Mercedes Pineda

List of Publications by Year
in descending order

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21 papers	1,384 citations	516215 16 h-index	752256 20 g-index
21 all docs	21 docs citations	21 times ranked	1736 citing authors

#	Article	IF	Citations
1	Cognitive stimulation has potential for brain activation in individuals with Rett syndrome. Journal of Intellectual Disability Research, 2022, 66, 213-224.	1.2	3
2	Endocrine and Growth Abnormalities in 4H Leukodystrophy Caused by Variants in POLR3A, POLR3B, and POLR1C. Journal of Clinical Endocrinology and Metabolism, 2021, 106, e660-e674.	1.8	26
3	Treatment outcomes following continuous miglustat therapy in patients with Niemann-Pick disease Type C: a final report of the NPC Registry. Orphanet Journal of Rare Diseases, 2020, 15, 104.	1.2	51
4	Evaluation of different suspicion indices in identifying patients with Niemann-Pick disease Type C in clinical practice: a post hoc analysis of a retrospective chart review. Orphanet Journal of Rare Diseases, 2019, 14, 161.	1.2	5
5	Disease characteristics, prognosis and miglustat treatment effects on disease progression in patients with Niemann-Pick disease Type C: an international, multicenter, retrospective chart review. Orphanet Journal of Rare Diseases, 2019, 14, 32.	1.2	37
6	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
7	Consensus clinical management guidelines for Niemann-Pick disease type C. Orphanet Journal of Rare Diseases, 2018, 13, 50.	1.2	200
8	Differences in Niemann-Pick disease Type C symptomatology observed in patients of different ages. Molecular Genetics and Metabolism, 2017, 120, 180-189.	0.5	32
9	GDF-15 Is Elevated in Children with Mitochondrial Diseases and Is Induced by Mitochondrial Dysfunction. PLoS ONE, 2016, 11, e0148709.	1.1	133
10	A Suspicion Index to aid screening of early-onset Niemann-Pick disease Type C (NP-C). BMC Pediatrics, 2016, 16, 107.	0.7	26
11	Clinical course of sly syndrome (mucopolysaccharidosis type VII). Journal of Medical Genetics, 2016, 53, 403-418.	1.5	133
12	Severe encephalopathy associated to pyruvate dehydrogenase mutations and unbalanced coenzyme Q10 content. European Journal of Human Genetics, 2016, 24, 367-372.	1.4	17
13	Cerebrospinal fluid synaptic proteins as useful biomarkers in tyrosine hydroxylase deficiency. Molecular Genetics and Metabolism, 2015, 114, 34-40.	0.5	14
14	Niemann-Pick type C Suspicion Index tool: analyses by age and association of manifestations. Journal of Inherited Metabolic Disease, 2014, 37, 93-101.	1.7	48
15	PO-0369â€…A Niemann-pick Disease Type C (np-c) Suspicion Index Tool To Aid Diagnosis In Paediatric Patients. Archives of Disease in Childhood, 2014, 99, A366.2-A366.	1.0	0
16	Development of a Suspicion Index to aid diagnosis of Niemann-Pick disease type C. Neurology, 2012, 78, 1560-1567.	1.5	127
17	Kearns-Sayre syndrome: Cerebral folate deficiency, MRI findings and new cerebrospinal fluid biochemical features. Mitochondrion, 2010, 10, 429-432.	1.6	53
18	Clinical experience with miglustat therapy in pediatric patients with Niemannâ€“Pick disease type C: A case series. Molecular Genetics and Metabolism, 2010, 99, 358-366.	0.5	117

#	Article	IF	Citations
19	Mental retardation and inborn errors of metabolism. Journal of Inherited Metabolic Disease, 2009, 32, 597-608.	1.7	43
20	Miglustat in patients with Niemann-Pick disease Type C (NP-C): A multicenter observational retrospective cohort study. Molecular Genetics and Metabolism, 2009, 98, 243-249.	0.5	175
21	Niemannâ€“Pick C disease in Spain: Clinical spectrum and development of a disability scale. Journal of the Neurological Sciences, 2006, 249, 1-6.	0.3	130