

Rosa Vargas-Poussou

List of Publications by Year in descending order

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Version: 2024-02-01

79
papers

3,323
citations

159585

30
h-index

149698

56
g-index

84
all docs

84
docs citations

84
times ranked

2565
citing authors

#	ARTICLE	IF	CITATIONS
1	Possible role for rare <i>TRPM7</i> variants in patients with hypomagnesaemia with secondary hypocalcaemia. <i>Nephrology Dialysis Transplantation</i> , 2023, 38, 679-690.	0.7	6
2	Pathophysiological aspects of the thick ascending limb and novel genetic defects: HELIX syndrome and transient antenatal Bartter syndrome. <i>Pediatric Nephrology</i> , 2022, 37, 239-252.	1.7	9
3	Genetic testing in the diagnosis of chronic kidney disease: recommendations for clinical practice. <i>Nephrology Dialysis Transplantation</i> , 2022, 37, 239-254.	0.7	63
4	Calcium pyrophosphate crystal deposition in a cohort of 57 patients with Gitelman syndrome. <i>Rheumatology</i> , 2022, 61, 2494-2503.	1.9	8
5	Gitelman-Like Syndrome Caused by Pathogenic Variants in mtDNA. <i>Journal of the American Society of Nephrology: JASN</i> , 2022, 33, 305-325.	6.1	26
6	Parathyroid hormone and phosphate homeostasis in patients with Bartter and Gitelman syndrome: an international cross-sectional study. <i>Nephrology Dialysis Transplantation</i> , 2022, 37, 2474-2486.	0.7	5
7	How the diagnosis and the management of genetic renal phosphate leak impact the life of kidney stone formers?. <i>Urolithiasis</i> , 2022, , 1.	2.0	0
8	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. <i>Kidney International</i> , 2021, 99, 324-335.	5.2	53
9	Diversity of functional alterations of the Cl ⁻ /5 exchanger in the region of the proton glutamate in patients with Dent disease 1. <i>Human Mutation</i> , 2021, 42, 537-550.	2.5	1
10	Distal renal tubular acidosis: ERKNet/ESPN clinical practice points. <i>Nephrology Dialysis Transplantation</i> , 2021, 36, 1585-1596.	0.7	18
11	Defects in <i>KCNJ16</i> 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.	6.1	46
12	When a maternal heterozygous mutation of the <i>CYP24A1</i> gene leads to infantile hypercalcemia through a maternal uniparental disomy of chromosome 20. <i>Molecular Cytogenetics</i> , 2021, 14, 23.	0.9	2
13	New insights into the role of endoplasmic reticulum-associated degradation in Bartter Syndrome Type 1. <i>Human Mutation</i> , 2021, 42, 947-968.	2.5	6
14	Recurrent Nephrolithiasis in a Patient With Hypercalcemia and Normal to Mildly Elevated Parathyroid Hormone. <i>American Journal of Kidney Diseases</i> , 2021, 77, A13-A15.	1.9	2
15	The variety of genetic defects explains the phenotypic heterogeneity of Familial Hyperkalemic Hypertension. <i>Kidney International Reports</i> , 2021, 6, 2639-2652.	0.8	13
16	mTOR-Activating Mutations in <i>RRAGD</i> Are Causative for Kidney Tubulopathy and Cardiomyopathy. <i>Journal of the American Society of Nephrology: JASN</i> , 2021, 32, 2885-2899.	6.1	24
17	Primary Hyperparathyroidism in Homozygous Sickle Cell Patients: A Hemolysis-Mediated Hypocalciuric Hypercalcemia Phenotype?. <i>Journal of Clinical Medicine</i> , 2021, 10, 5179.	2.4	1
18	Aldosterone-Related Myocardial Extracellular Matrix Expansion in Hypertension in Humans. <i>JACC: Cardiovascular Imaging</i> , 2020, 13, 2149-2159.	5.3	23

