Rosa Vargas-Poussou

List of Publications by Year
in descending order

79	3,323	159585 30	149698 56
papers	citations	h-index	g-index

84	84	84	2565
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Functional Characterization of a Calcium-Sensing Receptor Mutation in Severe Autosomal Dominant Hypocalcemia with a Bartter-Like Syndrome. Journal of the American Society of Nephrology: JASN, 2002, 13, 2259-2266.	6.1	309
2	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
3	Gitelman syndrome: consensus and guidance from a Kidney Disease: Improving Global Outcomes (KDIGO) Controversies Conference. Kidney International, 2017, 91, 24-33.	5.2	230
4	Spectrum of Mutations in Gitelman Syndrome. Journal of the American Society of Nephrology: JASN, 2011, 22, 693-703.	6.1	190
5	Novel Molecular Variants of the Na-K-2Cl Cotransporter Gene Are Responsible for Antenatal Bartter Syndrome. American Journal of Human Genetics, 1998, 62, 1332-1340.	6.2	146
6	Phenotypeâ€“genotype correlation in antenatal and neonatal variants ofÂBartter syndrome. Nephrology Dialysis Transplantation, 2009, 24, 1455-1464.	0.7	137
7	Genetic Investigation of Autosomal Recessive Distal Renal Tubular Acidosis. Journal of the American Society of Nephrology: JASN, 2006, 17, 1437-1443.	6.1	119
8	Familial Hypocalciuric Hypercalcemia Types 1 and 3 and Primary Hyperparathyroidism: Similarities and Differences. Journal of Clinical Endocrinology and Metabolism, 2016, 101, 2185-2195.	3.6	97
9	Clinical and Genetic Spectrum of Bartter Syndrome Type 3. Journal of the American Society of Nephrology: JASN, 2017, 28, 2540-2552.	6.1	92
10	Familial Hypomagnesemia with Hypercalciuria and Nephrocalcinosis. Clinical Journal of the American Society of Nephrology: CJASN, 2012, 7, 801-809.	4.5	82
11	Simultaneous sequencing of 37 genes identified causative mutations in the majority of children with renal tubulopathies. Kidney International, 2018, 93, 961-967.	5.2	77
12	Multiplex epithelium dysfunction due to CLDN10 mutation: the HELIX syndrome. Genetics in Medicine, 2018, 20, 190-201.	2.4	75
13	Treatment and long-term outcome in primary distal renal tubular acidosis. Nephrology Dialysis Transplantation, 2019, 34, 981-991.	0.7	75
14	Observations of a large Dent disease cohort. Kidney International, 2016, 90, 430-439.	5.2	71
15	Indomethacin, Amiloride, or Eplerenone for Treating Hypokalemia in Gitelman Syndrome. Journal of the American Society of Nephrology: JASN, 2015, 26, 468-475.	6.1	69
16	Mutation Update of theCLCN5Gene Responsible for Dent Disease 1. Human Mutation, 2015, 36, 743-752.	2.5	66
17	Genetic testing in the diagnosis of chronic kidney disease: recommendations for clinical practice. Nephrology Dialysis Transplantation, 2022, 37, 239-254.	0.7	63
18	A mouse model for distal renal tubular acidosis reveals a previously unrecognized role of the Vâ€ATPase a4 subunit in the proximal tubule. EMBO Molecular Medicine, 2012, 4, 1057-1071.	6.9	58

