Robert Kleta

List of Publications by Year
in descending order

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102 papers	6,089 citations	61984 43 h-index	74163 75 g-index
102 all docs	102 docs citations	102 times ranked	7500 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	Gitelman-Like Syndrome Caused by Pathogenic Variants in mtDNA. Journal of the American Society of Nephrology: JASN, 2022, 33, 305-325.	6.1	26
3	A Founder Mutation in EHD1 Presents with Tubular Proteinuria and Deafness. Journal of the American Society of Nephrology: JASN, 2022, 33, 732-745.	6.1	7
4	Novel insights in the genetics of steroid-sensitive nephrotic syndrome in childhood. Pediatric Nephrology, 2021, 36, 2165-2175.	1.7	11
5	Inherited Tubulopathies of the Kidney. Clinical Journal of the American Society of Nephrology: CJASN, 2021, 16, 620-630.	4.5	49
6	Defects in KCNJ16 Cause a Novel Tubulopathy with Hypokalemia, Salt Wasting, Disturbed Acid-Base Homeostasis, and Sensorineural Deafness. Journal of the American Society of Nephrology: JASN, 2021, 32, 1498-1512.	6.1	46
7	Founder mutation in the PMM2 promotor causes hyperinsulinemic hypoglycaemia/polycystic kidney disease (HIPKD). Molecular Genetics & Genomic Medicine, 2021, , e1674.	1.2	2
8	Identification of a Locus on the X Chromosome Linked to Familial Membranous Nephropathy. Kidney International Reports, 2021, 6, 1669-1676.	0.8	3
9	Tubulopathy meets Sherlock Holmes: biochemical fingerprinting of disorders of altered kidney tubular salt handling. Pediatric Nephrology, 2021, 36, 2553-2561.	1.7	11
10	Distinct Mitochondrial Pathologies Caused by Mutations of the Proximal Tubular Enzymes EHHADH and GATM. Frontiers in Physiology, 2021, 12, 715485.	2.8	10
11	Quantification of FAM20A in human milk and identification of calcium metabolism proteins. Physiological Reports, 2021, 9, e15150.	1.7	1
12	Bartter and Gitelman syndromes: Questions of class. Pediatric Nephrology, 2020, 35, 1815-1824.	1.7	30
13	Genetics of renovascular hypertension in children. Journal of Hypertension, 2020, 38, 1964-1970.	0.5	15
14	HEROIC: a 5-year observational cohort study aimed at identifying novel factors that drive diabetic kidney disease: rationale and study protocol. BMJ Open, 2020, 10, e033923.	1.9	1
15	Pseudouridylation defect due to DKC1 and NOP10 mutations causes nephrotic syndrome with cataracts, hearing impairment, and enterocolitis. Proceedings of the National Academy of Sciences of the United States of America, 2020, 117, 15137-15147.	7.1	32
16	The genetic architecture of membranous nephropathy and its potential to improve non-invasive diagnosis. Nature Communications, 2020, 11, 1600.	12.8	120
17	TRAP1 chaperone protein mutations and autoinflammation. Life Science Alliance, 2020, 3, e201900376.	2.8	9
18	Genetic Identification of Two Novel Loci Associated with Steroid-Sensitive Nephrotic Syndrome. Journal of the American Society of Nephrology: JASN, 2019, 30, 1375-1384.	6.1	40

