Rosa J Torres

List of Publications by Year
in descending order

98 papers	2,542 citations	236925 25 h-index	214800 47 g-index
108 all docs	108 docs citations	108 times ranked	2148 citing authors

#	Article	IF	Citations
1	Safety and Efficacy of Botulinum Toxin in the Treatment of Self-Biting Behavior in Lesch-Nyhan Disease. Pediatric Neurology, 2022, 127, 6-10.	2.1	1
2	Hereditary spastic paraplegia associated with a novel homozygous intronic noncanonical splice site variant in the AP4B1 gene. Annals of Human Genetics, 2022, 86, 109-118.	0.8	0
3	Symptomatic heterozygous X-Linked myotubular myopathy female patient with a large deletion at Xq28 and decrease expression of normal allele. European Journal of Medical Genetics, 2021, 64, 104170.	1.3	1
4	Genetic and Clinical Predictors of Age of ESKD in Individuals With Autosomal Dominant Tubulointerstitial Kidney Disease Due to UMOD Mutations. Kidney International Reports, 2020, 5, 1472-1485.	0.8	30
5	Physiological levels of folic acid reveal purine alterations in Lesch-Nyhan disease. Proceedings of the National Academy of Sciences of the United States of America, 2020, 117, 12071-12079.	7.1	13
6	Toll-Like receptor 4 (TLR4) polymorphism rs2149356 and risk of gout in a Spanish cohort. Nucleosides, Nucleotides and Nucleic Acids, 2020, 39, 1424-1431.	1.1	5
7	Systematic genetic analysis of early-onset gout: ABCG2 is the only associated locus. Rheumatology, 2020, 59, 2544-2549.	1.9	30
8	Macrocytic anemia in Leschâ€“Nyhan disease and its variants. Genetics in Medicine, 2019, 21, 353-360.	2.4	14
9	Current understanding of Lesch-Nyhan disease and potential therapeutic targets. Expert Opinion on Orphan Drugs, 2019, 7, 349-361.	0.8	4
10	The genetic etiology in cerebral palsy mimics: The results from a Greek tertiary care center. European Journal of Paediatric Neurology, 2019, 23, 427-437.	1.6	26
11	Aicar effect in early neuronal development. Nucleosides, Nucleotides and Nucleic Acids, 2018, 37, 261-272.	1.1	3
12	AutomediciÃ³n en el domicilio frente a lectura de la presiÃ³n arterial en la consulta en el seguimiento de diabÃ©ticos tipo ii : efecto sobre la presiÃ³n arterial ambulatoria y la albuminuria. Estudio aleatorizado. Medicina ClÃ¬nica, 2018, 150, 413-420.	0.6	0
13	<scp>GLUT</scp>9 influences uric acid concentration in patients with Leschâ€Nyhan disease. International Journal of Rheumatic Diseases, 2018, 21, 1270-1276.	1.9	7
14	RÃ©plica a Â«Tratamiento de la hiperuricemia asintomÃ¡ticaÂ». Revista Clinica Espanola, 2017, 217, 244.	0.6	0
15	Skewed X inactivation in Leschâ€“Nyhan disease carrier females. Journal of Human Genetics, 2017, 62, 1079-1083.	2.3	14
16	Unapparent hypoxanthine-guanine phosphoribosyltransferase deficiency. Clinica Chimica Acta, 2017, 472, 136-138.	1.1	5
17	Development of new forms of self-injurious behavior following total dental extraction in Leschâ€“Nyhan disease. Nucleosides, Nucleotides and Nucleic Acids, 2016, 35, 524-528.	1.1	6
18	Ultrasonography in the diagnosis of asymptomatic hyperuricemia and gout. Nucleosides, Nucleotides and Nucleic Acids, 2016, 35, 517-523.	1.1	13

