

# Bertrand Knebelmann

## List of Publications by Year in descending order

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Version: 2024-02-01

65  
papers

5,272  
citations

94269

37  
h-index

98622

67  
g-index

70  
all docs

70  
docs citations

70  
times ranked

5284  
citing authors

#	ARTICLE	IF	CITATIONS
1	X-linked Alport Syndrome. Journal of the American Society of Nephrology: JASN, 2000, 11, 649-657.	3.0	455
2	X-Linked Alport Syndrome: Natural History and Genotype-Phenotype Correlations in Girls and Women Belonging to 195 Families: A "European Community Alport Syndrome Concerted Action" Study. Journal of the American Society of Nephrology: JASN, 2003, 14, 2603-2610.	3.0	394
3	Lipocalin 2 is essential for chronic kidney disease progression in mice and humans. Journal of Clinical Investigation, 2010, 120, 4065-4076.	3.9	310
4	Early angiotensin-converting enzyme inhibition in Alport syndrome delays renal failure and improves life expectancy. Kidney International, 2012, 81, 494-501.	2.6	275
5	Spectrum of HNF1B Mutations in a Large Cohort of Patients Who Harbor Renal Diseases. Clinical Journal of the American Society of Nephrology: CJASN, 2010, 5, 1079-1090.	2.2	236
6	Autosomal dominant polycystic kidney disease: the changing face of clinical management. Lancet, The, 2015, 385, 1993-2002.	6.3	227
7	Recommendations for the use of tolvaptan in autosomal dominant polycystic kidney disease: a position statement on behalf of the ERA-EDTA Working Groups on Inherited Kidney Disorders and European Renal Best Practice. Nephrology Dialysis Transplantation, 2016, 31, 337-348.	0.4	206
8	Cyst Infections in Patients with Autosomal Dominant Polycystic Kidney Disease. Clinical Journal of the American Society of Nephrology: CJASN, 2009, 4, 1183-1189.	2.2	186
9	Autosomal recessive Alport syndrome: Immunohistochemical study of type IV collagen chain distribution. Kidney International, 1995, 47, 1142-1147.	2.6	155
10	Rituximab in Severe Lupus Nephritis. Clinical Journal of the American Society of Nephrology: CJASN, 2009, 4, 579-587.	2.2	151
11	Improving Mutation Screening in Familial Hematuric Nephropathies through Next Generation Sequencing. Journal of the American Society of Nephrology: JASN, 2014, 25, 2740-2751.	3.0	130
12	Evidence of digenic inheritance in Alport syndrome. Journal of Medical Genetics, 2015, 52, 163-174.	1.5	129
13	Effect of High-Cutoff Hemodialysis vs Conventional Hemodialysis on Hemodialysis Independence Among Patients With Myeloma Cast Nephropathy. JAMA - Journal of the American Medical Association, 2017, 318, 2099.	3.8	120
14	Clinical practice recommendations for the treatment of Alport syndrome: a statement of the Alport Syndrome Research Collaborative. Pediatric Nephrology, 2013, 28, 5-11.	0.9	118
15	Splice-mediated insertion of an Alu sequence in the COL4A3 mRNA causing autosomal recessive Alport syndrome. Human Molecular Genetics, 1995, 4, 675-679.	1.4	114
16	Phenotype and Genotype Characterization of Adenine Phosphoribosyltransferase Deficiency. Journal of the American Society of Nephrology: JASN, 2010, 21, 679-688.	3.0	112
17	Phenotype and Outcome in Hereditary Tubulointerstitial Nephritis Secondary to UMOD Mutations. Clinical Journal of the American Society of Nephrology: CJASN, 2011, 6, 2429-2438.	2.2	109
18	Bortezomib produces high hematological response rates with prolonged renal survival in monoclonal immunoglobulin deposition disease. Kidney International, 2015, 88, 1135-1143.	2.6	104

