Olivier Gribouval

List of Publications by Year
in descending order

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59 papers	6,753 citations	101384 36 h-index	128067 60 g-index
62 all docs	62 docs citations	62 times ranked	6403 citing authors

#	Article	IF	Citations
1	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.	3.3	32
2	APOL1 risk genotype in European steroid-resistant nephrotic syndrome and/or focal segmental glomerulosclerosis patients of different African ancestries. Nephrology Dialysis Transplantation, 2019, 34, 1885-1893.	0.4	12
3	Defects in t6A tRNA modification due to GON7 and YRDC mutations lead to Galloway-Mowat syndrome. Nature Communications, 2019, 10, 3967.	5.8	66
4	TBC1D8B Loss-of-Function Mutations Lead to X-Linked Nephrotic Syndrome via Defective Trafficking Pathways. American Journal of Human Genetics, 2019, 104, 348-355.	2.6	40
5	APOL1 risk genotype in Europe: Data in patients with focal segmental glomerulosclerosis and after renal transplantation. Nephrologie Et Therapeutique, 2019, 15, S85-S89.	0.2	2
6	Treatment and outcome of congenital nephrotic syndrome. Nephrology Dialysis Transplantation, 2019, 34, 458-467.	0.4	42
7	Human C-terminal CUBN variants associate with chronic proteinuria and normal renal function. Journal of Clinical Investigation, 2019, 130, 335-344.	3.9	54
8	Clinical and genetic heterogeneity in familial steroid-sensitive nephrotic syndrome. Pediatric Nephrology, 2018, 33, 473-483.	0.9	34
9	Comparison of Postdonation Kidney Function Between Caucasian Donors and Low-risk APOL1 Genotype Living Kidney Donors of African Ancestry. Transplantation, 2018, 102, e462-e463.	0.5	5
10	Identification of genetic causes for sporadic steroid-resistant nephrotic syndrome in adults. Kidney International, 2018, 94, 1013-1022.	2.6	51
11	A homozygous KAT2B variant modulates the clinical phenotype of ADD3 deficiency in humans and flies. PLoS Genetics, 2018, 14, e1007386.	1.5	17
12	Mutations in KEOPS-complex genes cause nephrotic syndrome with primary microcephaly. Nature Genetics, 2017, 49, 1529-1538.	9.4	164
13	Low renal but high extrarenal phenotype variability in Schimke immuno-osseous dysplasia. PLoS ONE, 2017, 12, e0180926.	1.1	25
14	Mutations in sphingosine-1-phosphate lyase cause nephrosis with ichthyosis and adrenal insufficiency. Journal of Clinical Investigation, 2017, 127, 912-928.	3.9	160
15	ADCK4-Associated Glomerulopathy Causes Adolescence-Onset FSGS. Journal of the American Society of Nephrology: JASN, 2016, 27, 63-68.	3.0	79
16	Loss-of-Function Mutations in WDR73 Are Responsible for Microcephaly and Steroid-Resistant Nephrotic Syndrome: Galloway-Mowat Syndrome. American Journal of Human Genetics, 2014, 95, 637-648.	2.6	108
17	A Homozygous Missense Mutation in the Ciliary Gene TTC21B Causes Familial FSGS. Journal of the American Society of Nephrology: JASN, 2014, 25, 2435-2443.	3.0	86
18	Absence of cell surface expression of human ACE leads to perinatal death. Human Molecular Genetics, 2014, 23, 1479-1491.	1.4	14

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19	Mutation-dependent recessive inheritance of NPHS2-associated steroid-resistant nephrotic syndrome. Nature Genetics, 2014, 46, 299-304.	9.4	134
20	NPHS2Mutations in Steroid-Resistant Nephrotic Syndrome: A Mutation Update and the Associated Phenotypic Spectrum. Human Mutation, 2014, 35, 178-186.	1.1	76
21	Integrin Alpha 8 Recessive Mutations Are Responsible for Bilateral Renal Agenesis in Humans. American Journal of Human Genetics, 2014, 94, 799.	2.6	1
22	Integrin Alpha 8 Recessive Mutations Are Responsible for Bilateral Renal Agenesis in Humans. American Journal of Human Genetics, 2014, 94, 288-294.	2.6	89
23	APOL1 Polymorphisms and Development of CKD in an IdenticalÂTwin Donor and Recipient Pair. American Journal of Kidney Diseases, 2014, 63, 816-819.	2.1	51
24	The Kidney as a Reservoir for HIV-1 after Renal Transplantation. Journal of the American Society of Nephrology: JASN, 2014, 25, 407-419.	3.0	121
25	DysgÃ©nÃ©sie tubulaire rÃ©nale et mutations des gÃ¨nes du systÃ¨me rÃ©nine angiotensine. Bulletin De L'Academie Nationale De Medecine, 2014, 198, 339-349.	0.0	1
26	LMX1B Mutations Cause Hereditary FSGS without Extrarenal Involvement. Journal of the American Society of Nephrology: JASN, 2013, 24, 1216-1222.	3.0	83
27	ARHGDIA mutations cause nephrotic syndrome via defective RHO GTPase signaling. Journal of Clinical Investigation, 2013, 123, 3243-3253.	3.9	196
28	Spectrum of mutations in the renin-angiotensin system genes in autosomal recessive renal tubular dysgenesis. Human Mutation, 2012, 33, 316-326.	1.1	86
29	INF2Mutations in Charcotâ€“Marieâ€“Tooth Disease with Glomerulopathy. New England Journal of Medicine, 2011, 365, 2377-2388.	13.9	235
30	Loss-of-function point mutations associated with renal tubular dysgenesis provide insights about renin function and cellular trafficking. Human Molecular Genetics, 2011, 20, 301-311.	1.4	13
31	Mutations in INF2 Are a Major Cause of Autosomal Dominant Focal Segmental Glomerulosclerosis. Journal of the American Society of Nephrology: JASN, 2011, 22, 239-245.	3.0	138
32	Analysis of recessive CD2AP and ACTN4 mutations in steroid-resistant nephrotic syndrome. Pediatric Nephrology, 2010, 25, 445-451.	0.9	27
33	Inherited renal tubular dysgenesis may not be universally fatal. Pediatric Nephrology, 2010, 25, 2531-2534.	0.9	21
34	Genotypeâ€“Phenotype Correlations in Non-Finnish Congenital Nephrotic Syndrome. Journal of the American Society of Nephrology: JASN, 2010, 21, 1209-1217.	3.0	127
35	Mutational analysis of the PLCE1 gene in steroid resistant nephrotic syndrome. Journal of Medical Genetics, 2010, 47, 445-452.	1.5	74
36	Angiotensin I-Converting Enzyme Gln1069Arg Mutation Impairs Trafficking to the Cell Surface Resulting in Selective Denaturation of the C-Domain. PLoS ONE, 2010, 5, e10438.	1.1	26

