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List of Publications by Year
in descending order

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61 papers	3,741 citations	172457 29 h-index	155660 55 g-index
63 all docs	63 docs citations	63 times ranked	5329 citing authors

#	Article	IF	Citations
1	A Single-Gene Cause in 29.5% of Cases of Steroid-Resistant Nephrotic Syndrome. Journal of the American Society of Nephrology: JASN, 2015, 26, 1279-1289.	6.1	499
2	Identification of 99 novel mutations in a worldwide cohort of 1,056 patients with a nephronophthisis-related ciliopathy. Human Genetics, 2013, 132, 865-884.	3.8	199
3	Defects in the IFT-B Component IFT172 Cause Jeune and Mainzer-Saldino Syndromes in Humans. American Journal of Human Genetics, 2013, 93, 915-925.	6.2	196
4	Mutations in RSPH1 Cause Primary Ciliary Dyskinesia with a Unique Clinical and Ciliary Phenotype. American Journal of Respiratory and Critical Care Medicine, 2014, 189, 707-717.	5.6	191
5	Zebrafish Ciliopathy Screen Plus Human Mutational Analysis Identifies C21orf59 and CCDC65 Defects as Causing Primary Ciliary Dyskinesia. American Journal of Human Genetics, 2013, 93, 672-686.	6.2	184
6	ANKS6 is a central component of a nephronophthisis module linking NEK8 to INVS and NPHP3. Nature Genetics, 2013, 45, 951-956.	21.4	183
7	ZMYND10 Is Mutated in Primary Ciliary Dyskinesia and Interacts with LRRC6. American Journal of Human Genetics, 2013, 93, 336-345.	6.2	183
8	Fourteen Monogenic Genes Account for 15% of Nephrolithiasis/Nephrocalcinosis. Journal of the American Society of Nephrology: JASN, 2015, 26, 543-551.	6.1	163
9	Mutations in SPAG1 Cause Primary Ciliary Dyskinesia Associated with Defective Outer and Inner Dynein Arms. American Journal of Human Genetics, 2013, 93, 711-720.	6.2	135
10	Whole exome sequencing frequently detects a monogenic cause in early onset nephrolithiasis andÂnephrocalcinosis. Kidney International, 2018, 93, 204-213.	5.2	133
11	High-throughput mutation analysis in patients with a nephronophthisis-associated ciliopathy applying multiplexed barcoded array-based PCR amplification and next-generation sequencing. Journal of Medical Genetics, 2012, 49, 756-767.	3.2	109
12	Prevalence of Monogenic Causes in Pediatric Patients with Nephrolithiasis or Nephrocalcinosis. Clinical Journal of the American Society of Nephrology: CJASN, 2016, 11, 664-672.	4.5	105
13	FAT1 mutations cause a glomerulotubular nephropathy. Nature Communications, 2016, 7, 10822.	12.8	99
14	DCDC2 Mutations Cause a Renal-Hepatic Ciliopathy by Disrupting Wnt Signaling. American Journal of Human Genetics, 2015, 96, 81-92.	6.2	98
15	TMEM231, mutated in orofaciodigital and Meckel syndromes, organizes the ciliary transition zone. Journal of Cell Biology, 2015, 209, 129-142.	5.2	95
16	Mutations of CEP83 Cause Infantile Nephronophthisis and Intellectual Disability. American Journal of Human Genetics, 2014, 94, 905-914.	6.2	90
17	Mutations in CSPP1 Lead to Classical Joubert Syndrome. American Journal of Human Genetics, 2014, 94, 80-86.	6.2	75
18	Rapid Detection of Monogenic Causes of Childhood-Onset Steroid-Resistant Nephrotic Syndrome. Clinical Journal of the American Society of Nephrology: CJASN, 2014, 9, 1109-1116.	4.5	74

