Virginia Nunes

List of Publications by Year
in descending order

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91 papers	5,855 citations	87888 38 h-index	76900 74 g-index
92 all docs	92 docs citations	92 times ranked	5126 citing authors

#	Article	IF	Citations
1	Treatment of Hepatic Fibrosis in Mice Based on Targeted Plasmonic Hyperthermia. ACS Nano, 2021, 15, 7547-7562.	14.6	25
2	Identification of the GlialCAM interactome: the G protein-coupled receptors GPRC5B and GPR37L1 modulate megalencephalic leukoencephalopathy proteins. Human Molecular Genetics, 2021, 30, 1649-1665.	2.9	12
3	S-Methyl-L-Ergothioneine to L-Ergothioneine Ratio in Urine Is a Marker of Cystine Lithiasis in a Cystinuria Mouse Model. Antioxidants, 2021, 10, 1424.	5.1	4
4	Cerebellar Astrocyte Transduction as Gene Therapy for Megalencephalic Leukoencephalopathy. Neurotherapeutics, 2020, 17, 2041-2053.	4.4	7
5	Choroid plexus LAT2 and SNAT3 as partners in CSF amino acid homeostasis maintenance. Fluids and Barriers of the CNS, 2020, 17, 17.	5.0	15
6	Dysfunctional LAT2 Amino Acid Transporter Is Associated With Cataract in Mouse and Humans. Frontiers in Physiology, 2019, 10, 688.	2.8	28
7	Comparison of zebrafish and mice knockouts for Megalencephalic Leukoencephalopathy proteins indicates that GlialCAM/MLC1 forms a functional unit. Orphanet Journal of Rare Diseases, 2019, 14, 268.	2.7	9
8	Cooperation of Antiporter LAT2/CD98hc with Uniporter TAT1 for Renal Reabsorption of Neutral Amino Acids. Journal of the American Society of Nephrology: JASN, 2018, 29, 1624-1635.	6.1	25
9	Megalencephalic leukoencephalopathy with subcortical cysts: A personal biochemical retrospective. European Journal of Medical Genetics, 2018, 61, 50-60.	1.3	19
10	GlialCAM/MLC1 modulates LRRC8/VRAC currents in an indirect manner: Implications for megalencephalic leukoencephalopathy. Neurobiology of Disease, 2018, 119, 88-99.	4.4	34
11	Mutations in L-type amino acid transporter-2 support SLC7A8 as a novel gene involved in age-related hearing loss. ELife, 2018, 7, .	6.0	38
12	Depolarization causes the formation of a ternary complex between GlialCAM, MLC1 and ClC-2 in astrocytes: implications in megalencephalic leukoencephalopathy. Human Molecular Genetics, 2017, 26, 2436-2450.	2.9	33
13	Monogenic diabetes syndromes: Locus-specific databases for AlstrÃ¶m, Wolfram, and Thiamine-responsive megaloblastic anemia. Human Mutation, 2017, 38, 764-777.	2.5	47
14	Novel cystine transporter in renal proximal tubule identified as a missing partner of cystinuria-related plasma membrane protein rBAT/SLC3A1. Proceedings of the National Academy of Sciences of the United States of America, 2016, 113, 775-780.	7.1	72
15	Digenic Inheritance in Cystinuria Mouse Model. PLoS ONE, 2015, 10, e0137277.	2.5	8
16	Identification of small-molecule inhibitors of calcineurin-NFATc signaling that mimic the PxIxIT motif of calcineurin binding partners. Science Signaling, 2015, 8, ra63.	3.6	22
17	Disrupting MLC1 and GlialCAM and ClC-2 interactions in leukodystrophy entails glial chloride channel dysfunction. Nature Communications, 2014, 5, 3475.	12.8	92
18	Two Novel Mutations in theBCKDK(Branched-Chain Keto-Acid Dehydrogenase Kinase) Gene Are Responsible for a Neurobehavioral Deficit in Two Pediatric Unrelated Patients. Human Mutation, 2014, 35, 470-477.	2.5	70

