

# Ángels García-Cazorla

## List of Publications by Year in descending order

Source: <https://exaly.com/author-pdf/2324321/publications.pdf>

Version: 2024-02-01

105  
papers

3,134  
citations

147726

31  
h-index

206029

48  
g-index

110  
all docs

110  
docs citations

110  
times ranked

4069  
citing authors

#	ARTICLE	IF	CITATIONS
1	Efficacy of baricitinib on chronic pericardial effusion in a patient with Aicardiâ€™GoutiÃƒres syndrome. <i>Rheumatology</i> , 2022, 61, e87-e89.	0.9	4
2	Cognitive stimulation has potential for brain activation in individuals with Rett syndrome. <i>Journal of Intellectual Disability Research</i> , 2022, 66, 213-224.	1.2	3
3	Diagnosis of Genetic White Matter Disorders by Singleton Whole-Exome and Genome Sequencing Using Interactome-Driven Prioritization. <i>Neurology</i> , 2022, , 10.1212/WNL.0000000000013278.	1.5	13
4	Cerebrospinal Fluid Ion Analysis in Neonatal Seizures. <i>Pediatric Neurology</i> , 2022, 128, 16-19.	1.0	1
5	Systematic Collaborative Reanalysis of Genomic Data Improves Diagnostic Yield in Neurologic Rare Diseases. <i>Journal of Molecular Diagnostics</i> , 2022, 24, 529-542.	1.2	6
6	Disease-associated GRIN protein truncating variants trigger NMDA receptor loss-of-function. <i>Human Molecular Genetics</i> , 2021, 29, 3859-3871.	1.4	16
7	Novel Protein Biomarkers of Monoamine Metabolism Defects Correlate with Disease Severity. <i>Movement Disorders</i> , 2021, 36, 690-703.	2.2	7
8	<i>GRIN</i> database: A unified and manually curated repertoire of <i>GRIN</i> variants. <i>Human Mutation</i> , 2021, 42, 8-18.	1.1	29
9	Unraveling Molecular Pathways Altered in MeCP2-Related Syndromes, in the Search for New Potential Avenues for Therapy. <i>Biomedicines</i> , 2021, 9, 148.	1.4	1
10	Copper Toxicity Associated With an ATP7A-Related Complex Phenotype. <i>Pediatric Neurology</i> , 2021, 119, 40-44.	1.0	4
11	Circulating Cell-Free Mitochondrial DNA in Cerebrospinal Fluid as a Biomarker for Mitochondrial Diseases. <i>Clinical Chemistry</i> , 2021, 67, 1113-1121.	1.5	7
12	Pediatric Gaucher disease with intermediate type 2â€™3 phenotype associated with parkinsonian features and levodopa responsiveness. <i>Parkinsonism and Related Disorders</i> , 2021, 91, 19-22.	1.1	2
13	Paradigmatic De Novo GRIN1 Variants Recapitulate Pathophysiological Mechanisms Underlying GRIN1-Related Disorder Clinical Spectrum. <i>International Journal of Molecular Sciences</i> , 2021, 22, 12656.	1.8	6
14	Clinical presentation and proteomic signature of patients with <i>TANGO2</i> mutations. <i>Journal of Inherited Metabolic Disease</i> , 2020, 43, 297-308.	1.7	43
15	Impairment of adenosinergic system in Rett syndrome: Novel therapeutic target to boost BDNF signalling. <i>Neurobiology of Disease</i> , 2020, 145, 105043.	2.1	9
16	Impairment of the mitochondrial one-carbon metabolism enzyme SHMT2 causes a novel brain and heart developmental syndrome. <i>Acta Neuropathologica</i> , 2020, 140, 971-975.	3.9	24
17	Neurotransmitter disorders. , 2020, , 917-929.		2
18	<sc><i>DNAJC6</i></sc> Mutations Disrupt Dopamine Homeostasis in Juvenile <sc>Parkinsonismâ€™Dystonia</sc>. <i>Movement Disorders</i> , 2020, 35, 1357-1368.	2.2	22

