Corinne Antignac

List of Publications by Year
in descending order

47 papers	5,073 citations	109321 35 h-index	223800 46 g-index
49 all docs	49 docs citations	49 times ranked	6213 citing authors

#	Article	IF	Citations
1	Agonists of prostaglandin E ₂ receptors as potential first in class treatment for nephronophthisis and related ciliopathies. Proceedings of the National Academy of Sciences of the United States of America, 2022, 119, e2115960119.	7.1	13
2	An international cohort study spanning five decades assessed outcomes of nephropathic cystinosis. Kidney International, 2021, 100, 1112-1123.	5.2	31
3	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
4	Renal tubular dysgenesis and microcolon, a novel association. Report of three cases. European Journal of Medical Genetics, 2019, 62, 254-258.	1.3	0
5	Defects in t6A tRNA modification due to GON7 and YRDC mutations lead to Galloway-Mowat syndrome. Nature Communications, 2019, 10, 3967.	12.8	66
6	TBC1D8B Loss-of-Function Mutations Lead to X-Linked Nephrotic Syndrome via Defective Trafficking Pathways. American Journal of Human Genetics, 2019, 104, 348-355.	6.2	40
7	Interaction between galectin-3 and cystinosin uncovers a pathogenic role of inflammation inÂkidney involvement of cystinosis. Kidney International, 2019, 96, 350-362.	5.2	23
8	Human C-terminal CUBN variants associate with chronic proteinuria and normal renal function. Journal of Clinical Investigation, 2019, 130, 335-344.	8.2	54
9	Endoplasmic reticulumâ€“retained podocin mutants are massively degraded by the proteasome. Journal of Biological Chemistry, 2018, 293, 4122-4133.	3.4	16
10	Abolishment of proximal tubule albumin endocytosis does not affect plasma albumin duringÂnephrotic syndrome in mice. Kidney International, 2018, 93, 335-342.	5.2	35
11	Mutations in multiple components of the nuclear pore complex cause nephrotic syndrome. Journal of Clinical Investigation, 2018, 128, 4313-4328.	8.2	89
12	Identification of genetic causes for sporadic steroid-resistant nephrotic syndrome in adults. Kidney International, 2018, 94, 1013-1022.	5.2	51
13	A homozygous KAT2B variant modulates the clinical phenotype of ADD3 deficiency in humans and flies. PLoS Genetics, 2018, 14, e1007386.	3.5	17
14	QMPSF is sensitive and specific in the detection of NPHP1 heterozygous deletions. Clinical Chemistry and Laboratory Medicine, 2017, 55, 809-816.	2.3	7
15	Mutations in KEOPS-complex genes cause nephrotic syndrome with primary microcephaly. Nature Genetics, 2017, 49, 1529-1538.	21.4	164
16	Mutations in sphingosine-1-phosphate lyase cause nephrosis with ichthyosis and adrenal insufficiency. Journal of Clinical Investigation, 2017, 127, 912-928.	8.2	160
17	MP031THE MUTATION-DEPENDENT PATHOGENICITY OF THE NPHS2 R229Q VARIANT. Nephrology Dialysis Transplantation, 2016, 31, i353-i353.	0.7	0
18	Cystinosin is a Component of the Vacuolar H+-ATPase-Ragulator-Rag Complex Controlling Mammalian Target of Rapamycin Complex 1 Signaling. Journal of the American Society of Nephrology: JASN, 2016, 27, 1678-1688.	6.1	71

