Ã€ngels GarcÃ-a-Cazorla

List of Publications by Year
in descending order

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105 papers	3,134 citations	147801 31 h-index	206112 48 g-index
110 all docs	110 docs citations	110 times ranked	4069 citing authors

#	Article	IF	Citations
1	Joubert syndrome: a model for untangling recessive disorders with extreme genetic heterogeneity. Journal of Medical Genetics, 2015, 52, 514-522.	3.2	219
2	Free-thiamine is a potential biomarker of thiamine transporter-2 deficiency: a treatable cause of Leigh syndrome. Brain, 2016, 139, 31-38.	7.6	174
3	Infantile parkinsonism and gabaergic hypotransmission in a patient with pyruvate carboxylase deficiency. Gene, 2013, 532, 302-306.	2.2	145
4	HPLC with electrochemical and fluorescence detection procedures for the diagnosis of inborn errors of biogenic amines and pterins. Journal of Neuroscience Methods, 2005, 142, 153-158.	2.5	114
5	Inborn Errors of Metabolism Overview. Pediatric Clinics of North America, 2018, 65, 179-208.	1.8	111
6	Development of the serotonergic cells in murine raphe nuclei and their relations with rhombomeric domains. Brain Structure and Function, 2013, 218, 1229-1277.	2.3	101
7	Abnormal Expression of Cerebrospinal Fluid Cation Chloride Cotransporters in Patients with Rett Syndrome. PLoS ONE, 2013, 8, e68851.	2.5	95
8	Pyruvate carboxylase deficiency: Metabolic characteristics and new neurological aspects. Annals of Neurology, 2006, 59, 121-127.	5.3	86
9	MITOCHONDRIAL DISEASES ASSOCIATED WITH CEREBRAL FOLATE DEFICIENCY. Neurology, 2008, 70, 1360-1362.	1.1	77
10	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
11	Secondary coenzyme Q 10 deficiencies in oxidative phosphorylation (OXPHOS) and non-OXPHOS disorders. Mitochondrion, 2016, 30, 51-58.	3.4	70
12	Long-term Follow-up of Neonatal Mitochondrial Cytopathies: A Study of 57 Patients. Pediatrics, 2005, 116, 1170-1177.	2.1	55
13	Pyridoxal 5â€²-phosphate values in cerebrospinal fluid: Reference values and diagnosis of PNPO deficiency in paediatric patients. Molecular Genetics and Metabolism, 2008, 94, 173-177.	1.1	54
14	<scp>l</scp> -Serine dietary supplementation is associated with clinical improvement of loss-of-function GRIN2B -related pediatric encephalopathy. Science Signaling, 2019, 12, .	3.6	53
15	Cerebral Folate Deficiency Syndromes in Childhood. Archives of Neurology, 2011, 68, 615-21.	4.5	52
16	Coenzyme Q10 deficiency in mitochondrial DNA depletion syndromes. Mitochondrion, 2013, 13, 337-341.	3.4	51
17	Phosphomannomutase deficiency (PMM2-CDG): ataxia and cerebellar assessment. Orphanet Journal of Rare Diseases, 2015, 10, 138.	2.7	49
18	Determination of 5-methyltetrahydrofolate in cerebrospinal fluid of paediatric patients: Reference values for a paediatric population. Clinica Chimica Acta, 2006, 371, 159-162.	1.1	48