#	ARTICLE	IF	CITATIONS
19	<sc>Serine dietary supplementation is associated with clinical improvement of loss-of-function GRIN2B-related pediatric encephalopathy. Science Signaling, 2019, 12, .	1.6	53
20	Discovery of Biomarker Panels for Neural Dysfunction in Inborn Errors of Amino Acid Metabolism. Scientific Reports, 2019, 9, 9128.	1.6	6
21	Muscle Involvement in a Large Cohort of Pediatric Patients with Genetic Diagnosis of Mitochondrial Disease. Journal of Clinical Medicine, 2019, 8, 68.	1.0	14
22	Molecular Characterization of New FBXL4 Mutations in Patients With mtDNA Depletion Syndrome. Frontiers in Genetics, 2019, 10, 1300.	1.1	7
23	Inborn Errors of Metabolism Overview. Pediatric Clinics of North America, 2018, 65, 179-208.	0.9	111
24	Gamma-aminobutyric acid levels in cerebrospinal fluid in neuropaediatric disorders. Developmental Medicine and Child Neurology, 2018, 60, 780-792.	1.1	8
25	Synaptic metabolism and brain circuitries in inborn errors of metabolism. Journal of Inherited Metabolic Disease, 2018, 41, 909-910.	1.7	5
26	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.	1.7	12
27	Synaptic metabolism: a new approach to inborn errors of neurotransmission. Journal of Inherited Metabolic Disease, 2018, 41, 1065-1075.	1.7	11
28	Cellular neurometabolism: a tentative to connect cell biology and metabolism in neurology. Journal of Inherited Metabolic Disease, 2018, 41, 1043-1054.	1.7	18
29	Presynaptic disorders: a clinical and pathophysiological approach focused on the synaptic vesicle. Journal of Inherited Metabolic Disease, 2018, 41, 1131-1145.	1.7	13
30	Severe infantile parkinsonism because of a de novo mutation on DLP1 mitochondrial peroxisomal protein. Movement Disorders, 2017, 32, 1108-1110.	2.2	17
31	Nonketotic hyperglycinemia: Functional assessment of missense variants in GLDC to understand phenotypes of the disease. Human Mutation, 2017, 38, 678-691.	1.1	13
32	The utility of Next Generation Sequencing for molecular diagnostics in Rett syndrome. Scientific Reports, 2017, 7, 12288.	1.6	23
33	Analysis of human cerebrospinal fluid monoamines and their cofactors by HPLC. Nature Protocols, 2017, 12, 2359-2366.	5.5	23
34	Biomarkers for the study of catecholamine and serotonin genetic diseases. , 2017, , 301-329.		1
35	Neurological Disease. , 2017, , 251-292.		2
36	Secondary coenzyme Q 10 deficiencies in oxidative phosphorylation (OXPHOS) and non-OXPHOS disorders. Mitochondrion, 2016, 30, 51-58.	1.6	70

