.8	66
8	Gene panel sequencing identifies a likely monogenic cause in 7% of 235 Pakistani families with nephrolithiasis. <i>Human Genetics</i> , 2019, 138, 211-219.	3.8	26
9	Monogenic causes of chronic kidney disease in adults. <i>Kidney International</i> , 2019, 95, 914-928.	5.2	174
10	Mutations of <i>ADAMTS9</i> Cause Nephronophthisis-Related Ciliopathy. <i>American Journal of Human Genetics</i> , 2019, 104, 45-54.	6.2	29
11	Whole-Exome Sequencing Enables a Precision Medicine Approach for Kidney Transplant Recipients. <i>Journal of the American Society of Nephrology: JASN</i> , 2019, 30, 201-215.	6.1	110
12	Genetic variants in the <i>LAMA5</i> gene in pediatric nephrotic syndrome. <i>Nephrology Dialysis Transplantation</i> , 2019, 34, 485-493.	0.7	22
13	Whole Exome Sequencing Reveals a Monogenic Cause of Disease in ~43% of 35 Families With Midaortic Syndrome. <i>Hypertension</i> , 2018, 71, 691-699.	2.7	22
14	Whole exome sequencing frequently detects a monogenic cause in early onset nephrolithiasis and nephrocalcinosis. <i>Kidney International</i> , 2018, 93, 204-213.	5.2	133
15	Whole Exome Sequencing of Patients with Steroid-Resistant Nephrotic Syndrome. <i>Clinical Journal of the American Society of Nephrology: CJASN</i> , 2018, 13, 53-62.	4.5	170
16	Mutations in multiple components of the nuclear pore complex cause nephrotic syndrome. <i>Journal of Clinical Investigation</i> , 2018, 128, 4313-4328.	8.2	89
17	A Multi-layered Quantitative In Vivo Expression Atlas of the Podocyte Unravels Kidney Disease Candidate Genes. <i>Cell Reports</i> , 2018, 23, 2495-2508.	6.4	81
18	Mutations in six nephrosis genes delineate a pathogenic pathway amenable to treatment. <i>Nature Communications</i> , 2018, 9, 1960.	12.8	90

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19	GAPVD1 and ANKFY1 Mutations Implicate RAB5 Regulation in Nephrotic Syndrome. <i>Journal of the American Society of Nephrology: JASN</i> , 2018, 29, 2123-2138.	6.1	42
20	Mutations in <i>WDR4</i> as a new cause of Galloway-Mowat syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 2460-2465.	1.2	56
21	The nucleoside-diphosphate kinase NME3 associates with nephronophthisis proteins and is required for ciliary function during renal development. <i>Journal of Biological Chemistry</i> , 2018, 293, 15243-15255.	3.4	13
22	Whole-Exome Sequencing Identifies Causative Mutations in Families with Congenital Anomalies of the Kidney and Urinary Tract. <i>Journal of the American Society of Nephrology: JASN</i> , 2018, 29, 2348-2361.	6.1	147
23	Acute multi-sgRNA knockdown of KEOPS complex genes reproduces the microcephaly phenotype of the stable knockout zebrafish model. <i>PLoS ONE</i> , 2018, 13, e0191503.	2.5	18
24	Mutations in <i>MAPKBP1</i> Cause Juvenile or Late-Onset Cilia-Independent Nephronophthisis. <i>American Journal of Human Genetics</i> , 2017, 100, 323-333.	6.2	29
25	Modeling Monogenic Human Nephrotic Syndrome in the <i>Drosophila</i> Garland Cell Nephrocyte. <i>Journal of the American Society of Nephrology: JASN</i> , 2017, 28, 1521-1533.	6.1	70
26	Cystic kidneys in fetal Walker-Warburg syndrome with <i>POMT2</i> mutation: Intrafamilial phenotypic variability in four siblings and review of literature. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 2697-2702.	1.2	11
27	Mutations in KEOPS-complex genes cause nephrotic syndrome with primary microcephaly. <i>Nature Genetics</i> , 2017, 49, 1529-1538.	21.4	164
28	Ciliopathies. <i>Cold Spring Harbor Perspectives in Biology</i> , 2017, 9, a028191.	5.5	325
29	Mutations in sphingosine-1-phosphate lyase cause nephrosis with ichthyosis and adrenal insufficiency. <i>Journal of Clinical Investigation</i> , 2017, 127, 912-928.	8.2	160