#	ARTICLE	IF	CITATIONS
19	Mitochondrial diseases mimicking neurotransmitter defects. Mitochondrion, 2008, 8, 273-278.	3.4	48
20	Targeted Next Generation Sequencing in Patients with Inborn Errors of Metabolism. PLoS ONE, 2016, 11, e0156359.	2.5	48
21	Epilepsy and inborn errors of metabolism in children. Journal of Inherited Metabolic Disease, 2009, 32, 609-617.	3.6	45
22	The clinical spectrum of inherited diseases involved in the synthesis and remodeling of complex lipids. A tentative overview. Journal of Inherited Metabolic Disease, 2015, 38, 19-40.	3.6	44
23	Mental retardation and inborn errors of metabolism. Journal of Inherited Metabolic Disease, 2009, 32, 597-608.	3.6	43
24	Progressive ataxia and myoclonic epilepsy in a patient with a homozygous mutation in the FOLR1 gene. Journal of Inherited Metabolic Disease, 2010, 33, 795-802.	3.6	43
25	Clinical presentation and proteomic signature of patients with TANGO2 mutations. Journal of Inherited Metabolic Disease, 2020, 43, 297-308.	3.6	43
26	Long-term evolution of eight Spanish patients with CDG type Ia: Typical and atypical manifestations. European Journal of Paediatric Neurology, 2009, 13, 444-451.	1.6	42
27	Study of inborn errors of metabolism in urine from patients with unexplained mental retardation. Journal of Inherited Metabolic Disease, 2010, 33, 1-7.	3.6	42
28	Inborn errors of metabolism and motor disturbances in children. Journal of Inherited Metabolic Disease, 2009, 32, 618-629.	3.6	41
29	Biochemical diagnosis of dopaminergic disturbances in paediatric patients: Analysis of cerebrospinal fluid homovanillic acid and other biogenic amines. Clinical Biochemistry, 2008, 41, 1306-1315.	1.9	39
30	Follow-up of folinic acid supplementation for patients with cerebral folate deficiency and Kearns-Sayre syndrome. Orphanet Journal of Rare Diseases, 2014, 9, 217.	2.7	39
31	Secondary abnormalities of neurotransmitters in infants with neurological disorders. Developmental Medicine and Child Neurology, 2007, 49, 740-744.	2.1	36
32	Homovanillic acid in cerebrospinal fluid of 1388 children with neurological disorders. Developmental Medicine and Child Neurology, 2013, 55, 559-566.	2.1	36
33	Cerebrospinal fluid alterations of the serotonin product, 5â€hydroxyindolacetic acid, in neurological disorders. Journal of Inherited Metabolic Disease, 2010, 33, 803-809.	3.6	34
34	A homozygous tyrosine hydroxylase gene promoter mutation in a patient with dopa-responsive encephalopathy: Clinical, biochemical and genetic analysis. Molecular Genetics and Metabolism, 2007, 92, 274-277.	1.1	31
35	Protein expression profiles in patients carrying NFU1 mutations. Contribution to the pathophysiology of the disease. Journal of Inherited Metabolic Disease, 2013, 36, 841-847.	3.6	31
36	Evolution of maple syrup urine disease in patients diagnosed by newborn screening versus late diagnosis. European Journal of Paediatric Neurology, 2015, 19, 652-659.	1.6	29

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37	Behavioural and emotional problems, intellectual impairment and healthâ€related quality of life in patients with organic acidurias and urea cycle disorders. Journal of Inherited Metabolic Disease, 2016, 39, 231-241.	3.6	29
38	GRIN database: A unified and manually curated repertoire of GRIN variants. Human Mutation, 2021, 42, 8-18.	2.5	29
39	Neural commitment of human pluripotent stem cells under defined conditions recapitulates neural development and generates patientâ€specific neural cells. Biotechnology Journal, 2015, 10, 1578-1588.	3.5	28
40	Hypokinetic-rigid syndrome in children and inborn errors of metabolism. European Journal of Paediatric Neurology, 2011, 15, 295-302.	1.6	27
41	Clinical, etiological and therapeutic aspects of cerebral folate deficiency. Expert Review of Neurotherapeutics, 2015, 15, 793-802.	2.8	26
42	Impairment of the mitochondrial one-carbon metabolism enzyme SHMT2 causes a novel brain and heart developmental syndrome. Acta Neuropathologica, 2020, 140, 971-975.	7.7	24
43	Diseases of the Synaptic Vesicle: A Potential New Group of Neurometabolic Disorders Affecting Neurotransmission. Seminars in Pediatric Neurology, 2016, 23, 306-320.	2.0	23
44	The utility of Next Generation Sequencing for molecular diagnostics in Rett syndrome. Scientific Reports, 2017, 7, 12288.	3.3	23
45	Analysis of human cerebrospinal fluid monoamines and their cofactors by HPLC. Nature Protocols, 2017, 12, 2359-2366.	12.0	23
46	Tyrosine hydroxylase deficiency in three Greek patients with a common ancestral mutation. Movement Disorders, 2010, 25, 1086-1090.	3.9	22
47	Cerebrospinal Fluid Neopterin Analysis in Neuropediatric Patients: Establishment of a New Cut Off-Value for the Identification of Inflammatory-Immune Mediated Processes. PLoS ONE, 2013, 8, e83237.	2.5	22
48	<scp>DNAJC6</scp> Mutations Disrupt Dopamine Homeostasis in Juvenile <scp>Parkinsonismâ€Dystonia</scp>. Movement Disorders, 2020, 35, 1357-1368.	3.9	22
49	White matter alterations associated with chromosomal disorders. Developmental Medicine and Child Neurology, 2004, 46, 148-153.	2.1	19
50	Cerebrospinal fluid pterins and neurotransmitters in early severe epileptic encephalopathies. Brain and Development, 2008, 30, 106-111.	1.1	19
51	Parkinsonism and inborn errors of metabolism. Journal of Inherited Metabolic Disease, 2014, 37, 627-642.	3.6	19
52	Review and evaluation of the methodological quality of the existing guidelines and recommendations for inherited neurometabolic disorders. Orphanet Journal of Rare Diseases, 2015, 10, 164.	2.7	19
53	Mitochondrial respiratory chain deficiencies expressing the enzymatic deficiency in the hepatic tissue: A study of 31 patients. Journal of Pediatrics, 2006, 149, 401-405.e3.	1.8	18
54	Cellular neurometabolism: a tentative to connect cell biology and metabolism in neurology. Journal of Inherited Metabolic Disease, 2018, 41, 1043-1054.	3.6	18

