John A. Sayer

List of Publications by Year
in descending order

220 papers	7,311 citations	61984 43 h-index	71685 76 g-index
236 all docs	236 docs citations	236 times ranked	7476 citing authors

#	Article	IF	Citations
1	The centrosomal protein nephrocystin-6 is mutated in Joubert syndrome and activates transcription factor ATF4. Nature Genetics, 2006, 38, 674-681.	21.4	535
2	Nephrocystin-5, a ciliary IQ domain protein, is mutated in Senior-Loken syndrome and interacts with RPGR and calmodulin. Nature Genetics, 2005, 37, 282-288.	21.4	367
3	In-frame deletion in a novel centrosomal/ciliary protein CEP290/NPHP6 perturbs its interaction with RPGR and results in early-onset retinal degeneration in the rd16 mouse. Human Molecular Genetics, 2006, 15, 1847-1857.	2.9	353
4	100,000 Genomes Pilot on Rare-Disease Diagnosis in Health Care â€" Preliminary Report. New England Journal of Medicine, 2021, 385, 1868-1880.	27.0	352
5	Exome Capture Reveals ZNF423 and CEP164 Mutations, Linking Renal Ciliopathies to DNA Damage Response Signaling. Cell, 2012, 150, 533-548.	28.9	347
6	Loss of GLIS2 causes nephronophthisis in humans and mice by increased apoptosis and fibrosis. Nature Genetics, 2007, 39, 1018-1024.	21.4	221
7	Germline selection shapes human mitochondrial DNA diversity. Science, 2019, 364, .	12.6	178
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	Expression analyses and interaction with the anaphase promoting complex protein Apc2 suggest a role for inversin in primary cilia and involvement in the cell cycle. Human Molecular Genetics, 2002, 11, 3345-3350.	2.9	133
10	Whole exome sequencing frequently detects a monogenic cause in early onset nephrolithiasis andÂnephrocalcinosis. Kidney International, 2018, 93, 204-213.	5.2	133
11	Evidence of digenic inheritance in Alport syndrome. Journal of Medical Genetics, 2015, 52, 163-174.	3.2	129
12	Nephrocalcinosis: molecular insights into calcium precipitation within the kidney. Clinical Science, 2004, 106, 549-561.	4.3	121
13	Update of PAX2 mutations in renal coloboma syndrome and establishment of a locus-specific database. Human Mutation, 2012, 33, 457-466.	2.5	109
14	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
15	Individuals with mutations in XPNPEP3, which encodes a mitochondrial protein, develop a nephronophthisis-like nephropathy. Journal of Clinical Investigation, 2010, 120, 791-802.	8.2	102
16	A meckelinâ€"filamin A interaction mediates ciliogenesis. Human Molecular Genetics, 2012, 21, 1272-1286.	2.9	96
17	TMEM231, mutated in orofaciodigital and Meckel syndromes, organizes the ciliary transition zone. Journal of Cell Biology, 2015, 209, 129-142.	5.2	95
18	Mutation analysis of NPHP6/CEP290 in patients with Joubert syndrome and Senior Loken syndrome. Journal of Medical Genetics, 2007, 44, 657-663.	3.2	93

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19	Many Genesâ€"One Disease? Genetics of Nephronophthisis (NPHP) and NPHP-Associated Disorders. Frontiers in Pediatrics, 2017, 5, 287.	1.9	89
20	Mutational analysis of the RPGRIP1L gene in patients with Joubert syndrome and nephronophthisis. Kidney International, 2007, 72, 1520-1526.	5.2	88
21	Identification of the first AHI1 gene mutations in nephronophthisis-associated Joubert syndrome. Pediatric Nephrology, 2006, 21, 32-35.	1.7	87
22	Acidosis and Deafness in Patients with Recessive Mutations in FOXI1. Journal of the American Society of Nephrology: JASN, 2018, 29, 1041-1048.	6.1	84
23	Clinical and genetic spectra of autosomal dominant tubulointerstitial kidney disease due to mutationsÂin UMOD and MUC1. Kidney International, 2020, 98, 717-731.	5.2	75
24	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
25	Genetic and physical interaction between the NPHP5 and NPHP6 gene products. Human Molecular Genetics, 2008, 17, 3655-3662.	2.9	72
