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	Epilepsy, Ataxia, Sensorineural Deafness, Tubulopathy, andKCNJ10Mutations. New England Journal of Medicine, 2009, 360, 1960-1970.	27.0	518
2	Risk HLA-DQA1 and PLA₂R1 Alleles in Idiopathic Membranous Nephropathy. New England Journal of Medicine, 2011, 364, 616-626.	27.0	442
3	Enabling the genomic revolution in Africa. Science, 2014, 344, 1346-1348.	12.6	361
4	Mutations in SLC6A19, encoding B0AT1, cause Hartnup disorder. Nature Genetics, 2004, 36, 999-1002.	21.4	272
5	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
6	Nephropathic Cystinosis in Adults: Natural History and Effects of Oral Cysteamine Therapy. Annals of Internal Medicine, 2007, 147, 242.	3.9	202
7	KCNJ10 gene mutations causing EAST syndrome (epilepsy, ataxia, sensorineural deafness, and) Tj ETQq1 1 0.784314 rgBT /Overlock 10 United States of America, 2010, 107, 14490-14495.	7.1	186
8	Genetic causes of hypomagnesemia, a clinical overview. Pediatric Nephrology, 2017, 32, 1123-1135.	1.7	123
9	The genetic architecture of membranous nephropathy and its potential to improve non-invasive diagnosis. Nature Communications, 2020, 11, 1600.	12.8	120
10	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
11	Long-term follow-up of well-treated nephropathic cystinosis patients. Journal of Pediatrics, 2004, 145, 555-560.	1.8	113
12	A Missense Mutation in KCTD17 Causes Autosomal Dominant Myoclonus-Dystonia. American Journal of Human Genetics, 2015, 96, 938-947.	6.2	109
13	Bartter Syndromes and Other Salt-Losing Tubulopathies. Nephron Physiology, 2006, 104, p73-p80.	1.2	104
14	Mistargeting of Peroxisomal EHHADH and Inherited Renal Fanconi's Syndrome. New England Journal of Medicine, 2014, 370, 129-138.	27.0	99
15	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
16	Renal involvement in children with malignancies. Pediatric Nephrology, 1999, 13, 153-162.	1.7	97
17	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
18	Swallowing Dysfunction in 101 Patients with Nephropathic Cystinosis. Medicine (United States), 2005, 84, 137-146.	1.0	87

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19	Nephrocalcinosis (Enamel Renal Syndrome) Caused by Autosomal Recessive FAM20A Mutations. Nephron Physiology, 2013, 122, 1-6.	1.2	84
20	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
21	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
22	Clinical and molecular aspects of distal renal tubular acidosis in children. Pediatric Nephrology, 2017, 32, 987-996.	1.7	76
23	The kinetochore protein,CENPF, is mutated in human ciliopathy and microcephaly phenotypes. Journal of Medical Genetics, 2015, 52, 147-156.	3.2	75
24	Treatment and long-term outcome in primary distal renal tubular acidosis. Nephrology Dialysis Transplantation, 2019, 34, 981-991.	0.7	75
25	Pharmacological treatment of nephropathic cystinosis with cysteamine. Expert Opinion on Pharmacotherapy, 2004, 5, 2255-2262.	1.8	74
26	Renal Fanconi syndrome: taking a proximal look at the nephron. Nephrology Dialysis Transplantation, 2015, 30, 1456-1460.	0.7	74
27	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
28	Altered electroretinograms in patients with KCNJ10 mutations and EAST syndrome. Journal of Physiology, 2011, 589, 1681-1689.	2.9	66
29	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
30	Zebrafish as a model for kidney function and disease. Pediatric Nephrology, 2019, 34, 751-762.	1.7	61
31	Epidemiology of paediatric renal stone disease: a 22-year single centre experience in the UK. BMC Nephrology, 2017, 18, 136.	1.8	59
32	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
33	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
34	Clinical and diagnostic features of Bartter and Gitelman syndromes. CKJ: Clinical Kidney Journal, 2018, 11, 302-309.	2.9	56
35	Neurological features of epilepsy, ataxia, sensorineural deafness, tubulopathy syndrome. Developmental Medicine and Child Neurology, 2013, 55, 846-856.	2.1	53
36	Glycine Amidinotransferase (GATM), Renal Fanconi Syndrome, and Kidney Failure. Journal of the American Society of Nephrology: JASN, 2018, 29, 1849-1858.	6.1	53