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19	High-throughput sequencing contributes to the diagnosis of tubulopathies and familial hypercalcemia hypocalciuria in adults. Kidney International, 2019, 96, 1408-1416.	5.2	36
20	HNF1B Mutations Are Associated With a Gitelman-like Tubulopathy That Develops During Childhood. Kidney International Reports, 2019, 4, 1304-1311.	0.8	39
21	Noncoding deletions reveal a gene that is critical for intestinal function. Nature, 2019, 571, 107-111.	27.8	24
22	Treatment and long-term outcome in primary distal renal tubular acidosis. Nephrology Dialysis Transplantation, 2019, 34, 981-991.	0.7	75
23	Molecular Basis for Autosomal-Dominant Renal Fanconi Syndrome Caused by HNF4A. Cell Reports, 2019, 29, 4407-4421.e5.	6.4	31
24	Enamel renal syndrome: A novel homozygous FAM20A founder mutation in 5 new Brazilian families. European Journal of Medical Genetics, 2019, 62, 103561.	1.3	16
25	Zebrafish as a model for kidney function and disease. Pediatric Nephrology, 2019, 34, 751-762.	1.7	61
26	Simultaneous sequencing of 37 genes identified causative mutations in the majority of children with renal tubulopathies. Kidney International, 2018, 93, 961-967.	5.2	77
27	Glycine Amidinotransferase (GATM), Renal Fanconi Syndrome, and Kidney Failure. Journal of the American Society of Nephrology: JASN, 2018, 29, 1849-1858.	6.1	53
28	Salt-Losing Tubulopathies in Children: Whatâ€™s New, Whatâ€™s Controversial?. Journal of the American Society of Nephrology: JASN, 2018, 29, 727-739.	6.1	57
29	Clinical and diagnostic features of Bartter and Gitelman syndromes. CKJ: Clinical Kidney Journal, 2018, 11, 302-309.	2.9	56
30	OVAS: an open-source variant analysis suite with inheritance modelling. BMC Bioinformatics, 2018, 19, 46.	2.6	1
31	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
32	Germline De Novo Mutations in ATP1A1 Cause Renal Hypomagnesemia, Refractory Seizures, and Intellectual Disability. American Journal of Human Genetics, 2018, 103, 808-816.	6.2	74
33	MUC1 Makes Me Miserable. Journal of the American Society of Nephrology: JASN, 2018, 29, 2257-2258.	6.1	6
34	Genetic risk variants for membranous nephropathy: extension of and association with other chronic kidney disease aetiologies. Nephrology Dialysis Transplantation, 2017, 32, 325-332.	0.7	63
35	Genetic causes of hypomagnesemia, a clinical overview. Pediatric Nephrology, 2017, 32, 1123-1135.	1.7	123
36	Autosomal dominant familial Mediterranean fever in Northern European Caucasians associated with deletion of p.M694 residueâ€”a case series and genetic exploration. Rheumatology, 2017, 56, 209-213.	1.9	49

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37	Of dogs and men. European Journal of Human Genetics, 2017, 25, 161-161.	2.8	2
38	Galactosylation of IgA1 Is Associated with Common Variation in C1GALT1. Journal of the American Society of Nephrology: JASN, 2017, 28, 2158-2166.	6.1	93
39	Clinical and molecular aspects of distal renal tubular acidosis in children. Pediatric Nephrology, 2017, 32, 987-996.	1.7	76
40	An example of the utility of genomic analysis for fast and accurate clinical diagnosis of complex rare phenotypes. Orphanet Journal of Rare Diseases, 2017, 12, 24.	2.7	10
41	Epidemiology of paediatric renal stone disease: a 22-year single centre experience in the UK. BMC Nephrology, 2017, 18, 136.	1.8	59
42	Polycystic Kidney Disease with Hyperinsulinemic Hypoglycemia Caused by a Promoter Mutation in Phosphomannomutase 2. Journal of the American Society of Nephrology: JASN, 2017, 28, 2529-2539.	6.1	99
43	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
44	Autoinflammatory periodic fever, immunodeficiency, and thrombocytopenia (PFIT) caused by mutation in actin-regulatory gene WDR1 . Journal of Experimental Medicine, 2017, 214, 59-71.	8.5	117
45	Enamel-renal syndrome in 2 patients with a mutation in FAM20 A and atypical hypertrichosis and hearing loss phenotypes. Oral Surgery, Oral Medicine, Oral Pathology and Oral Radiology, 2017, 123, 229-234.e2.	0.4	16
46	Pathophysiology, current treatments and future targets in hereditary forms of renal Fanconi syndrome. Expert Opinion on Orphan Drugs, 2017, 5, 45-54.	0.8	3
47	HaploForge: a comprehensive pedigree drawing and haplotype visualization web application. Bioinformatics, 2017, 33, 3871-3877.	4.1	3
48	Membranous nephropathy: a retrospective observational study of membranous nephropathy in north east and central London. BMC Nephrology, 2017, 18, 201.	1.8	10
49	Mutations in linker for activation of TÂcells (LAT) lead to a novel form of severe combined immunodeficiency. Journal of Allergy and Clinical Immunology, 2017, 139, 634-642.e5.	2.9	38
50	Fainting Fanconi syndrome clarified by proxy: a case report. BMC Nephrology, 2017, 18, 230.	1.8	12
51	TO029EPIDEMIOLOGY OF PAEDIATRIC UROLITHIASIS IN A LARGE COHORT IN UK. Nephrology Dialysis Transplantation, 2016, 31, i73-i74.	0.7	0
52	EAST syndrome: Clinical, pathophysiological, and genetic aspects of mutations in KCNJ10. Rare Diseases (Austin, Tex), 2016, 4, e1195043.	1.8	36
53	Founder mutation inKCNJ10in Pakistani patients with EAST syndrome. Molecular Genetics & Genomic Medicine, 2016, 4, 521-526.	1.2	11
54	Renal Fanconi Syndrome Is Caused by a Mistargeting-Based Mitochondriopathy. Cell Reports, 2016, 15, 1423-1429.	6.4	27

