Juan M Pascual

List of Publications by Year
in descending order

92 papers	5,625 citations	101543 36 h-index	88630 70 g-index
97 all docs	97 docs citations	97 times ranked	7901 citing authors

#	Article	IF	Citations
1	Mitochondrial disease manifestations in relation to transcriptome location and function. Molecular Genetics and Metabolism, 2022, 135, 82-92.	1.1	3
2	A subset of synaptic transmission events is coupled to acetyl coenzyme A production. Journal of Neurophysiology, 2022, 127, 623-636.	1.8	5
3	Elucidating the Role of Glycogen in Glucose Transporter 1 Deficiency Syndrome. FASEB Journal, 2022, 36, .	0.5	0
4	Metabolismâ€based therapies for epilepsy: new directions for future cures. Annals of Clinical and Translational Neurology, 2021, 8, 1730-1737.	3.7	6
5	Quantification of early learning and movement sub-structure predictive of motor performance. Scientific Reports, 2021, 11, 14405.	3.3	7
6	Development and validation of a LC-MS/MS method for quantitation of 3-hydroxypentanoic acid and 3-oxopentanoic acid in human plasma and its application to a clinical study of glucose transporter type I deficiency (G1D) syndrome. Journal of Pharmaceutical and Biomedical Analysis, 2021, 205, 114335.	2.8	2
7	Large Animal Models of Glioma: Current Status and Future Prospects. Anticancer Research, 2021, 41, 5343-5353.	1.1	18
8	Triheptanoin Mitigates Brain ATP Depletion and Mitochondrial Dysfunction in a Mouse Model of Alzheimerâ€™s Disease. Journal of Alzheimer's Disease, 2020, 78, 425-437.	2.6	8
9	GLUT1 deficiency. Neurology: Genetics, 2020, 6, e472.	1.9	5
10	Assessment of Interlaboratory Variation in the Interpretation of Genomic Test Results in Patients With Epilepsy. JAMA Network Open, 2020, 3, e203812.	5.9	7
11	Glut1 Deficiency Syndrome (Glut1DS): State of the art in 2020 and recommendations of the international Glut1DS study group. Epilepsia Open, 2020, 5, 354-365.	2.4	142
12	Glucose transporter type I deficiency and other glucose flux disorders. , 2020, , 849-863.		0
13	Pyruvate dehydrogenase, pyruvate carboxylase, Krebs cycle, and mitochondrial transport disorders. , 2020, , 427-436.		1
14	Exosomes in disease: Epigenetic signals from the nervous system to the rest of the organism. Neuroscience Letters, 2019, 708, 134293.	2.1	0
15	Functional Assessment of Lipoyltransferase-1 Deficiency in Cells, Mice, and Humans. Cell Reports, 2019, 27, 1376-1386.e6.	6.4	55
16	Brain metabolism modulates neuronal excitability in a mouse model of pyruvate dehydrogenase deficiency. Science Translational Medicine, 2019, 11, .	12.4	53
17	Predicting seizure by modeling synaptic plasticity based on EEG signals - a case study of inherited epilepsy. Communications in Nonlinear Science and Numerical Simulation, 2018, 56, 330-343.	3.3	18
18	Mutations in Disordered Regions Can Cause Disease by Creating Dileucine Motifs. Cell, 2018, 175, 239-253.e17.	28.9	97