#	Article	IF	Citations
19	ADCK4-Associated Glomerulopathy Causes Adolescence-Onset FSGS. Journal of the American Society of Nephrology: JASN, 2016, 27, 63-68.	6.1	79
20	Novel NEK8 Mutations Cause Severe Syndromic Renal Cystic Dysplasia through YAP Dysregulation. PLoS Genetics, 2016, 12, e1005894.	3.5	77
21	Evidence of digenic inheritance in Alport syndrome. Journal of Medical Genetics, 2015, 52, 163-174.	3.2	129
22	Mutations in TRAF3IP1/IFT54 reveal a new role for IFT proteins in microtubule stabilization. Nature Communications, 2015, 6, 8666.	12.8	84
23	Clinical utility gene card for: Cystinosis. European Journal of Human Genetics, 2014, 22, 713-713.	2.8	27
24	Renal fibrosis is the common feature of autosomal dominant tubulointerstitial kidney diseases caused by mutations in mucin 1 or uromodulin. Kidney International, 2014, 86, 589-599.	5.2	86
25	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.	6.2	108
26	A Homozygous Missense Mutation in the Ciliary Gene TTC21B Causes Familial FSGS. Journal of the American Society of Nephrology: JASN, 2014, 25, 2435-2443.	6.1	86
27	Mutation-dependent recessive inheritance of NPHS2-associated steroid-resistant nephrotic syndrome. Nature Genetics, 2014, 46, 299-304.	21.4	134
28	Nephropathic cystinosis: an international consensus document. Nephrology Dialysis Transplantation, 2014, 29, iv87-iv94.	0.7	164
29	Initial Steroid Sensitivity in Children with Steroid-Resistant Nephrotic Syndrome Predicts Post-Transplant Recurrence. Journal of the American Society of Nephrology: JASN, 2014, 25, 1342-1348.	6.1	93
30	Mutations of CEP83 Cause Infantile Nephronophthisis and Intellectual Disability. American Journal of Human Genetics, 2014, 94, 905-914.	6.2	90
31	ADCK4 mutations promote steroid-resistant nephrotic syndrome through CoQ10 biosynthesis disruption. Journal of Clinical Investigation, 2013, 123, 5179-5189.	8.2	275
32	ARHGDIA mutations cause nephrotic syndrome via defective RHO GTPase signaling. Journal of Clinical Investigation, 2013, 123, 3243-3253.	8.2	196
33	Exome Capture Reveals ZNF423 and CEP164 Mutations, Linking Renal Ciliopathies to DNA Damage Response Signaling. Cell, 2012, 150, 533-548.	28.9	347
34	Studying nonobstructive azoospermia in cystinosis: histologicÂexamination of testes andÂepididymis and sperm analysis inÂa Ctnsâ¯/â¯ mouse model. Fertility and Sterility, 2012, 98, 162-165.	1.0	2
35	INF2Mutations in Charcotâ€“Marieâ€“Tooth Disease with Glomerulopathy. New England Journal of Medicine, 2011, 365, 2377-2388.	27.0	235
36	Control of the Wnt pathways by nephrocystin-4 is required for morphogenesis of the zebrafish pronephros. Human Molecular Genetics, 2011, 20, 2611-2627.	2.9	53

#	Article	IF	Citations
37	Renal phenotype of the cystinosis mouse model is dependent upon genetic background. Nephrology Dialysis Transplantation, 2010, 25, 1059-1066.	0.7	82
38	Dominant Renin Gene Mutations Associated with Early-Onset Hyperuricemia, Anemia, and Chronic Kidney Failure. American Journal of Human Genetics, 2009, 85, 204-213.	6.2	146
39	Genetics of nephrotic syndrome: connecting molecular genetics to podocyte physiology. Human Molecular Genetics, 2009, 18, R185-R194.	2.9	108
40	Nephrocystin-1 and nephrocystin-4 are required for epithelial morphogenesis and associate with PALS1/PATJ and Par6. Human Molecular Genetics, 2009, 18, 4711-4723.	2.9	94
41	Mutations in genes in the renin-angiotensin system are associated with autosomal recessive renal tubular dysgenesis. Nature Genetics, 2005, 37, 964-968.	21.4	244
42	Intralysosomal Cystine Accumulation in Mice Lacking Cystinosin, the Protein Defective in Cystinosis. Molecular and Cellular Biology, 2002, 22, 7622-7632.	2.3	151
43	Genetic models: clues for understanding the pathogenesis of idiopathic nephrotic syndrome. Journal of Clinical Investigation, 2002, 109, 447-449.	8.2	29
44	Mutations in theCOL4A4 and COL4A3 Genes Cause Familial Benign Hematuria. Journal of the American Society of Nephrology: JASN, 2002, 13, 1248-1254.	6.1	106
45	Characterization of the NPHP1 Locus: Mutational Mechanism Involved in Deletions in Familial Juvenile Nephronophthisis. American Journal of Human Genetics, 2000, 66, 778-789.	6.2	125
46	Mutations in the Chloride Channel Gene CLCNKB as a Cause of Classic Bartter Syndrome. Journal of the American Society of Nephrology: JASN, 2000, 11, 1449-1459.	6.1	255
47	A novel gene encoding an integral membrane protein is mutated in nephropathic cystinosis. Nature Genetics, 1998, 18, 319-324.	21.4	562