#	ARTICLE	IF	CITATIONS
37	Neuromuscular Manifestations in Mitochondrial Diseases in Children. <i>Seminars in Pediatric Neurology</i> , 2016, 23, 290-305.	1.0	6
38	Pyridoxal Phosphate Supplementation in Neuropediatric Disorders. <i>Seminars in Pediatric Neurology</i> , 2016, 23, 351-358.	1.0	7
39	Diseases of the Synaptic Vesicle: A Potential New Group of Neurometabolic Disorders Affecting Neurotransmission. <i>Seminars in Pediatric Neurology</i> , 2016, 23, 306-320.	1.0	23
40	Study of a fetal brain affected by a severe form of tyrosine hydroxylase deficiency, a rare cause of early parkinsonism. <i>Metabolic Brain Disease</i> , 2016, 31, 705-709.	1.4	7
41	Free-thiamine is a potential biomarker of thiamine transporter-2 deficiency: a treatable cause of Leigh syndrome. <i>Brain</i> , 2016, 139, 31-38.	3.7	174
42	Behavioural and emotional problems, intellectual impairment and health-related quality of life in patients with organic acidurias and urea cycle disorders. <i>Journal of Inherited Metabolic Disease</i> , 2016, 39, 231-241.	1.7	29
43	Clinical Approach to Inborn Errors of Metabolism in Pediatrics. , 2016, , 3-70.		15
44	Disorders of Pyruvate Metabolism and the Tricarboxylic Acid Cycle. , 2016, , 187-199.		4
45	Targeted Next Generation Sequencing in Patients with Inborn Errors of Metabolism. <i>PLoS ONE</i> , 2016, 11, e0156359.	1.1	48
46	Diagnosis of Biogenic Amines Synthesis Defects. <i>Journal of Pediatric Neurology</i> , 2015, 13, 186-197.	0.0	2
47	Phosphomannomutase deficiency (PMM2-CDG): ataxia and cerebellar assessment. <i>Orphanet Journal of Rare Diseases</i> , 2015, 10, 138.	1.2	49
48	Neural commitment of human pluripotent stem cells under defined conditions recapitulates neural development and generates patient-specific neural cells. <i>Biotechnology Journal</i> , 2015, 10, 1578-1588.	1.8	28
49	Review and evaluation of the methodological quality of the existing guidelines and recommendations for inherited neurometabolic disorders. <i>Orphanet Journal of Rare Diseases</i> , 2015, 10, 164.	1.2	19
50	The clinical spectrum of inherited diseases involved in the synthesis and remodeling of complex lipids. A tentative overview. <i>Journal of Inherited Metabolic Disease</i> , 2015, 38, 19-40.	1.7	44
51	Joubert syndrome: a model for untangling recessive disorders with extreme genetic heterogeneity. <i>Journal of Medical Genetics</i> , 2015, 52, 514-522.	1.5	219
52	Mutation loads in different tissues from six pathogenic mtDNA point mutations. <i>Mitochondrion</i> , 2015, 22, 17-22.	1.6	15
53	Evolution of maple syrup urine disease in patients diagnosed by newborn screening versus late diagnosis. <i>European Journal of Paediatric Neurology</i> , 2015, 19, 652-659.	0.7	29
54	Efficacy of the Ketogenic Diet for the Treatment of Refractory Childhood Epilepsy: Cerebrospinal Fluid Neurotransmitters and Amino Acid Levels. <i>Pediatric Neurology</i> , 2015, 53, 422-426.	1.0	13