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55	Levodopa therapy in a Leschâ€Nyhan disease patient: Pathological, biochemical, neuroimaging, and therapeutic remarks. Movement Disorders, 2008, 23, 1297-1300.	3.9	17
56	Severe infantile parkinsonism because of a de novo mutation on DLP1 mitochondrialâ€peroxisomal protein. Movement Disorders, 2017, 32, 1108-1110.	3.9	17
57	Infectious Acute Hemicerebellitis. Journal of Child Neurology, 2004, 19, 390-392.	1.4	16
58	Disease-associated GRIN protein truncating variants trigger NMDA receptor loss-of-function. Human Molecular Genetics, 2021, 29, 3859-3871.	2.9	16
59	Mutation loads in different tissues from six pathogenic mtDNA point mutations. Mitochondrion, 2015, 22, 17-22.	3.4	15
60	Clinical Approach to Inborn Errors of Metabolism in Pediatrics. , 2016, , 3-70.		15
61	Cerebrospinal fluid synaptic proteins as useful biomarkers in tyrosine hydroxylase deficiency. Molecular Genetics and Metabolism, 2015, 114, 34-40.	1.1	14
62	Muscle Involvement in a Large Cohort of Pediatric Patients with Genetic Diagnosis of Mitochondrial Disease. Journal of Clinical Medicine, 2019, 8, 68.	2.4	14
63	Efficacy of the Ketogenic Diet for the Treatment of Refractory Childhood Epilepsy: Cerebrospinal Fluid Neurotransmitters and Amino Acid Levels. Pediatric Neurology, 2015, 53, 422-426.	2.1	13
64	Nonketotic hyperglycinemia: Functional assessment of missense variants inGLDCto understand phenotypes of the disease. Human Mutation, 2017, 38, 678-691.	2.5	13
65	Presynaptic disorders: a clinical and pathophysiological approach focused on the synaptic vesicle. Journal of Inherited Metabolic Disease, 2018, 41, 1131-1145.	3.6	13
66	Diagnosis of Genetic White Matter Disorders by Singleton Whole-Exome and Genome Sequencing Using Interactome-Driven Prioritization. Neurology, 2022, , 10.1212/WNL.0000000000013278.	1.1	13
67	Deletion in the tyrosine hydroxylase gene in a patient with a mild phenotype. Movement Disorders, 2011, 26, 1558-1560.	3.9	12
68	Analysis of cerebrospinal fluid Î³â€aminobutyric acid by capillary electrophoresis with laserâ€induced fluorescence detection. Electrophoresis, 2014, 35, 1181-1187.	2.4	12
69	Cerebrospinal fluid monoamines, pterins, and folate in patients with mitochondrial diseases: systematic review and hospital experience. Journal of Inherited Metabolic Disease, 2018, 41, 1147-1158.	3.6	12
70	White matter alterations associated with chromosomal disorders. Developmental Medicine and Child Neurology, 2004, 46, 148-53.	2.1	12
71	Folate analysis for the differential diagnosis of profound cerebrospinal fluid folate deficiency. Clinical Biochemistry, 2011, 44, 719-721.	1.9	11
72	Synaptic metabolism: a new approach to inborn errors of neurotransmission. Journal of Inherited Metabolic Disease, 2018, 41, 1065-1075.	3.6	11