26	Successful treatment of hypercalcaemia associated with aCYP24A1mutation with fluconazole: Fig.Â1.. CKJ: Clinical Kidney Journal, 2015, 8, 453-455.	2.9	72
27	Murine Joubert syndrome reveals Hedgehog signaling defects as a potential therapeutic target for nephronophthisis. Proceedings of the National Academy of Sciences of the United States of America, 2014, 111, 9893-9898.	7.1	71
28	Progress in Understanding the Genetics of Calcium-Containing Nephrolithiasis. Journal of the American Society of Nephrology: JASN, 2017, 28, 748-759.	6.1	70
29	ARL3 Mutations Cause Joubert Syndrome by Disrupting Ciliary Protein Composition. American Journal of Human Genetics, 2018, 103, 612-620.	6.2	70
30	Healthcare recommendations for Joubert syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 229-249.	1.2	66
31	Monoallelic IFT140 pathogenic variants are an important cause of the autosomal dominant polycystic kidney-spectrum phenotype. American Journal of Human Genetics, 2022, 109, 136-156.	6.2	62
32	Characterization of CSF2RA mutation related juvenile pulmonary alveolar proteinosis. Orphanet Journal of Rare Diseases, 2014, 9, 171.	2.7	61
33	Cystinuria: clinical practice recommendation. Kidney International, 2021, 99, 48-58.	5.2	58
34	Renal calcium stones: insights from the control of bone mineralization. Experimental Physiology, 2008, 93, 43-49.	2.0	57
35	Prospective Evaluation of Kidney Disease in Joubert Syndrome. Clinical Journal of the American Society of Nephrology: CJASN, 2017, 12, 1962-1973.	4.5	56
36	A role for CBS domain 2 in trafficking of chloride channel CLC-5. Biochemical and Biophysical Research Communications, 2003, 310, 600-605.	2.1	54

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37	Nephronophthisis. European Journal of Human Genetics, 2009, 17, 406-416.	2.8	54
38	Clinical and Genetic Analysis of Patients with Cystinuria in the United Kingdom. Clinical Journal of the American Society of Nephrology: CJASN, 2015, 10, 1235-1245.	4.5	54
39	A human patient-derived cellular model of Joubert syndrome reveals ciliary defects which can be rescued with targeted therapies. Human Molecular Genetics, 2017, 26, 4657-4667.	2.9	53
40	Uromodulin is expressed in renal primary cilia and UMOD mutations result in decreased ciliary uromodulin expression. Human Molecular Genetics, 2010, 19, 1985-1997.	2.9	52
41	Mutations in mitochondrial DNA causing tubulointerstitial kidney disease. PLoS Genetics, 2017, 13, e1006620.	3.5	52
42	DNA replication stress underlies renal phenotypes in CEP290-associated Joubert syndrome. Journal of Clinical Investigation, 2015, 125, 3657-3666.	8.2	48
43	Clinical and Functional Characterization of URAT1 Variants. PLoS ONE, 2011, 6, e28641.	2.5	48
44	Jouberin localizes to collecting ducts and interacts with nephrocystin-1. Kidney International, 2008, 74, 1139-1149.	5.2	46
45	Naturally occurring antisense RNA: function and mechanisms of action. Current Opinion in Nephrology and Hypertension, 2009, 18, 343-349.	2.0	45
46	Nephronophthisis: A Genetically Diverse Ciliopathy. International Journal of Nephrology, 2011, 2011, 1-10.	1.3	44
47	Targeted exon skipping of a CEP290 mutation rescues Joubert syndrome phenotypes in vitro and in a murine model. Proceedings of the National Academy of Sciences of the United States of America, 2018, 115, 12489-12494.	7.1	44
48	An update on the use of tolvaptan for autosomal dominant polycystic kidney disease: consensus statement on behalf of the ERA Working Group on Inherited Kidney Disorders, the European Rare Kidney Disease Reference Network and Polycystic Kidney Disease International. Nephrology Dialysis Transplantation, 2022, 37, 825-839.	0.7	44
49	Dysfunction of the ciliary ARMC9/TOGARAM1 protein module causes Joubert syndrome. Journal of Clinical Investigation, 2020, 130, 4423-4439.	8.2	43
50	A molecular genetic analysis of childhood nephrotic syndrome in a cohort of Saudi Arabian families. Journal of Human Genetics, 2013, 58, 480-489.	2.3	42