#	ARTICLE	IF	CITATIONS
55	Clinical, etiological and therapeutic aspects of cerebral folate deficiency. <i>Expert Review of Neurotherapeutics</i> , 2015, 15, 793-802.	1.4	26
56	Cerebrospinal fluid synaptic proteins as useful biomarkers in tyrosine hydroxylase deficiency. <i>Molecular Genetics and Metabolism</i> , 2015, 114, 34-40.	0.5	14
57	Follow-up of folinic acid supplementation for patients with cerebral folate deficiency and Kearns-Sayre syndrome. <i>Orphanet Journal of Rare Diseases</i> , 2014, 9, 217.	1.2	39
58	Analysis of cerebrospinal fluid $\gamma$ -aminobutyric acid by capillary electrophoresis with laser-induced fluorescence detection. <i>Electrophoresis</i> , 2014, 35, 1181-1187.	1.3	12
59	Two Novel Mutations in the <i>BCKDK</i> (Branched-Chain Keto-Acid Dehydrogenase Kinase) Gene Are Responsible for a Neurobehavioral Deficit in Two Pediatric Unrelated Patients. <i>Human Mutation</i> , 2014, 35, 470-477.	1.1	70
60	Parkinsonism and inborn errors of metabolism. <i>Journal of Inherited Metabolic Disease</i> , 2014, 37, 627-642.	1.7	19
61	Protein expression profiles in patients carrying <i>NFU1</i> mutations. Contribution to the pathophysiology of the disease. <i>Journal of Inherited Metabolic Disease</i> , 2013, 36, 841-847.	1.7	31
62	Coenzyme Q10 deficiency in mitochondrial DNA depletion syndromes. <i>Mitochondrion</i> , 2013, 13, 337-341.	1.6	51
63	Homovanillic acid in cerebrospinal fluid of 1388 children with neurological disorders. <i>Developmental Medicine and Child Neurology</i> , 2013, 55, 559-566.	1.1	36
64	Infantile parkinsonism and gabaergic hypotransmission in a patient with pyruvate carboxylase deficiency. <i>Gene</i> , 2013, 532, 302-306.	1.0	145
65	Development of the serotonergic cells in murine raphe nuclei and their relations with rhombomeric domains. <i>Brain Structure and Function</i> , 2013, 218, 1229-1277.	1.2	101
66	Cerebrospinal Fluid Neopterin Analysis in Neuropediatric Patients: Establishment of a New Cut Off-Value for the Identification of Inflammatory-Immune Mediated Processes. <i>PLoS ONE</i> , 2013, 8, e83237.	1.1	22
67	Abnormal Expression of Cerebrospinal Fluid Cation Chloride Cotransporters in Patients with Rett Syndrome. <i>PLoS ONE</i> , 2013, 8, e68851.	1.1	95
68	Disorders of Pyruvate Metabolism and the Tricarboxylic Acid Cycle. , 2012, , 187-200.		1
69	Disorders of Neurotransmission. , 2012, , 405-422.		3
70	Biochemical parameters to assess choroid plexus dysfunction in Kearns-Sayre syndrome patients. <i>Mitochondrion</i> , 2011, 11, 867-870.	1.6	8
71	Hypokinetic-rigid syndrome in children and inborn errors of metabolism. <i>European Journal of Paediatric Neurology</i> , 2011, 15, 295-302.	0.7	27
72	Analysis of synaptic proteins in the cerebrospinal fluid as a new tool in the study of inborn errors of neurotransmission. <i>Journal of Inherited Metabolic Disease</i> , 2011, 34, 523-528.	1.7	6

#	ARTICLE	IF	CITATIONS
73	Deletion in the tyrosine hydroxylase gene in a patient with a mild phenotype. <i>Movement Disorders</i> , 2011, 26, 1558-1560.	2.2	12
74	Folate analysis for the differential diagnosis of profound cerebrospinal fluid folate deficiency. <i>Clinical Biochemistry</i> , 2011, 44, 719-721.	0.8	11
75	Cerebral Folate Deficiency Syndromes in Childhood. <i>Archives of Neurology</i> , 2011, 68, 615-21.	4.9	52
76	Study of inborn errors of metabolism in urine from patients with unexplained mental retardation. <i>Journal of Inherited Metabolic Disease</i> , 2010, 33, 1-7.	1.7	42
77	Progressive ataxia and myoclonic epilepsy in a patient with a homozygous mutation in the <i>FOLR1</i> gene. <i>Journal of Inherited Metabolic Disease</i> , 2010, 33, 795-802.	1.7	43
78	Cerebrospinal fluid alterations of the serotonin product, 5-hydroxyindolacetic acid, in neurological disorders. <i>Journal of Inherited Metabolic Disease</i> , 2010, 33, 803-809.	1.7	34
79	Tyrosine hydroxylase deficiency in three Greek patients with a common ancestral mutation. <i>Movement Disorders</i> , 2010, 25, 1086-1090.	2.2	22
80	Propionic Acidemia. , 2010, , 485-488.		2
81	Neurological Disease. , 2010, , 127-159.		1
82	Palmoplantar Keratoderma Vörner-Unna-Thost. , 2009, , 1560-1561.		0
83	Long-term evolution of eight Spanish patients with CDG type Ia: Typical and atypical manifestations. <i>European Journal of Paediatric Neurology</i> , 2009, 13, 444-451.	0.7	42
84	Mental retardation and inborn errors of metabolism. <i>Journal of Inherited Metabolic Disease</i> , 2009, 32, 597-608.	1.7	43
85	Epilepsy and inborn errors of metabolism in children. <i>Journal of Inherited Metabolic Disease</i> , 2009, 32, 609-617.	1.7	45
86	Inborn errors of metabolism and motor disturbances in children. <i>Journal of Inherited Metabolic Disease</i> , 2009, 32, 618-629.	1.7	41
87	Levodopa therapy in a Lesch-Nyhan disease patient: Pathological, biochemical, neuroimaging, and therapeutic remarks. <i>Movement Disorders</i> , 2008, 23, 1297-1300.	2.2	17
88	Biochemical diagnosis of dopaminergic disturbances in paediatric patients: Analysis of cerebrospinal fluid homovanillic acid and other biogenic amines. <i>Clinical Biochemistry</i> , 2008, 41, 1306-1315.	0.8	39
89	Cerebrospinal fluid pterins and neurotransmitters in early severe epileptic encephalopathies. <i>Brain and Development</i> , 2008, 30, 106-111.	0.6	19
90	Mitochondrial diseases mimicking neurotransmitter defects. <i>Mitochondrion</i> , 2008, 8, 273-278.	1.6	48

