Rafael Artuch

List of Publications by Year
in descending order

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135 papers	4,520 citations	108046 37 h-index	169272 56 g-index
142 all docs	142 docs citations	142 times ranked	5252 citing authors

#	Article	IF	Citations
1	Cerebrospinal Fluid Ion Analysis in Neonatal Seizures. Pediatric Neurology, 2022, 128, 16-19.	1.0	1
2	Technical Aspects of Coenzyme Q10 Analysis: Validation of a New HPLC-ED Method. Antioxidants, 2022, 11, 528.	2.2	3
3	The clinical and biochemical hallmarks generally associated with <scp>GLUT1DS</scp> may be caused by defects in genes other than <scp>SLC2A1</scp>. Clinical Genetics, 2022, 102, 40-55.	1.0	3
4	Biomedical point-of-care microanalyzer for potentiometric determination of ammonium ion in plasma and whole blood. Analytica Chimica Acta, 2022, 1205, 339782.	2.6	4
5	Systematic Collaborative Reanalysis of Genomic Data Improves Diagnostic Yield in Neurologic Rare Diseases. Journal of Molecular Diagnostics, 2022, 24, 529-542.	1.2	6
6	Biochemical Diagnosis of Mitochondrial Disorders. , 2021, , 201-214.		0
7	Generation of mitochondrial reactive oxygen species is controlled by ATPase inhibitory factor 1 and regulates cognition. PLoS Biology, 2021, 19, e3001252.	2.6	22
8	CERKL, a retinal dystrophy gene, regulates mitochondrial function and dynamics in the mammalian retina. Neurobiology of Disease, 2021, 156, 105405.	2.1	17
9	Circulating Cell-Free Mitochondrial DNA in Cerebrospinal Fluid as a Biomarker for Mitochondrial Diseases. Clinical Chemistry, 2021, 67, 1113-1121.	1.5	7
10	CPEB alteration and aberrant transcriptome-polyadenylation lead to a treatable SLC19A3 deficiency in Huntingtonâ€™s disease. Science Translational Medicine, 2021, 13, eabe7104.	5.8	14
11	Pediatric Gaucher disease with intermediate type 2â€“3 phenotype associated with parkinsonian features and levodopa responsiveness. Parkinsonism and Related Disorders, 2021, 91, 19-22.	1.1	2
12	Plasma idebenone monitoring in Friedreichâ€™s ataxia patients during a long-term follow-up. Biomedicine and Pharmacotherapy, 2021, 143, 112143.	2.5	5
13	Clinical presentation and proteomic signature of patients with TANGO2 mutations. Journal of Inherited Metabolic Disease, 2020, 43, 297-308.	1.7	43
14	Infectious stress triggers a POLG-related mitochondrial disease. Neurogenetics, 2020, 21, 19-27.	0.7	9
15	The Value of Mouse Models of Rare Diseases: A Spanish Experience. Frontiers in Genetics, 2020, 11, 583932.	1.1	12
16	Neurotransmitter disorders. , 2020, , 917-929.		2
17	Laboratory Diagnosis of a Case with Coenzyme Q10 Deficiency. Clinical Chemistry, 2020, 66, 1465-1467.	1.5	1
18	Cerebrospinal fluid neopterin as a biomarker of neuroinflammatory diseases. Scientific Reports, 2020, 10, 18291.	1.6	30

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19	Coenzyme Q10 Treatment Monitoring in Different Human Biological Samples. Antioxidants, 2020, 9, 979.	2.2	12
20	Disrupted Mitochondrial and Metabolic Plasticity Underlie Comorbidity between Age-Related and Degenerative Disorders as Parkinson Disease and Type 2 Diabetes Mellitus. Antioxidants, 2020, 9, 1063.	2.2	8
21	Inducible Slc7a7 Knockout Mouse Model Recapitulates Lysinuric Protein Intolerance Disease. International Journal of Molecular Sciences, 2019, 20, 5294.	1.8	21
