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	Treatment and long-term outcome in primary nephrogenic diabetes insipidus. Nephrology Dialysis Transplantation, 2023, 38, 2120-2130.	0.7	9
2	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
3	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
4	Kidney traits on repeatâ€"the role of MUC1 VNTR. Kidney International, 2022, 101, 863-866.	5.2	1
5	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
6	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
7	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
8	Cutaneous manifestations of acute kidney injury. CKJ: Clinical Kidney Journal, 2022, 15, 855-864.	2.9	1
9	Pseudodominant Alport syndrome caused by pathogenic homozygous and compound heterozygous COL4A3 splicing variants. Annals of Human Genetics, 2022, 86, 145-152.	0.8	3
10	MO042: Biallelic variants in TTC21B as a rare cause of early-onset arterial hypertension and tubuloglomerular kidney disease. Nephrology Dialysis Transplantation, 2022, 37, .	0.7	0
11	Diverse molecular causes of unsolved autosomal dominant tubulointerstitial kidney diseases. Kidney International, 2022, 102, 405-420.	5.2	10
12	Research priorities for autosomal dominant polycystic kidney disease: a UK priority setting partnership. BMJ Open, 2022, 12, e055780.	1.9	3
13	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
14	Novel pathogenic MAPKBP1 variant in a family with nephronophthisis. CKJ: Clinical Kidney Journal, 2021, 14, 728-730.	2.9	0
15	Cystinuria: clinical practice recommendation. Kidney International, 2021, 99, 48-58.	5.2	58
16	ARL3, a small GTPase with a functionally conserved role in primary cilia and immune synapses. Small GTPases, 2021, 12, 167-176.	1.6	10
17	The diagnostic yield of whole exome sequencing as a first approach in consanguineous Omani renal ciliopathy syndrome patients. F1000Research, 2021, 10, 207.	1.6	1
18	Allele frequency of variants reported to cause adenine phosphoribosyltransferase deficiency. European Journal of Human Genetics, 2021, 29, 1061-1070.	2.8	5

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19	Cell preservation methods and its application to studying rare disease. Molecular and Cellular Probes, 2021, 56, 101694.	2.1	3
20	FC 014INFLUENCE OF GENETIC VARIATION IN SLC7A13/AGT1 IN HUMAN CYSTINURIA. Nephrology Dialysis Transplantation, 2021, 36, .	0.7	0
21	Molecular genetics of renal ciliopathies. Biochemical Society Transactions, 2021, 49, 1205-1220.	3.4	15
22	MO004PRIMARY BILIARY CHOLANGITIS PRESENTING WITH RENAL FANCONI SYNDROME: A FORGOTTEN PHENOTYPE. Nephrology Dialysis Transplantation, 2021, 36, .	0.7	0
23	Clinical versus research genomics in kidney disease. Nature Reviews Nephrology, 2021, 17, 570-571.	9.6	4
24	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
25	A discarded synonymous variant in NPHP3 explains nephronophthisis and congenital hepatic fibrosis in several families. Human Mutation, 2021, 42, 1221-1228.	2.5	12
26	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
27	Genetic compensation for cilia defects in cep290 mutants by upregulation of cilia-associated small GTPases. Journal of Cell Science, 2021, 134, .	2.0	14
28	Clinical and genetic spectra of autosomal dominant tubulointerstitial kidney disease. Nephrology Dialysis Transplantation, 2021, , .	0.7	12
29	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
30	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
31	Gene and epigenetic editing in the treatment of primary ciliopathies. Progress in Molecular Biology and Translational Science, 2021, 182, 353-401.	1.7	3
32	A mutant wfs1 zebrafish model of Wolfram syndrome manifesting visual dysfunction and developmental delay. Scientific Reports, 2021, 11, 20491.	3.3	11
33	Use of whole genome sequencing to determine genetic basis of suspected mitochondrial disorders: cohort study. BMJ, The, 2021, 375, e066288.	6.0	42
34	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
35	Case Report: A Novel In-Frame Deletion of GLIS2 Leading to Nephronophthisis and Early Onset Kidney Failure. Frontiers in Genetics, 2021, 12, 791495.	2.3	2
36	The challenges of diagnosis and management of Gitelman syndrome. Clinical Endocrinology, 2020, 92, 3-10.	2.4	19

