Mercedes Serrano

List of Publications by Citations

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	Mitochondrial diseases mimicking neurotransmitter defects. *Mitochondrion*, **2008**, 8, 273-8	4.9	48
41	International clinical guidelines for the management of phosphomannomutase 2-congenital disorders of glycosylation: Diagnosis, treatment and follow up. *Journal of Inherited Metabolic Disease*, **2019**, 42, 5-28	5.4	45
40	Free-thiamine is a potential biomarker of thiamine transporter-2 deficiency: a treatable cause of Leigh syndrome. *Brain*, **2016**, 139, 31-8	11.2	45
39	Kearns-Sayre syndrome: cerebral folate deficiency, MRI findings and new cerebrospinal fluid biochemical features. *Mitochondrion*, **2010**, 10, 429-32	4.9	45
38	Reversible generalized dystonia and encephalopathy from thiamine transporter 2 deficiency. *Movement Disorders*, **2012**, 27, 1295-8	7	36
37	Homovanillic acid in cerebrospinal fluid of 1388 children with neurological disorders. *Developmental Medicine and Child Neurology*, **2013**, 55, 559-66	3.3	35
36	Biochemical diagnosis of dopaminergic disturbances in paediatric patients: analysis of cerebrospinal fluid homovanillic acid and other biogenic amines. *Clinical Biochemistry*, **2008**, 41, 1306-15	3.5	33
35	AZATAX: Acetazolamide safety and efficacy in cerebellar syndrome in PMM2 congenital disorder of glycosylation (PMM2-CDG). *Annals of Neurology*, **2019**, 85, 740-751	9.4	32
34	Phosphomannomutase deficiency (PMM2-CDG): ataxia and cerebellar assessment. *Orphanet Journal of Rare Diseases*, **2015**, 10, 138	4.2	31
33	Cerebrospinal fluid alterations of the serotonin product, 5-hydroxyindolacetic acid, in neurological disorders. *Journal of Inherited Metabolic Disease*, **2010**, 33, 803-9	5.4	29
32	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, 274-7	3.7	28
31	Neuropsychiatric manifestations in late-onset urea cycle disorder patients. *Journal of Child Neurology*, **2010**, 25, 352-8	2.5	26
30	Stroke-Like Episodes and Cerebellar Syndrome in Phosphomannomutase Deficiency (PMM2-CDG): Evidence for Hypoglycosylation-Driven Channelopathy. *International Journal of Molecular Sciences*, **2018**, 19,	6.3	25
29	Hyaline fibromatosis syndrome: Clinical update and phenotype-genotype correlations. *Human Mutation*, **2018**, 39, 1752-1763	4.7	22
28	Tyrosine hydroxylase deficiency in three Greek patients with a common ancestral mutation. *Movement Disorders*, **2010**, 25, 1086-90	7	20
27	Environmental circumstances influencing tic expression in children. *European Journal of Paediatric Neurology*, **2014**, 18, 157-62	3.8	19
26	A Population-Based Study on Congenital Disorders of Protein N- and Combined with O-Glycosylation Experience in Clinical and Genetic Diagnosis. *Journal of Pediatrics*, **2017**, 183, 170-177.e1	3.6	17

25	PLA2G6-associated neurodegeneration: New insights into brain abnormalities and disease progression. *Parkinsonism and Related Disorders*, **2019**, 61, 179-186	3.6	17
24	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	3.7	16
23	Levodopa therapy in a Lesch-Nyhan disease patient: pathological, biochemical, neuroimaging, and therapeutic remarks. *Movement Disorders*, **2008**, 23, 1297-300	7	15
22	International consensus guidelines for phosphoglucomutase 1 deficiency (PGM1-CDG): Diagnosis, follow-up, and management. *Journal of Inherited Metabolic Disease*, **2021**, 44, 148-163	5.4	15
21	Epigenetic cerebellar diseases. *Handbook of Clinical Neurology / Edited By P J Vinken and G W Bruyn*, **2018**, 155, 227-244	3	12
20	Cranial ultrasound and chronological changes in molybdenum cofactor deficiency. *Pediatric Radiology*, **2007**, 37, 1043-6	2.8	12
19	Retained central venous lines in the newborn: report of one case and systematic review of the literature. *Neonatal Network: NN*, **2007**, 26, 105-10	0.8	12
18	Longitudinal volumetric and 2D assessment of cerebellar atrophy in a large cohort of children with phosphomannomutase deficiency (PMM2-CDG). *Journal of Inherited Metabolic Disease*, **2017**, 40, 709-713	5.4	11
17	Rare CACNA1A mutations leading to congenital ataxia. *Pflugers Archiv European Journal of Physiology*, **2020**, 472, 791-809	4.6	10
16	Deletion in the tyrosine hydroxylase gene in a patient with a mild phenotype. *Movement Disorders*, **2011**, 26, 1558-60	7	10
15	Cerebrospinal fluid neopterin and cryopyrin-associated periodic syndrome. *Pediatric Neurology*, **2009**, 41, 448-50	2.9	9
14	From gestalt to gene: early predictive dysmorphic features of PMM2-CDG. *Journal of Medical Genetics*, **2019**, 56, 236-245	5.8	9
13	A quantitative assessment of the evolution of cerebellar syndrome in children with phosphomannomutase-deficiency (PMM2-CDG). *Orphanet Journal of Rare Diseases*, **2017**, 12, 155	4.2	7
12	Elevated thrombin generation in patients with congenital disorder of glycosylation and combined coagulation factor deficiencies. *Journal of Thrombosis and Haemostasis*, **2019**, 17, 1798-1807	15.4	6
11	New and potential strategies for the treatment of PMM2-CDG. *Biochimica Et Biophysica Acta - General Subjects*, **2020**, 1864, 129686	4	6
10	Okur-Chung neurodevelopmental syndrome in a patient from Spain. *American Journal of Medical Genetics, Part A*, **2020**, 182, 20-24	2.5	5
9	Life-threatening secondary hemophagocytic lymphohistiocytosis following vagal nerve stimulator infection in a child with CHD2 myoclonic encephalopathy: a case report. *Childls Nervous System*, **2020**, 36, 2851-2856	1.7	1
8	Expanding the phenotype of X-linked SSR4-CDG: Connective tissue implications. *Human Mutation*, **2021**, 42, 142-149	4.7	1

7	Early-onset severe spinocerebellar ataxia 42 with neurodevelopmental deficits (SCA42ND): Case report, pharmacological trial, and literature review. *American Journal of Medical Genetics, Part A*, **2021**, 185, 256-260	2.5	1
6	Clinical Assessment of Dysarthria in Children with Cerebellar Syndrome Associated with PMM2-CDG. *Neuropediatrics*, **2018**, 49, 408-413	1.6	1
5	Should patients with Phosphomannomutase 2-CDG (PMM2-CDG) be screened for adrenal insufficiency?. *Molecular Genetics and Metabolism*, **2021**, 133, 397-399	3.7	1
4	Platelet Dysfunction in Noonan and 22q11.2 Deletion Syndromes in Childhood. *Thrombosis and Haemostasis*, **2020**, 120, 457-465	7	0
3	Stroke-Like Episodes in PMM2-CDG: When the Lack of Other Evidence Is the Only Evidence. *Frontiers in Pediatrics*, **2021**, 9, 717864	3.4	0
2	Mutations Causing Early Onset Ataxia: Profiling Clinical, Dysmorphic and Structural-Functional Findings. *International Journal of Molecular Sciences*, **2021**, 22,	6.3	0
1	La sociedad civil y las enfermedades raras. *Arbor*, **2018**, 194, 459	0.2	