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	2-hydroxyglutarate detection by magnetic resonance spectroscopy in IDH-mutated patients with gliomas. Nature Medicine, 2012, 18, 624-629.	30.7	711
2	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
3	Glut-1 deficiency syndrome: Clinical, genetic, and therapeutic aspects. Annals of Neurology, 2005, 57, 111-118.	5.3	298
4	Metabolism of [Uâ€¹³C]glucose in human brain tumors in vivo. NMR in Biomedicine, 2012, 25, 1234-1244.	2.8	282
5	Altered cerebellar connectivity in autism and cerebellar-mediated rescue of autism-related behaviors in mice. Nature Neuroscience, 2017, 20, 1744-1751.	14.8	275
6	Pyruvate carboxylase deficiency: Mechanisms, mimics and anaplerosis. Molecular Genetics and Metabolism, 2010, 101, 9-17.	1.1	242
7	A mouse model for Glut-1 haploinsufficiency. Human Molecular Genetics, 2006, 15, 1169-1179.	2.9	165
8	Autosomal dominant Glutâ€1 deficiency syndrome and familial epilepsy. Annals of Neurology, 2001, 50, 476-485.	5.3	153
9	Rectal Biopsy in the Diagnosis of Neuronal Intranuclear Hyaline Inclusion Disease. Journal of Child Neurology, 2004, 19, 59-62.	1.4	147
10	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
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	Imaging the metabolic footprint of Glut1 deficiency on the brain. Annals of Neurology, 2002, 52, 458-464.	5.3	140
13	Systemic Metabolic Abnormalities in Adult-onset Acid Maltase Deficiency. JAMA Neurology, 2013, 70, 756.	9.0	136
14	Deregulation of mitochondrial F1FO-ATP synthase via OSCP in Alzheimerâ€™s disease. Nature Communications, 2016, 7, 11483.	12.8	127
15	GLUT1 deficiency and other glucose transporter diseases. European Journal of Endocrinology, 2004, 150, 627-633.	3.7	125
16	Clinical Spectrum of Mitochondrial DNA Depletion Due to Mutations in the Thymidine Kinase 2 Gene. Archives of Neurology, 2006, 63, 1122.	4.5	112
17	K+ pore structure revealed by reporter cysteines at inner and outer surfaces. Neuron, 1995, 14, 1055-1063.	8.1	110
18	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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19	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
20	Mutations in Disordered Regions Can Cause Disease by Creating Dileucine Motifs. Cell, 2018, 175, 239-253.e17.	28.9	97
21	Triheptanoin for Glucose Transporter Type I Deficiency (G1D). JAMA Neurology, 2014, 71, 1255.	9.0	91
22	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
23	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
24	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
25	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
26	Metabolic plasticity maintains proliferation in pyruvate dehydrogenase deficient cells. Cancer & Metabolism, 2015, 3, 7.	5.0	56
27	Functional Assessment of Lipoyltransferase-1 Deficiency in Cells, Mice, and Humans. Cell Reports, 2019, 27, 1376-1386.e6.	6.4	55
28	Diagnostic Yield of Clinical Next-Generation Sequencing Panels for Epilepsy. JAMA Neurology, 2014, 71, 650.	9.0	54
29	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
30	Protean Phenotypic Features of the A3243G Mitochondrial DNA Mutation. Archives of Neurology, 2009, 66, 85-91.	4.5	53
31	Brain metabolism modulates neuronal excitability in a mouse model of pyruvate dehydrogenase deficiency. Science Translational Medicine, 2019, 11, .	12.4	53
32	Cerebral Folate Deficiency Syndromes in Childhood. Archives of Neurology, 2011, 68, 615-21.	4.5	52
33	Brain Glucose Supply and the Syndrome of Infantile Neuroglycopenia. Archives of Neurology, 2007, 64, 507.	4.5	49
34	Structural Signatures and Membrane Helix 4 in GLUT1. Journal of Biological Chemistry, 2008, 283, 16732-16742.	3.4	49
35	Nerve Conduction Abnormalities in Patients With MELAS and the A3243G Mutation. Archives of Neurology, 2006, 63, 746.	4.5	47
36	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