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19	Diagnosis and management of Bartter syndrome: executive summary of the consensus and recommendations from the European Rare Kidney Disease Reference Network Working Group for Tubular Disorders. Kidney International, 2021, 99, 324-335.	5.2	53
20	ClC-K chloride channels: emerging pathophysiology of Bartter syndrome type 3. American Journal of Physiology - Renal Physiology, 2015, 308, F1324-F1334.	2.7	52
21	Claudin-16 Deficiency Impairs Tight Junction Function in Ameloblasts, Leading to Abnormal Enamel Formation. Journal of Bone and Mineral Research, 2016, 31, 498-513.	2.8	50
22	Effect of Hydrochlorothiazide on Urinary Calcium Excretion in Dent Disease: An Uncontrolled Trial. American Journal of Kidney Diseases, 2008, 52, 1084-1095.	1.9	48
23	Defects in KCNJ16 Cause a Novel Tubulopathy with Hypokalemia, Salt Wasting, Disturbed Acid-Base Homeostasis, and Sensorineural Deafness. Journal of the American Society of Nephrology: JASN, 2021, 32, 1498-1512.	6.1	46
24	Amelogenesis imperfecta in familial hypomagnesaemia and hypercalciuria with nephrocalcinosis caused byCLDN19gene mutations. Journal of Medical Genetics, 2017, 54, 26-37.	3.2	45
25	Prevalence of Novel MAGED2 Mutations in Antenatal Bartter Syndrome. Clinical Journal of the American Society of Nephrology: CJASN, 2018, 13, 242-250.	4.5	45
26	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
27	Novel CLCN5 mutations in patients with Dentâ€™s disease result in altered ion currents or impaired exchanger processing. Kidney International, 2009, 76, 999-1005.	5.2	36
28	Resistance to Insulin in Patients with Gitelman Syndrome and a Subtle Intermediate Phenotype in Heterozygous Carriers: A Cross-Sectional Study. Journal of the American Society of Nephrology: JASN, 2019, 30, 1534-1545.	6.1	36
29	High-throughput sequencing contributes to the diagnosis of tubulopathies and familial hypercalcemia hypocalciuria in adults. Kidney International, 2019, 96, 1408-1416.	5.2	36
30	Genome-Wide Meta-Analysis Unravels Interactions between Magnesium Homeostasis and Metabolic Phenotypes. Journal of the American Society of Nephrology: JASN, 2018, 29, 335-348.	6.1	34
31	Pro-FHH: A Risk Equation to Facilitate the Diagnosis of Parathyroid-Related Hypercalcemia. Journal of Clinical Endocrinology and Metabolism, 2018, 103, 2534-2542.	3.6	34
32	Mutations in the Vasopressin V2 Receptor and Aquaporin-2 Genes in Twelve Families with Congenital Nephrogenic Diabetes Insipidus. Advances in Experimental Medicine and Biology, 1998, 449, 387-390.	1.6	32
33	Mutation affecting the conserved acidic WNK1 motif causes inherited hyperkalemic hyperchloremic acidosis. Journal of Clinical Investigation, 2020, 130, 6379-6394.	8.2	32
34	Bartter Syndrome Prenatal Diagnosis Based on Amniotic Fluid Biochemical Analysis. Pediatric Research, 2010, 67, 300-303.	2.3	29
35	Renal, Ocular, and Neuromuscular Involvements in Patients with CLDN19 Mutations. Clinical Journal of the American Society of Nephrology: CJASN, 2011, 6, 355-360.	4.5	27
36	NovelCLCNKBMutations Causing Bartter Syndrome Affect Channel Surface Expression. Human Mutation, 2013, 34, 1269-1278.	2.5	27

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37	Gitelman-Like Syndrome Caused by Pathogenic Variants in mtDNA. Journal of the American Society of Nephrology: JASN, 2022, 33, 305-325.	6.1	26
38	mTOR-Activating Mutations in RRAGD Are Causative for Kidney Tubulopathy and Cardiomyopathy. Journal of the American Society of Nephrology: JASN, 2021, 32, 2885-2899.	6.1	24
39	CLCNKB mutations causing mild Bartter syndrome profoundly alter the pH and Ca2+ dependence of ClC-Kb channels. Pflugers Archiv European Journal of Physiology, 2014, 466, 1713-1723.	2.8	23
40	Aldosterone-Related Myocardial Extracellular Matrix Expansion in Hypertension in Humans. JACC: Cardiovascular Imaging, 2020, 13, 2149-2159.	5.3	23
41	Functionomics of NCC mutations in Gitelman syndrome using a novel mammalian cell-based activity assay. American Journal of Physiology - Renal Physiology, 2016, 311, F1159-F1167.	2.7	22
42	Evaluating PVALB as a candidate gene for SLC12A3-negative cases of Gitelman's syndrome. Nephrology Dialysis Transplantation, 2008, 23, 3120-3125.	0.7	21
43	A novel CLCN5 pathogenic mutation supports Dent disease with normal endosomal acidification. Human Mutation, 2018, 39, 1139-1149.	2.5	19
44	Prenatal hyperechogenic kidneys in three cases of infantile hypercalcemia associated with SLC34A1 mutations. Pediatric Nephrology, 2018, 33, 1723-1729.	1.7	19
45	Distal renal tubular acidosis: ERKNet/ESPN clinical practice points. Nephrology Dialysis Transplantation, 2021, 36, 1585-1596.	0.7	18
46	Mutations in ATP6V1B1 and ATP6V0A4 genes cause recessive distal renal tubular acidosis in Mexican families. Molecular Genetics & Genomic Medicine, 2016, 4, 303-311.	1.2	17
47	Report of a family with two different hereditary diseases leading to early nephrocalcinosis. Pediatric Nephrology, 2008, 23, 149-153.	1.7	16
48	Localization of Tubular Adaptation to Renal Sodium Loss in Gitelman Syndrome. Clinical Journal of the American Society of Nephrology: CJASN, 2012, 7, 472-478.	4.5	14
49	Clinical characteristics of familial hypocalciuric hypercalcaemia type 1: A multicentre study of 77 adult patients. Clinical Endocrinology, 2020, 93, 248-260.	2.4	14
50	The variety of genetic defects explains the phenotypic heterogeneity of Familial Hyperkalemic Hypertension. Kidney International Reports, 2021, 6, 2639-2652.	0.8	13
51	Clinical utility gene card for: familial hypomagnesemia with hypercalciuria and nephrocalcinosis with/without severe ocular involvement. European Journal of Human Genetics, 2015, 23, 889-889.	2.8	11
52	Red Blood Cell AE1/Band 3 Transports in Dominant Distal Renal Tubular Acidosis Patients. Kidney International Reports, 2020, 5, 348-357.	0.8	11
53	p.Ala541Thr variant of MEN1 gene: A non deleterious polymorphism or a pathogenic mutation?. Annales D'Endocrinologie, 2014, 75, 133-140.	1.4	10
54	A new SLC12A3 founder mutation (p.Val647Met) in Gitelman's syndrome patients of Roma ancestry. Nefrologia, 2017, 37, 423-428.	0.4	9