51	Use of whole genome sequencing to determine genetic basis of suspected mitochondrial disorders: cohort study. BMJ, The, 2021, 375, e066288.	6.0	42
52	Mutations in SLC26A1 Cause Nephrolithiasis. American Journal of Human Genetics, 2016, 98, 1228-1234.	6.2	41
53	The Molecular Genetics of Gordon Syndrome. Genes, 2019, 10, 986.	2.4	41
54	Disruption of clc-5 leads to a redistribution of annexin A2 and promotes calcium crystal agglomeration in collecting duct epithelial cells. Cellular and Molecular Life Sciences, 2006, 63, 367-377.	5.4	38

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55	Clinical spectrum, prognosis and estimated prevalence of DNAJB11-kidney disease. Kidney International, 2020, 98, 476-487.	5.2	38
56	Renal ciliopathies. Current Opinion in Genetics and Development, 2019, 56, 49-60.	3.3	37
57	A CEP104-CSPP1 Complex Is Required for Formation of Primary Cilia Competent in Hedgehog Signaling. Cell Reports, 2019, 28, 1907-1922.e6.	6.4	34
58	The Genetics of Nephrolithiasis. Nephron Experimental Nephrology, 2008, 110, e37-e43.	2.2	32
59	Lessons learned from a multidisciplinary renal genetics clinic. QJM - Monthly Journal of the Association of Physicians, 2017, 110, 453-457.	0.5	32
60	Clinical, biochemical, and pathophysiological analysis of SLC34A1 mutations. Physiological Reports, 2018, 6, e13715.	1.7	32
61	Genetic testing can resolve diagnostic confusion in Alport syndrome. CKJ: Clinical Kidney Journal, 2014, 7, 197-200.	2.9	31
62	A novel LMX1B mutation in a family with end-stage renal disease of 'unknown cause'. CKJ: Clinical Kidney Journal, 2015, 8, 113-119.	2.9	30
63	Modelling a ciliopathy: Ahi1 knockdown in model systems reveals an essential role in brain, retinal, and renal development. Cellular and Molecular Life Sciences, 2012, 69, 993-1009.	5.4	29
64	Genetic spectrum of Saudi Arabian patients with antenatal cystic kidney disease and ciliopathy phenotypes using a targeted renal gene panel. Journal of Medical Genetics, 2016, 53, 338-347.	3.2	28
65	An international cohort study of autosomal dominant tubulointerstitial kidney disease due to mutations identifies distinct clinical subtypes. Kidney International, 2020, 98, 1589-1604.	5.2	27
66	Urinary Concentration Defects and Mechanisms Underlying Nephronophthisis. Kidney and Blood Pressure Research, 2008, 31, 152-162.	2.0	26
67	Autosomal Dominant Mutation in the Signal Peptide of Renin in a Kindred With Anemia, Hyperuricemia, and CKD. American Journal of Kidney Diseases, 2011, 58, 821-825.	1.9	26
68	Tenofovir disoproxil fumarate-associated renal tubular dysfunction. Aids, 2017, 31, 1297-1301.	2.2	26
69	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
70	Diagnosis and Clinical Biochemistry of Inherited Tubulopathies. Annals of Clinical Biochemistry, 2001, 38, 459-470.	1.6	25
71	Practical approaches to the management of autosomal dominant polycystic kidney disease patients in the era of tolvaptan. CKJ: Clinical Kidney Journal, 2018, 11, 62-69.	2.9	25
72	Nephronophthisis. Journal of Pediatric Genetics, 2015, 03, 103-114.	0.7	24

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73	Human urine-derived renal epithelial cells provide insights into kidney-specific alternate splicing variants. European Journal of Human Genetics, 2018, 26, 1791-1796.	2.8	22
74	Mouse genetics reveals Barttin as a genetic modifier of Joubert syndrome. Proceedings of the National Academy of Sciences of the United States of America, 2020, 117, 1113-1118.	7.1	22
75	Electrolyte Disturbances in SARS-CoV-2 Infection. F1000Research, 2020, 9, 587.	1.6	22
76	Progressive liver, kidney, and heart degeneration in children and adults affected by TULP3 mutations. American Journal of Human Genetics, 2022, 109, 928-943.	6.2	22