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19	Clinical and Genetic Spectrum of Bartter Syndrome Type 3. <i>Journal of the American Society of Nephrology: JASN</i> , 2017, 28, 2540-2552.	3.0	92
20	Alport syndrome and diffuse leiomyomatosis: Deletions in the 5' end of the COL4A5 collagen gene. <i>Kidney International</i> , 1992, 42, 1178-1183.	2.6	91
21	Endoplasmic reticulum stress drives proteinuria-induced kidney lesions via Lipocalin 2. <i>Nature Communications</i> , 2016, 7, 10330.	5.8	88
22	Adenine Phosphoribosyltransferase Deficiency. <i>Clinical Journal of the American Society of Nephrology: CJASN</i> , 2012, 7, 1521-1527.	2.2	87
23	Randall-type monoclonal immunoglobulin deposition disease: novel insights from a nationwide cohort study. <i>Blood</i> , 2019, 133, 576-587.	0.6	78
24	Multiplex epithelium dysfunction due to CLDN10 mutation: the HELIX syndrome. <i>Genetics in Medicine</i> , 2018, 20, 190-201.	1.1	75
25	CKD and Its Risk Factors among Patients with Cystinuria. <i>Clinical Journal of the American Society of Nephrology: CJASN</i> , 2015, 10, 842-851.	2.2	71
26	Observations of a large Dent disease cohort. <i>Kidney International</i> , 2016, 90, 430-439.	2.6	71
27	Mutation Update of the <i>CLCN5</i> Gene Responsible for Dent Disease 1. <i>Human Mutation</i> , 2015, 36, 743-752.	1.1	66
28	Cystine crystal volume determination: a useful tool in the management of cystinuric patients. <i>Urological Research</i> , 2003, 31, 207-211.	1.5	58
29	$\alpha$ 24 Integrin and Laminin 5 Are Aberrantly Expressed in Polycystic Kidney Disease. <i>American Journal of Pathology</i> , 2003, 163, 1791-1800.	1.9	58
30	Rapamycin inhibits human renal epithelial cell proliferation: Effect on cyclin D3 mRNA expression and stability. <i>Kidney International</i> , 2005, 67, 2422-2433.	2.6	58
31	Cystinuria: clinical practice recommendation. <i>Kidney International</i> , 2021, 99, 48-58.	2.6	58
32	Screening for intracranial aneurysms in autosomal dominant polycystic kidney disease is cost-effective. <i>Kidney International</i> , 2018, 93, 716-726.	2.6	46
33	Defects in KCNJ16 Cause a Novel Tubulopathy with Hypokalemia, Salt Wasting, Disturbed Acid-Base Homeostasis, and Sensorineural Deafness. <i>Journal of the American Society of Nephrology: JASN</i> , 2021, 32, 1498-1512.	3.0	46
34	Thalidomide in patients with multiple myeloma and renal failure. <i>British Journal of Haematology</i> , 2004, 125, 96-97.	1.2	45
35	mTOR inhibitors may benefit kidney transplant recipients with mitochondrial diseases. <i>Kidney International</i> , 2019, 95, 455-466.	2.6	44
36	The clinicopathologic characteristics of kidney diseases related to monotypic IgA deposits. <i>Kidney International</i> , 2017, 91, 720-728.	2.6	43

