Roser Torra

List of Publications by Year
in descending order

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111	6,957	76196	62479
papers	citations	40 h-index	80 g-index

119	119	119	5712
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Unified Criteria for Ultrasonographic Diagnosis of ADPKD. Journal of the American Society of Nephrology: JASN, 2009, 20, 205-212.	3.0	590
2	Comparison of phenotypes of polycystic kidney disease types 1 and 2. Lancet, The, 1999, 353, 103-107.	6.3	547
3	Treatment of Fabryâ€™s Disease with the Pharmacologic Chaperone Migalastat. New England Journal of Medicine, 2016, 375, 545-555.	13.9	390
4	Cellular and subcellular localization of the ARPKD protein; fibrocystin is expressed on primary cilia. Human Molecular Genetics, 2003, 12, 2703-2710.	1.4	287
5	Early angiotensin-converting enzyme inhibition in Alport syndrome delays renal failure and improves life expectancy. Kidney International, 2012, 81, 494-501.	2.6	275
6	Chronic kidney disease is a key risk factor for severe COVID-19: a call to action by the ERA-EDTA. Nephrology Dialysis Transplantation, 2021, 36, 87-94.	0.4	259
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	Clinical Utility of Genetic Testing in Children and Adults with Steroid-Resistant Nephrotic Syndrome. Clinical Journal of the American Society of Nephrology: CJASN, 2011, 6, 1139-1148.	2.2	189
9	Coordinate Expression of the Autosomal Dominant Polycystic Kidney Disease Proteins, Polycystin-2 And Polycystin-1, in Normal and Cystic Tissue. American Journal of Pathology, 1999, 154, 1721-1729.	1.9	174
10	Genotype-Renal Function Correlation in Type 2 Autosomal Dominant Polycystic Kidney Disease. Journal of the American Society of Nephrology: JASN, 2003, 14, 1164-1174.	3.0	129
11	Incompletely Penetrant PKD1 Alleles Mimic the Renal Manifestations of ARPKD. Journal of the American Society of Nephrology: JASN, 2010, 21, 1097-1102.	3.0	126
12	A complete mutation screen of PKHD1 in autosomal-recessive polycystic kidney disease (ARPKD) pedigrees. Kidney International, 2003, 64, 391-403.	2.6	113
13	Incidence of renal failure and nephroprotection by RAAS inhibition in heterozygous carriers of X-chromosomal and autosomal recessive Alport mutations. Kidney International, 2012, 81, 779-783.	2.6	113
14	Nephrin mutations cause childhood- and adult-onset focal segmental glomerulosclerosis. Kidney International, 2009, 76, 1268-1276.	2.6	111
15	A kidney-disease gene panel allows a comprehensive genetic diagnosis of cystic andÂglomerular inherited kidney diseases. Kidney International, 2018, 94, 363-371.	2.6	109
16	Mutations in theCOL4A4 and COL4A3 Genes Cause Familial Benign Hematuria. Journal of the American Society of Nephrology: JASN, 2002, 13, 1248-1254.	3.0	106
17	TRPC6 mutational analysis in a large cohort of patients with focal segmental glomerulosclerosis. Nephrology Dialysis Transplantation, 2009, 24, 3089-3096.	0.4	99
18	Expert consensus guidelines for the genetic diagnosis of Alport syndrome. Pediatric Nephrology, 2019, 34, 1175-1189.	0.9	97