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55	Genetic, pathophysiological, and clinical aspects of nephrocalcinosis. American Journal of Physiology - Renal Physiology, 2016, 311, F1243-F1252.	2.7	46
56	Renal apnoea: extreme disturbance of homoeostasis in a child with Bartter syndrome type IV. Lancet, The, 2016, 388, 631-632.	13.7	20
57	Autosomal-Recessive Mutations in SLC34A1 Encoding Sodium-Phosphate Cotransporter 2A Cause Idiopathic Infantile Hypercalcemia. Journal of the American Society of Nephrology: JASN, 2016, 27, 604-614.	6.1	207
58	A Missense Mutation in KCTD17 Causes Autosomal Dominant Myoclonus-Dystonia. American Journal of Human Genetics, 2015, 96, 938-947.	6.2	109
59	Insulin Receptor and the Kidney: Nephrocalcinosis in Patients with Recessive INSR Mutations. Nephron Physiology, 2015, 128, 55-61.	1.2	18
60	The kinetochore protein,CENPF, is mutated in human ciliopathy and microcephaly phenotypes. Journal of Medical Genetics, 2015, 52, 147-156.	3.2	75
61	Renal Fanconi syndrome: taking a proximal look at the nephron. Nephrology Dialysis Transplantation, 2015, 30, 1456-1460.	0.7	74
62	Consanguinity in Saudi Arabia: A Unique Opportunity for Pediatric Kidney Research. American Journal of Kidney Diseases, 2014, 63, 304-310.	1.9	19
63	Mistargeting of Peroxisomal EHHADH and Inherited Renal Fanconi's Syndrome. New England Journal of Medicine, 2014, 370, 129-138.	27.0	99
64	Enabling the genomic revolution in Africa. Science, 2014, 344, 1346-1348.	12.6	361
65	The Case | Renal tubular acidosis and eye findings. Kidney International, 2014, 86, 217-218.	5.2	20
66	A novel claudin-16 mutation, severe bone disease, and nephrocalcinosis. Lancet, The, 2014, 383, 98.	13.7	8
67	Nephrocalcinosis (Enamel Renal Syndrome) Caused by Autosomal Recessive FAM20A Mutations. Nephron Physiology, 2013, 122, 1-6.	1.2	84
68	Generation and validation of a zebrafish model of EAST (Epilepsy, ataxia, sensorineural deafness and) Tj ETQq0 0 0 rgBT /Overlock 10 Tf	2.4	48
69	KCNJ10 Mutations Display Differential Sensitivity to Heteromerisation with KCNJ16. Nephron Physiology, 2013, 123, 7-14.	1.2	34
70	Neurological features of epilepsy, ataxia, sensorineural deafness, tubulopathy syndrome. Developmental Medicine and Child Neurology, 2013, 55, 846-856.	2.1	53
71	Epilepsy in kcnj10 Morphant Zebrafish Assessed with a Novel Method for Long-Term EEG Recordings. PLoS ONE, 2013, 8, e79765.	2.5	49
72	Genetic testing in renal disease. Pediatric Nephrology, 2012, 27, 873-883.	1.7	45