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37	Healthcare recommendations for Joubert syndrome. American Journal of Medical Genetics, Part A, 2020, 182, 229-249.	1.2	66
38	Use of patient derived urine renal epithelial cells to confirm pathogenicity of PKHD1 alleles. BMC Nephrology, 2020, 21, 435.	1.8	13
39	Early B-cell Factor 3â€“Related Genetic Disease Can Mimic Urofacial Syndrome. Kidney International Reports, 2020, 5, 1823-1827.	0.8	7
40	RE: Clinical and Molecular Diagnosis of Joubert Syndrome and Related Disorders. Pediatric Neurology, 2020, 112, 10.	2.1	5
41	Calcium oxalate crystal deposition in the kidney: identification, causes and consequences. Urolithiasis, 2020, 48, 377-384.	2.0	21
42	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
43	Regarding â€œDerivation and validation of genome-wide polygenic score for urinary tractÂstone diagnosisâ€. Kidney International, 2020, 98, 1347.	5.2	1
44	Fetal Anomalies Associated with Novel Pathogenic Variants in TMEM94. Genes, 2020, 11, 967.	2.4	4
45	Clinical and genetic characteristics of autosomal recessive polycystic kidney disease in Oman. BMC Nephrology, 2020, 21, 347.	1.8	10
46	Expression patterns of ciliopathy genes ARL3 and CEP120 reveal roles in multisystem development. BMC Developmental Biology, 2020, 20, 26.	2.1	5
47	Are conventional stone analysis techniques reliable for the identification of 2,8-dihydroxyadenine kidney stones? A case series. Urolithiasis, 2020, 48, 337-344.	2.0	1
48	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
49	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
50	Variable Phenotypes Seen with a Homozygous CYP24A1 Mutation: Case Report. SN Comprehensive Clinical Medicine, 2020, 2, 995-1002.	0.6	1
51	Embryonic and foetal expression patterns of the ciliopathy gene CEP164. PLoS ONE, 2020, 15, e0221914.	2.5	5
52	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
53	Clinical spectrum, prognosis and estimated prevalence of DNAJB11-kidney disease. Kidney International, 2020, 98, 476-487.	5.2	38
54	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

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55	SGLT2 inhibitors â€“ a potential treatment for Alport syndrome. Clinical Science, 2020, 134, 379-388.	4.3	15
56	Dysfunction of the ciliary ARMC9/TOGARAM1 protein module causes Joubert syndrome. Journal of Clinical Investigation, 2020, 130, 4423-4439.	8.2	43
57	Electrolyte Disturbances in SARS-CoV-2 Infection. F1000Research, 2020, 9, 587.	1.6	22
58	Case Report: Renal potassium wasting in SARS-CoV-2 infection. F1000Research, 2020, 9, 659.	1.6	3
59	Biallelic CYP24A1 variants presenting during pregnancy: clinical and biochemical phenotypes. Endocrine Connections, 2020, 9, 530-541.	1.9	14
60	SARS-CoV-2 and hypokalaemia: evidence and implications. F1000Research, 2020, 9, 587.	1.6	10
61	Case Report: Renal potassium wasting in SARS-CoV-2 infection. F1000Research, 2020, 9, 659.	1.6	2
62	Embryonic and foetal expression patterns of the ciliopathy gene CEP164. , 2020, 15, e0221914.		0
63	Embryonic and foetal expression patterns of the ciliopathy gene CEP164. , 2020, 15, e0221914.		0
64	Embryonic and foetal expression patterns of the ciliopathy gene CEP164. , 2020, 15, e0221914.		0
65	Embryonic and foetal expression patterns of the ciliopathy gene CEP164. , 2020, 15, e0221914.		0
66	Embryonic and foetal expression patterns of the ciliopathy gene CEP164. , 2020, 15, e0221914.		0
67	Embryonic and foetal expression patterns of the ciliopathy gene CEP164. , 2020, 15, e0221914.		0
68	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
69	Novel CYP24A1 Mutation in a Young Male Patient with Nephrolithiasis: Case Report. Kidney and Blood Pressure Research, 2019, 44, 870-877.	2.0	10
70	Using human urine-derived renal epithelial cells to model kidney disease in inherited ciliopathies. Translational Science of Rare Diseases, 2019, 4, 87-95.	1.5	1
71	Targeted exon skipping rescues ciliary protein composition defects in Joubert syndrome patient fibroblasts. Scientific Reports, 2019, 9, 10828.	3.3	15
72	Molecular Genetic Diagnosis of Omani Patients With Inherited Cystic Kidney Disease. Kidney International Reports, 2019, 4, 1751-1759.	0.8	5