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19	Whole exome sequencing identifies causative mutations in the majority of consanguineous or familial cases with childhood-onset increased renal echogenicity. Kidney International, 2016, 89, 468-475.	5.2	74
20	Mutation of the Mg2+ Transporter SLC41A1 Results in a Nephronophthisis-Like Phenotype. Journal of the American Society of Nephrology: JASN, 2013, 24, 967-977.	6.1	63
21	Cystinuria: clinical practice recommendation. Kidney International, 2021, 99, 48-58.	5.2	58
22	Value of renal gene panel diagnostics in adults waiting for kidney transplantation due to undetermined end-stage renal disease. Kidney International, 2019, 96, 222-230.	5.2	47
23	Targeted Resequencing of 29 Candidate Genes and Mouse Expression Studies ImplicateZIC3andFOXF1in Human VATER/VACTERL Association. Human Mutation, 2015, 36, 1150-1154.	2.5	46
24	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
25	Mutations in SLC26A1 Cause Nephrolithiasis. American Journal of Human Genetics, 2016, 98, 1228-1234.	6.2	41
26	Large-scale targeted sequencing comparison highlights extreme genetic heterogeneity in nephronophthisis-related ciliopathies. Journal of Medical Genetics, 2016, 53, 208-214.	3.2	39
27	Refining genotypeâ€“phenotype correlations in 304 patients with autosomal recessive polycystic kidney disease and PKHD1 gene variants. Kidney International, 2021, 100, 650-659.	5.2	38
28	Genetic and Evolutionary Analyses of the Human Bone Morphogenetic Protein Receptor 2 (BMPR2) in the Pathophysiology of Obesity. PLoS ONE, 2011, 6, e16155.	2.5	38
29	Role of genetic variation in the human sodiumâ€“glucose cotransporter 2 gene (SGLT2) in glucose homeostasis. Pharmacogenomics, 2011, 12, 1119-1126.	1.3	37
30	IFT81, encoding an IFT-B core protein, as a very rare cause of a ciliopathy phenotype. Journal of Medical Genetics, 2015, 52, 657-665.	3.2	32
31	Clinical, biochemical, and pathophysiological analysis of SLC34A1 mutations. Physiological Reports, 2018, 6, e13715.	1.7	32
32	Matching clinical and genetic diagnoses in autosomal dominant polycystic kidney disease reveals novel phenocopies and potential candidate genes. Genetics in Medicine, 2020, 22, 1374-1383.	2.4	30
33	Mutations in MAPKBP1 Cause Juvenile or Late-Onset Cilia-Independent Nephronophthisis. American Journal of Human Genetics, 2017, 100, 323-333.	6.2	29
34	Mutations of ADAMTS9 Cause Nephronophthisis-Related Ciliopathy. American Journal of Human Genetics, 2019, 104, 45-54.	6.2	29
35	Gene panel sequencing identifies a likely monogenic cause in 7% of 235 Pakistani families with nephrolithiasis. Human Genetics, 2019, 138, 211-219.	3.8	26
36	Biallelic Expression of Mucin-1 in Autosomal Dominant Tubulointerstitial Kidney Disease: Implications for Nongenetic Disease Recognition. Journal of the American Society of Nephrology: JASN, 2018, 29, 2298-2309.	6.1	25