22	ADCK2 Haploinsufficiency Reduces Mitochondrial Lipid Oxidation and Causes Myopathy Associated with CoQ Deficiency. Journal of Clinical Medicine, 2019, 8, 1374.	1.0	27
23	Prevalence of sleep disorders in early-treated phenylketonuric children and adolescents. Correlation with dopamine and serotonin status. European Journal of Paediatric Neurology, 2019, 23, 685-691.	0.7	6
24	Discovery of Biomarker Panels for Neural Dysfunction in Inborn Errors of Amino Acid Metabolism. Scientific Reports, 2019, 9, 9128.	1.6	6
25	Cerebral folate deficiency: Analytical tests and differential diagnosis. Journal of Inherited Metabolic Disease, 2019, 42, 655-672.	1.7	69
26	AZATAX: Acetazolamide safety and efficacy in cerebellar syndrome in PMM2 congenital disorder of glycosylation (PMM2â€CDG). Annals of Neurology, 2019, 85, 740-751.	2.8	50
27	Cerebrospinal Fluid Neopterin in Children With Enterovirus-Related Brainstem Encephalitis. Pediatric Neurology, 2019, 96, 70-73.	1.0	8
28	Plasma coenzyme Q10 status is impaired in selected genetic conditions. Scientific Reports, 2019, 9, 793.	1.6	27
29	Effect of blood contamination of cerebrospinal fluid on amino acids, biogenic amines, pterins and vitamins. Fluids and Barriers of the CNS, 2019, 16, 34.	2.4	8
30	From gestalt to gene: early predictive dysmorphic features of PMM2-CDG. Journal of Medical Genetics, 2019, 56, 236-245.	1.5	19
31	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
32	Mutations in the mitochondrial complex I assembly factor NDUFAF6 cause isolated bilateral striatal necrosis and progressive dystonia in childhood. Molecular Genetics and Metabolism, 2019, 126, 250-258.	0.5	20
33	Molecular Characterization of New FBXL4 Mutations in Patients With mtDNA Depletion Syndrome. Frontiers in Genetics, 2019, 10, 1300.	1.1	7
34	Gammaâ€aminobutyric acid levels in cerebrospinal fluid in neuropaediatric disorders. Developmental Medicine and Child Neurology, 2018, 60, 780-792.	1.1	8
35	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.	3.0	25
36	Transcriptional alterations in skin fibroblasts from Parkinson's disease patients with parkin mutations. Neurobiology of Aging, 2018, 65, 206-216.	1.5	13

#	Article	IF	Citations
37	Molecular diagnosis of coenzyme Q₁₀ deficiency: an update. Expert Review of Molecular Diagnostics, 2018, 18, 491-498.	1.5	33
38	A targeted metabolomic procedure for amino acid analysis in different biological specimens by ultra-high-performance liquid chromatographyâ€“tandem mass spectrometry. Metabolomics, 2018, 14, 76.	1.4	30
39	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
40	Stroke-Like Episodes and Cerebellar Syndrome in Phosphomannomutase Deficiency (PMM2-CDG): Evidence for Hypoglycosylation-Driven Channelopathy. International Journal of Molecular Sciences, 2018, 19, 619.	1.8	40
41	Consensus guideline for the diagnosis and treatment of aromatic l-amino acid decarboxylase (AADC) deficiency. Orphanet Journal of Rare Diseases, 2017, 12, 12.	1.2	172
42	Genetic Rescue of Mitochondrial and Skeletal Muscle Impairment in an Induced Pluripotent Stem Cells Model of Coenzyme Q10 Deficiency. Stem Cells, 2017, 35, 1687-1703.	1.4	24
43	Longitudinal volumetric and 2D assessment of cerebellar atrophy in a large cohort of children with phosphomannomutase deficiency (PMM2â€CDG). Journal of Inherited Metabolic Disease, 2017, 40, 709-713.	1.7	16