77	Expression and Localisation of the Pyrophosphate Transporter, ANK, in Murine Kidney Cells. Cellular Physiology and Biochemistry, 2007, 20, 507-516.	1.6	21
78	Bialleleic PKD1 mutations underlie early-onset autosomal dominant polycystic kidney disease in Saudi Arabian families. Pediatric Nephrology, 2019, 34, 1615-1623.	1.7	21
79	Calcium oxalate crystal deposition in the kidney: identification, causes and consequences. Urolithiasis, 2020, 48, 377-384.	2.0	21
80	Pantoprazole-induced acute interstitial nephritis. Journal of Nephrology, 2004, 17, 580-1.	2.0	20
81	CYP24A1 mutation leading to nephrocalcinosis. Kidney International, 2014, 85, 1475.	5.2	19
82	A novel homozygous UMOD mutation reveals gene dosage effects on uromodulin processing and urinary excretion. Nephrology Dialysis Transplantation, 2017, 32, 1994-1999.	0.7	19
83	Molecular genetic investigations identify new clinical phenotypes associated with BCS1L-related mitochondrial disease. Human Molecular Genetics, 2019, 28, 3766-3776.	2.9	19
84	The challenges of diagnosis and management of Gitelman syndrome. Clinical Endocrinology, 2020, 92, 3-10.	2.4	19
85	Nephrocalcinosis: A Review of Monogenic Causes and Insights They Provide into This Heterogeneous Condition. International Journal of Molecular Sciences, 2020, 21, 369.	4.1	19
86	The voltageâ€dependent Cl âˆ' channel ClCâ€5 and plasma membrane Cl âˆ' conductances of mouse renal collecting duct cells (mIMCDâ€3). Journal of Physiology, 2001, 536, 769-783.	2.9	18
87	Calcium phosphate and calcium oxalate crystal handling is dependent upon CLC-5 expression in mouse collecting duct cells. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2004, 1689, 83-90.	3.8	18
88	Genome-wide linkage and association study implicates the 10q26 region as a major genetic contributor to primary nonsyndromic vesicoureteric reflux. Scientific Reports, 2017, 7, 14595.	3.3	17
89	Disordered calcium crystal handling in antisense CLC-5-treated collecting duct cells. Biochemical and Biophysical Research Communications, 2003, 300, 305-310.	2.1	16
90	Glanzmann thrombasthenia in Pakistan: molecular analysis and identification of novel mutations. Clinical Genetics, 2016, 89, 187-192.	2.0	16

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91	End-Stage Kidney Failure in Oman: An Analysis of Registry Data with an Emphasis on Congenital and Inherited Renal Diseases. International Journal of Nephrology, 2017, 2017, 1-7.	1.3	16
92	The Medical Management of Urolithiasis. British Journal of Medical and Surgical Urology, 2010, 3, 87-95.	0.2	15
93	Investigating Embryonic Expression Patterns and Evolution of AHI1 and CEP290 Genes, Implicated in Joubert Syndrome. PLoS ONE, 2012, 7, e44975.	2.5	15
94	Novel compound heterozygous mutations in AMN cause Imerslund-GrÃsbeck syndrome in two half-sisters: a case report. BMC Medical Genetics, 2015, 16, 35.	2.1	15
95	Targeted exon skipping rescues ciliary protein composition defects in Joubert syndrome patient fibroblasts. Scientific Reports, 2019, 9, 10828.	3.3	15
96	Evaluating pathogenicity of SLC34A3-Ser192Leu, a frequent European missense variant in disorders of renal phosphate wasting. Urolithiasis, 2019, 47, 511-519.	2.0	15
97	Molecular genetics of renal ciliopathies. Biochemical Society Transactions, 2021, 49, 1205-1220.	3.4	15
98	SGLT2 inhibitors â€“ a potential treatment for Alport syndrome. Clinical Science, 2020, 134, 379-388.	4.3	15
99	Genetic compensation for cilia defects in cep290 mutants by upregulation of cilia-associated small GTPases. Journal of Cell Science, 2021, 134, .	2.0	14
100	Biallelic CYP24A1 variants presenting during pregnancy: clinical and biochemical phenotypes. Endocrine Connections, 2020, 9, 530-541.	1.9	14
101	Urinary Stone Formation: Dentâ€™s Disease Moves Understanding Forward. Nephron Experimental Nephrology, 2002, 10, 176-181.	2.2	13