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37	A Deregulated Stress Response Underlies Distinct INF2-Associated Disease Profiles. Journal of the American Society of Nephrology: JASN, 2020, 31, 1296-1313.	6.1	20
38	Claudin-10a Deficiency Shifts Proximal Tubular Cl- Permeability to Cation Selectivity via Claudin-2 Redistribution. Journal of the American Society of Nephrology: JASN, 2022, 33, 699-717.	6.1	20
39	Diagnosing FSGS without kidney biopsy â€“ a novel INF2-mutation in a family with ESRD of unknown origin. BMC Medical Genetics, 2016, 17, 73.	2.1	16
40	THOC5: a novel gene involved in HDL-cholesterol metabolism. Journal of Lipid Research, 2013, 54, 3170-3176.	4.2	15
41	Evaluating pathogenicity of SLC34A3-Ser192Leu, a frequent European missense variant in disorders of renal phosphate wasting. Urolithiasis, 2019, 47, 511-519.	2.0	15
42	Update on Hereditary Kidney Stone Disease and Introduction of a New Clinical Patient Registry in Germany. Frontiers in Pediatrics, 2018, 6, 47.	1.9	14
43	Posttransplant nephrotic syndrome resulting from NELL1-positive membranous nephropathy. American Journal of Transplantation, 2021, 21, 3175-3179.	4.7	14
44	The nucleoside-diphosphate kinase NME3 associates with nephronophthisis proteins and is required for ciliary function during renal development. Journal of Biological Chemistry, 2018, 293, 15243-15255.	3.4	13
45	Genetics of kidney stone diseaseâ€”Polygenic meets monogenic. Nephrologie Et Therapeutique, 2021, 17, S88-S94.	0.5	10
46	Defective claudin-10 causes a novel variation of HELIX syndrome through compromised tight junction strand assembly. Genes and Diseases, 2022, 9, 1301-1314.	3.4	9
47	Acute kidney injury and its progression in hospitalized patientsâ€”Results from a retrospective multicentre cohort study with a digital decision support system. PLoS ONE, 2021, 16, e0254608.	2.5	9
48	Biallelic pathogenic variants in roundabout guidance receptor 1 associate with syndromic congenital anomalies of the kidney and urinary tract. Kidney International, 2022, 101, 1039-1053.	5.2	8
49	Isolated Renal Relapse of Sarcoidosis under Low-Dose Glucocorticoid Therapy. Journal of General Internal Medicine, 2008, 23, 879-882.	2.6	7
50	Retrospective genetic analysis illustrates the spectrum of autosomal Alport syndrome in a case of living-related donor kidney transplantation. BMC Nephrology, 2019, 20, 340.	1.8	7
51	Novel nephronophthisis-associated variants reveal functional importance of MAPKBP1 dimerization for centriolar recruitment. Kidney International, 2020, 98, 958-969.	5.2	6
52	Effective immunosuppressive management with belatacept and eculizumab in post-transplant aHUS due to a homozygous deletion of CFHR1/CFHR3 and the presence of CFH antibodies. CKJ: Clinical Kidney Journal, 2017, 10, 742-746.	2.9	5
53	Successful Simultaneous Pancreas Kidney Transplantation in Maturity-Onset Diabetes of the Young Type 5. Transplantation, 2011, 92, e45-e47.	1.0	3
54	Deleterious Impact of a Novel CFH Splice Site Variant in Atypical Hemolytic Uremic Syndrome. Frontiers in Genetics, 2019, 10, 465.	2.3	3

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55	Autosomal dominant polycystic kidney disease in absence of renal cyst formation illustrates genetic interaction between WT1 and PKD1. Journal of Medical Genetics, 2021, 58, 140-144.	3.2	2
56	Challenging Disease Ontology by Instances of Atypical PKHD1 and PKD1 Genetics. Frontiers in Genetics, 2021, 12, 682565.	2.3	2
57	Novel somatic PBX1 mosaicism likely masking syndromic CAKUT in an adult with bilateral kidney hypoplasia. CKJ: Clinical Kidney Journal, 0, , .	2.9	2
58	Early Flow Disturbances of Tunnelled Haemodialysis Catheters and Topographic Landmarks in Chest X-Ray. Blood Purification, 2018, 46, 70-76.	1.8	0
59	The Case | Atypical cysts in a patient with autosomal dominant polycystic kidney disease. Kidney International, 2019, 96, 1043-1044.	5.2	0
60	â€˜Case of the Monthâ€™ from the University Medicine Mannheim: managing a complex stone patient with recurrent stone formation. BJU International, 2021, 127, 402-408.	2.5	0
61	MO047: Biallelic pathogenic variants in ROBO1 associate with syndromic CAKUT. Nephrology Dialysis Transplantation, 2022, 37, .	0.7	0