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	Consensus clinical management guidelines for Niemann-Pick disease type C. Orphanet Journal of Rare Diseases, 2018, 13, 50.	1.2	200
2	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
3	GDF-15 Is Elevated in Children with Mitochondrial Diseases and Is Induced by Mitochondrial Dysfunction. PLoS ONE, 2016, 11, e0148709.	1.1	133
4	Clinical course of sly syndrome (mucopolysaccharidosis type VII). Journal of Medical Genetics, 2016, 53, 403-418.	1.5	133
5	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
6	Development of a Suspicion Index to aid diagnosis of Niemann-Pick disease type C. Neurology, 2012, 78, 1560-1567.	1.5	127
7	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
8	Kearns-Sayre syndrome: Cerebral folate deficiency, MRI findings and new cerebrospinal fluid biochemical features. Mitochondrion, 2010, 10, 429-432.	1.6	53
9	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
10	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
11	Mental retardation and inborn errors of metabolism. Journal of Inherited Metabolic Disease, 2009, 32, 597-608.	1.7	43
12	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
13	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
14	A Suspicion Index to aid screening of early-onset Niemann-Pick disease Type C (NP-C). BMC Pediatrics, 2016, 16, 107.	0.7	26
15	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
16	Severe encephalopathy associated to pyruvate dehydrogenase mutations and unbalanced coenzyme Q10 content. European Journal of Human Genetics, 2016, 24, 367-372.	1.4	17
17	Cerebrospinal fluid synaptic proteins as useful biomarkers in tyrosine hydroxylase deficiency. Molecular Genetics and Metabolism, 2015, 114, 34-40.	0.5	14
18	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

#	Article	IF	Citations
19	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
20	Cognitive stimulation has potential for brain activation in individuals with Rett syndrome. Journal of Intellectual Disability Research, 2022, 66, 213-224.	1.2	3
21	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