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19	Age-dependent changes of cerebral copper metabolism in Atp7b â^'/â^' knockout mouse model of Wilsonâ€™s disease by [64Cu]CuCl2-PET/CT. Metabolic Brain Disease, 2017, 32, 717-726.	2.9	16
20	Clinical Aspects of Glucose Transporter Type 1 Deficiency. JAMA Neurology, 2017, 74, 727.	9.0	35
21	Frontotemporal Degeneration in a Child. Pediatric Neurology, 2017, 72, 62-64.	2.1	0
22	Intramyocellular lipid excess in the mitochondrial disorder MELAS. Neurology: Genetics, 2017, 3, e160.	1.9	9
23	Oxidation of [Uâ€‹sup›13‹/sup›C]glucose in the human brain at 7T under steady state conditions. Magnetic Resonance in Medicine, 2017, 78, 2065-2071.	3.0	25
24	Altered cerebellar connectivity in autism and cerebellar-mediated rescue of autism-related behaviors in mice. Nature Neuroscience, 2017, 20, 1744-1751.	14.8	275
25	The life, times and work of Charles R. Roe, M.D.. Neuroscience Letters, 2017, 637, 1-3.	2.1	0
26	Deregulation of mitochondrial F1FO-ATP synthase via OSCP in Alzheimerâ€™s disease. Nature Communications, 2016, 7, 11483.	12.8	127
27	Genetic Gradients in Epileptic Brain Malformations. JAMA Neurology, 2016, 73, 787.	9.0	2
28	Understanding Atomic Interactions to Achieve Well-being. JAMA Neurology, 2016, 73, 626.	9.0	2
29	The expanding clinical phenotype of Bosch-Boonstra-Schaaf optic atrophy syndrome: 20 new cases and possible genotypeâ€“phenotype correlations. Genetics in Medicine, 2016, 18, 1143-1150.	2.4	64
30	Menkes Disease and Other ATP7A Disorders. , 2015, , 455-462.		1
31	Metabolic plasticity maintains proliferation in pyruvate dehydrogenase deficient cells. Cancer & Metabolism, 2015, 3, 7.	5.0	56
32	Wolman Disease. , 2015, , 403-409.		0
33	Pyruvate Dehydrogenase, Pyruvate Carboxylase, Krebs Cycle and Mitochondrial Transport Disorders. , 2015, , 291-297.		2
34	Acute effect of glucose on cerebral blood flow, blood oxygenation, and oxidative metabolism. Human Brain Mapping, 2015, 36, 707-716.	3.6	24
35	A Protein Kinase C Phosphorylation Motif in GLUT1 Affects Glucose Transport and is Mutated in GLUT1 Deficiency Syndrome. Molecular Cell, 2015, 58, 845-853.	9.7	108
36	IKBKGMutation With Incontinentia Pigmenti and Ring-Enhancing Encephalopathy. JAMA Neurology, 2015, 72, 1533.	9.0	2