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19	A review of the implication of hypoxanthine excess in the physiopathology of Leschâ€“Nyhan disease. Nucleosides, Nucleotides and Nucleic Acids, 2016, 35, 507-516.	1.1	13
20	A double-blind, placebo-controlled, crossover trial of the selective dopamine D1 receptor antagonist ecopipam in patients with Lesch-Nyhan disease. Molecular Genetics and Metabolism, 2016, 118, 160-166.	1.1	23
21	La ecografÃa en el diagnÃ³stico de la hiperuricemia asintomÃ¡tica y la gota. Revista Clinica Espanola, 2016, 216, 445-450.	0.6	6
22	Arts syndrome with a novel missense mutation in the PRPS1 gene: A case report. Brain and Development, 2016, 38, 954-958.	1.1	15
23	Hypoxanthine deregulates genes involved in early neuronal development. Implications in Leschâ€Nyhan disease pathogenesis. Journal of Inherited Metabolic Disease, 2015, 38, 1109-1118.	3.6	23
24	Consequences of Delayed Dental Extraction in Leschâ€Nyhan Disease. Movement Disorders Clinical Practice, 2014, 1, 225-229.	1.5	19
25	HPRT Deficiency in Spain: What Have We Learned in the Past 30 Years (1984â€“2013)?. Nucleosides, Nucleotides and Nucleic Acids, 2014, 33, 223-232.	1.1	9
26	Expanding the phenotype of PRPS1 syndromes in females: neuropathy, hearing loss and retinopathy. Orphanet Journal of Rare Diseases, 2014, 9, 190.	2.7	31
27	Metabolic Syndrome in Primary Gout. Nucleosides, Nucleotides and Nucleic Acids, 2014, 33, 185-191.	1.1	17
28	Fenofibrate Increases Serum Creatinine in a Patient with Familial Nephropathy Associated to Hyperuricemia. Nucleosides, Nucleotides and Nucleic Acids, 2014, 33, 181-184.	1.1	7
29	Efficacy and Safety of a Urate Lowering Regimen in Primary Gout. Nucleosides, Nucleotides and Nucleic Acids, 2014, 33, 174-180.	1.1	8
30	Genotypeâ€“phenotype correlations in neurogenetics: Lesch-Nyhan disease as a model disorder. Brain, 2014, 137, 1282-1303.	7.6	105
31	Novel Developments in Metabolic Disorders of Purine and Pyrimidine Metabolism and Therapeutic Applications of Their Analogs. Nucleosides, Nucleotides and Nucleic Acids, 2014, 33, 165-173.	1.1	8
32	Tubular Urate Transporter Gene Polymorphisms Differentiate Patients with Gout Who Have Normal and Decreased Urinary Uric Acid Excretion. Journal of Rheumatology, 2014, 41, 1863-1870.	2.0	29
33	De la mutaciÃ³n al fenotipo; variabilidad clÃnica en la enfermedad de Lesch-Nyhan. El papel de la epigenÃ©tica. Revista Clinica Espanola, 2014, 214, 461-465.	0.6	4
34	Enzyme activity and brain anatomy: lessons from HPRT deficiency. Lancet Neurology, The, 2013, 12, 1129-1131.	10.2	2
35	Resolution of Massive Tophaceous Gout with Three Urate-lowering Drugs. American Journal of Medicine, 2013, 126, e9-e10.	1.5	14
36	Clinical utility gene card for: Leschâ€“Nyhan Syndrome - update 2013. European Journal of Human Genetics, 2013, 21, 1187-1187.	2.8	11

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37	Absence of SLC22A12/URAT1 Gene Mutations in Patients with Primary Gout. Journal of Rheumatology, 2012, 39, 1901-1901.	2.0	5
38	Diagnosis of gout in patients with asymptomatic hyperuricaemia: a pilot ultrasound study: Table 1. Annals of the Rheumatic Diseases, 2012, 71, 157-158.	0.9	163
39	Adenosine, dopamine and serotonin receptors imbalance in lymphocytes of Leschâ€Nyhan patients. Journal of Inherited Metabolic Disease, 2012, 35, 1129-1135.	3.6	22
40	Carrier and prenatal diagnosis of Leschâ€“Nyhan disease due to a defect in HPRT gene expression regulation. Gene, 2012, 511, 306-307.	2.2	9
41	Update on the Phenotypic Spectrum of Lesch-Nyhan Disease and its Attenuated Variants. Current Rheumatology Reports, 2012, 14, 189-194.	4.7	61
42	Uric acid excretion in healthy subjects: a nomogram to assess the mechanisms underlying purine metabolic disorders. Metabolism: Clinical and Experimental, 2012, 61, 512-518.	3.4	46
43	Clinical utility gene card for: Leschâ€“Nyhan syndrome. European Journal of Human Genetics, 2011, 19, 2-3.	2.8	7
44	Genome-wide study of familial juvenile hyperuricaemic (gouty) nephropathy (FJHN) indicates a new locus, FJHN3, linked to chromosome 2p22.1-p21. Human Genetics, 2011, 129, 51-58.	3.8	25
45	Mechanisms for phenotypic variation in Leschâ€“Nyhan disease and its variants. Human Genetics, 2011, 129, 71-78.	3.8	21
46	Attenuated variants of Lesch-Nyhan disease. Brain, 2010, 133, 671-689.	7.6	147
47	Metabolic Syndrome Characteristics in Gout Patients. Nucleosides, Nucleotides and Nucleic Acids, 2010, 29, 325-329.	1.1	12
48	Uric Acid Metabolism in Patients with Primary Gout and the Metabolic Syndrome. Nucleosides, Nucleotides and Nucleic Acids, 2010, 29, 330-334.	1.1	18
49	Partial HPRT Deficiency Phenotype and Incomplete Splicing Mutation. Nucleosides, Nucleotides and Nucleic Acids, 2010, 29, 295-300.	1.1	8
50	Methylation Status of HPRT1 Promoter in HPRT Deficiency with Normal Coding Region. Nucleosides, Nucleotides and Nucleic Acids, 2010, 29, 301-305.	1.1	7
51	Lesch-Nyhan syndrome. Drugs of the Future, 2010, 35, 421.	0.1	5
52	Efficacy of Rasburicase in Hyperuricemia Secondary to Lesch-Nyhan Syndrome. American Journal of Kidney Diseases, 2009, 53, 677-680.	1.9	33
53	Abnormal adenosine and dopamine receptor expression in lymphocytes of Leschâ€“Nyhan patients. Brain, Behavior, and Immunity, 2009, 23, 1125-1131.	4.1	17
54	Levodopa therapy in a Leschâ€Nyhan disease patient: Pathological, biochemical, neuroimaging, and therapeutic remarks. Movement Disorders, 2008, 23, 1297-1300.	3.9	17