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19	Renin-aldosterone system evaluation over four decades in an extended family with autosomal dominant pseudohypoaldosteronism due to a deletion in the NR3C2 gene. <i>Journal of Steroid Biochemistry and Molecular Biology</i> , 2020, 204, 105755.	2.5	8
20	A Rare Cause of Chronic Hypokalemia with Metabolic Alkalosis: Case Report and Differential Diagnosis. <i>Children</i> , 2020, 7, 212.	1.5	5
21	Investigation of Vestibular Function in Adult Patients with Gitelman Syndrome: Results of an Observational Study. <i>Journal of Clinical Medicine</i> , 2020, 9, 3790.	2.4	4
22	QT Interval in Adult with Chronic Hypokalemia due to Gitelman Syndrome. <i>Clinical Journal of the American Society of Nephrology: CJASN</i> , 2020, 15, 1640-1642.	4.5	3
23	Red Blood Cell AE1/Band 3 Transports in Dominant Distal Renal Tubular Acidosis Patients. <i>Kidney International Reports</i> , 2020, 5, 348-357.	0.8	11
24	Clinical characteristics of familial hypocalciuric hypercalcaemia type 1: A multicentre study of 77 adult patients. <i>Clinical Endocrinology</i> , 2020, 93, 248-260.	2.4	14
25	Mutation affecting the conserved acidic WNK1 motif causes inherited hyperkalemic hyperchloremic acidosis. <i>Journal of Clinical Investigation</i> , 2020, 130, 6379-6394.	8.2	32
26	The Case Severe hypertension and hyperkalemia in a kidney transplant recipient. <i>Kidney International</i> , 2019, 96, 529-530.	5.2	2
27	Resistance to Insulin in Patients with Gitelman Syndrome and a Subtle Intermediate Phenotype in Heterozygous Carriers: A Cross-Sectional Study. <i>Journal of the American Society of Nephrology: JASN</i> , 2019, 30, 1534-1545.	6.1	36
28	High-throughput sequencing contributes to the diagnosis of tubulopathies and familial hypercalcemia hypocalciuria in adults. <i>Kidney International</i> , 2019, 96, 1408-1416.	5.2	36
29	Treatment and long-term outcome in primary distal renal tubular acidosis. <i>Nephrology Dialysis Transplantation</i> , 2019, 34, 981-991.	0.7	75
30	Simultaneous sequencing of 37 genes identified causative mutations in the majority of children with renal tubulopathies. <i>Kidney International</i> , 2018, 93, 961-967.	5.2	77
31	Acidosis tubular renal distal en dos niÃ±as diagnosticadas de hipotiroidismo adquirido. <i>Nefrologia</i> , 2018, 38, 655-659.	0.4	0
32	Genome-Wide Meta-Analysis Unravels Interactions between Magnesium Homeostasis and Metabolic Phenotypes. <i>Journal of the American Society of Nephrology: JASN</i> , 2018, 29, 335-348.	6.1	34
33	Prevalence of Novel MAGED2 Mutations in Antenatal Bartter Syndrome. <i>Clinical Journal of the American Society of Nephrology: CJASN</i> , 2018, 13, 242-250.	4.5	45
34	Multiplex epithelium dysfunction due to CLDN10 mutation: the HELIX syndrome. <i>Genetics in Medicine</i> , 2018, 20, 190-201.	2.4	75
35	A novel <i>CLCN5</i> pathogenic mutation supports Dent disease with normal endosomal acidification. <i>Human Mutation</i> , 2018, 39, 1139-1149.	2.5	19
36	Prenatal hyperechogenic kidneys in three cases of infantile hypercalcemia associated with SLC34A1 mutations. <i>Pediatric Nephrology</i> , 2018, 33, 1723-1729.	1.7	19