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73	Renal ciliopathies. Current Opinion in Genetics and Development, 2019, 56, 49-60.	3.3	37
74	Molecular genetic investigations identify new clinical phenotypes associated with BCS1L-related mitochondrial disease. Human Molecular Genetics, 2019, 28, 3766-3776.	2.9	19
75	CYP24A1 mutations and hypervitaminosis D. Clinical Medicine, 2019, 19, 92.2-93.	1.9	0
76	Germline selection shapes human mitochondrial DNA diversity. Science, 2019, 364, .	12.6	178
77	Bialleleic PKD1 mutations underlie early-onset autosomal dominant polycystic kidney disease in Saudi Arabian families. Pediatric Nephrology, 2019, 34, 1615-1623.	1.7	21
78	A case of ocular cystinosis associated with two potentially severe CTNS mutations. Ophthalmic Genetics, 2019, 40, 157-160.	1.2	2
79	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
80	Evaluating pathogenicity of SLC34A3-Ser192Leu, a frequent European missense variant in disorders of renal phosphate wasting. Urolithiasis, 2019, 47, 511-519.	2.0	15
81	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
82	Acquired C1-inhibitor deficiency presenting with nephrotic syndrome. BMJ Case Reports, 2019, 12, e230388.	0.5	2
83	Homozygous Variant inARL3Causes Autosomal Recessive Cone Rod Dystrophy. , 2019, 60, 4811.		12
84	The Molecular Genetics of Gordon Syndrome. Genes, 2019, 10, 986.	2.4	41
85	Seasonal hypercalcaemia; consider CYP24A1 mutation. QJM - Monthly Journal of the Association of Physicians, 2019, 112, 393-393.	0.5	3
86	Precision medicine in renal stone-formers. Urolithiasis, 2019, 47, 99-105.	2.0	9
87	Case Report: Investigation and molecular genetic diagnosis of familial hypomagnesaemia: a case report. F1000Research, 2019, 8, 666.	1.6	2
88	Case Report: Investigation and molecular genetic diagnosis of familial hypomagnesaemia. F1000Research, 2019, 8, 666.	1.6	2
89	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
90	Editorial Comment. Journal of Urology, 2018, 199, 632-632.	0.4	0

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91	Whole exome sequencing frequently detects a monogenic cause in early onset nephrolithiasis andÂnephrocalcinosis. Kidney International, 2018, 93, 204-213.	5.2	133
92	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
93	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
94	ARL3 Mutations Cause Joubert Syndrome by Disrupting Ciliary Protein Composition. American Journal of Human Genetics, 2018, 103, 612-620.	6.2	70
95	Clinical, biochemical, and pathophysiological analysis of SLC34A1 mutations. Physiological Reports, 2018, 6, e13715.	1.7	32
96	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
97	Re: Sagy I, Zeldetz V, Halerin D, Abu Tailakh M, Novack V. The effect of Ramadan fast on the incidence of renal colic emergency department visits1. QJM - Monthly Journal of the Association of Physicians, 2018, 111, 353-354.	0.5	1
98	Variable phenotypic presentations of renal involvement in Fabry disease: a case series. F1000Research, 2018, 7, 356.	1.6	5
99	Using zebrafish to study the function of nephronophthisis and related ciliopathy genes. F1000Research, 2018, 7, 1133.	1.6	6
100	Using zebrafish to study the function of nephronophthisis and related ciliopathy genes. F1000Research, 2018, 7, 1133.	1.6	5
101	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
102	Tenofovir disoproxil fumarate-associated renal tubular dysfunction. Aids, 2017, 31, 1297-1301.	2.2	26
103	Progress in Understanding the Genetics of Calcium-Containing Nephrolithiasis. Journal of the American Society of Nephrology: JASN, 2017, 28, 748-759.	6.1	70
104	Urolithiasis as an extraarticular manifestation of ankylosing spondylitis. Rheumatology International, 2017, 37, 1949-1956.	3.0	12
105	Emerging treatments and personalised medicine for ciliopathies associated with cystic kidney disease. Expert Opinion on Orphan Drugs, 2017, 5, 785-798.	0.8	9
106	Lessons learned from a multidisciplinary renal genetics clinic. QJM - Monthly Journal of the Association of Physicians, 2017, 110, 453-457.	0.5	32
107	Prospective Evaluation of Kidney Disease in Joubert Syndrome. Clinical Journal of the American Society of Nephrology: CJASN, 2017, 12, 1962-1973.	4.5	56
108	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