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37	Clinicopathologic predictors of renal outcomes in light chain cast nephropathy: a multicenter retrospective study. <i>Blood</i> , 2020, 135, 1833-1846.	0.6	42
38	Laminin 5 Regulates Polycystic Kidney Cell Proliferation and Cyst Formation. <i>Journal of Biological Chemistry</i> , 2006, 281, 29181-29189.	1.6	40
39	A New Workflow for Proteomic Analysis of Urinary Exosomes and Assessment in Cystinuria Patients. <i>Journal of Proteome Research</i> , 2015, 14, 567-577.	1.8	39
40	Aberrant splicing of the COL4A5 gene in patients with Alport syndrome. <i>Human Molecular Genetics</i> , 1994, 3, 317-322.	1.4	37
41	Impaired formation of desmosomal junctions in ADPKD epithelia. <i>Histochemistry and Cell Biology</i> , 2005, 124, 487-497.	0.8	35
42	Study Design and Baseline Characteristics of the CARDINAL Trial: A Phase 3 Study of Bardoxolone Methyl in Patients with Alport Syndrome. <i>American Journal of Nephrology</i> , 2021, 52, 180-189.	1.4	31
43	Adverse events associated with currently used medical treatments for cystinuria and treatment goals: results from a series of 442 patients in France. <i>BJU International</i> , 2019, 124, 849-861.	1.3	30
44	An international cohort study of autosomal dominant tubulointerstitial kidney disease due to mutations identifies distinct clinical subtypes. <i>Kidney International</i> , 2020, 98, 1589-1604.	2.6	27
45	Challenging the traditional approach for interpreting genetic variants: Lessons from Fabry disease. <i>Clinical Genetics</i> , 2022, 101, 390-402.	1.0	26
46	Complete Remission of Lupus Nephritis With Rituximab and Steroids for Induction and Rituximab Alone for Maintenance Therapy. <i>American Journal of Kidney Diseases</i> , 2008, 52, 346-352.	2.1	25
47	Clinical and molecular characterization of cystinuria in a French cohort: relevance of assessing large-scale rearrangements and splicing variants. <i>Molecular Genetics &amp; Genomic Medicine</i> , 2017, 5, 373-389.	0.6	22
48	A molecular approach to inherited kidney disorders. <i>Kidney International</i> , 1993, 44, 1205-1216.	2.6	21
49	Screening for Unruptured Intracranial Aneurysms in Autosomal Dominant Polycystic Kidney Disease: A Survey of 420 Nephrologists. <i>PLoS ONE</i> , 2016, 11, e0153176.	1.1	17
50	COVID-19 outbreak in vaccinated patients from a haemodialysis unit: antibody titres as a marker of protection from infection. <i>Nephrology Dialysis Transplantation</i> , 2022, 37, 1357-1365.	0.4	17
51	2,8-Dihydroxyadenine Urolithiasis: A Not So Rare Inborn Error of Purine Metabolism. <i>Nucleosides, Nucleotides and Nucleic Acids</i> , 2014, 33, 241-252.	0.4	16
52	Mesangial IgG Glomerulonephritis. <i>Journal of the American Society of Nephrology: JASN</i> , 2002, 13, 379-387.	3.0	15
53	Light and heavy chain deposition disease associated with CH1 deletion. <i>CKJ: Clinical Kidney Journal</i> , 2015, 8, 237-239.	1.4	12
54	APOL1 risk genotype in European steroid-resistant nephrotic syndrome and/or focal segmental glomerulosclerosis patients of different African ancestries. <i>Nephrology Dialysis Transplantation</i> , 2019, 34, 1885-1893.	0.4	12

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55	The 2019 and 2021 International Workshops on Alport Syndrome. <i>European Journal of Human Genetics</i> , 2022, 30, 507-516.	1.4	12
56	Red Blood Cell AE1/Band 3 Transports in Dominant Distal Renal Tubular Acidosis Patients. <i>Kidney International Reports</i> , 2020, 5, 348-357.	0.4	11
57	Kidney Transplant Outcomes in Patients With Adenine Phosphoribosyltransferase Deficiency. <i>Transplantation</i> , 2020, 104, 2120-2128.	0.5	10
58	Reversible paraparesis in multiple myeloma with renal failure. <i>Nephrology Dialysis Transplantation</i> , 2006, 21, 1439-1440.	0.4	4
59	Enzyme therapy for Fabry's disease: registered for success?. <i>Lancet, The</i> , 2009, 374, 1950-1951.	6.3	3
60	Establishing a core outcome measure for pain in patients with autosomal dominant polycystic kidney disease: a consensus workshop report. <i>CKJ: Clinical Kidney Journal</i> , 2022, 15, 407-416.	1.4	3
61	Erythrocytosis associated with IgA nephropathy. <i>EBioMedicine</i> , 2022, 75, 103785.	2.7	2
62	AA amyloidosis associated with Fabry disease. <i>International Journal of Clinical Practice</i> , 2020, 74, e13577.	0.8	1
63	FC 0104-WEEK ANTIBIOTIC THERAPY PREVENTS RECURRENT RENAL CYST INFECTIONS IN AUTOSOMAL DOMINANT POLYCYSTIC KIDNEY DISEASE. <i>Nephrology Dialysis Transplantation</i> , 2021, 36, .	0.4	0
64	Genetic Diagnosis and Counseling in Inherited Renal Diseases. , 1998, , 685-694.		0
65	Efficacy of Prolonged Antibiotic Therapy for Renal Cyst Infections in Polycystic Kidney Disease. <i>Mayo Clinic Proceedings</i> , 2022, 97, 1305-1317.	1.4	0