## **Eric Olinger**

List of Publications by Year in descending order

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#	Article	lF	CITATIONS
1	Kidney traits on repeatâ $\in$ "the role of MUC1 VNTR. Kidney International, 2022, 101, 863-866.	5.2	1
2	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
3	Meta-GWAS Reveals Novel Genetic Variants Associated with Urinary Excretion of Uromodulin. Journal of the American Society of Nephrology: JASN, 2022, 33, 511-529.	6.1	14
4	Biallelic variants in <scp><i>TTC21B</i></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
5	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
6	Pseudodominant Alport syndrome caused by pathogenic homozygous and compound heterozygous <i>COL4A3</i> splicing variants. Annals of Human Genetics, 2022, 86, 145-152.	0.8	3
7	What Does Uromodulin Do?. Clinical Journal of the American Society of Nephrology: CJASN, 2021, 16, 150-153.	4.5	18
8	Uromodulin, Salt, and 24-Hour Blood Pressure in the General Population. Clinical Journal of the American Society of Nephrology: CJASN, 2021, 16, 787-789.	4.5	11
9	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
10	Molecular genetics of renal ciliopathies. Biochemical Society Transactions, 2021, 49, 1205-1220.	3.4	15
11	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
12	A discarded synonymous variant in <i>NPHP3</i> explains nephronophthisis and congenital hepatic fibrosis in several families. Human Mutation, 2021, 42, 1221-1228.	2.5	12
13	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
14	Clinical and genetic spectra of autosomal dominant tubulointerstitial kidney disease. Nephrology Dialysis Transplantation, 2021, , .	0.7	12
15	Identification of <i>LAMA1</i> mutations ends diagnostic odyssey and has prognostic implications for patients with presumed Joubert syndrome. Brain Communications, 2021, 3, fcab163.	3.3	8
16	Update of genetic variants in <i>CEP120</i> and <i>CC2D2A</i> —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
17	Mendelian randomization to assess causality between uromodulin, blood pressure and chronic kidney disease. Kidney International, 2021, 100, 1282-1291.	5.2	20
18	Clinical and genetic spectra of kidney disease caused by REN mutations. Kidney International, 2020, 98, 1397-1400.	5.2	7

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19	Genetic and Clinical Predictors of Age of ESKD in Individuals With Autosomal Dominant Tubulointerstitial Kidney Disease Due to UMOD Mutations. Kidney International Reports, 2020, 5, 1472-1485.	0.8	30
20	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
21	The cryo-EM structure of the human uromodulin filament core reveals a unique assembly mechanism. ELife, 2020, 9, .	6.0	26
22	Hepsin-mediated Processing of Uromodulin is Crucial for Salt-sensitivity and Thick Ascending Limb Homeostasis. Scientific Reports, 2019, 9, 12287.	3.3	41
23	Autosomal dominant tubulointerstitial kidney disease. Nature Reviews Disease Primers, 2019, 5, 60.	30.5	139
24	The Urinary Excretion of Uromodulin is Regulated by the Potassium Channel ROMK. Scientific Reports, 2019, 9, 19517.	3.3	21
25	Claudins: a tale of interactions in the thick ascending limb. Kidney International, 2018, 93, 535-537.	5.2	15
26	Multiplex epithelium dysfunction due to CLDN10 mutation: the HELIX syndrome. Genetics in Medicine, 2018, 20, 190-201.	2.4	75
27	The excretion of uromodulin is modulated by the calcium-sensing receptor. Kidney International, 2018, 94, 882-886.	5.2	20
28	Uromodulin and Nephron Mass. Clinical Journal of the American Society of Nephrology: CJASN, 2018, 13, 1556-1557.	4.5	44
29	Common variants in CLDN14 are associated with differential excretion of magnesium over calcium in urine. Pflugers Archiv European Journal of Physiology, 2017, 469, 91-103.	2.8	27
30	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
31	Mouse model for inherited renal fibrosis associated with endoplasmic reticulum stress. DMM Disease Models and Mechanisms, 2017, 10, 773-786.	2.4	34
32	Common Elements in Rare Kidney Diseases: Conclusions from a Kidney Disease: Improving Global Outcomes (KDIGO) Controversies Conference. Kidney International, 2017, 92, 796-808.	5.2	40
33	Uromodulin: from physiology to rare and complex kidney disorders. Nature Reviews Nephrology, 2017, 13, 525-544.	9.6	220
34	The value of ECG parameters as markers of treatment response in Fabry cardiomyopathy. Heart, 2016, 102, 1309-1314.	2.9	17
35	Tubular proteinuria in patients with HNF1α mutations: HNF1α drives endocytosis in the proximal tubule. Kidney International, 2016, 89, 1075-1089.	5.2	29
36	Paradoxical response to furosemide in uromodulin-associated kidney disease. Nephrology Dialysis Transplantation, 2015, 30, 330-335.	0.7	23

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37	The serine protease hepsin mediates urinary secretion and polymerisation of Zona Pellucida domain protein uromodulin. ELife, 2015, 4, e08887.	6.0	92