44	Analysis of human cerebrospinal fluid monoamines and their cofactors by HPLC. Nature Protocols, 2017, 12, 2359-2366.	5.5	23
45	Coenzyme Q₁₀ in the Treatment of Mitochondrial Disease. FIRE Forum for International Research in Education, 2017, 5, 232640981770777.	0.7	22
46	Thiamine deficiency in childhood with attention to genetic causes: Survival and outcome predictors. Annals of Neurology, 2017, 82, 317-330.	2.8	65
47	Urinary sulphatoxymelatonin as a biomarker of serotonin status in biogenic amine-deficient patients. Scientific Reports, 2017, 7, 14675.	1.6	8
48	Urine oligosaccharide tests for the diagnosis of oligosaccharidoses. Reviews in Analytical Chemistry, 2017, 36, .	1.5	7
49	The Value of Coenzyme Q10 Determination in Mitochondrial Patients. Journal of Clinical Medicine, 2017, 6, 37.	1.0	21
50	GDF-15 Is Elevated in Children with Mitochondrial Diseases and Is Induced by Mitochondrial Dysfunction. PLoS ONE, 2016, 11, e0148709.	1.1	133
51	Secondary coenzyme Q 10 deficiencies in oxidative phosphorylation (OXPHOS) and non-OXPHOS disorders. Mitochondrion, 2016, 30, 51-58.	1.6	70
52	Inborn error metabolic screening in individuals with nonsyndromic autism spectrum disorders. Developmental Medicine and Child Neurology, 2016, 58, 842-847.	1.1	34
53	Pyridoxal Phosphate Supplementation in Neuropediatric Disorders. Seminars in Pediatric Neurology, 2016, 23, 351-358.	1.0	7
54	Treatment of genetic defects of thiamine transport and metabolism. Expert Review of Neurotherapeutics, 2016, 16, 755-763.	1.4	33

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55	Ndufs4 related Leigh syndrome: A case report and review of the literature. Mitochondrion, 2016, 28, 73-78.	1.6	59
56	Impaired Neurotransmission in Early-treated Phenylketonuria Patients. Seminars in Pediatric Neurology, 2016, 23, 332-340.	1.0	18
57	Biochemical Analyses of Cerebrospinal Fluid for the Diagnosis of Neurometabolic Conditions. What Can We Expect?. Seminars in Pediatric Neurology, 2016, 23, 273-284.	1.0	10
58	Severe encephalopathy associated to pyruvate dehydrogenase mutations and unbalanced coenzyme Q10 content. European Journal of Human Genetics, 2016, 24, 367-372.	1.4	17
59	Free-thiamine is a potential biomarker of thiamine transporter-2 deficiency: a treatable cause of Leigh syndrome. Brain, 2016, 139, 31-38.	3.7	174
60	Targeted Next Generation Sequencing in Patients with Inborn Errors of Metabolism. PLoS ONE, 2016, 11, e0156359.	1.1	48
61	Determination of urinary coenzyme <scp>Q</scp>₁₀ by <scp>HPLC</scp> with electrochemical detection: Reference values for a paediatric population. BioFactors, 2015, 41, 424-430.	2.6	20
62	Phosphomannomutase deficiency (PMM2-CDG): ataxia and cerebellar assessment. Orphanet Journal of Rare Diseases, 2015, 10, 138.	1.2	49
63	Neurotransmitter Disorders. , 2015, , 703-712.		4
64	Long-term survival in a child with severe encephalopathy, multiple respiratory chain deficiency and GFM1 mutations. Frontiers in Genetics, 2015, 6, 102.	1.1	13
65	Molecular diagnosis of coenzyme Q₁₀deficiency. Expert Review of Molecular Diagnostics, 2015, 15, 1049-1059.	1.5	16
66	Mutation loads in different tissues from six pathogenic mtDNA point mutations. Mitochondrion, 2015, 22, 17-22.	1.6	15
67	Cerebrospinal Fluid Selenium Concentrations in Pediatric Patients with Neurologic Disorders. Journal of Pediatric Biochemistry, 2015, 05, 015-020.	0.2	1