30	Advillin acts upstream of phospholipase C β 1 in steroid-resistant nephrotic syndrome. <i>Journal of Clinical Investigation</i> , 2017, 127, 4257-4269.	8.2	39
31	Mutations in <i>SLC26A1</i> Cause Nephrolithiasis. <i>American Journal of Human Genetics</i> , 2016, 98, 1228-1234.	6.2	41
32	The ciliopathy-associated <i>CPLANE</i> proteins direct basal body recruitment of intraflagellar transport machinery. <i>Nature Genetics</i> , 2016, 48, 648-656.	21.4	119
33	<i>FAT1</i> mutations cause a glomerulotubular nephropathy. <i>Nature Communications</i> , 2016, 7, 10822.	12.8	99
34	Prevalence of Monogenic Causes in Pediatric Patients with Nephrolithiasis or Nephrocalcinosis. <i>Clinical Journal of the American Society of Nephrology: CJASN</i> , 2016, 11, 664-672.	4.5	105
35	Mutations in nuclear pore genes <i>NUP93</i> , <i>NUP205</i> and <i>XPO5</i> cause steroid-resistant nephrotic syndrome. <i>Nature Genetics</i> , 2016, 48, 457-465.	21.4	149
36	Large-scale targeted sequencing comparison highlights extreme genetic heterogeneity in nephronophthisis-related ciliopathies. <i>Journal of Medical Genetics</i> , 2016, 53, 208-214.	3.2	39

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37	Whole exome sequencing identifies causative mutations in the majority of consanguineous or familial cases with childhood-onset increased renal echogenicity. <i>Kidney International</i> , 2016, 89, 468-475.	5.2	74
38	Targeted Resequencing of 29 Candidate Genes and Mouse Expression Studies Implicate <i>ZIC3</i> and <i>FOXF1</i> in Human VATER/VACTERL Association. <i>Human Mutation</i> , 2015, 36, 1150-1154.	2.5	46
39	DCDC2 Mutations Cause a Renal-Hepatic Ciliopathy by Disrupting Wnt Signaling. <i>American Journal of Human Genetics</i> , 2015, 96, 81-92.	6.2	98
40	<i>IFT81</i> , encoding an IFT-B core protein, as a very rare cause of a ciliopathy phenotype. <i>Journal of Medical Genetics</i> , 2015, 52, 657-665.	3.2	32
41	Fourteen Monogenic Genes Account for 15% of Nephrolithiasis/Nephrocalcinosis. <i>Journal of the American Society of Nephrology: JASN</i> , 2015, 26, 543-551.	6.1	163
42	Gene mutation analysis in Iranian children with nephronophthisis: a two-center study. <i>Iranian Journal of Kidney Diseases</i> , 2015, 9, 119-25.	0.1	2
43	The Vac14-interaction Network Is Linked to Regulators of the Endolysosomal and Autophagic Pathway. <i>Molecular and Cellular Proteomics</i> , 2014, 13, 1397-1411.	3.8	51
44	Mutations of CEP83 Cause Infantile Nephronophthisis and Intellectual Disability. <i>American Journal of Human Genetics</i> , 2014, 94, 905-914.	6.2	90
45	Identification of 99 novel mutations in a worldwide cohort of 1,056 patients with a nephronophthisis-related ciliopathy. <i>Human Genetics</i> , 2013, 132, 865-884.	3.8	199
46	Zebrafish Ciliopathy Screen Plus Human Mutational Analysis Identifies C21orf59 and CCDC65 Defects as Causing Primary Ciliary Dyskinesia. <i>American Journal of Human Genetics</i> , 2013, 93, 672-686.	6.2	184
47	Mutations in SPAG1 Cause Primary Ciliary Dyskinesia Associated with Defective Outer and Inner Dynein Arms. <i>American Journal of Human Genetics</i> , 2013, 93, 711-720.	6.2	135
48	Defects in the IFT-B Component IFT172 Cause Jeune and Mainzer-Saldino Syndromes in Humans. <i>American Journal of Human Genetics</i> , 2013, 93, 915-925.	6.2	196
49	High-throughput mutation analysis in patients with a nephronophthisis-associated ciliopathy applying multiplexed barcoded array-based PCR amplification and next-generation sequencing. <i>Journal of Medical Genetics</i> , 2012, 49, 756-767.	3.2	109
50	Decreased number of circulating progenitor cells in obesity: beneficial effects of weight reduction. <i>European Heart Journal</i> , 2008, 29, 1560-1568.	2.2	104