102	Extracellular calcium-sensing receptor dysfunction is associated with two new phenotypes. Clinical Endocrinology, 2003, 59, 419-421.	2.4	13
103	The Pyrophosphate Transporter ANKH is Expressed in Kidney and Bone Cells and Colocalises to the Primary Cilium/Basal Body Complex. Cellular Physiology and Biochemistry, 2009, 24, 595-604.	1.6	13
104	Functional modelling of a novel mutation in BBS5. Cilia, 2014, 3, 3.	1.8	13
105	Use of patient derived urine renal epithelial cells to confirm pathogenicity of PKHD1 alleles. BMC Nephrology, 2020, 21, 435.	1.8	13
106	Nephrocystin-1 interacts directly with Ack1 and is expressed in human collecting duct. Biochemical and Biophysical Research Communications, 2008, 371, 877-882.	2.1	12
107	Novel mutations of the CLCN5 gene including a complex allele and A 5â€² UTR mutation in Dent disease 1. Clinical Genetics, 2009, 76, 413-416.	2.0	12
108	Identification of compound heterozygous KCNJ1 mutations (encoding ROMK) in a kindred with Bartter's syndrome and a functional analysis of their pathogenicity. Physiological Reports, 2013, 1, e00160.	1.7	12

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109	Urolithiasis as an extraarticular manifestation of ankylosing spondylitis. Rheumatology International, 2017, 37, 1949-1956.	3.0	12
110	Homozygous Variant inARL3Causes Autosomal Recessive Cone Rod Dystrophy. , 2019, 60, 4811.		12
111	A discarded synonymous variant in NPHP3 explains nephronophthisis and congenital hepatic fibrosis in several families. Human Mutation, 2021, 42, 1221-1228.	2.5	12
112	Clinical and genetic spectra of autosomal dominant tubulointerstitial kidney disease. Nephrology Dialysis Transplantation, 2021, , .	0.7	12
113	Disease Modeling To Understand the Pathomechanisms of Human Genetic Kidney Disorders. Clinical Journal of the American Society of Nephrology: CJASN, 2020, 15, 855-872.	4.5	11
114	Novel loss of function variants in FRAS1 AND FREM2 underlie renal agenesis in consanguineous families. Journal of Nephrology, 2021, 34, 893-900.	2.0	11
115	Murine Cep290 phenotypes are modified by genetic backgrounds and provide an impetus for investigating disease modifier alleles. F1000Research, 2015, 4, 590.	1.6	11
116	A mutant wfs1 zebrafish model of Wolfram syndrome manifesting visual dysfunction and developmental delay. Scientific Reports, 2021, 11, 20491.	3.3	11
117	Novel CYP24A1 Mutation in a Young Male Patient with Nephrolithiasis: Case Report. Kidney and Blood Pressure Research, 2019, 44, 870-877.	2.0	10
118	Clinical and genetic characteristics of autosomal recessive polycystic kidney disease in Oman. BMC Nephrology, 2020, 21, 347.	1.8	10
119	ARL3, a small GTPase with a functionally conserved role in primary cilia and immune synapses. Small GTPases, 2021, 12, 167-176.	1.6	10
120	Diagnosis and clinical biochemistry of inherited tubulopathies. Annals of Clinical Biochemistry, 2001, 38, 459-470.	1.6	10
121	SARS-CoV-2 and hypokalaemia: evidence and implications. F1000Research, 2020, 9, 587.	1.6	10
122	Diverse molecular causes of unsolved autosomal dominant tubulointerstitial kidney diseases. Kidney International, 2022, 102, 405-420.	5.2	10
123	Parapelvic cysts leading to a diagnosis of Fabry disease. Kidney International, 2008, 74, 1366.	5.2	9
124	Renal Stone Disease. Nephron Physiology, 2011, 118, p35-p44.	1.2	9
125	A novel CLDN16 mutation in a large family with familial hypomagnesaemia with hypercalciuria and nephrocalcinosis. BMC Research Notes, 2013, 6, 527.	1.4	9
126	Emerging treatments and personalised medicine for ciliopathies associated with cystic kidney disease. Expert Opinion on Orphan Drugs, 2017, 5, 785-798.	0.8	9

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127	Precision medicine in renal stone-formers. Urolithiasis, 2019, 47, 99-105.	2.0	9
128	From disease modelling to personalised therapy in patients with CEP290 mutations. F1000Research, 2017, 6, 669.	1.6	9