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37	Pro-FHH: A Risk Equation to Facilitate the Diagnosis of Parathyroid-Related Hypercalcemia. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2018, 103, 2534-2542.	3.6	34
38	Amelogenesis imperfecta in familial hypomagnesaemia and hypercalciuria with nephrocalcinosis caused by <i>CLDN19</i> gene mutations. <i>Journal of Medical Genetics</i> , 2017, 54, 26-37.	3.2	45
39	A new <i>SLC12A3</i> founder mutation (p.Val647Met) in Gitelman's syndrome patients of Roma ancestry. <i>Nefrologia</i> , 2017, 37, 423-428.	0.4	9
40	Clinical and Genetic Spectrum of Bartter Syndrome Type 3. <i>Journal of the American Society of Nephrology: JASN</i> , 2017, 28, 2540-2552.	6.1	92
41	Gitelman syndrome: consensus and guidance from a Kidney Disease: Improving Global Outcomes (KDIGO) Controversies Conference. <i>Kidney International</i> , 2017, 91, 24-33.	5.2	230
42	Common Elements in Rare Kidney Diseases: Conclusions from a Kidney Disease: Improving Global Outcomes (KDIGO) Controversies Conference. <i>Kidney International</i> , 2017, 92, 796-808.	5.2	40
43	A new <i>SLC12A3</i> founder mutation (p.Val647Met) in Gitelman's syndrome patients of Roma ancestry. <i>Nefrologia</i> , 2017, 37, 423-428.	0.4	3
44	Signification of distal urinary acidification defects in hypocitraturic patients. <i>PLoS ONE</i> , 2017, 12, e0177329.	2.5	5
45	Prenatal diagnosis of Bartter syndrome: amniotic fluid aldosterone. <i>Annales De Biologie Clinique</i> , 2017, 75, 204-208.	0.1	1
46	Claudin-16 Deficiency Impairs Tight Junction Function in Ameloblasts, Leading to Abnormal Enamel Formation. <i>Journal of Bone and Mineral Research</i> , 2016, 31, 498-513.	2.8	50
47	Prenatal diagnosis of Bartter syndrome: amniotic fluid aldosterone. <i>Prenatal Diagnosis</i> , 2016, 36, 88-91.	2.3	3
48	Functionomics of NCC mutations in Gitelman syndrome using a novel mammalian cell-based activity assay. <i>American Journal of Physiology - Renal Physiology</i> , 2016, 311, F1159-F1167.	2.7	22
49	Familial Hypocalciuric Hypercalcemia Types 1 and 3 and Primary Hyperparathyroidism: Similarities and Differences. <i>Journal of Clinical Endocrinology and Metabolism</i> , 2016, 101, 2185-2195.	3.6	97
50	Fetal urine biochemistry in antenatal Bartter syndrome: a case report. <i>Clinical Case Reports (discontinued)</i> , 2016, 4, 876-878.	0.5	3
51	Mutations in <i>ATP6V1B1</i> and <i>ATP6V0A4</i> genes cause recessive distal renal tubular acidosis in Mexican families. <i>Molecular Genetics & Genomic Medicine</i> , 2016, 4, 303-311.	1.2	17
52	Observations of a large Dent disease cohort. <i>Kidney International</i> , 2016, 90, 430-439.	5.2	71
53	ClC-K chloride channels: emerging pathophysiology of Bartter syndrome type 3. <i>American Journal of Physiology - Renal Physiology</i> , 2015, 308, F1324-F1334.	2.7	52
54	Indomethacin, Amiloride, or Eplerenone for Treating Hypokalemia in Gitelman Syndrome. <i>Journal of the American Society of Nephrology: JASN</i> , 2015, 26, 468-475.	6.1	69

