BelÃ©n PÃ©rez-DueÃ±as

List of Publications by Year
in descending order

37 papers	1,522 citations	361413 20 h-index	377865 34 g-index
37 all docs	37 docs citations	37 times ranked	2360 citing authors

#	ARTICLE	IF	CITATIONS
1	Mutations in the histone methyltransferase gene KMT2B cause complex early-onset dystonia. Nature Genetics, 2017, 49, 223-237.	21.4	186
2	Reversible Lactic Acidosis in a Newborn With Thiamine Transporter-2 Deficiency. Pediatrics, 2013, 131, e1670-e1675.	2.1	181
3	Free-thiamine is a potential biomarker of thiamine transporter-2 deficiency: a treatable cause of Leigh syndrome. Brain, 2016, 139, 31-38.	7.6	174
4	Genetic defects of thiamine transport and metabolism: A review of clinical phenotypes, genetics, and functional studies. Journal of Inherited Metabolic Disease, 2019, 42, 581-597.	3.6	77
5	Secondary coenzyme Q 10 deficiencies in oxidative phosphorylation (OXPHOS) and non-OXPHOS disorders. Mitochondrion, 2016, 30, 51-58.	3.4	70
6	Thiamine deficiency in childhood with attention to genetic causes: Survival and outcome predictors. Annals of Neurology, 2017, 82, 317-330.	5.3	65
7	Genetic and phenotypic spectrum associated with IFIH1 gainâ€ofâ€function. Human Mutation, 2020, 41, 837-849.	2.5	63
8	Genetic, Phenotypic, and Interferon Biomarker Status in ADAR1-Related Neurological Disease. Neuropediatrics, 2017, 48, 166-184.	0.6	62
9	Ndufs4 related Leigh syndrome: A case report and review of the literature. Mitochondrion, 2016, 28, 73-78.	3.4	59
10	Thiamine transporter-2 deficiency: outcome and treatment monitoring. Orphanet Journal of Rare Diseases, 2014, 9, 92.	2.7	55
11	Phosphomannomutase deficiency (PMM2-CDG): ataxia and cerebellar assessment. Orphanet Journal of Rare Diseases, 2015, 10, 138.	2.7	49
12	Reversible generalized dystonia and encephalopathy from thiamine transporter 2 deficiency. Movement Disorders, 2012, 27, 1295-1298.	3.9	42
13	PLA2G6-associated neurodegeneration: New insights into brain abnormalities and disease progression. Parkinsonism and Related Disorders, 2019, 61, 179-186.	2.2	41
14	Tetrahydrobiopterin responsiveness in patients with phenylketonuria. Clinical Biochemistry, 2004, 37, 1083-1090.	1.9	35
15	Childhood onset progressive myoclonic dystonia due to a de novo KCTD17 splicing mutation. Parkinsonism and Related Disorders, 2019, 61, 7-9.	2.2	34
16	Treatment of genetic defects of thiamine transport and metabolism. Expert Review of Neurotherapeutics, 2016, 16, 755-763.	2.8	33
17	Hyaline fibromatosis syndrome: Clinical update and phenotype-genotype correlations. Human Mutation, 2018, 39, 1752-1763.	2.5	32
18	Environmental circumstances influencing tic expression in children. European Journal of Paediatric Neurology, 2014, 18, 157-162.	1.6	28

#	Article	IF	Citations
19	The genetic etiology in cerebral palsy mimics: The results from a Greek tertiary care center. European Journal of Paediatric Neurology, 2019, 23, 427-437.	1.6	26
20	The European Reference Network for Rare Neurological Diseases. Frontiers in Neurology, 2020, 11, 616569.	2.4	26
21	Delineating the neurological phenotype in children with defects in the <scp>ECHS1</scp> or <scp>HIBCH</scp> gene. Journal of Inherited Metabolic Disease, 2021, 44, 401-414.	3.6	23
22	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.	1.1	20
23	Brain injury in glutaric aciduria type I: The value of functional techniques in magnetic resonance imaging. European Journal of Paediatric Neurology, 2009, 13, 534-540.	1.6	19
24	From gestalt to gene: early predictive dysmorphic features of PMM2-CDG. Journal of Medical Genetics, 2019, 56, 236-245.	3.2	19
25	Encephalopathies with intracranial calcification in children: clinical and genetic characterization. Orphanet Journal of Rare Diseases, 2018, 13, 135.	2.7	17
26	Magnetic resonance imaging pattern recognition in childhood bilateral basal ganglia disorders. Brain Communications, 2020, 2, fcaa178.	3.3	17
27	Clinical phenotypes of infantile onset CACNA1A-related disorder. European Journal of Paediatric Neurology, 2021, 30, 144-154.	1.6	13
28	PRKRAâ€Related Disorders: Bilateral Striatal Degeneration in Addition to DYT16 Spectrum. Movement Disorders, 2021, 36, 1038-1040.	3.9	10
29	Comparison of methylation episignatures in KMT2B- and KMT2D-related human disorders. Epigenomics, 2022, 14, 537-547.	2.1	10
30	Expanding the Î²-III Spectrin-Associated Phenotypes toward Non-Progressive Congenital Ataxias with Neurodegeneration. International Journal of Molecular Sciences, 2021, 22, 2505.	4.1	8
31	Delineating the motor phenotype of SGCE-myoclonus dystonia syndrome. Parkinsonism and Related Disorders, 2020, 80, 165-174.	2.2	7
32	Îµ-Sarcoglycan: Unraveling the Myoclonus-Dystonia Gene. Molecular Neurobiology, 2021, 58, 3938-3952.	4.0	7
33	Treatable Inborn Errors of Metabolism Due to Membrane Vitamin Transporters Deficiency. Seminars in Pediatric Neurology, 2016, 23, 341-350.	2.0	6
34	Systematic Collaborative Reanalysis of Genomic Data Improves Diagnostic Yield in Neurologic Rare Diseases. Journal of Molecular Diagnostics, 2022, 24, 529-542.	2.8	6
35	A framework for paediatric neuromodulation â€“ Recognising the challenges and a platform for data sharing. European Journal of Paediatric Neurology, 2017, 21, 18-19.	1.6	1
36	Generation of three human iPSC lines from PLAN (PLA2G6-associated neurodegeneration) patients. Stem Cell Research, 2021, 53, 102338.	0.7	1

#	Article	IF	Citations
37	Genetic diagnosis of basal ganglia disease in childhood. Developmental Medicine and Child Neurology, 2022, 64, 743-752.	2.1	0