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37	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
38	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
39	Clinical Aspects of Glucose Transporter Type 1 Deficiency. JAMA Neurology, 2017, 74, 727.	9.0	35
40	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
41	Epilepsy in Inherited Metabolic Disorders. Neurologist, 2008, 14, S2-S14.	0.7	31
42	Childhood Chorea With Cerebral Hypotrophy. Archives of Neurology, 2009, 66, 1410-4.	4.5	31
43	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
44	Contribution of the NH2terminus of Kv2.1 to channel activation. American Journal of Physiology - Cell Physiology, 1997, 273, C1849-C1858.	4.6	28
45	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
46	Glucose transporter protein syndromes. International Review of Neurobiology, 2002, 51, 259-IN10.	2.0	25
47	Oxidation of [Uâ€¹³C]glucose in the human brain at 7T under steady state conditions. Magnetic Resonance in Medicine, 2017, 78, 2065-2071.	3.0	25
48	Functional Studies of the T295M Mutation Causing Glut1 Deficiency: Glucose Efflux Preferentially Affected by T295M. Pediatric Research, 2008, 64, 538-543.	2.3	24
49	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
50	Acute effect of glucose on cerebral blood flow, blood oxygenation, and oxidative metabolism. Human Brain Mapping, 2015, 36, 707-716.	3.6	24
51	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
52	Large Animal Models of Glioma: Current Status and Future Prospects. Anticancer Research, 2021, 41, 5343-5353.	1.1	18
53	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
54	Functional Studies of Threonine 310 Mutations in Glut1. Journal of Biological Chemistry, 2003, 278, 49015-49021.	3.4	15

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55	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
56	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
57	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
58	Intramyocellular lipid excess in the mitochondrial disorder MELAS. Neurology: Genetics, 2017, 3, e160.	1.9	9
59	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
60	Assessment of Interlaboratory Variation in the Interpretation of Genomic Test Results in Patients With Epilepsy. JAMA Network Open, 2020, 3, e203812.	5.9	7
61	Quantification of early learning and movement sub-structure predictive of motor performance. Scientific Reports, 2021, 11, 14405.	3.3	7
62	Metabolismâ€based therapies for epilepsy: new directions for future cures. Annals of Clinical and Translational Neurology, 2021, 8, 1730-1737.	3.7	6
63	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
64	GLUT1 deficiency. Neurology: Genetics, 2020, 6, e472.	1.9	5
65	A subset of synaptic transmission events is coupled to acetyl coenzyme A production. Journal of Neurophysiology, 2022, 127, 623-636.	1.8	5
66	Metabolic Diseases of the Nervous System. , 2007, , 149-161.		3
67	Mitochondrial disease manifestations in relation to transcriptome location and function. Molecular Genetics and Metabolism, 2022, 135, 82-92.	1.1	3
68	Ataxia and Weakness in a Young Woman. Archives of Neurology, 2012, 69, 924.	4.5	2
69	Unsuspected stroke signals: From extravascular blood to vessel lumen. Neuroscience Letters, 2012, 514, 1.	2.1	2
70	Pyruvate Dehydrogenase, Pyruvate Carboxylase, Krebs Cycle and Mitochondrial Transport Disorders. , 2015, , 291-297.		2
71	IKBKGMutation With Incontinentia Pigmenti and Ring-Enhancing Encephalopathy. JAMA Neurology, 2015, 72, 1533.	9.0	2
72	Genetic Gradients in Epileptic Brain Malformations. JAMA Neurology, 2016, 73, 787.	9.0	2

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73	Understanding Atomic Interactions to Achieve Well-being. JAMA Neurology, 2016, 73, 626.	9.0	2
74	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
75	ENCEPHALOPATHIES. , 2007, , 1434-1446.		1
76	Menkes Disease and Other ATP7A Disorders. , 2015, , 455-462.		1
77	Glucose Transporter Type I Deficiency and Other Glucose Flux Disorders. , 2015, , 649-662.		1
78	Developmental Neurology: A Molecular Perspective. , 2007, , 145-147.		1
79	Pyruvate dehydrogenase, pyruvate carboxylase, Krebs cycle, and mitochondrial transport disorders. , 2020, , 427-436.		1
80	Fever, molecular excitability and epilepsy. Neuroscience Letters, 2004, 368, 1.	2.1	0
81	Preventing misfolded neuronal protein aggregation by molecular diplomacy. Neuroscience Letters, 2009, 449, 85-86.	2.1	0
82	Synaptic transporters are deceived if they think themselves free. Neuroscience Letters, 2011, 487, 1-2.	2.1	0
83	Animal models of the human mind: Is there anything like being autistic?. Neuroscience Letters, 2011, 505, 59-60.	2.1	0
84	Disorders of Muscle Excitability. , 2012, , 783-800.		0
85	Wolman Disease. , 2015, , 403-409.		0
86	Glut1 Deficiency (G1D). , 2015, , 785-795.		0
87	Frontotemporal Degeneration in a Child. Pediatric Neurology, 2017, 72, 62-64.	2.1	0
88	The life, times and work of Charles R. Roe, M.D.. Neuroscience Letters, 2017, 637, 1-3.	2.1	0
89	Exosomes in disease: Epigenetic signals from the nervous system to the rest of the organism. Neuroscience Letters, 2019, 708, 134293.	2.1	0
90	Genetic Disorders of Neuromuscular Development. , 2007, , 163-176.		0

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91	Glucose transporter type I deficiency and other glucose flux disorders. , 2020, , 849-863.		0
92	Elucidating the Role of Glycogen in Glucose Transporter 1 Deficiency Syndrome. FASEB Journal, 2022, 36, .	0.5	0