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37	New Treatment Options for Bartter's Syndrome. New England Journal of Medicine, 2000, 343, 661-662.	27.0	52
38	Renal glucosuria due to SGLT2 mutations. Molecular Genetics and Metabolism, 2004, 82, 56-58.	1.1	50
39	KCNJ10 Mutations Disrupt Function in Patients with EAST Syndrome. Nephron Physiology, 2011, 119, p40-p48.	1.2	49
40	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
41	Inherited Tubulopathies of the Kidney. Clinical Journal of the American Society of Nephrology: CJASN, 2021, 16, 620-630.	4.5	49
42	Epilepsy in kcnj10 Morphant Zebrafish Assessed with a Novel Method for Long-Term EEG Recordings. PLoS ONE, 2013, 8, e79765.	2.5	49
43	Generation and validation of a zebrafish model of EAST (Epilepsy, ataxia, sensorineural deafness and) Tj ETQq1 1 0.784314 rgBT /Over	2.4	48
44	Genetic, pathophysiological, and clinical aspects of nephrocalcinosis. American Journal of Physiology - Renal Physiology, 2016, 311, F1243-F1252.	2.7	46
45	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
46	Genetic testing in renal disease. Pediatric Nephrology, 2012, 27, 873-883.	1.7	45
47	Genetic basis of cystinosis in Turkish patients: a single-center experience. Pediatric Nephrology, 2012, 27, 115-121.	1.7	41
48	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
49	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
50	HNF1B Mutations Are Associated With a Gitelman-like Tubulopathy That Develops During Childhood. Kidney International Reports, 2019, 4, 1304-1311.	0.8	39
51	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
52	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
53	FISH diagnosis of the common 57-kb deletion in CTNS causing cystinosis. Human Genetics, 2004, 115, 510-514.	3.8	36
54	Nodular Regenerative Hyperplasia and Severe Portal Hypertension in Cystinosis. Clinical Gastroenterology and Hepatology, 2006, 4, 387-394.	4.4	36

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55	EAST syndrome: Clinical, pathophysiological, and genetic aspects of mutations in KCNJ10. Rare Diseases (Austin, Tex), 2016, 4, e1195043.	1.8	36
56	High-throughput sequencing contributes to the diagnosis of tubulopathies and familial hypercalcemia hypocalciuria in adults. Kidney International, 2019, 96, 1408-1416.	5.2	36
57	KCNJ10 Mutations Display Differential Sensitivity to Heteromerisation with KCNJ16. Nephron Physiology, 2013, 123, 7-14.	1.2	34
58	CTNS Mutations in African American Patients with Cystinosis. Molecular Genetics and Metabolism, 2001, 74, 332-337.	1.1	33
59	Fanconi or not Fanconi? Lowe Syndrome Revisited. Clinical Journal of the American Society of Nephrology: CJASN, 2008, 3, 1244-1245.	4.5	32
60	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
61	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
62	Molecular Basis for Autosomal-Dominant Renal Fanconi Syndrome Caused by HNF4A. Cell Reports, 2019, 29, 4407-4421.e5.	6.4	31
63	Bartter and Gitelman syndromes: Questions of class. Pediatric Nephrology, 2020, 35, 1815-1824.	1.7	30
64	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
65	First NIH/Office of Rare Diseases Conference on Cystinosis: past, present, and future. Pediatric Nephrology, 2005, 20, 452-454.	1.7	29
66	Renal Fanconi Syndrome Is Caused by a Mistargeting-Based Mitochondriopathy. Cell Reports, 2016, 15, 1423-1429.	6.4	27
67	Keratopathy of Multiple Myeloma Masquerading as Corneal Crystals of Ocular Cystinosis. Mayo Clinic Proceedings, 2004, 79, 410-412.	3.0	26
68	Gitelman-Like Syndrome Caused by Pathogenic Variants in mtDNA. Journal of the American Society of Nephrology: JASN, 2022, 33, 305-325.	6.1	26
69	Noncoding deletions reveal a gene that is critical for intestinal function. Nature, 2019, 571, 107-111.	27.8	24
70	The Case | Renal tubular acidosis and eye findings. Kidney International, 2014, 86, 217-218.	5.2	20
71	Renal apnoea: extreme disturbance of homoeostasis in a child with Bartter syndrome type IV. Lancet, The, 2016, 388, 631-632.	13.7	20
72	Consanguinity in Saudi Arabia: A Unique Opportunity for Pediatric Kidney Research. American Journal of Kidney Diseases, 2014, 63, 304-310.	1.9	19

