

Mercedes Serrano

List of Publications by Year in Descending Order

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The third column is the impact factor (IF) of the journal, and the fourth column is the number of citations of the article.

42
papers

713
citations

17
h-index

25
g-index

45
ext. papers

885
ext. citations

4.5
avg, IF

3.48
L-index

#	Paper	IF	Citations
42	Stroke-Like Episodes in PMM2-CDG: When the Lack of Other Evidence Is the Only Evidence. <i>Frontiers in Pediatrics</i> , 2021 , 9, 717864	3.4	0
41	Mutations Causing Early Onset Ataxia: Profiling Clinical, Dysmorphic and Structural-Functional Findings. <i>International Journal of Molecular Sciences</i> , 2021 , 22,	6.3	0
40	International consensus guidelines for phosphoglucomutase 1 deficiency (PGM1-CDG): Diagnosis, follow-up, and management. <i>Journal of Inherited Metabolic Disease</i> , 2021 , 44, 148-163	5.4	15
39	Expanding the phenotype of X-linked SSR4-CDG: Connective tissue implications. <i>Human Mutation</i> , 2021 , 42, 142-149	4.7	1
38	Early-onset severe spinocerebellar ataxia 42 with neurodevelopmental deficits (SCA42ND): Case report, pharmacological trial, and literature review. <i>American Journal of Medical Genetics, Part A</i> , 2021 , 185, 256-260	2.5	1
37	Should patients with Phosphomannomutase 2-CDG (PMM2-CDG) be screened for adrenal insufficiency?. <i>Molecular Genetics and Metabolism</i> , 2021 , 133, 397-399	3.7	1
36	Rare CACNA1A mutations leading to congenital ataxia. <i>Pflugers Archiv European Journal of Physiology</i> , 2020 , 472, 791-809	4.6	10
35	Platelet Dysfunction in Noonan and 22q11.2 Deletion Syndromes in Childhood. <i>Thrombosis and Haemostasis</i> , 2020 , 120, 457-465	7	0
34	Life-threatening secondary hemophagocytic lymphohistiocytosis following vagal nerve stimulator infection in a child with CHD2 myoclonic encephalopathy: a case report. <i>Childs Nervous System</i> , 2020 , 36, 2851-2856	1.7	1
33	Okur-Chung neurodevelopmental syndrome in a patient from Spain. <i>American Journal of Medical Genetics, Part A</i> , 2020 , 182, 20-24	2.5	5
32	New and potential strategies for the treatment of PMM2-CDG. <i>Biochimica Et Biophysica Acta - General Subjects</i> , 2020 , 1864, 129686	4	6
31	AZATAX: Acetazolamide safety and efficacy in cerebellar syndrome in PMM2 congenital disorder of glycosylation (PMM2-CDG). <i>Annals of Neurology</i> , 2019 , 85, 740-751	9.4	32
30	International clinical guidelines for the management of phosphomannomutase 2-congenital disorders of glycosylation: Diagnosis, treatment and follow up. <i>Journal of Inherited Metabolic Disease</i> , 2019 , 42, 5-28	5.4	45
29	Elevated thrombin generation in patients with congenital disorder of glycosylation and combined coagulation factor deficiencies. <i>Journal of Thrombosis and Haemostasis</i> , 2019 , 17, 1798-1807	15.4	6
28	From gestalt to gene: early predictive dysmorphic features of PMM2-CDG. <i>Journal of Medical Genetics</i> , 2019 , 56, 236-245	5.8	9
27	PLA2G6-associated neurodegeneration: New insights into brain abnormalities and disease progression. <i>Parkinsonism and Related Disorders</i> , 2019 , 61, 179-186	3.6	17
26	Stroke-Like Episodes and Cerebellar Syndrome in Phosphomannomutase Deficiency (PMM2-CDG): Evidence for Hypoglycosylation-Driven Channelopathy. <i>International Journal of Molecular Sciences</i> , 2018 , 19,	6.3	25