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55	Metabolic syndrome: prevalence, associated factors, and C-reactive protein. Metabolism: Clinical and Experimental, 2008, 57, 1232-1240.	3.4	43
56	Normal HPRT coding region in complete and partial HPRT deficiency. Molecular Genetics and Metabolism, 2008, 94, 167-172.	1.1	24
57	Serum Urate, Metabolic Syndrome, and Cardiovascular Risk Factors. A Population-Based Study. Nucleosides, Nucleotides and Nucleic Acids, 2008, 27, 620-623.	1.1	29
58	Asymptomatic Hyperuricemia: Impact of Ultrasonography. Nucleosides, Nucleotides and Nucleic Acids, 2008, 27, 592-595.	1.1	127
59	The Diagnosis Of HPRT Deficiency in the 21St Century. Nucleosides, Nucleotides and Nucleic Acids, 2008, 27, 564-569.	1.1	10
60	An Unusual Patient with Hypothyroidism, Tophaceous Gout, and Marked Joint Destruction. Nucleosides, Nucleotides and Nucleic Acids, 2008, 27, 604-607.	1.1	2
61	Urinary Guanidinoacetate and Creatine Levels in Patients with HPRT Deficiency. Nucleosides, Nucleotides and Nucleic Acids, 2008, 27, 575-577.	1.1	0
62	An unusual cause of stuttering. , 2008, , 349-351.		0
63	Hypoxanthine-guanine phosophoribosyltransferase (HPRT) deficiency: Lesch-Nyhan syndrome. Orphanet Journal of Rare Diseases, 2007, 2, 48.	2.7	224
64	Efficacy and safety of allopurinol in patients with hypoxanthine-guanine phosphoribosyltransferase deficiency. Metabolism: Clinical and Experimental, 2007, 56, 1179-1186.	3.4	47
65	Hypoxanthine decreases equilibrative type of adenosine transport in lymphocytes from Leschâ€“Nyhan patients. European Journal of Clinical Investigation, 2007, 37, 905-911.	3.4	15
66	Familial Nephropathy Associated with Hyperuricemia in Spain: Our Experience with 3 Families Harbouring a UMOD Mutation. Nucleosides, Nucleotides and Nucleic Acids, 2006, 25, 1295-1300.	1.1	8
67	Losartan improves resistance artery lesions and prevents CTGF and TGF-Î² production in mild hypertensive patients. Kidney International, 2006, 69, 1237-1244.	5.2	29
68	Delineation of the motor disorder of Leschâ€“Nyhan disease. Brain, 2006, 129, 1201-1217.	7.6	247
69	Hypoxanthine Effect on Equilibrative and Concentrative Adenosine Transport in Human Lymphocytes. Implications in the Phatogenesis of Lesch-Nyhan Syndrome. Nucleosides, Nucleotides and Nucleic Acids, 2006, 25, 1065-1069.	1.1	3
70	Efficacy and Safety of Allopurinol in Patients with the Lesch-Nyhan Syndrome and Partial Hypoxanthine- Phosphoribosyltransferase Deficiency: a Follow-up Study of 18 Spanish Patients. Nucleosides, Nucleotides and Nucleic Acids, 2006, 25, 1077-1082.	1.1	15
71	Adenosine Transport in HPRT Deficient Lymphocytes from Leschâ€Nyhan Disease Patients. Nucleosides, Nucleotides and Nucleic Acids, 2004, 23, 1193-1196.	1.1	4
72	Familial juvenile hyperuricaemic nephropathy. QJM - Monthly Journal of the Association of Physicians, 2004, 97, 457-458.	0.5	5