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73	Insulin Receptor and the Kidney: Nephrocalcinosis in Patients with Recessive INSR Mutations. Nephron Physiology, 2015, 128, 55-61.	1.2	18
74	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
75	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
76	Genetics of renovascular hypertension in children. Journal of Hypertension, 2020, 38, 1964-1970.	0.5	15
77	Cystinosis. Journal of the American Society of Nephrology: JASN, 2002, 13, 2189-2191.	6.1	12
78	Fainting Fanconi syndrome clarified by proxy: a case report. BMC Nephrology, 2017, 18, 230.	1.8	12
79	Founder mutation inKCNJ10in Pakistani patients with EAST syndrome. Molecular Genetics & Genomic Medicine, 2016, 4, 521-526.	1.2	11
80	Novel insights in the genetics of steroid-sensitive nephrotic syndrome in childhood. Pediatric Nephrology, 2021, 36, 2165-2175.	1.7	11
81	Tubulopathy meets Sherlock Holmes: biochemical fingerprinting of disorders of altered kidney tubular salt handling. Pediatric Nephrology, 2021, 36, 2553-2561.	1.7	11
82	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
83	Membranous nephropathy: a retrospective observational study of membranous nephropathy in north east and central London. BMC Nephrology, 2017, 18, 201.	1.8	10
84	Distinct Mitochondrial Pathologies Caused by Mutations of the Proximal Tubular Enzymes EHHADH and GATM. Frontiers in Physiology, 2021, 12, 715485.	2.8	10
85	TRAP1 chaperone protein mutations and autoinflammation. Life Science Alliance, 2020, 3, e201900376.	2.8	9
86	Treatment and long-term outcome in primary nephrogenic diabetes insipidus. Nephrology Dialysis Transplantation, 2023, 38, 2120-2130.	0.7	9
87	A novel claudin-16 mutation, severe bone disease, and nephrocalcinosis. Lancet, The, 2014, 383, 98.	13.7	8
88	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
89	MUC1 Makes Me Miserable. Journal of the American Society of Nephrology: JASN, 2018, 29, 2257-2258.	6.1	6
90	A deeper look into cysteamine absorption for the treatment of cystinosis. Journal of Pediatrics, 2006, 148, 718-719.	1.8	5

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91	A patient with polyuria and hydronephrosis: answer. Pediatric Nephrology, 2011, 26, 1979-1980.	1.7	3
92	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
93	HaploForge: a comprehensive pedigree drawing and haplotype visualization web application. Bioinformatics, 2017, 33, 3871-3877.	4.1	3
94	Identification of a Locus on the X Chromosome Linked to Familial Membranous Nephropathy. Kidney International Reports, 2021, 6, 1669-1676.	0.8	3
95	Cystinosis and Mickey Mouse. Nephrology Dialysis Transplantation, 2010, 25, 1032-1033.	0.7	2
96	Of dogs and men. European Journal of Human Genetics, 2017, 25, 161-161.	2.8	2
97	Founder mutation in the PMM2 promotor causes hyperinsulinemic hypoglycaemia/polycystic kidney disease (HIPKD). Molecular Genetics & Genomic Medicine, 2021, , e1674.	1.2	2
98	Filtering the genes and sorting the glomerular filter: a new piece in the puzzle?. Nephrology Dialysis Transplantation, 2011, 26, 2743-2745.	0.7	1
99	OVAS: an open-source variant analysis suite with inheritance modelling. BMC Bioinformatics, 2018, 19, 46.	2.6	1
100	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
101	Quantification of FAM20A in human milk and identification of calcium metabolism proteins. Physiological Reports, 2021, 9, e15150.	1.7	1
102	TO029EPIDEMIOLOGY OF PAEDIATRIC UROLITHIASIS IN A LARGE COHORT IN UK. Nephrology Dialysis Transplantation, 2016, 31, i73-i74.	0.7	0