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19	Autosomal Dominant Polycystic Kidney Disease Types 1 and 2: Assessment of US Sensitivity for Diagnosis. Radiology, 1999, 213, 273-276.	3.6	89
20	International consensus statement on the diagnosis and management of autosomal dominant polycystic kidney disease in children and young people. Nature Reviews Nephrology, 2019, 15, 713-726.	4.1	86
21	X-Linked and Autosomal Recessive Alport Syndrome: Pathogenic Variant Features and Further Genotype-Phenotype Correlations. PLoS ONE, 2016, 11, e0161802.	1.1	75
22	Clinical and genetic spectra of autosomal dominant tubulointerstitial kidney disease due to mutationsÂin UMOD and MUC1. Kidney International, 2020, 98, 717-731.	2.6	75
23	Targeted next-generation sequencing in steroid-resistant nephrotic syndrome: mutations in multiple glomerular genes may influence disease severity. European Journal of Human Genetics, 2015, 23, 1192-1199.	1.4	72
24	Location of mutations within the PKD2 gene influences clinical outcome. Kidney International, 2000, 57, 1444-1451.	2.6	70
25	Imaging of Kidney Cysts and Cystic Kidney Diseases in Children: An International Working Group Consensus Statement. Radiology, 2019, 290, 769-782.	3.6	69
26	Diagnosis of autosomal dominant polycystic kidney disease using efficient PKD1 and PKD2 targeted nextâ€generation sequencing. Molecular Genetics & Genomic Medicine, 2014, 2, 412-421.	0.6	67
27	Clinical Value ofNPHS2Analysis in Early- and Adult-Onset Steroid-Resistant Nephrotic Syndrome. Clinical Journal of the American Society of Nephrology: CJASN, 2011, 6, 344-354.	2.2	65
28	Autosomal dominant polycystic kidney disease with anticipation and Caroli's disease associated with a PKD1 mutation Rapid Communication. Kidney International, 1997, 52, 33-38.	2.6	59
29	Genetic Variation of DKK3 May Modify Renal Disease Severity in ADPKD. Journal of the American Society of Nephrology: JASN, 2010, 21, 1510-1520.	3.0	59
30	Influence of the ACE gene polymorphism in the progression of renal failure in autosomal dominant polycystic kidney disease. American Journal of Kidney Diseases, 1999, 34, 273-278.	2.1	57
31	Prevalence of Cysts in Seminal Tract and Abnormal Semen Parameters in Patients with Autosomal Dominant Polycystic Kidney Disease. Clinical Journal of the American Society of Nephrology: CJASN, 2008, 3, 790-793.	2.2	57
32	Spanish guidelines for the management of autosomal dominant polycystic kidney disease. Nephrology Dialysis Transplantation, 2014, 29, iv95-iv105.	0.4	56
33	A Loss-of-Function Model for Cystogenesis in Human Autosomal Dominant Polycystic Kidney Disease Type 2. American Journal of Human Genetics, 1999, 65, 345-352.	2.6	51
34	A Review of the Imaging Techniques for Measuring Kidney and Cyst Volume in Establishing Autosomal Dominant Polycystic Kidney Disease Progression. American Journal of Nephrology, 2018, 48, 67-78.	1.4	51
35	Abdominal sonographic study of autosomal dominant polycystic kidney disease. Journal of Clinical Ultrasound, 2000, 28, 277-282.	0.4	50
36	Increased prevalence of polycystic kidney disease type 2 among elderly polycystic patients. American Journal of Kidney Diseases, 2000, 36, 728-734.	2.1	48