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19	Megalencephalic leukoencephalopathy with subcortical cysts protein 1 regulates glial surface localization of GLIALCAM from fish to humans. Human Molecular Genetics, 2014, 23, 5069-5086.	2.9	34
20	Cerebral Cortex Hyperthyroidism of Newborn Mct8-Deficient Mice Transiently Suppressed by Lat2 Inactivation. PLoS ONE, 2014, 9, e96915.	2.5	25
21	Genotypic classification of patients with Wolfram syndrome: insights into the natural history of the disease and correlation with phenotype. Genetics in Medicine, 2013, 15, 497-506.	2.4	128
22	EURO-WABB: an EU rare diseases registry for Wolfram syndrome, AlstrÃ¶m syndrome and Bardet-Biedl syndrome. BMC Pediatrics, 2013, 13, 130.	1.7	43
23	Protein kinase CK2-dependent phosphorylation of the human Regulators of Calcineurin reveals a novel mechanism regulating the calcineurinâ€“NFATc signaling pathway. Biochimica Et Biophysica Acta - Molecular Cell Research, 2013, 1833, 2311-2321.	4.1	15
24	Differential cystine and dibasic amino acid handling after loss of function of the amino acid transporter b^{0,+}AT (Slc7a9) in mice. American Journal of Physiology - Renal Physiology, 2013, 305, F1645-F1655.	2.7	13
25	Insights into MLC pathogenesis: GlialCAM is an MLC1 chaperone required for proper activation of volume-regulated anion currents. Human Molecular Genetics, 2013, 22, 4405-4416.	2.9	50
26	Clinical utility gene card for: Cystinuria. European Journal of Human Genetics, 2012, 20, 3-3.	2.8	12
27	GlialCAM, a Protein Defective in a Leukodystrophy, Serves as a ClC-2 Clâ¯ Channel Auxiliary Subunit. Neuron, 2012, 73, 951-961.	8.1	118
28	BootstRatio: A web-based statistical analysis of fold-change in qPCR and RT-qPCR data using resampling methods. Computers in Biology and Medicine, 2012, 42, 438-445.	7.0	56
29	Molecular mechanisms of MLC1 and GLIALCAM mutations in megalencephalic leukoencephalopathy with subcortical cysts. Human Molecular Genetics, 2011, 20, 3266-3277.	2.9	80
30	Mutant GlialCAM Causes Megalencephalic Leukoencephalopathy with Subcortical Cysts, Benign Familial Macrocephaly, and Macrocephaly with Retardation and Autism. American Journal of Human Genetics, 2011, 88, 422-432.	6.2	148
31	Knockdown of MLC1 in primary astrocytes causes cell vacuolation: A MLC disease cell model. Neurobiology of Disease, 2011, 43, 228-238.	4.4	60
32	Metabonomics and population studies: age-related amino acids excretion and inferring networks through the study of urine samples in two Italian isolated populations. Amino Acids, 2010, 38, 65-73.	2.7	18
33	Pathophysiology and treatment of cystinuria. Nature Reviews Nephrology, 2010, 6, 424-434.	9.6	183
34	Novel SLC7A7 large rearrangements in lysinuric protein intolerance patients involving the same AluY repeat. European Journal of Human Genetics, 2009, 17, 71-79.	2.8	29
35	Altered expression of 12S/MTâ€RNR1, MTâ€CO2/COX2, and MTâ€ATP6 mitochondrial genes in prostate cancer. Prostate, 2008, 68, 1086-1096.	2.3	40
36	Genetic and genomic analysis modeling of germline c-MYC overexpression and cancer susceptibility. BMC Genomics, 2008, 9, 12.	2.8	27