25	Epigenetic cerebellar diseases. <i>Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn, 2018, 155, 227-244</i>	3	12
24	La sociedad civil y las enfermedades raras. <i>Arbor, 2018, 194, 459</i>	0.2	
23	Clinical Assessment of Dysarthria in Children with Cerebellar Syndrome Associated with PMM2-CDG. <i>Neuropediatrics, 2018, 49, 408-413</i>	1.6	1
22	Hyaline fibromatosis syndrome: Clinical update and phenotype-genotype correlations. <i>Human Mutation, 2018, 39, 1752-1763</i>	4.7	22
21	A Population-Based Study on Congenital Disorders of Protein N- and Combined with O-Glycosylation Experience in Clinical and Genetic Diagnosis. <i>Journal of Pediatrics, 2017, 183, 170-177.e13</i>	3.6	17
20	Longitudinal volumetric and 2D assessment of cerebellar atrophy in a large cohort of children with phosphomannomutase deficiency (PMM2-CDG). <i>Journal of Inherited Metabolic Disease, 2017, 40, 709-713</i>	5.4	11
19	A quantitative assessment of the evolution of cerebellar syndrome in children with phosphomannomutase-deficiency (PMM2-CDG). <i>Orphanet Journal of Rare Diseases, 2017, 12, 155</i>	4.2	7
18	Free-thiamine is a potential biomarker of thiamine transporter-2 deficiency: a treatable cause of Leigh syndrome. <i>Brain, 2016, 139, 31-8</i>	11.2	45
17	Phosphomannomutase deficiency (PMM2-CDG): ataxia and cerebellar assessment. <i>Orphanet Journal of Rare Diseases, 2015, 10, 138</i>	4.2	31
16	Environmental circumstances influencing tic expression in children. <i>European Journal of Paediatric Neurology, 2014, 18, 157-62</i>	3.8	19
15	Homovanillic acid in cerebrospinal fluid of 1388 children with neurological disorders. <i>Developmental Medicine and Child Neurology, 2013, 55, 559-66</i>	3.3	35
14	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, 2013, 8, e83237</i>	3.7	16
13	Reversible generalized dystonia and encephalopathy from thiamine transporter 2 deficiency. <i>Movement Disorders, 2012, 27, 1295-8</i>	7	36
12	Deletion in the tyrosine hydroxylase gene in a patient with a mild phenotype. <i>Movement Disorders, 2011, 26, 1558-60</i>	7	10
11	Neuropsychiatric manifestations in late-onset urea cycle disorder patients. <i>Journal of Child Neurology, 2010, 25, 352-8</i>	2.5	26
10	Kearns-Sayre syndrome: cerebral folate deficiency, MRI findings and new cerebrospinal fluid biochemical features. <i>Mitochondrion, 2010, 10, 429-32</i>	4.9	45
9	Cerebrospinal fluid alterations of the serotonin product, 5-hydroxyindolacetic acid, in neurological disorders. <i>Journal of Inherited Metabolic Disease, 2010, 33, 803-9</i>	5.4	29
8	Tyrosine hydroxylase deficiency in three Greek patients with a common ancestral mutation. <i>Movement Disorders, 2010, 25, 1086-90</i>	7	20

7	Cerebrospinal fluid neopterin and cryopyrin-associated periodic syndrome. <i>Pediatric Neurology</i> , 2009 , 41, 448-50	2.9	9
6	Mitochondrial diseases mimicking neurotransmitter defects. <i>Mitochondrion</i> , 2008 , 8, 273-8	4.9	48
5	Levodopa therapy in a Lesch-Nyhan disease patient: pathological, biochemical, neuroimaging, and therapeutic remarks. <i>Movement Disorders</i> , 2008 , 23, 1297-300	7	15
4	Biochemical diagnosis of dopaminergic disturbances in paediatric patients: analysis of cerebrospinal fluid homovanillic acid and other biogenic amines. <i>Clinical Biochemistry</i> , 2008 , 41, 1306-15 ^{3.5}		33
3	Cranial ultrasound and chronological changes in molybdenum cofactor deficiency. <i>Pediatric Radiology</i> , 2007 , 37, 1043-6	2.8	12
2	Retained central venous lines in the newborn: report of one case and systematic review of the literature. <i>Neonatal Network: NN</i> , 2007 , 26, 105-10	0.8	12
1	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, 274-7	3.7	28