68	Can folic acid have a role in mitochondrial disorders?. Drug Discovery Today, 2015, 20, 1349-1354.	3.2	38
69	Clinical, etiological and therapeutic aspects of cerebral folate deficiency. Expert Review of Neurotherapeutics, 2015, 15, 793-802.	1.4	26
70	Coenzyme Q10 and Pyridoxal Phosphate Deficiency Is a Common Feature in Mucopolysaccharidosis Type III. JIMD Reports, 2015, 25, 1-7.	0.7	8
71	Follow-up of folinic acid supplementation for patients with cerebral folate deficiency and Kearns-Sayre syndrome. Orphanet Journal of Rare Diseases, 2014, 9, 217.	1.2	39
72	Association between coenzyme Q10 and glucose transporter (GLUT1) deficiency. BMC Pediatrics, 2014, 14, 284.	0.7	15

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73	Biochemical Diagnosis of Coenzyme Q₁₀ Deficiency. Molecular Syndromology, 2014, 5, 147-155.	0.3	35
74	Analysis of cerebrospinal fluid Î³â€aminobutyric acid by capillary electrophoresis with laserâ€induced fluorescence detection. Electrophoresis, 2014, 35, 1181-1187.	1.3	12
75	Characterization of CoQ₁₀ biosynthesis in fibroblasts of patients with primary and secondary CoQ₁₀ deficiency. Journal of Inherited Metabolic Disease, 2014, 37, 53-62.	1.7	24
76	Transcriptomic profiling of TK2 deficient human skeletal muscle suggests a role for the p53 signalling pathway and identifies growth and differentiation factor-15 as a potential novel biomarker for mitochondrial myopathies. BMC Genomics, 2014, 15, 91.	1.2	104
77	A capillary electrophoresis procedure for the screening of oligosaccharidoses and related diseases. Analytical and Bioanalytical Chemistry, 2014, 406, 4337-4343.	1.9	11
78	Coenzyme Q10 deficiency in mitochondrial DNA depletion syndromes. Mitochondrion, 2013, 13, 337-341.	1.6	51
79	Homovanillic acid in cerebrospinal fluid of 1388 children with neurological disorders. Developmental Medicine and Child Neurology, 2013, 55, 559-566.	1.1	36
80	Survival transcriptome in the coenzyme Q₁₀deficiency syndrome is acquired by epigenetic modifications: a modelling study for human coenzyme Q₁₀deficiencies. BMJ Open, 2013, 3, e002524.	0.8	19
81	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.	1.1	22
82	Haploinsufficiency ofCOQ4causes coenzyme Q₁₀deficiency. Journal of Medical Genetics, 2012, 49, 187-191.	1.5	95
83	New mitochondrial DNA mutations in tRNA associated with three severe encephalopamyopathic phenotypes: neonatal, infantile, and childhood onset. Neurogenetics, 2012, 13, 245-250.	0.7	23
84	Dyskinesias as a Limiting Factor in the Treatment of Segawa Disease. Pediatric Neurology, 2012, 46, 404-406.	1.0	17
85	Genetic causes of cerebral folate deficiency: clinical, biochemical and therapeutic aspects. Drug Discovery Today, 2012, 17, 1299-1306.	3.2	161
86	Reversible generalized dystonia and encephalopathy from thiamine transporter 2 deficiency. Movement Disorders, 2012, 27, 1295-1298.	2.2	42
87	Novel features in the evolution of adenylosuccinate lyase deficiency. European Journal of Paediatric Neurology, 2012, 16, 343-348.	0.7	14
88	Biochemical parameters to assess choroid plexus dysfunction in Kearnsâ€“Sayre syndrome patients. Mitochondrion, 2011, 11, 867-870.	1.6	8
89	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.	1.7	6
90	Genistein supplementation in patients affected by Sanfilippo disease. Journal of Inherited Metabolic Disease, 2011, 34, 1039-1044.	1.7	67