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37	Clinical and Genetic Features of Autosomal Dominant Alport Syndrome: A Cohort Study. American Journal of Kidney Diseases, 2021, 78, 560-570.e1.	2.1	48
38	Autosomal recessive Alportâ€™s syndrome and benign familial hematuria are collagen type IV diseases. American Journal of Kidney Diseases, 2003, 42, 952-959.	2.1	47
39	Genetics in chronic kidney disease: conclusions from a Kidney Disease: Improving Global Outcomes (KDIGO) Controversies Conference. Kidney International, 2022, 101, 1126-1141.	2.6	46
40	Are Sodium Transporters in Urinary Exosomes Reliable Markers of Tubular Sodium Reabsorption in Hypertensive Patients?. Nephron Physiology, 2010, 114, p25-p34.	1.5	45
41	Clinical utility of genetic testing in early-onset kidney disease: seven genes are the main players. Nephrology Dialysis Transplantation, 2022, 37, 687-696.	0.4	44
42	An update on the use of tolvaptan for autosomal dominant polycystic kidney disease: consensus statement on behalf of the ERA Working Group on Inherited Kidney Disorders, the European Rare Kidney Disease Reference Network and Polycystic Kidney Disease International. Nephrology Dialysis Transplantation, 2022, 37, 825-839.	0.4	44
43	Ciclosporin-induced hypertension is associated with increased sodium transporter of the loop of Henle (NKCC2). Nephrology Dialysis Transplantation, 2007, 22, 2810-2816.	0.4	42
44	Autosomal Dominant Tubulointerstitial Kidney Disease: Clinical Presentation of Patients With ADTKD-UMOD and ADTKD-MUC1. American Journal of Kidney Diseases, 2018, 72, 411-418.	2.1	42
45	Mutational analysis within the 3â€² region of the PKD1 gene. Kidney International, 1999, 55, 1225-1233.	2.6	41
46	Haplotype analysis improves molecular diagnostics of autosomal recessive polycystic kidney disease. American Journal of Kidney Diseases, 2005, 45, 77-87.	2.1	41
47	Assessing the effectiveness of rapamycin on angiomyolipoma in tuberous sclerosis: a two years trial. Orphanet Journal of Rare Diseases, 2012, 7, 87.	1.2	41
48	Stem cell therapy for Alport syndrome: the hope beyond the hype. Nephrology Dialysis Transplantation, 2008, 24, 731-734.	0.4	40
49	Advances and unmet needs in genetic, basic and clinical science in Alport syndrome: report from the 2015 International Workshop on Alport Syndrome. Nephrology Dialysis Transplantation, 2017, 32, gfw095.	0.4	40
50	Collagen type IV (Â3-Â4) nephropathy: from isolated haematuria to renal failure. Nephrology Dialysis Transplantation, 2004, 19, 2429-2432.	0.4	39
51	New therapeutic options for Alport syndrome. Nephrology Dialysis Transplantation, 2019, 34, 1272-1279.	0.4	37
52	Genetic kidney diseases as an underrecognized cause of chronic kidney disease: the key role of international registry reports. CKJ: Clinical Kidney Journal, 2021, 14, 1879-1885.	1.4	36
53	Fabry Nephropathy: An Evidence-Based Narrative Review. Kidney and Blood Pressure Research, 2018, 43, 406-421.	0.9	35
54	Sonographic pattern of recessive polycystic kidney disease in young adults. Differences from the dominant form. Nephrology Dialysis Transplantation, 2000, 15, 1373-1378.	0.4	34

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55	Renal replacement therapy in ADPKD patients: a 25-year survey based on the Catalan registry. BMC Nephrology, 2013, 14, 186.	0.8	33
56	Loss of heterozygosity in renal and hepatic epithelial cystic cells from ADPKD1 patients. European Journal of Human Genetics, 2000, 8, 487-492.	1.4	31
57	Study Design and Baseline Characteristics of the CARDINAL Trial: A Phase 3 Study of Bardoxolone Methyl in Patients with Alport Syndrome. American Journal of Nephrology, 2021, 52, 180-189.	1.4	31
58	Renal manifestations in Fabry disease and therapeutic options. Kidney International, 2008, 74, S29-S32.	2.6	30
59	Seven novel mutations of the PKD2 gene in families with autosomal dominant polycystic kidney disease. Kidney International, 1999, 56, 28-33.	2.6	29
60	Genetic Testing for X-Linked Alport Syndrome by Direct Sequencing of COL4A5 cDNA From Hair Root RNA Samples. American Journal of Kidney Diseases, 2007, 50, 257.e1-257.e14.	2.1	27
61	Contribution of theTTC21Bgene to glomerular and cystic kidney diseases. Nephrology Dialysis Transplantation, 2017, 32, gfv453.	0.4	26
62	Study of candidate genes affecting the progression of renal disease in autosomal dominant polycystic kidney disease type 1. Nephrology Dialysis Transplantation, 2007, 22, 1567-1577.	0.4	25
63	Recommendations for imaging-based diagnosis and management of renal angiomyolipoma associated with tuberous sclerosis complex. CKJ: Clinical Kidney Journal, 2017, 10, 728-737.	1.4	25
64	DNA variant databases improve test accuracy and phenotype prediction in Alport syndrome. Pediatric Nephrology, 2014, 29, 971-977.	0.9	22
65	Male-to-male transmission of X-linked Alport syndrome in a boy with a 47,XXY karyotype. European Journal of Human Genetics, 2005, 13, 1040-1046.	1.4	21
66	Establishing a Core Outcome Set for Autosomal Dominant Polycystic Kidney Disease: Report of the Standardized Outcomes in Nephrologyâ€“Polycystic Kidney Disease (SONG-PKD) Consensus Workshop. American Journal of Kidney Diseases, 2021, 77, 255-263.	2.1	21
67	Very Low-Molecular-Mass Fragments of Albumin in the Plasma of Patients With Focal Segmental Glomerulosclerosis. American Journal of Kidney Diseases, 2009, 54, 871-880.	2.1	20
68	MYH9-related disease: it does exist, may be more frequent than you think and requires specific therapy. CKJ: Clinical Kidney Journal, 2019, 12, 488-493.	1.4	20
69	Analysis of published PKD1 gene sequence variants. Nature Genetics, 2007, 39, 427-428.	9.4	19
70	Hypertension in autosomal-dominant polycystic kidney disease (ADPKD). CKJ: Clinical Kidney Journal, 2013, 6, 457-463.	1.4	17
71	Rare diseases, rare presentations: recognizing atypical inherited kidney disease phenotypes in the age of genomics. CKJ: Clinical Kidney Journal, 2017, 10, 586-593.	1.4	17
72	Recurrence of the PKD1 nonsense mutation Q4041X in Spanish, Italian, and British families. Human Mutation, 1998, 11, S117-S120.	1.1	16