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37	Glucose Transporter Type I Deficiency and Other Glucose Flux Disorders. , 2015, , 649-662.		1
38	Glut1 Deficiency (G1D). , 2015, , 785-795.		0
39	Glucose Transporter Type I Deficiency (G1D) at 25 (1990-2015): Presumptions, Facts, and the Lives of Persons With This Rare Disease. Pediatric Neurology, 2015, 53, 379-393.	2.1	33
40	Diagnostic Yield of Clinical Next-Generation Sequencing Panels for Epilepsy. JAMA Neurology, 2014, 71, 650.	9.0	54
41	Triheptanoin for Glucose Transporter Type I Deficiency (G1D). JAMA Neurology, 2014, 71, 1255.	9.0	91
42	Alternating Hemiplegia of Childhood With a de Novo Mutation in ATP1A3 and Changes in SLC2A1 Responsive to a Ketogenic Diet. Pediatric Neurology, 2014, 50, 377-379.	2.1	27
43	A novel de novo KIF21A mutation in a patient with congenital fibrosis of the extraocular muscles and MÃ¶bius syndrome. Molecular Vision, 2014, 20, 368-75.	1.1	6
44	Modeling of Brain Metabolism and Pyruvate Compartmentation Using ¹³C NMR in Vivo: Caution Required. Journal of Cerebral Blood Flow and Metabolism, 2013, 33, 1160-1167.	4.3	24
45	Molecular and clinical characterization of the myopathic form of mitochondrial DNA depletion syndrome caused by mutations in the thymidine kinase (TK2) gene. Molecular Genetics and Metabolism, 2013, 110, 153-161.	1.1	40
46	Heptanoate as a Neural Fuel: Energetic and Neurotransmitter Precursors in Normal and Glucose Transporter I-Deficient (G1D) Brain. Journal of Cerebral Blood Flow and Metabolism, 2013, 33, 175-182.	4.3	83
47	Systemic Metabolic Abnormalities in Adult-onset Acid Maltase Deficiency. JAMA Neurology, 2013, 70, 756.	9.0	136
48	Valproic Acid Enhances Glucose Transport in the Cultured Brain Astrocytes of Glucose Transporter 1 Heterozygous Mice. Journal of Child Neurology, 2013, 28, 70-76.	1.4	9
49	Effect of Hypoxia and Hyperoxia on Cerebral Blood Flow, Blood Oxygenation, and Oxidative Metabolism. Journal of Cerebral Blood Flow and Metabolism, 2012, 32, 1909-1918.	4.3	145
50	Disorders of Muscle Excitability. , 2012, , 783-800.		0
51	Ataxia and Weakness in a Young Woman. Archives of Neurology, 2012, 69, 924.	4.5	2
52	Cortical metabolism in pyruvate dehydrogenase deficiency revealed by ex vivo multiplet 13C NMR of the adult mouse brain. Neurochemistry International, 2012, 61, 1036-1043.	3.8	12
53	Unsuspected stroke signals: From extravascular blood to vessel lumen. Neuroscience Letters, 2012, 514, 1.	2.1	2
54	Analysis of Tumor Metabolism Reveals Mitochondrial Glucose Oxidation in Genetically Diverse Human Glioblastomas in the Mouse Brain InÂVivo. Cell Metabolism, 2012, 15, 827-837.	16.2	459

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55	2-hydroxyglutarate detection by magnetic resonance spectroscopy in IDH-mutated patients with gliomas. Nature Medicine, 2012, 18, 624-629.	30.7	711
56	High-resolution detection of 13C multiplets from the conscious mouse brain by ex vivo NMR spectroscopy. Journal of Neuroscience Methods, 2012, 203, 50-55.	2.5	14
57	Glut1 deficiency (G1D): Epilepsy and metabolic dysfunction in a mouse model of the most common human phenotype. Neurobiology of Disease, 2012, 48, 92-101.	4.4	59
58	Glucose metabolism via the pentose phosphate pathway, glycolysis and Krebs cycle in an orthotopic mouse model of human brain tumors. NMR in Biomedicine, 2012, 25, 1177-1186.	2.8	66
59	Metabolism of [Uâ€¹³C]glucose in human brain tumors in vivo. NMR in Biomedicine, 2012, 25, 1234-1244.	2.8	282
60	Synaptic transporters are deceived if they think themselves free. Neuroscience Letters, 2011, 487, 1-2.	2.1	0
61	Animal models of the human mind: Is there anything like being autistic?. Neuroscience Letters, 2011, 505, 59-60.	2.1	0
62	Measurement of glycine in the human brain in vivo by ¹Hâ€MRS at 3 T: application in brain tumors. Magnetic Resonance in Medicine, 2011, 66, 609-618.	3.0	44
63	Cerebral Folate Deficiency Syndromes in Childhood. Archives of Neurology, 2011, 68, 615-21.	4.5	52
64	Pyruvate carboxylase deficiency: Mechanisms, mimics and anaplerosis. Molecular Genetics and Metabolism, 2010, 101, 9-17.	1.1	242
65	Protean Phenotypic Features of the A3243G Mitochondrial DNA Mutation. Archives of Neurology, 2009, 66, 85-91.	4.5	53
66	Preventing misfolded neuronal protein aggregation by molecular diplomacy. Neuroscience Letters, 2009, 449, 85-86.	2.1	0
67	Childhood Chorea With Cerebral Hypotrophy. Archives of Neurology, 2009, 66, 1410-4.	4.5	31
68	Structural Signatures and Membrane Helix 4 in GLUT1. Journal of Biological Chemistry, 2008, 283, 16732-16742.	3.4	49
69	Functional Studies of the T295M Mutation Causing Glut1 Deficiency: Glucose Efflux Preferentially Affected by T295M. Pediatric Research, 2008, 64, 538-543.	2.3	24
70	Epilepsy in Inherited Metabolic Disorders. Neurologist, 2008, 14, S2-S14.	0.7	31
71	Brain Glucose Supply and the Syndrome of Infantile Neuroglycopenia. Archives of Neurology, 2007, 64, 507.	4.5	49
72	ENCEPHALOPATHIES. , 2007, , 1434-1446.		1

