

Carlos E Prada

List of Publications by Year in descending order

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Version: 2024-02-01

54
papers

1,774
citations

430754

18
h-index

302012

39
g-index

56
all docs

56
docs citations

56
times ranked

3835
citing authors

#	ARTICLE	IF	CITATIONS
1	The diagnosis and management of Gaucher disease in pediatric patients: Where do we go from here?. <i>Molecular Genetics and Metabolism</i> , 2022, 136, 4-21.	0.5	18
2	Craniosynostosis is a feature of Costello syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 1280-1286.	0.7	3
3	NAA10 p.(N101K) disrupts N-terminal acetyltransferase complex NatA and is associated with developmental delay and hemihypertrophy. <i>European Journal of Human Genetics</i> , 2021, 29, 280-288.	1.4	7
4	CHARGE syndrome in the era of molecular diagnosis: Similar outcomes in those without coloboma or choanal atresia. <i>European Journal of Medical Genetics</i> , 2021, 64, 104103.	0.7	4
5	DLG4-related synaptopathy: a new rare brain disorder. <i>Genetics in Medicine</i> , 2021, 23, 888-899.	1.1	16
6	Everolimus for severe arrhythmias in tuberous sclerosis complex related cardiac rhabdomyomas. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 1525-1531.	0.7	6
7	POLRMT mutations impair mitochondrial transcription causing neurological disease. <i>Nature Communications</i> , 2021, 12, 1135.	5.8	21
8	UBA2 variants underlie a recognizable syndrome with variable aplasia cutis congenita and ectrodactyly. <i>Genetics in Medicine</i> , 2021, 23, 1624-1635.	1.1	7
9	Generation of a Mouse Model to Study the Noonan Syndrome Gene Lztr1 in the Telencephalon. <i>Frontiers in Cell and Developmental Biology</i> , 2021, 9, 673995.	1.8	1
10	Hyperleucinosis during infections in maple syrup urine disease post liver transplantation. <i>Molecular Genetics and Metabolism Reports</i> , 2021, 27, 100763.	0.4	3
11	Neutropenia and intellectual disability are hallmarks of biallelic and de novo CLPB deficiency. <i>Genetics in Medicine</i> , 2021, 23, 1705-1714.	1.1	22
12	A role for sustained MAPK activity in the mouse ventral telencephalon. <i>Developmental Biology</i> , 2021, 476, 137-147.	0.9	6
13	Molecular characterization of mucopolysaccharidosis type IVA patients in the Andean region of Colombia. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2021, 187, 388-395.	0.7	7
14	Acute liver dysfunction with delayed peak of serum aminotransferase levels as a presentation of ornithine transcarbamylase deficiency in females. <i>American Journal of Medical Genetics, Part A</i> , 2021, 185, 909-915.	0.7	0
15	A recurrent, de novo pathogenic variant in ARPC4 disrupts actin filament formation and causes a neurodevelopmental disorder with microcephaly and speech delay. <i>Human Genetics and Genomics Advances</i> , 2021, 3, 100072.	1.0	4
16	725. Complete Blood Count Values Vary in Degree of Change with Day of Fever in Children with Dengue Fever. <i>Open Forum Infectious Diseases</i> , 2021, 8, S461-S462.	0.4	1
17	Enhanced MAPK1 Function Causes a Neurodevelopmental Disorder within the RASopathy Clinical Spectrum. <i>American Journal of Human Genetics</i> , 2020, 107, 499-513.	2.6	48
18	Novel progressive acrodysostosis-like skeletal dysplasia, cerebellar atrophy, and ichthyosis. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 2214-2221.	0.7	1