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37	Molecular pathogenesis of megalencephalic leukoencephalopathy with subcortical cysts: mutations in MLC1 cause folding defects. Human Molecular Genetics, 2008, 17, 3728-3739.	2.9	60
38	Slc7a9knockout mouse is a good cystinuria model for antilithiasic pharmacological studies. American Journal of Physiology - Renal Physiology, 2007, 293, F732-F740.	2.7	25
39	Aneuploidy of chromosome Y in prostate tumors and seminal vesicles: A possible sign of aging rather than an indicator of carcinogenesis?. Molecular Carcinogenesis, 2007, 46, 543-552.	2.7	14
40	Mitochondrial DNA haplogroups in Spanish patients with hypertrophic cardiomyopathy. International Journal of Cardiology, 2006, 112, 202-206.	1.7	57
41	Identification of somatic and germline mitochondrial DNA sequence variants in prostate cancer patients. Mutation Research - Fundamental and Molecular Mechanisms of Mutagenesis, 2006, 595, 42-51.	1.0	52
42	Tissue Imprints or Primary Cultures. Diagnostic Molecular Pathology, 2005, 14, 243-246.	2.1	2
43	The Genetics of Heteromeric Amino Acid Transporters. Physiology, 2005, 20, 112-124.	3.1	112
44	Expression of mitochondrial genes and transcription estimation in different brain areas in Alzheimer's disease patients. Neurobiology of Disease, 2005, 18, 296-304.	4.4	4
45	Mitochondrial respiratory chain in brain homogenates: activities in different brain areas in patients with Alzheimerâ€™s disease. Aging Clinical and Experimental Research, 2005, 17, 1-7.	2.9	11
46	Thiol Modification of Cysteine 327 in the Eighth Transmembrane Domain of the Light Subunit xCT of the Heteromeric Cystine/Glutamate Antiporter Suggests Close Proximity to the Substrate Binding Site/Permeation Pathway. Journal of Biological Chemistry, 2004, 279, 11214-11221.	3.4	34
47	The amino acid transporter asc-1 is not involved in cystinuria. Kidney International, 2004, 66, 1453-1464.	5.2	25
48	First prenatal diagnosis for Wolfram syndrome by molecular analysis of theWFS1gene. Prenatal Diagnosis, 2004, 24, 787-789.	2.3	17
49	Mitochondrial Effects of Antiretroviral Therapies in Asymptomatic Patients. Antiviral Therapy, 2004, 9, 47-55.	1.0	65
50	Suitability of oligonucleotide-mediated cystic fibrosis gene repair in airway epithelial cells. Journal of Gene Medicine, 2003, 5, 625-639.	2.8	5
51	Slc7a9-deficient mice develop cystinuria non-I and cystine urolithiasis. Human Molecular Genetics, 2003, 12, 2097-2108.	2.9	74
52	Short Communication: Reversible Mitochondrial Respiratory Chain Impairment During Symptomatic Hyperlactatemia Associated with Antiretroviral Therapy. AIDS Research and Human Retroviruses, 2003, 19, 1027-1032.	1.1	19
53	The WFS1 (Wolfram syndrome 1) is not a major susceptibility gene for the development of psychiatric disorders. Psychiatric Genetics, 2003, 13, 29-32.	1.1	13
54	Mitochondrial Dna Depletion and Respiratory Chain Enzyme Deficiencies are Present in Peripheral Blood Mononuclear Cells of HIV-Infected Patients with Haart-Related Lipodystrophy. Antiviral Therapy, 2003, 8, 333-338.	1.0	67

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55	Comparison between SLC3A1 and SLC7A9 Cystinuria Patients and Carriers. Journal of the American Society of Nephrology: JASN, 2002, 13, 2547-2553.	6.1	234
56	Non-viral vector-mediated uptake, distribution, and stability of chimeraplasts in human airway epithelial cells. Journal of Gene Medicine, 2002, 4, 308-322.	2.8	15
57	WFS1 mutations in Spanish patients with diabetes mellitus and deafness. European Journal of Human Genetics, 2002, 10, 421-426.	2.8	35
58	Presence of a Major WFS1 Mutation in Spanish Wolfram Syndrome Pedigrees. Molecular Genetics and Metabolism, 2001, 72, 72-81.	1.1	67
59	Mitochondrial involvement in antiretroviral therapy-related lipodystrophy. Aids, 2001, 15, 1643-1651.	2.2	81
60	Cystinuria type I: Identification of eight new mutations in SLC3A1. Kidney International, 2001, 59, 1250-1256.	5.2	24
61	Is mitochondrial DNA depletion involved in Alzheimer's disease?. European Journal of Human Genetics, 2001, 9, 279-285.	2.8	48
62	ATB0/SLC1A5 gene. Fine localisation and exclusion of association with the intestinal phenotype of cystic fibrosis. European Journal of Human Genetics, 2001, 9, 860-866.	2.8	10
63	Functional analysis of mutations in SLC7A9, and genotypeâ€“phenotype correlation in non-Type I cystinuria. Human Molecular Genetics, 2001, 10, 305-316.	2.9	125
64	Detection of two novel large deletions inSLC3A1 by semi-quantitative fluorescent multiplex PCR. Human Mutation, 2000, 15, 373-379.	2.5	24
65	Respiratory chain dysfunction associated with multiple mitochondrial DNA deletions in antiretroviral therapy-related lipodystrophy. Aids, 2000, 14, 1855-1857.	2.2	31
66	Non-type I cystinuria caused by mutations in SLC7A9, encoding a subunit (bo,+AT) of rBAT. Nature Genetics, 1999, 23, 52-57.	21.4	280
67	Identification of SLC7A7, encoding y+LAT-1, as the lysinuric protein intolerance gene. Nature Genetics, 1999, 21, 293-296.	21.4	286
68	Recombinant Families Locate the Gene for Non-Type I Cystinuria between Markers C13 and D19S587 on Chromosome 19q13.1. Genomics, 1999, 60, 362-365.	2.9	9
69	Paternal origin of a de novo novel CFTR mutation (L1065R) causing cystic fibrosis. Human Mutation, 1998, 11, S99-S102.	2.5	7
70	Biochemical and molecular effects of chronic haloperidol administration on brain and muscle mitochondria of rats. Journal of Neuroscience Research, 1998, 53, 475-481.	2.9	30
71	Reduced steady-state levels of mitochondrial RNA and increased mitochondrial DNA amount in human brain with aging. Molecular Brain Research, 1997, 52, 284-289.	2.3	140
72	Qualitative and Quantitative Changes in Skeletal Muscle mtDNA and Expression of Mitochondrial-Encoded Genes in the Human Aging Process. Biochemical and Molecular Medicine, 1997, 62, 165-171.	1.4	77