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73	Impairment of adenosinergic system in Rett syndrome: Novel therapeutic target to boost BDNF signalling. Neurobiology of Disease, 2020, 145, 105043.	4.4	9
74	Biochemical parameters to assess choroid plexus dysfunction in Kearnsâ€“Sayre syndrome patients. Mitochondrion, 2011, 11, 867-870.	3.4	8
75	Gammaâ€aminobutyric acid levels in cerebrospinal fluid in neuropaediatric disorders. Developmental Medicine and Child Neurology, 2018, 60, 780-792.	2.1	8
76	Pyridoxal Phosphate Supplementation in Neuropediatric Disorders. Seminars in Pediatric Neurology, 2016, 23, 351-358.	2.0	7
77	Study of a fetal brain affected by a severe form of tyrosine hydroxylase deficiency, a rare cause of early parkinsonism. Metabolic Brain Disease, 2016, 31, 705-709.	2.9	7
78	Molecular Characterization of New FBXL4 Mutations in Patients With mtDNA Depletion Syndrome. Frontiers in Genetics, 2019, 10, 1300.	2.3	7
79	Novel Protein Biomarkers of Monoamine Metabolism Defects Correlate with Disease Severity. Movement Disorders, 2021, 36, 690-703.	3.9	7
80	Circulating Cell-Free Mitochondrial DNA in Cerebrospinal Fluid as a Biomarker for Mitochondrial Diseases. Clinical Chemistry, 2021, 67, 1113-1121.	3.2	7
81	Analysis of synaptic proteins in the cerebrospinal fluid as a new tool in the study of inborn errors of neurotransmission. Journal of Inherited Metabolic Disease, 2011, 34, 523-528.	3.6	6
82	Neuromuscular Manifestations in Mitochondrial Diseases in Children. Seminars in Pediatric Neurology, 2016, 23, 290-305.	2.0	6
83	Discovery of Biomarker Panels for Neural Dysfunction in Inborn Errors of Amino Acid Metabolism. Scientific Reports, 2019, 9, 9128.	3.3	6
84	Paradigmatic De Novo GRIN1 Variants Recapitulate Pathophysiological Mechanisms Underlying GRIN1-Related Disorder Clinical Spectrum. International Journal of Molecular Sciences, 2021, 22, 12656.	4.1	6
85	Systematic Collaborative Reanalysis of Genomic Data Improves Diagnostic Yield in Neurologic Rare Diseases. Journal of Molecular Diagnostics, 2022, 24, 529-542.	2.8	6
86	PERINATAL ASPHYXIA MAY CAUSE REDUCTION IN CSF DOPAMINE METABOLITE CONCENTRATIONS. Neurology, 2007, 69, 311-313.	1.1	5
87	Synaptic metabolism and brain circuitries in inborn errors of metabolism. Journal of Inherited Metabolic Disease, 2018, 41, 909-910.	3.6	5
88	Copper Toxicity Associated With an ATP7A-Related Complex Phenotype. Pediatric Neurology, 2021, 119, 40-44.	2.1	4
89	Disorders of Pyruvate Metabolism and the Tricarboxylic Acid Cycle. , 2016, , 187-199.		4
90	Efficacy of baricitinib on chronic pericardial effusion in a patient with Aicardiâ€“GoutiÃ¨res syndrome. Rheumatology, 2022, 61, e87-e89.	1.9	4

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91	Disorders of Neurotransmission. , 2012, , 405-422.		3
92	Cognitive stimulation has potential for brain activation in individuals with Rett syndrome. Journal of Intellectual Disability Research, 2022, 66, 213-224.	2.0	3
93	Monoamine neurotransmitters in early epileptic encephalopathies: New insights into pathophysiology and therapy. Developmental Medicine and Child Neurology, 0, , .	2.1	3
94	Propionic Acidemia. , 2010, , 485-488.		2
95	Diagnosis of Biogenic Amines Synthesis Defects. Journal of Pediatric Neurology, 2015, 13, 186-197.	0.2	2
96	Neurological Disease. , 2017, , 251-292.		2
97	Neurotransmitter disorders. , 2020, , 917-929.		2
98	Pediatric Gaucher disease with intermediate type 2â€“3 phenotype associated with parkinsonian features and levodopa responsiveness. Parkinsonism and Related Disorders, 2021, 91, 19-22.	2.2	2
99	Disorders of Pyruvate Metabolism and the Tricarboxylic Acid Cycle. , 2012, , 187-200.		1
100	Biomarkers for the study of catecholamine and serotonin genetic diseases. , 2017, , 301-329.		1
101	Unraveling Molecular Pathways Altered in MeCP2-Related Syndromes, in the Search for New Potential Avenues for Therapy. Biomedicines, 2021, 9, 148.	3.2	1
102	Neurological Disease. , 2010, , 127-159.		1
103	Cerebrospinal Fluid Ion Analysis in Neonatal Seizures. Pediatric Neurology, 2022, 128, 16-19.	2.1	1
104	Volumetric study of brain MRI in a cohort of patients with neurotransmitter disorders. Neuroradiology, 0, , .	2.2	1
105	Palmoplantar Keratoderma VÃ¶rner-Unna-Thost. , 2009, , 1560-1561.		0