129	Treatment and long-term outcome in primary nephrogenic diabetes insipidus. Nephrology Dialysis Transplantation, 2023, 38, 2120-2130.	0.7	9
130	Pseudohypoaldosteronism type 2 presenting with hypertension and hyperkalaemia due to a novel mutation in the WNK4 gene. QJM - Monthly Journal of the Association of Physicians, 2012, 105, 791-794.	0.5	8
131	A preliminary survey of practice patterns across several European kidney stone centers and a call for action in developing shared practice. Urolithiasis, 2019, 47, 219-224.	2.0	8
132	Identification of LAMA1 mutations ends diagnostic odyssey and has prognostic implications for patients with presumed Joubert syndrome. Brain Communications, 2021, 3, fcab163.	3.3	8
133	Update of genetic variants in CEP120 and CC2D2Aâ€"With an emphasis on genotypeâ€phenotype correlations, tissue specific transcripts and exploring mutation specific exon skipping therapies. Molecular Genetics & Genomic Medicine, 2021, 9, e1603.	1.2	8
134	Rapid onset intratubular calcification following renal transplantation requiring urgent parathyroidectomy. Clinical Nephrology, 2007, 68, 47-51.	0.7	8
135	A wide spectrum of phenotypes in a family with renal coloboma syndrome caused by aPAX2mutation. CKJ: Clinical Kidney Journal, 2013, 6, 410-413.	2.9	7
136	Chondrocalcinosis and Gitelman syndrome. QJM - Monthly Journal of the Association of Physicians, 2016, 109, 563-564.	0.5	7
137	Early B-cell Factor 3â€"Related Genetic Disease Can Mimic Urofacial Syndrome. Kidney International Reports, 2020, 5, 1823-1827.	0.8	7
138	Kidney Disease in Oman: a View of the Current and Future Landscapes. Iranian Journal of Kidney Diseases, 2017, 11, 263-270.	0.1	7
139	Acute renal failure from contrast medium: Beware patients taking metforminâ€¦. BMJ: British Medical Journal, 2006, 333, 653.2.	2.3	6
140	Using zebrafish to study the function of nephronophthisis and related ciliopathy genes. F1000Research, 2018, 7, 1133.	1.6	6
141	Prenatal exome sequencing and chromosomal microarray analysis in fetal structural anomalies in a highly consanguineous population reveals a propensity of ciliopathy genes causing multisystem phenotypes. Human Genetics, 2022, 141, 101-126.	3.8	6
142	Biallelic variants in <scp>TTC21B</scp> as a rare cause of earlyâ€onset arterial hypertension and tubuloglomerular kidney disease. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2022, 190, 109-120.	1.6	6
143	Digital ischaemia in a renal transplant patient. Nephrology Dialysis Transplantation, 2004, 19, 1656-1657.	0.7	5
144	Senior-Loken syndrome secondary to NPHP5/IQCB1 mutation in an Iranian family. CKJ: Clinical Kidney Journal, 2011, 4, 421-423.	2.9	5

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145	Monogenic diabetes, renal dysplasia and hypopituitarism: a patient with a HNF1A mutation. QJM - Monthly Journal of the Association of Physicians, 2011, 104, 881-883.	0.5	5
146	Case Report Whole-exome analysis of a child with polycystic kidney disease and ventriculomegaly. Genetics and Molecular Research, 2015, 14, 3618-3624.	0.2	5
147	Molecular Genetic Diagnosis of Omani Patients With Inherited Cystic Kidney Disease. Kidney International Reports, 2019, 4, 1751-1759.	0.8	5
148	RE: Clinical and Molecular Diagnosis of Joubert Syndrome and Related Disorders. Pediatric Neurology, 2020, 112, 10.	2.1	5
149	Expression patterns of ciliopathy genes ARL3 and CEP120 reveal roles in multisystem development. BMC Developmental Biology, 2020, 20, 26.	2.1	5
150	Embryonic and foetal expression patterns of the ciliopathy gene CEP164. PLoS ONE, 2020, 15, e0221914.	2.5	5
151	Allele frequency of variants reported to cause adenine phosphoribosyltransferase deficiency. European Journal of Human Genetics, 2021, 29, 1061-1070.	2.8	5
152	Variable phenotypic presentations of renal involvement in Fabry disease: a case series. F1000Research, 2018, 7, 356.	1.6	5