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73	Metabolic Diseases of the Nervous System. , 2007, , 149-161.		3
74	Developmental Neurology: A Molecular Perspective. , 2007, , 145-147.		1
75	Genetic Disorders of Neuromuscular Development. , 2007, , 163-176.		0
76	Clinical Spectrum of Mitochondrial DNA Depletion Due to Mutations in the Thymidine Kinase 2 Gene. Archives of Neurology, 2006, 63, 1122.	4.5	112
77	Nerve Conduction Abnormalities in Patients With MELAS and the A3243G Mutation. Archives of Neurology, 2006, 63, 746.	4.5	47
78	A mouse model for Glut-1 haploinsufficiency. Human Molecular Genetics, 2006, 15, 1169-1179.	2.9	165
79	Glut-1 deficiency syndrome: Clinical, genetic, and therapeutic aspects. Annals of Neurology, 2005, 57, 111-118.	5.3	298
80	GLUT1 deficiency and other glucose transporter diseases. European Journal of Endocrinology, 2004, 150, 627-633.	3.7	125
81	Rectal Biopsy in the Diagnosis of Neuronal Intranuclear Hyaline Inclusion Disease. Journal of Child Neurology, 2004, 19, 59-62.	1.4	147
82	Fever, molecular excitability and epilepsy. Neuroscience Letters, 2004, 368, 1.	2.1	0
83	Functional Studies of Threonine 310 Mutations in Glut1. Journal of Biological Chemistry, 2003, 278, 49015-49021.	3.4	15
84	Changes in Glucose Transport and Water Permeability Resulting from the T310I Pathogenic Mutation in Glut1 Are Consistent with Two Transport Channels per Monomer. Journal of Biological Chemistry, 2002, 277, 30991-30997.	3.4	29
85	Glucose transporter protein syndromes. International Review of Neurobiology, 2002, 51, 259-IN10.	2.0	25
86	Imaging the metabolic footprint of Glut1 deficiency on the brain. Annals of Neurology, 2002, 52, 458-464.	5.3	140
87	Autosomal dominant Glutâ€1 deficiency syndrome and familial epilepsy. Annals of Neurology, 2001, 50, 476-485.	5.3	153
88	The Intrinsic Electrostatic Potential and the Intermediate Ring of Charge in the Acetylcholine Receptor Channel. Journal of General Physiology, 2000, 115, 93-106.	1.9	53
89	Delimiting the Binding Site for Quaternary Ammonium Lidocaine Derivatives in the Acetylcholine Receptor Channel. Journal of General Physiology, 1998, 112, 611-621.	1.9	40
90	State-dependent Accessibility and Electrostatic Potential in the Channel of the Acetylcholine Receptor. Journal of General Physiology, 1998, 111, 717-739.	1.9	110

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91	Contribution of the NH2terminus of Kv2.1 to channel activation. American Journal of Physiology - Cell Physiology, 1997, 273, C1849-C1858.	4.6	28
92	K+ pore structure revealed by reporter cysteines at inner and outer surfaces. Neuron, 1995, 14, 1055-1063.	8.1	110