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73	The Pathophysiology of Hyperuricemia in Essential Hypertension: A Pilot Study. Nucleosides, Nucleotides and Nucleic Acids, 2004, 23, 1197-1199.	1.1	5
74	Hypoxanthine Effects on Cyclic AMP Levels in Human Lymphocytes. Nucleosides, Nucleotides and Nucleic Acids, 2004, 23, 1181-1183.	1.1	4
75	Effects of Hypoxanthine on Adenosine Transport in Human Lymphocytes. Implications in the Phatogenesis of Leschâ€“Nyhan Syndrome. Nucleosides, Nucleotides and Nucleic Acids, 2004, 23, 1177-1179.	1.1	3
76	Adenosine transport in peripheral blood lymphocytes from Lesch-Nyhan patients. Biochemical Journal, 2004, 377, 733-739.	3.7	14
77	Citocinas y marcadores de activaciÃ³n coagulativa en ancianos con fiebre de causa infecciosa. Medicina ClÃnica, 2004, 122, 358-358.	0.6	0
78	Phosphoribosylpyrophosphate synthetase overactivity as a cause of uric acid overproduction in a young woman. Arthritis and Rheumatism, 2003, 48, 2036-2041.	6.7	29
79	Familial juvenile hyperuricaemic nephropathy (FJHN): linkage analysis in 15 families, physical and transcriptional characterisation of the FJHN critical region on chromosome 16p11.2 and the analysis of seven candidate genes. European Journal of Human Genetics, 2003, 11, 145-154.	2.8	25
80	Prevalence of left ventricular hypertrophy in patients with mild hypertension in primary care: impact of echocardiography on cardiovascular risk stratification. American Journal of Hypertension, 2003, 16, 556-563.	2.0	51
81	The trials of alternative medicine. QJM - Monthly Journal of the Association of Physicians, 2003, 96, 172-172.	0.5	2
82	UROMODULIN Mutations Cause Familial Juvenile Hyperuricemic Nephropathy. Journal of Clinical Endocrinology and Metabolism, 2003, 88, 1398-1401.	3.6	99
83	Genetic Mapping Studies of Familial Juvenile Hyperuricemic Nephropathy on Chromosome 16p11-p13. Journal of Clinical Endocrinology and Metabolism, 2003, 88, 464-470.	3.6	26
84	The Spectrum of Hypoxanthine-Guanine Phosphoribosyltransferase (HPRT) Deficiency. Medicine (United) Tj ETQq0 0 0 rgBT /Overlock 10	1.0	108
85	Molecular basis of hypoxanthine-guanine phosphoribosyltransferase deficiency in thirteen Spanish families. Human Mutation, 2000, 15, 383-383.	2.5	35
86	The Spectrum of Hypoxanthine-Guanine Phosphoribosyltransferase (HPRT) Deficiency. , 2000, 486, 15-21.		8
87	Purine metabolism in female heterozygotes for hypoxanthine-guanine phosphoribosyltransferase deficiency. European Journal of Clinical Investigation, 1998, 28, 950-957.	3.4	13
88	Gout New Questions for an Ancient Disease. Advances in Experimental Medicine and Biology, 1998, 431, 1-5.	1.6	3
89	Genetic Diagnosis of Hypoxanthine-Guanine Phosphoribosyltransferase (Hgprt) Carrier Status by Restriction Analysis and Directed Mutagenesis. Advances in Experimental Medicine and Biology, 1998, 431, 201-204.	1.6	3
90	HGPRT and Aprt Activities in Hemolysates During the First Year of Life. Advances in Experimental Medicine and Biology, 1998, 431, 141-145.	1.6	0

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91	Determination of phosphoribosylpyrophosphate synthetase activity in human cells by a non-isotopic, one step method. Clinica Chimica Acta, 1996, 245, 105-112.	1.1	5
92	Purine Metabolism in Female Patients with Primary Gout. Advances in Experimental Medicine and Biology, 1995, 370, 69-72.	1.6	3
93	Determination of the Activity of Recombinant Human Phosphoribosylpyrophosphate Synthetase Isoform 1 by a Non-Isotopic, One-Step Method. Advances in Experimental Medicine and Biology, 1995, 370, 821-824.	1.6	2
94	Purine metabolism in women with primary gout. American Journal of Medicine, 1994, 97, 332-338.	1.5	16
95	A simplified method for the determination of phosphoribosylpyrophosphate synthetase activity in hemolysates. Clinica Chimica Acta, 1994, 224, 55-63.	1.1	12
96	Osseous and Intestinal Compartments in the Humoral Hypercalcemia of Malignancy Associated to Walker 256 Tumor in Rats. Oncology, 1993, 50, 148-152.	1.9	4
97	Binding of serum osteocalcin to hydroxyapatite in Paget's disease of bone. Bone and Mineral, 1991, 14, 55-65.	1.9	4
98	Clinical usefulness of serum tartrate-resistant acid phosphatase in paget's disease of bone: Correlation with other biochemical markers of bone demodelling. Calcified Tissue International, 1991, 49, 14-16.	3.1	25