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73	High heterogeneity for cystic fibrosis in Spanish families: 75 mutations account for 90% of chromosomes. Human Genetics, 1997, 101, 365-370.	3.8	94
74	Sporadic heteroplasmic single 5.5 Kb mitochondrial DNA deletion associated with cerebellar ataxia, hypogonadotropic hypogonadism, choroidal dystrophy, and mitochondrial respiratory chain complex I deficiency. Human Mutation, 1997, 10, 212-216.	2.5	16
75	Genomic Structure and Organization of the HumanrBATGene (SLC3A1). Genomics, 1996, 37, 249-252.	2.9	22
76	Molecular analysis of the cystinuria disease gene: identification of four new mutations, one large deletion, and one polymorphism. Human Genetics, 1996, 98, 447-451.	3.8	44
77	PRENATAL DIAGNOSIS OF CYSTIC FIBROSIS IN A HIGHLY HETEROGENEOUS POPULATION. , 1996, 16, 215-222.		12
78	The rBAT Gene Is Responsible for L-Cystine Uptake via the bo,+-like Amino Acid Transport System in a â€œRenal Proximal Tubularâ€•Cell Line (OK Cells). Journal of Biological Chemistry, 1996, 271, 10569-10576.	3.4	48
79	Assignment of the gene responsible for cystinuria (rBAT) and of markers D2S119 and D2S177 to 2p16 by fluorescence in situ hybridization. Human Genetics, 1995, 95, 633-6.	3.8	30
80	Mutations in the Cystic Fibrosis Gene in Patients with Congenital Absence of the Vas Deferens. New England Journal of Medicine, 1995, 332, 1475-1480.	27.0	959
81	Analysis of the CFTR gene confirms the high genetic heterogeneity of the Spanish population: 43 mutations account for only 78% of CF chromosomes. Human Genetics, 1994, 93, 447-51.	3.8	65
82	Analysis of the CFTR gene in the Spanish population: SSCP-screening for 60 known mutations and identification of four new mutations (Q30X, A120T, 1812-1 Gâ†'A, and 3667de14). Human Mutation, 1994, 3, 223-230.	2.5	31
83	Cystinuria caused by mutations in rBAT, a gene involved in the transport of cystine. Nature Genetics, 1994, 6, 420-425.	21.4	366
84	Cystic fibrosis in Spain: high frequency of mutation G542X in the Mediterranean coastal area. Human Genetics, 1993, 91, 66-70.	3.8	35
85	Microsatellite haplotypes for cystic fibrosis: mutation frameworks and evolutionary tracers. Human Molecular Genetics, 1993, 2, 1015-1022.	2.9	97
86	Haplotype analysis to determine the position of a mutation among closely linked DNA markers. Human Molecular Genetics, 1993, 2, 1007-1014.	2.9	19
87	Identification of a new missense mutation (P205S) in the first transmembrane domain of the CFTR gene associated with a mild cystic fibrosis phenotype. Human Molecular Genetics, 1993, 2, 1741-1742.	2.9	18
88	Identification of a 31-bp insertion (3860ins31) in exon 20 of the cysticfibrosis (CFTR) gene. Human Molecular Genetics, 1993, 2, 1317-1318.	2.9	7
89	Uniparental inheritance of microsatellite alleles of the cystic fibrosis gene (CFTR): identification of a 50 kilobase deletion. Human Molecular Genetics, 1993, 2, 677-681.	2.9	32
90	Identification of a frameshift mutation (1609delCA) in exon 10 of the CFTR gene in seven Spanish cystic fibrosis patients. Human Mutation, 1992, 1, 75-76.	2.5	12

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91	CAGT Microsatellite alleles within the cystic fibrosis transmembrane conductance regulator (CFTR) gene are not generated by unequal crossingover. Genomics, 1991, 10, 692-698.	2.9	129