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73	RevisiÃ³n de la nefropatÃa tubulointersticial autosÃ³mica dominante. Nefrologia, 2017, 37, 235-243.	0.2	16
74	Novel homozygous OSGEP gene pathogenic variants in two unrelated patients with Galloway-Mowat syndrome: case report and review of the literature. BMC Nephrology, 2019, 20, 126.	0.8	16
75	Insight into response to mTOR inhibition when PKD1 and TSC2 are mutated. BMC Medical Genetics, 2015, 16, 39.	2.1	15
76	Autosomal Dominant Polycystic Kidney Disease: Clinical Assessment of Rapid Progression. American Journal of Nephrology, 2018, 48, 308-317.	1.4	15
77	Cost-effective PKHD1 genetic testing for autosomal recessive polycystic kidney disease. Pediatric Nephrology, 2014, 29, 223-234.	0.9	14
78	Long-term follow-up of renal function in patients treated with migalastat for Fabry disease. Molecular Genetics and Metabolism Reports, 2021, 28, 100786.	0.4	14
79	Generation of integration-free induced pluripotent stem cell lines derived from two patients with X-linked Alport syndrome (XLAS). Stem Cell Research, 2017, 25, 291-295.	0.3	13
80	Sangrado de angiomiolipoma renal en paciente con sÃndrome de genes contiguos (TSC2/PKD1) tras 17 aÃ±os de tratamiento renal sustitutivo. Nefrologia, 2017, 37, 87-92.	0.2	12
81	Podocyturia: why it may have added value in rare diseases. CKJ: Clinical Kidney Journal, 2019, 12, 49-52.	1.4	12
82	The 2019 and 2021 International Workshops on Alport Syndrome. European Journal of Human Genetics, 2022, 30, 507-516.	1.4	12
83	Building a network of ADPKD reference centres across Europe: the EuroCYST initiative. Nephrology Dialysis Transplantation, 2014, 29, iv26-iv32.	0.4	11
84	Everolimus safety and efficacy for renal angiomyolipomas associated with tuberous sclerosis complex: a Spanish expanded access trial. Orphanet Journal of Rare Diseases, 2016, 11, 128.	1.2	11
85	Renal angiomyolipoma bleeding in a patient with TSC2/PKD1 contiguous gene syndrome after 17 years of renal replacement therapy. Nefrologia, 2017, 37, 87-92.	0.2	10
86	NefropatÃa asociada a mutaciÃ³n del gen MYH9. Nefrologia, 2019, 39, 133-140.	0.2	9
87	Integration-free induced pluripotent stem cells derived from a patient with autosomal recessive Alport syndrome (ARAS). Stem Cell Research, 2017, 25, 1-5.	0.3	8
88	Clinical and genetic characterization of a cohort of proteinuric patients with biallelic CUBN variants. Nephrology Dialysis Transplantation, 2022, 37, 1906-1915.	0.4	8
89	Clinical trial recommendations for potential Alport syndrome therapies. Kidney International, 2020, 97, 1109-1116.	2.6	7
90	A review on autosomal dominant tubulointerstitial kidney disease. Nefrologia, 2017, 37, 235-243.	0.2	6