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55	Pathophysiological aspects of the thick ascending limb and novel genetic defects: HELIX syndrome and transient antenatal Bartter syndrome. Pediatric Nephrology, 2022, 37, 239-252.	1.7	9
56	Renin-aldosterone system evaluation over four decades in an extended family with autosomal dominant pseudohypoaldosteronism due to a deletion in the NR3C2 gene. Journal of Steroid Biochemistry and Molecular Biology, 2020, 204, 105755.	2.5	8
57	Calcium pyrophosphate crystal deposition in a cohort of 57 patients with Gitelman syndrome. Rheumatology, 2022, 61, 2494-2503.	1.9	8
58	The mutation c.1196_1202dup7bp (p.Ser402X) in the SLC12A3 gene clusters in Italian Gitelman syndrome patients and reflects the presence of a common ancestor. Nephrology Dialysis Transplantation, 2011, 26, 557-561.	0.7	7
59	How Bartterâ€™s and Gitelmanâ€™s Syndromes, and Dentâ€™s Disease Have Provided Important Insights into the Function of Three Renal Chloride Channels: ClC-Ka/b and ClC-5. Nephron Physiology, 2006, 103, p7-p13.	1.2	6
60	New insights into the role of endoplasmic reticulumâ€associated degradation in Bartter Syndrome Type 1. Human Mutation, 2021, 42, 947-968.	2.5	6
61	Possible role for rareTRPM7variants in patients with hypomagnesaemia with secondary hypocalcaemia. Nephrology Dialysis Transplantation, 2023, 38, 679-690.	0.7	6
62	A Rare Cause of Chronic Hypokalemia with Metabolic Alkalosis: Case Report and Differential Diagnosis. Children, 2020, 7, 212.	1.5	5
63	Signification of distal urinary acidification defects in hypocitraturic patients. PLoS ONE, 2017, 12, e0177329.	2.5	5
64	Parathyroid hormone and phosphate homeostasis in patients with Bartter and Gitelman syndrome: an international cross-sectional study. Nephrology Dialysis Transplantation, 2022, 37, 2474-2486.	0.7	5
65	Investigation of Vestibular Function in Adult Patients with Gitelman Syndrome: Results of an Observational Study. Journal of Clinical Medicine, 2020, 9, 3790.	2.4	4
66	A pseudo-dominant form of Gitelman's syndrome. CKJ: Clinical Kidney Journal, 2011, 4, 386-389.	2.9	3
67	Prenatal diagnosis of Bartter syndrome: amniotic fluid aldosterone. Prenatal Diagnosis, 2016, 36, 88-91.	2.3	3
68	Fetal urine biochemistry in antenatal Bartter syndrome: a case report. Clinical Case Reports (discontinued), 2016, 4, 876-878.	0.5	3
69	A new SLC12A3 founder mutation (p.Val647Met) in Gitelman's syndrome patients of Roma ancestry. Nefrologia, 2017, 37, 423-428.	0.4	3
70	QT Interval in Adult with Chronic Hypokalemia due to Gitelman Syndrome. Clinical Journal of the American Society of Nephrology: CJASN, 2020, 15, 1640-1642.	4.5	3
71	The Case | Severe hypertension and hyperkalemia in a kidney transplant recipient. Kidney International, 2019, 96, 529-530.	5.2	2
72	When a maternal heterozygous mutation of the CYP24A1 gene leads to infantile hypercalcemia through a maternal uniparental disomy of chromosome 20. Molecular Cytogenetics, 2021, 14, 23.	0.9	2

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73	Recurrent Nephrolithiasis in a Patient With Hypercalcemia and Normal to Mildly Elevated Parathyroid Hormone. American Journal of Kidney Diseases, 2021, 77, A13-A15.	1.9	2
74	Diversity of functional alterations of the ClCâ€5 exchanger in the region of the proton glutamate in patients with Dent disease 1. Human Mutation, 2021, 42, 537-550.	2.5	1
75	Prenatal diagnosis of Bartter syndrome: amniotic fluid aldosterone. Annales De Biologie Clinique, 2017, 75, 204-208.	0.1	1
76	Primary Hyperparathyroidism in Homozygous Sickle Cell Patients: A Hemolysis-Mediated Hypocalciuric Hypercalcemia Phenotype?. Journal of Clinical Medicine, 2021, 10, 5179.	2.4	1
77	Functional Characterization Of Clc-5 Mutations Associated With Dent's Disease. Biophysical Journal, 2009, 96, 270a.	0.5	0
78	Acidosis tubular renal distal en dos niÃ±as diagnosticadas de hipotiroidismo adquirido. Nefrologia, 2018, 38, 655-659.	0.4	0
79	How the diagnosis and the management of genetic renal phosphate leak impact the life of kidney stone formers?. Urolithiasis, 2022, , 1.	2.0	0