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91	Neuropsychiatric symptoms and intelligence quotient in autosomal dominant Segawa disease. Journal of Neurology, 2011, 258, 2155-2162.	1.8	31
92	Genotypeâ€“phenotype correlations in sepiapterin reductase deficiency. A splicing defect accounts for a new phenotypic variant. Neurogenetics, 2011, 12, 183-191.	0.7	30
93	Defining the pathogenicity of creatine deficiency syndrome. Human Mutation, 2011, 32, 282-291.	1.1	26
94	Folate analysis for the differential diagnosis of profound cerebrospinal fluid folate deficiency. Clinical Biochemistry, 2011, 44, 719-721.	0.8	11
95	Cerebral Folate Deficiency Syndromes in Childhood. Archives of Neurology, 2011, 68, 615-21.	4.9	52
96	Study of inborn errors of metabolism in urine from patients with unexplained mental retardation. Journal of Inherited Metabolic Disease, 2010, 33, 1-7.	1.7	42
97	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.	1.7	43
98	Cerebrospinal fluid alterations of the serotonin product, 5â€hydroxyindolacetic acid, in neurological disorders. Journal of Inherited Metabolic Disease, 2010, 33, 803-809.	1.7	34
99	Selenium Concentration in Cerebrospinal Fluid Samples from a Paediatric Population. Neurochemical Research, 2010, 35, 1290-1293.	1.6	10
100	Coenzyme Q₁₀â€responsive ataxia: 2â€yearâ€treatment followâ€up. Movement Disorders, 2010, 25, 1262-1268.	2.2	56
101	The monitoring of trace elements in blood samples from patients with inborn errors of metabolism. Journal of Inherited Metabolic Disease, 2010, 33, 43-49.	1.7	24
102	Kearns-Sayre syndrome: Cerebral folate deficiency, MRI findings and new cerebrospinal fluid biochemical features. Mitochondrion, 2010, 10, 429-432.	1.6	53
103	Coenzyme Q deficiency triggers mitochondria degradation by mitophagy. Autophagy, 2009, 5, 19-32.	4.3	179
104	Coenzyme Q10 deficiency associated with a mitochondrial DNA depletion syndrome: A case report. Clinical Biochemistry, 2009, 42, 742-745.	0.8	25
105	Coenzyme Q10 Deficiencies in Neuromuscular Diseases. Advances in Experimental Medicine and Biology, 2009, 652, 117-128.	0.8	21
106	Analysis of Coenzyme Q10 in muscle and fibroblasts for the diagnosis of CoQ10 deficiency syndromes. Clinical Biochemistry, 2008, 41, 697-700.	0.8	65
107	Levodopa therapy in a Leschâ€Nyhan disease patient: Pathological, biochemical, neuroimaging, and therapeutic remarks. Movement Disorders, 2008, 23, 1297-1300.	2.2	17
108	Biochemical diagnosis of dopaminergic disturbances in paediatric patients: Analysis of cerebrospinal fluid homovanillic acid and other biogenic amines. Clinical Biochemistry, 2008, 41, 1306-1315.	0.8	39

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109	Cerebrospinal fluid pterins and neurotransmitters in early severe epileptic encephalopathies. Brain and Development, 2008, 30, 106-111.	0.6	19
110	Idebenone treatment in paediatric and adult patients with Friedreich ataxia: Long-term follow-up. European Journal of Paediatric Neurology, 2008, 12, 470-475.	0.7	89
111	Cognitive functions and the antioxidant system in phenylketonuric patients.. Neuropsychology, 2008, 22, 426-431.	1.0	31
112	Mitochondrial diseases mimicking neurotransmitter defects. Mitochondrion, 2008, 8, 273-278.	1.6	48
113	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.	0.5	54