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91	MYH9 Associated nephropathy. Nefrologia, 2019, 39, 133-140.	0.2	6
92	Do clinical guidelines facilitate or impede drivers of treatment in Fabry disease?. Orphanet Journal of Rare Diseases, 2022, 17, 42.	1.2	6
93	Can ketogenic dietary interventions slow disease progression in ADPKD: what we know and what we don't. CKJ: Clinical Kidney Journal, 2022, 15, 1034-1036.	1.4	6
94	Clinical profile of women diagnosed with Fabry disease non receiving enzyme replacement therapy. Medicina ClÃnica, 2019, 153, 47-55.	0.3	5
95	Recent advances in the clinical management of autosomal dominant polycystic kidney disease. F1000Research, 2019, 8, 116.	0.8	5
96	Comparative analysis of tools to predict rapid progression in autosomal dominant polycystic kidney disease. CKJ: Clinical Kidney Journal, 2022, 15, 912-921.	1.4	5
97	Recomendaciones de manejo de la afectaciÃ³n renal en el complejo esclerosis tuberosa. Nefrologia, 2020, 40, 142-151.	0.2	4
98	How genomics reclassifies diseases: the case of Alport syndrome. CKJ: Clinical Kidney Journal, 2020, 13, 933-935.	1.4	4
99	Cardiovascular risk factors and the impact on prognosis in patients with chronic kidney disease secondary to autosomal dominant polycystic kidney disease. BMC Nephrology, 2021, 22, 110.	0.8	4
100	UGA hopping: a sport for nephrologists too?. Nephrology Dialysis Transplantation, 2010, 25, 2391-2395.	0.4	3
101	Autosomal dominant polycystic kidney disease: possibly the least silent cause of chronic kidney disease. CKJ: Clinical Kidney Journal, 2021, 14, 2281-2284.	1.4	3
102	Cystatin C estimated glomerular filtration rate to assess renal function in early stages of autosomal dominant polycystic kidney disease. PLoS ONE, 2017, 12, e0174583.	1.1	3
103	Flank pain has a significant adverse impact on quality of life in ADPKD: the CYSTic-QoL study. CKJ: Clinical Kidney Journal, 2022, 15, 2063-2071.	1.4	3
104	Recommendations for the multidisciplinary management of tuberous sclerosis complex. Medicina ClÃnica (English Edition), 2016, 147, 211-216.	0.1	2
105	Recommendations for the management of renal involvement in tuberous sclerosis complex. Nefrologia, 2020, 40, 142-151.	0.2	2
106	Response to â€˜Is standard GLA gene mutation analysis definitive for the diagnosis of Fabry disease?â€™. Kidney International, 2009, 75, 1116.	2.6	0
107	Fabry disease in untreated women with enzyme replacement therapy: Symptomatology and clinical profile. Molecular Genetics and Metabolism, 2015, 114, S18-S19.	0.5	0
108	TO037DEFINING RAPID DISEASE PROGRESSION IN A SPANISH ADPKD COHORT. Nephrology Dialysis Transplantation, 2017, 32, iii95-iii95.	0.4	0

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109	MO070CLINICAL AND GENETIC FEATURES IN A LARGE SPANISH COHORT WITH HETEROZYGOUS MUTATIONS IN COL4A3-COL4A4 GENES. Nephrology Dialysis Transplantation, 2017, 32, iii76-iii76.	0.4	0
110	Foreword. CKJ: Clinical Kidney Journal, 2018, 11, i1-i1.	1.4	0
111	Clinical profile of women diagnosed with Fabry disease non receiving enzyme replacement therapy. Medicina ClÃnica (English Edition), 2019, 153, 47-55.	0.1	0