#	ARTICLE	IF	CITATIONS
91	Pyridoxal 5â€²-phosphate values in cerebrospinal fluid: Reference values and diagnosis of PNPO deficiency in paediatric patients. <i>Molecular Genetics and Metabolism</i> , 2008, 94, 173-177.	0.5	54
92	MITOCHONDRIAL DISEASES ASSOCIATED WITH CEREBRAL FOLATE DEFICIENCY. <i>Neurology</i> , 2008, 70, 1360-1362.	1.5	77
93	PERINATAL ASPHYXIA MAY CAUSE REDUCTION IN CSF DOPAMINE METABOLITE CONCENTRATIONS. <i>Neurology</i> , 2007, 69, 311-313.	1.5	5
94	A homozygous tyrosine hydroxylase gene promoter mutation in a patient with dopa-responsive encephalopathy: Clinical, biochemical and genetic analysis. <i>Molecular Genetics and Metabolism</i> , 2007, 92, 274-277.	0.5	31
95	Secondary abnormalities of neurotransmitters in infants with neurological disorders. <i>Developmental Medicine and Child Neurology</i> , 2007, 49, 740-744.	1.1	36
96	Determination of 5-methyltetrahydrofolate in cerebrospinal fluid of paediatric patients: Reference values for a paediatric population. <i>Clinica Chimica Acta</i> , 2006, 371, 159-162.	0.5	48
97	Mitochondrial respiratory chain deficiencies expressing the enzymatic deficiency in the hepatic tissue: A study of 31 patients. <i>Journal of Pediatrics</i> , 2006, 149, 401-405.e3.	0.9	18
98	Pyruvate carboxylase deficiency: Metabolic characteristics and new neurological aspects. <i>Annals of Neurology</i> , 2006, 59, 121-127.	2.8	86
99	HPLC with electrochemical and fluorescence detection procedures for the diagnosis of inborn errors of biogenic amines and pterins. <i>Journal of Neuroscience Methods</i> , 2005, 142, 153-158.	1.3	114
100	Long-term Follow-up of Neonatal Mitochondrial Cytopathies: A Study of 57 Patients. <i>Pediatrics</i> , 2005, 116, 1170-1177.	1.0	55
101	Infectious Acute Hemicerebellitis. <i>Journal of Child Neurology</i> , 2004, 19, 390-392.	0.7	16
102	White matter alterations associated with chromosomal disorders. <i>Developmental Medicine and Child Neurology</i> , 2004, 46, 148-153.	1.1	19
103	White matter alterations associated with chromosomal disorders. <i>Developmental Medicine and Child Neurology</i> , 2004, 46, 148-53.	1.1	12
104	Monoamine neurotransmitters in early epileptic encephalopathies: New insights into pathophysiology and therapy. <i>Developmental Medicine and Child Neurology</i> , 0, , .	1.1	3
105	Volumetric study of brain MRI in a cohort of patients with neurotransmitter disorders. <i>Neuroradiology</i> , 0, , .	1.1	1