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19	An international telemedicine program for diagnosis of genetic disorders: Partnership of pediatrician and geneticist. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2020, 184, 996-1008.	0.7	9
20	Outcomes of screening for gammopathies in children and adults with Gaucher disease type 1 in a cohort from Brazil and the United States. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2020, 184, 1052-1059.	0.7	4
21	Clinical and molecular analysis of 26 individuals with Noonan syndrome in a reference institution in Colombia. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2020, 184, 1042-1051.	0.7	6
22	Gaucher disease and SARS-CoV-2 infection: Emerging management challenges. <i>Molecular Genetics and Metabolism</i> , 2020, 130, 164-169.	0.5	25
23	Loss- or Gain-of-Function Mutations in ACOX1 Cause Axonal Loss via Different Mechanisms. <i>Neuron</i> , 2020, 106, 589-606.e6.	3.8	71
24	Introduction to the special issue on Clinical Genetics in Latin America. <i>American Journal of Medical Genetics, Part C: Seminars in Medical Genetics</i> , 2020, 184, 873-875.	0.7	0
25	Disruptive variants of <i>CSDE1</i> associate with autism and interfere with neuronal development and synaptic transmission. <i>Science Advances</i> , 2019, 5, eaax2166.	4.7	35
26	Phenotypic and biochemical analysis of an international cohort of individuals with variants in NAA10 and NAA15. <i>Human Molecular Genetics</i> , 2019, 28, 2900-2919.	1.4	46
27	Expanding and Underscoring the Hepatoencephalopathic Phenotype of QIL1/MIC13. <i>Hepatology</i> , 2019, 70, 1066-1070.	3.6	17
28	Mutations in the Neuronal Vesicular SNARE VAMP2 Affect Synaptic Membrane Fusion and Impair Human Neurodevelopment. <i>American Journal of Human Genetics</i> , 2019, 104, 721-730.	2.6	88
29	Substantial pain burden in frequency, intensity, interference and chronicity among children and adults with neurofibromatosis Type 1. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 602-607.	0.7	20
30	Prevalence of pathogenic and likely pathogenic variants in the RASopathy genes in patients who have had panel testing for cardiomyopathy. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 608-614.	0.7	9
31	Correlating liver stiffness with disease severity scoring system (DS3) values in Gaucher disease type 1 (GD1) patients. <i>Molecular Genetics and Metabolism</i> , 2018, 123, 357-363.	0.5	11
32	Vitamin D deficiency and pre-eclampsia in Colombia: PREVitD study. <i>Pregnancy Hypertension</i> , 2018, 14, 240-244.	0.6	20
33	Cardiac Rhabdomyomas in Tuberous Sclerosis Complex. <i>Journal of Pediatrics</i> , 2018, 192, 264-264.e1.	0.9	13
34	The Tatton-Brown-Rahman Syndrome: A clinical study of 55 individuals with de novo constitutive DNMT3A variants. <i>Wellcome Open Research</i> , 2018, 3, 46.	0.9	75
35	The role of IQSEC2 in syndromic intellectual disability: Narrowing the diagnostic odyssey. , 2017, 173, 2814-2820.		14
36	Outcomes of four patients with homocysteine remethylation disorders detected by newborn screening. <i>Genetics in Medicine</i> , 2016, 18, 162-167.	1.1	21

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37	Determination of Anti-Adeno-Associated Viral Vector Neutralizing Antibodies in Patients With Heart Failure in the Cardiovascular Foundation of Colombia (ANVIAS): Study Protocol. <i>JMIR Research Protocols</i> , 2016, 5, e102.	0.5	6
38	TAF1 Variants Are Associated with Dysmorphic Features, Intellectual Disability, and Neurological Manifestations. <i>American Journal of Human Genetics</i> , 2015, 97, 922-932.	2.6	101
39	Neurological and cardiac responses after treatment with miglustat and a ketogenic diet in a patient with Sandhoff disease. <i>European Journal of Medical Genetics</i> , 2015, 58, 180-183.	0.7	16
40	The Use of Magnetic Resonance Imaging Screening for Optic Pathway Gliomas in Children with Neurofibromatosis Type 1. <i>Journal of Pediatrics</i> , 2015, 167, 851-856.e1.	0.9	88
41	Mutations in SLC25A46, encoding a UGO1-like protein, cause an optic atrophy spectrum disorder. <i>Nature Genetics</i> , 2015, 47, 926-932.	9.4	166
42	CNS, lung, and lymph node involvement in Gaucher disease type 3 after 11years of therapy: Clinical, histopathologic, and biochemical findings. <i>Molecular Genetics and Metabolism</i> , 2015, 114, 233-241.	0.5	54
43	Globus pallidus involvement as initial presentation of methylmalonic acidemia. <i>Movement Disorders</i> , 2014, 29, 870-870.	2.2	4
44	Endothelial dysfunction and preeclampsia: role of oxidative stress. <i>Frontiers in Physiology</i> , 2014, 5, 372.	1.3	306
45	Neuronopathic lysosomal storage diseases: Clinical and pathologic findings. <i>Developmental Disabilities Research Reviews</i> , 2013, 17, 226-246.	2.9	12
46	Neurofibroma-associated macrophages play roles in tumor growth and response to pharmacological inhibition. <i>Acta Neuropathologica</i> , 2013, 125, 159-168.	3.9	104
47	Malonyl Coenzyme A Decarboxylase Deficiency: Early Dietary Restriction and Time Course of Cardiomyopathy. <i>Pediatrics</i> , 2012, 130, e456-e460.	1.0	26
48	Genetic Causes of Macroglossia: Diagnostic Approach. <i>Pediatrics</i> , 2012, 129, e431-e437.	1.0	38
49	Recurrent pancreatitis in ornithine transcarbamylase deficiency. <i>Molecular Genetics and Metabolism</i> , 2012, 106, 482-484.	0.5	3
50	Pediatric Plexiform Neurofibromas: Impact on Morbidity and Mortality in Neurofibromatosis Type 1. <i>Journal of Pediatrics</i> , 2012, 160, 461-467.	0.9	122
51	Cardiac Disease in Methylmalonic Acidemia. <i>Journal of Pediatrics</i> , 2011, 159, 862-864.	0.9	40
52	Severe cervical scoliosis in the fetus. <i>Prenatal Diagnosis</i> , 2011, 31, 1198-1202.	1.1	6
53	Lethal presentation of neurofibromatosis and Noonan syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2011, 155, 1360-1366.	0.7	17
54	Prevalence of Genetic Diagnoses in a Cohort With Valvar Pulmonary Stenosis. <i>Circulation Genomic and Precision Medicine</i> , 0, , .	1.6	0