153	Using zebrafish to study the function of nephronophthisis and related ciliopathy genes. F1000Research, 2018, 7, 1133.	1.6	5
154	Diagnosing Fabry disease--delays and difficulties within discordant siblings. QJM - Monthly Journal of the Association of Physicians, 2015, 108, 585-590.	0.5	4
155	Renal tubular dysgenesis: antenatal ultrasound scanning and molecular investigations in a Saudi Arabian family. CKJ: Clinical Kidney Journal, 2016, 9, 807-810.	2.9	4
156	Clinical and Biochemical Features of Patients with CYP24A1 Mutations. , 0, , .		4
157	Large Retroperitoneal Haemorrhage Following Cyst Rupture in a Patient with Autosomal Dominant Polycystic Kidney Disease. Case Reports in Nephrology, 2017, 2017, 1-3.	0.4	4
158	Fetal Anomalies Associated with Novel Pathogenic Variants in TMEM94. Genes, 2020, 11, 967.	2.4	4
159	Clinical versus research genomics in kidney disease. Nature Reviews Nephrology, 2021, 17, 570-571.	9.6	4
160	Sarcoidosis presenting with hypercalcaemia following withdrawal of long-term immunosuppression in renal transplantation. Oxford Medical Case Reports, 2014, 2014, 86-88.	0.4	3
161	Seasonal hypercalcaemia; consider CYP24A1 mutation. QJM - Monthly Journal of the Association of Physicians, 2019, 112, 393-393.	0.5	3
162	Cell preservation methods and its application to studying rare disease. Molecular and Cellular Probes, 2021, 56, 101694.	2.1	3

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163	Gene and epigenetic editing in the treatment of primary ciliopathies. Progress in Molecular Biology and Translational Science, 2021, 182, 353-401.	1.7	3
164	Case Report: Renal potassium wasting in SARS-CoV-2 infection. F1000Research, 2020, 9, 659.	1.6	3
165	Pseudodominant Alport syndrome caused by pathogenic homozygous and compound heterozygous COL4A3 splicing variants. Annals of Human Genetics, 2022, 86, 145-152.	0.8	3
166	Research priorities for autosomal dominant polycystic kidney disease: a UK priority setting partnership. BMJ Open, 2022, 12, e055780.	1.9	3
167	Renal expression of Ca2+-activated Clâ^' channels. Current Topics in Membranes, 2002, 53, 283-307.	0.9	2
168	Multiple thyroid cysts as an extra-renal manifestation of ADPKD. CKJ: Clinical Kidney Journal, 2008, 1, 266-267.	2.9	2
169	ANKH and Renal Stone Formation in Ankylosing Spondylitis. Journal of Rheumatology, 2012, 39, 1756-1756.	2.0	2
170	The challenges and surprises of a definitive molecular genetic diagnosis. Kidney International, 2014, 85, 748-749.	5.2	2
171	Re: Loss-of-Function Mutations of CYP24A1, the Vitamin D 24-hydroxylase Gene, Cause Long-standing Hypercalciuric Nephrolithiasis and Nephrocalcinosis. European Urology, 2015, 68, 164-165.	1.9	2
172	A case of ocular cystinosis associated with two potentially severe CTNS mutations. Ophthalmic Genetics, 2019, 40, 157-160.	1.2	2
173	Acquired C1-inhibitor deficiency presenting with nephrotic syndrome. BMJ Case Reports, 2019, 12, e230388.	0.5	2
174	Whole exome sequencing of large populations: identification of loss of function alleles and implications for inherited kidney diseases. Kidney International, 2021, 99, 1255-1259.	5.2	2
175	The diagnostic yield of whole exome sequencing as a first approach in consanguineous Omani renal ciliopathy syndrome patients. F1000Research, 2021, 10, 207.	1.6	2
176	Case Report: Making a diagnosis of familial renal disease â€“ clinical and patient perspectives. F1000Research, 2017, 6, 470.	1.6	2
177	Case Report: Investigation and molecular genetic diagnosis of familial hypomagnesaemia: a case report. F1000Research, 2019, 8, 666.	1.6	2
178	Case Report: Investigation and molecular genetic diagnosis of familial hypomagnesaemia. F1000Research, 2019, 8, 666.	1.6	2
179	Case Report: Renal potassium wasting in SARS-CoV-2 infection. F1000Research, 2020, 9, 659.	1.6	2
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