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109	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
110	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
111	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
112	Mutations in mitochondrial DNA causing tubulointerstitial kidney disease. PLoS Genetics, 2017, 13, e1006620.	3.5	52
113	Cystinuria: A Review of Inheritance Patterns, Diagnosis, Medical Treatment and Prevention of Stones. , 2017, , .		1
114	Re: Ghebre Y., Raghu G. Proton pump inhibitors in IPF: beyond mere suppression of gastric acidity. QJM: An International Journal of Medicine 2016; 109:577â€“9. QJM - Monthly Journal of the Association of Physicians, 2017, 110, 261-261.	0.5	0
115	Many Genesâ€”One Disease? Genetics of Nephronophthisis (NPHP) and NPHP-Associated Disorders. Frontiers in Pediatrics, 2017, 5, 287.	1.9	89
116	Case Report: Making a diagnosis of familial renal disease â€“ clinical and patient perspectives. F1000Research, 2017, 6, 470.	1.6	2
117	From disease modelling to personalised therapy in patients with CEP290 mutations. F1000Research, 2017, 6, 669.	1.6	9
118	Kidney Disease in Oman: a View of the Current and Future Landscapes. Iranian Journal of Kidney Diseases, 2017, 11, 263-270.	0.1	7
119	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
120	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
121	Chondrocalcinosis and Gitelman syndrome. QJM - Monthly Journal of the Association of Physicians, 2016, 109, 563-564.	0.5	7
122	Mutations in SLC26A1 Cause Nephrolithiasis. American Journal of Human Genetics, 2016, 98, 1228-1234.	6.2	41
123	Glanzmann thrombasthenia in Pakistan: molecular analysis and identification of novel mutations. Clinical Genetics, 2016, 89, 187-192.	2.0	16
124	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
125	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
126	Case Report: Cervical chondrocalcinosis as a complication of Gitelman syndrome. F1000Research, 2016, 5, 875.	1.6	0

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127	Nephronophthisis. Journal of Pediatric Genetics, 2015, 03, 103-114.	0.7	24
128	FP054PHENOTYPIC ANALYSIS OF A COHORT OF PATIENTS WITH HEPATOCYTE NUCLEAR FACTOR 1 BETA (HNF1b) MUTATIONS. Nephrology Dialysis Transplantation, 2015, 30, iii82-iii83.	0.7	0
129	Yersinia pseudotuberculosis aortitis in a patient with diverticulosis and polycystic kidney disease. Oxford Medical Case Reports, 2015, 2015, 269-271.	0.4	0
130	Successful treatment of hypercalcaemia associated with aCYP24A1mutation with fluconazole: Fig.Â1.. CKJ: Clinical Kidney Journal, 2015, 8, 453-455.	2.9	72
131	Diagnosing Fabry disease–delays and difficulties within discordant siblings. QJM - Monthly Journal of the Association of Physicians, 2015, 108, 585-590.	0.5	4
132	Evidence of digenic inheritance in Alport syndrome. Journal of Medical Genetics, 2015, 52, 163-174.	3.2	129
133	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
134	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
135	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
136	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
137	TMEM231, mutated in orofaciodigital and Meckel syndromes, organizes the ciliary transition zone. Journal of Cell Biology, 2015, 209, 129-142.	5.2	95
138	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
139	Fourteen Monogenic Genes Account for 15% of Nephrolithiasis/Nephrocalcinosis. Journal of the American Society of Nephrology: JASN, 2015, 26, 543-551.	6.1	163
140	DNA replication stress underlies renal phenotypes in CEP290-associated Joubert syndrome. Journal of Clinical Investigation, 2015, 125, 3657-3666.	8.2	48
141	Murine Cep290 phenotypes are modified by genetic backgrounds and provide an impetus for investigating disease modifier alleles. F1000Research, 2015, 4, 590.	1.6	11
142	The challenges and surprises of a definitive molecular genetic diagnosis. Kidney International, 2014, 85, 748-749.	5.2	2
143	Sarcoidosis presenting with hypercalcaemia following withdrawal of long-term immunosuppression in renal transplantation. Oxford Medical Case Reports, 2014, 2014, 86-88.	0.4	3
144	CYP24A1 mutation leading to nephrocalcinosis. Kidney International, 2014, 85, 1475.	5.2	19