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73	Genetic basis of cystinosis in Turkish patients: a single-center experience. Pediatric Nephrology, 2012, 27, 115-121.	1.7	41
74	Altered electroretinograms in patients with KCNJ10 mutations and EAST syndrome. Journal of Physiology, 2011, 589, 1681-1689.	2.9	66
75	A patient with polyuria and hydronephrosis: answer. Pediatric Nephrology, 2011, 26, 1979-1980.	1.7	3
76	The salt-wasting phenotype of EAST syndrome, a disease with multifaceted symptoms linked to the KCNJ10 K+ channel. Pflugers Archiv European Journal of Physiology, 2011, 461, 423-435.	2.8	57
77	KCNJ10 Mutations Disrupt Function in Patients with EAST Syndrome. Nephron Physiology, 2011, 119, p40-p48.	1.2	49
78	Filtering the genes and sorting the glomerular filter: a new piece in the puzzle?. Nephrology Dialysis Transplantation, 2011, 26, 2743-2745.	0.7	1
79	Risk HLA-DQA1 and PLA₂R1 Alleles in Idiopathic Membranous Nephropathy. New England Journal of Medicine, 2011, 364, 616-626.	27.0	442
80	KCNJ10 gene mutations causing EAST syndrome (epilepsy, ataxia, sensorineural deafness, and) Tj ETQq0 0 0 rgBT /Overlock 10 Tf 50 46 United States of America, 2010, 107, 14490-14495.	7.1	186
81	Cystinosis and Mickey Mouse. Nephrology Dialysis Transplantation, 2010, 25, 1032-1033.	0.7	2
82	Epilepsy, Ataxia, Sensorineural Deafness, Tubulopathy, andKCNJ10Mutations. New England Journal of Medicine, 2009, 360, 1960-1970.	27.0	518
83	Fanconi or not Fanconi? Lowe Syndrome Revisited. Clinical Journal of the American Society of Nephrology: CJASN, 2008, 3, 1244-1245.	4.5	32
84	Nephropathic Cystinosis in Adults: Natural History and Effects of Oral Cysteamine Therapy. Annals of Internal Medicine, 2007, 147, 242.	3.9	202
85	Nodular Regenerative Hyperplasia and Severe Portal Hypertension in Cystinosis. Clinical Gastroenterology and Hepatology, 2006, 4, 387-394.	4.4	36
86	A deeper look into cysteamine absorption for the treatment of cystinosis. Journal of Pediatrics, 2006, 148, 718-719.	1.8	5
87	Bartter Syndromes and Other Salt-Losing Tubulopathies. Nephron Physiology, 2006, 104, p73-p80.	1.2	104
88	Swallowing Dysfunction in 101 Patients with Nephropathic Cystinosis. Medicine (United States), 2005, 84, 137-146.	1.0	87
89	First NIH/Office of Rare Diseases Conference on Cystinosis: past, present, and future. Pediatric Nephrology, 2005, 20, 452-454.	1.7	29
90	Pharmacological treatment of nephropathic cystinosis with cysteamine. Expert Opinion on Pharmacotherapy, 2004, 5, 2255-2262.	1.8	74

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91	Mutations in SLC6A19, encoding B0AT1, cause Hartnup disorder. Nature Genetics, 2004, 36, 999-1002.	21.4	272
92	FISH diagnosis of the common 57-kb deletion in CTNS causing cystinosis. Human Genetics, 2004, 115, 510-514.	3.8	36
93	Keratopathy of Multiple Myeloma Masquerading as Corneal Crystals of Ocular Cystinosis. Mayo Clinic Proceedings, 2004, 79, 410-412.	3.0	26
94	Renal glucosuria due to SGLT2 mutations. Molecular Genetics and Metabolism, 2004, 82, 56-58.	1.1	50
95	Long-term follow-up of well-treated nephropathic cystinosis patients. Journal of Pediatrics, 2004, 145, 555-560.	1.8	113
96	Biochemical and molecular analyses of infantile free sialic acid storage disease in North American children. American Journal of Medical Genetics Part A, 2003, 120A, 28-33.	2.4	31
97	Inhibition of Na+-Dependent Transporters in Cystine-Loaded Human Renal Cells: Electrophysiological Studies on the Fanconi Syndrome of Cystinosis. Journal of the American Society of Nephrology: JASN, 2002, 13, 2085-2093.	6.1	29
98	Cystinosis. Journal of the American Society of Nephrology: JASN, 2002, 13, 2189-2191.	6.1	12
99	3-Methylglutaconic aciduria type III in a non-Iraqi-Jewish kindred: clinical and molecular findings. Molecular Genetics and Metabolism, 2002, 76, 201-206.	1.1	36
100	CTNS Mutations in African American Patients with Cystinosis. Molecular Genetics and Metabolism, 2001, 74, 332-337.	1.1	33
101	New Treatment Options for Bartter's Syndrome. New England Journal of Medicine, 2000, 343, 661-662.	27.0	52
102	Renal involvement in children with malignancies. Pediatric Nephrology, 1999, 13, 153-162.	1.7	97