114	Oral Insulin-Mimetic Compounds That Act Independently of Insulin. Diabetes, 2007, 56, 486-493.	0.3	60
115	Dopa-responsive infantile hypokinetic rigid syndrome due to dominant guanosine triphosphate cyclohydrolase 1 deficiency. Journal of the Neurological Sciences, 2007, 256, 90-93.	0.3	15
116	Clinical, biochemical and molecular aspects of cerebellar ataxia and Coenzyme Q10 deficiency. Cerebellum, 2007, 6, 118-122.	1.4	51
117	Creatine transporter deficiency: Prevalence among patients with mental retardation and pitfalls in metabolite screening. Clinical Biochemistry, 2007, 40, 1328-1331.	0.8	67
118	Cerebellar ataxia with coenzyme Q10 deficiency: Diagnosis and follow-up after coenzyme Q10 supplementation. Journal of the Neurological Sciences, 2006, 246, 153-158.	0.3	94
119	Determination of 5-methyltetrahydrofolate in cerebrospinal fluid of paediatric patients: Reference values for a paediatric population. Clinica Chimica Acta, 2006, 371, 159-162.	0.5	48
120	Oral phenylalanine loading test for the diagnosis of dominant guanosine triphosphate cyclohydrolase 1 deficiency. Clinical Biochemistry, 2006, 39, 893-897.	0.8	7
121	Methods for the diagnosis of creatine deficiency syndromes: A comparative study. Journal of Neuroscience Methods, 2006, 156, 305-309.	1.3	34
122	Cerebral folate deficiency and leukoencephalopathy caused by a mitochondrial DNA deletion. Annals of Neurology, 2006, 59, 394-398.	2.8	122
123	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.	1.3	114
124	Pre- and postnatal diagnosis of tyrosine hydroxylase deficiency. Prenatal Diagnosis, 2005, 25, 671-675.	1.1	17
125	Cognitive functions in classic phenylketonuria and mild hyperphenyl-alaninaemia: experience in a paediatric population. Developmental Medicine and Child Neurology, 2005, 47, 443-448.	1.1	6
126	A longitudinal study of antioxidant status in phenylketonuric patients. Clinical Biochemistry, 2004, 37, 198-203.	0.8	57

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127	Western blotting with diaminobenzidine detection for the diagnosis of congenital disorders of glycosylation. Journal of Neuroscience Methods, 2003, 125, 167-171.	1.3	9
128	Lipophilic antioxidants in patients with phenylketonuria. American Journal of Clinical Nutrition, 2003, 77, 185-188.	2.2	58
129	Monitoring of idebenone treatment in patients with Friedreich's ataxia by high-pressure liquid chromatography with electrochemical detection. Journal of Neuroscience Methods, 2002, 115, 63-66.	1.3	10
130	Is there a relationship between plasma phenylalanine and cholesterol in phenylketonuric patients under dietary treatment?. Clinical Biochemistry, 2001, 34, 373-376.	0.8	26
131	Personal Experience with the Application of Carbohydrate-Deficient Transferrin (CDT) Assays to the Detection of Congenital Disorders of Glycosylation. Clinical Chemistry and Laboratory Medicine, 2000, 38, 965-9.	1.4	13
132	Decreased serum ubiquinone-10 concentrations in phenylketonuria. American Journal of Clinical Nutrition, 1999, 70, 892-895.	2.2	56
133	Serum Ubiquinone-10 in a Pediatric Population. Clinical Chemistry, 1998, 44, 2378-2379.	1.5	11
134	Tocopherol in inborn errors of intermediary metabolism. Clinica Chimica Acta, 1997, 263, 147-155.	0.5	34
135	Monoamine neurotransmitters in early epileptic encephalopathies: New insights into pathophysiology and therapy. Developmental Medicine and Child Neurology, 0, , .	1.1	3