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145	Characterization of CSF2RA mutation related juvenile pulmonary alveolar proteinosis. Orphanet Journal of Rare Diseases, 2014, 9, 171.	2.7	61
146	Genetic testing can resolve diagnostic confusion in Alport syndrome. CKJ: Clinical Kidney Journal, 2014, 7, 197-200.	2.9	31
147	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
148	Functional modelling of a novel mutation in BBS5. Cilia, 2014, 3, 3.	1.8	13
149	A novel CLDN16 mutation in a large family with familial hypomagnesaemia with hypercalciuria and nephrocalcinosis. BMC Research Notes, 2013, 6, 527.	1.4	9
150	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
151	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
152	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
153	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
154	ANKH and Renal Stone Formation in Ankylosing Spondylitis. Journal of Rheumatology, 2012, 39, 1756-1756.	2.0	2
155	A meckelinâ€“filamin A interaction mediates ciliogenesis. Human Molecular Genetics, 2012, 21, 1272-1286.	2.9	96
156	Is there anything good in uric acid?. QJM - Monthly Journal of the Association of Physicians, 2012, 105, 395-395.	0.5	1
157	Exome Capture Reveals ZNF423 and CEP164 Mutations, Linking Renal Ciliopathies to DNA Damage Response Signaling. Cell, 2012, 150, 533-548.	28.9	347
158	Investigating Embryonic Expression Patterns and Evolution of AHI1 and CEP290 Genes, Implicated in Joubert Syndrome. PLoS ONE, 2012, 7, e44975.	2.5	15
159	Update of PAX2 mutations in renal coloboma syndrome and establishment of a locus-specific database. Human Mutation, 2012, 33, 457-466.	2.5	109
160	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
161	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
162	Failure to thrive and nephrolithiasis in a boy with congenital cyanotic heart anomalyâ€”questions. Pediatric Nephrology, 2011, 26, 2153-2154.	1.7	1

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163	Failure to thrive and nephrolithiasis in a boy with congenital cyanotic heart anomaly: answers. Pediatric Nephrology, 2011, 26, 2155-2157.	1.7	0
164	Senior-Loken syndrome secondary to NPHP5/IQCB1 mutation in an Iranian family. CKJ: Clinical Kidney Journal, 2011, 4, 421-423.	2.9	5
165	Nephronophthisis: A Genetically Diverse Ciliopathy. International Journal of Nephrology, 2011, 2011, 1-10.	1.3	44
166	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
167	Renal Stone Disease. Nephron Physiology, 2011, 118, p35-p44.	1.2	9
168	Clinical and Functional Characterization of URAT1 Variants. PLoS ONE, 2011, 6, e28641.	2.5	48
169	Juvenile nephronophthisis on MRIâ€"a potential case of Joubert syndrome?. Pediatric Radiology, 2010, 40, 1581-1581.	2.0	1
170	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
171	A novel mutation in NPHS2 causing nephrotic syndrome in a Saudi Arabian family. CKJ: Clinical Kidney Journal, 2010, 3, 545-548.	2.9	0
172	Primary hyperparathyroidism: just how â€˜primaryâ€™ is it really?. Therapeutic Advances in Endocrinology and Metabolism, 2010, 1, 191-196.	3.2	1
173	Secondary hyperparathyroidism in a poorly compliant patient. QJM - Monthly Journal of the Association of Physicians, 2010, 103, 125-125.	0.5	0
174	The Medical Management of Urolithiasis. British Journal of Medical and Surgical Urology, 2010, 3, 87-95.	0.2	15
175	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
176	Individuals with mutations in XPNPEP3, which encodes a mitochondrial protein, develop a nephronophthisis-like nephropathy. Journal of Clinical Investigation, 2010, 120, 1362-1362.	8.2	0
177	Genetic and physical interaction between the NPHP5 and NPHP6 gene products. Human Molecular Genetics, 2009, 18, 4226-4226.	2.9	1
178	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
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