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55	Clinical utility gene card for: familial hypomagnesemia with hypercalciuria and nephrocalcinosis with/without severe ocular involvement. <i>European Journal of Human Genetics</i> , 2015, 23, 889-889.	2.8	11
56	Mutation Update of the <i>CLCN5</i> Gene Responsible for Dent Disease 1. <i>Human Mutation</i> , 2015, 36, 743-752.	2.5	66
57	CLCNKB mutations causing mild Bartter syndrome profoundly alter the pH and Ca ²⁺ dependence of ClC-Kb channels. <i>Pflugers Archiv European Journal of Physiology</i> , 2014, 466, 1713-1723.	2.8	23
58	p.Ala541Thr variant of MEN1 gene: A non deleterious polymorphism or a pathogenic mutation?. <i>Annales D'Endocrinologie</i> , 2014, 75, 133-140.	1.4	10
59	Novel <i>CLCNKB</i> Mutations Causing Bartter Syndrome Affect Channel Surface Expression. <i>Human Mutation</i> , 2013, 34, 1269-1278.	2.5	27
60	Localization of Tubular Adaptation to Renal Sodium Loss in Gitelman Syndrome. <i>Clinical Journal of the American Society of Nephrology: CJASN</i> , 2012, 7, 472-478.	4.5	14
61	Familial Hypomagnesemia with Hypercalciuria and Nephrocalcinosis. <i>Clinical Journal of the American Society of Nephrology: CJASN</i> , 2012, 7, 801-809.	4.5	82
62	A mouse model for distal renal tubular acidosis reveals a previously unrecognized role of the V _a ATPase α 4 subunit in the proximal tubule. <i>EMBO Molecular Medicine</i> , 2012, 4, 1057-1071.	6.9	58
63	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. <i>Nephrology Dialysis Transplantation</i> , 2011, 26, 557-561.	0.7	7
64	Renal, Ocular, and Neuromuscular Involvements in Patients with CLDN19 Mutations. <i>Clinical Journal of the American Society of Nephrology: CJASN</i> , 2011, 6, 355-360.	4.5	27
65	Spectrum of Mutations in Gitelman Syndrome. <i>Journal of the American Society of Nephrology: JASN</i> , 2011, 22, 693-703.	6.1	190
66	A pseudo-dominant form of Gitelman's syndrome. <i>CKJ: Clinical Kidney Journal</i> , 2011, 4, 386-389.	2.9	3
67	Bartter Syndrome Prenatal Diagnosis Based on Amniotic Fluid Biochemical Analysis. <i>Pediatric Research</i> , 2010, 67, 300-303.	2.3	29
68	Phenotype-genotype correlation in antenatal and neonatal variants of Bartter syndrome. <i>Nephrology Dialysis Transplantation</i> , 2009, 24, 1455-1464.	0.7	137
69	Novel CLCN5 mutations in patients with Dent's disease result in altered ion currents or impaired exchanger processing. <i>Kidney International</i> , 2009, 76, 999-1005.	5.2	36
70	Functional Characterization Of Clc-5 Mutations Associated With Dent's Disease. <i>Biophysical Journal</i> , 2009, 96, 270a.	0.5	0
71	Report of a family with two different hereditary diseases leading to early nephrocalcinosis. <i>Pediatric Nephrology</i> , 2008, 23, 149-153.	1.7	16
72	Effect of Hydrochlorothiazide on Urinary Calcium Excretion in Dent Disease: An Uncontrolled Trial. <i>American Journal of Kidney Diseases</i> , 2008, 52, 1084-1095.	1.9	48

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73	Evaluating PVALB as a candidate gene for SLC12A3-negative cases of Gitelman's syndrome. <i>Nephrology Dialysis Transplantation</i> , 2008, 23, 3120-3125.	0.7	21
74	How Bartter's and Gitelman's Syndromes, and Dent's Disease Have Provided Important Insights into the Function of Three Renal Chloride Channels: ClC-Ka/b and ClC-5. <i>Nephron Physiology</i> , 2006, 103, p7-p13.	1.2	6
75	Genetic Investigation of Autosomal Recessive Distal Renal Tubular Acidosis. <i>Journal of the American Society of Nephrology: JASN</i> , 2006, 17, 1437-1443.	6.1	119
76	Functional Characterization of a Calcium-Sensing Receptor Mutation in Severe Autosomal Dominant Hypocalcemia with a Bartter-Like Syndrome. <i>Journal of the American Society of Nephrology: JASN</i> , 2002, 13, 2259-2266.	6.1	309
77	Mutations in the Chloride Channel Gene CLCNKB as a Cause of Classic Bartter Syndrome. <i>Journal of the American Society of Nephrology: JASN</i> , 2000, 11, 1449-1459.	6.1	255
78	Novel Molecular Variants of the Na-K-2Cl Cotransporter Gene Are Responsible for Antenatal Bartter Syndrome. <i>American Journal of Human Genetics</i> , 1998, 62, 1332-1340.	6.2	146
79	Mutations in the Vasopressin V2 Receptor and Aquaporin-2 Genes in Twelve Families with Congenital Nephrogenic Diabetes Insipidus. <i>Advances in Experimental Medicine and Biology</i> , 1998, 449, 387-390.	1.6	32