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37	Inherited renal tubular dysgenesis: the first patients surviving the neonatal period. European Journal of Pediatrics, 2008, 167, 311-316.	1.3	28
38	CAN WE LIVE WITHOUT A FUNCTIONAL RENINâ€ANGIOTENSIN SYSTEM?. Clinical and Experimental Pharmacology and Physiology, 2008, 35, 431-433.	0.9	13
39	Nephrin Mutations Can Cause Childhood-Onset Steroid-Resistant Nephrotic Syndrome. Journal of the American Society of Nephrology: JASN, 2008, 19, 1871-1878.	3.0	119
40	COL4A1Mutations and Hereditary Angiopathy, Nephropathy, Aneurysms, and Muscle Cramps. New England Journal of Medicine, 2007, 357, 2687-2695.	13.9	305
41	Autosomal-dominant familial hematuria with retinal arteriolar tortuosity and contractures: A novel syndrome. Kidney International, 2005, 67, 2354-2360.	2.6	36
42	Mutations in genes in the renin-angiotensin system are associated with autosomal recessive renal tubular dysgenesis. Nature Genetics, 2005, 37, 964-968.	9.4	244
43	Case Report. A novel NPHS2 gene mutation in Turkish children with familial steroid-resistant nephrotic syndrome. Nephrology, 2004, 9, 310-312.	0.7	3
44	NPHS2 mutation analysis shows genetic heterogeneityof steroid-resistant nephrotic syndrome and lowpost-transplant recurrence. Kidney International, 2004, 66, 571-579.	2.6	313
45	In vivo expression of podocyte slit diaphragm-associated proteins in nephrotic patients with NPHS2 mutation. Kidney International, 2004, 66, 945-954.	2.6	37
46	Plasma Membrane Targeting of Podocin Through the Classical Exocytic Pathway: Effect of NPHS2 Mutations. Traffic, 2004, 5, 37-44.	1.3	86
47	Gamma-D crystallin gene (CRYGD) mutation causes autosomal dominant congenital cerulean cataracts. Journal of Medical Genetics, 2003, 40, 262-267.	1.5	78
48	Podocin Localizes in the Kidney to the Slit Diaphragm Area. American Journal of Pathology, 2002, 160, 131-139.	1.9	284
49	Evidence of clinical and genetic heterogeneity in autosomal dominant congenital cerulean cataracts. Ophthalmic Genetics, 2002, 23, 199-208.	0.5	11
50	The gene mutated in juvenile nephronophthisis type 4 encodes a novel protein that interacts with nephrocystin. Nature Genetics, 2002, 32, 300-305.	9.4	210
51	Clinical and Genetic Evaluation of Familial Steroid-Responsive Nephrotic Syndrome in Childhood. Journal of the American Society of Nephrology: JASN, 2001, 12, 374-378.	3.0	70
52	NPHS2, encoding the glomerular protein podocin, is mutated in autosomal recessive steroid-resistant nephrotic syndrome. Nature Genetics, 2000, 24, 349-354.	9.4	1,270
53	The photoreceptor cell-specific nuclear receptor gene (PNR) accounts for retinitis pigmentosa in the Crypto-Jews from Portugal (Marranos), survivors from the Spanish Inquisition. Human Genetics, 2000, 107, 276-284.	1.8	86
54	Structure of the Gene for Congenital Nephrotic Syndrome of the Finnish Type (NPHS1) and Characterization of Mutations. American Journal of Human Genetics, 1999, 64, 51-61.	2.6	346

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55	Clinical and Genetic Features of Familial Nephrotic Syndromes. Pediatric Research, 1999, 45, 332A-332A.	1.1	0
56	A novel gene encoding an integral membrane protein is mutated in nephropathic cystinosis. Nature Genetics, 1998, 18, 319-324.	9.4	562
57	Presymptomatic diagnosis of familial steroid-resistant riephrotic syndrome. Lancet, The, 1996, 347, 1050-1051.	6.3	8
58	Congenital nephrotic syndrome of the Finnish type: linkage to the locus in a non-Finnish population. Pediatric Nephrology, 1996, 10, 135-138.	0.9	38
59	Mapping a gene (SRN1) to chromosome 1q25-q31 in idiopathic nephrotic syndrome confirms a distinct entity of autosomal recessive nephrosis. Human Molecular Genetics, 1995, 4, 2155-2158.	1.4	114