Carlos E Prada

List of Publications by Year
in descending order

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54 papers	1,774 citations	430874 18 h-index	302126 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?. Molecular Genetics and Metabolism, 2022, 136, 4-21.	1.1	18
2	Craniosynostosis is a feature of Costello syndrome. American Journal of Medical Genetics, Part A, 2022, 188, 1280-1286.	1.2	3
3	NAA10 p.(N101K) disrupts N-terminal acetyltransferase complex NatA and is associated with developmental delay and hemihypertrophy. European Journal of Human Genetics, 2021, 29, 280-288.	2.8	7
4	CHARGE syndrome in the era of molecular diagnosis: Similar outcomes in those without coloboma or choanal atresia. European Journal of Medical Genetics, 2021, 64, 104103.	1.3	4
5	DLG4-related synaptopathy: a new rare brain disorder. Genetics in Medicine, 2021, 23, 888-899.	2.4	16
6	Everolimus for severe arrhythmias in tuberous sclerosis complex related cardiac rhabdomyomas. American Journal of Medical Genetics, Part A, 2021, 185, 1525-1531.	1.2	6
7	POLRMT mutations impair mitochondrial transcription causing neurological disease. Nature Communications, 2021, 12, 1135.	12.8	21
8	UBA2 variants underlie a recognizable syndrome with variable aplasia cutis congenita and ectrodactyly. Genetics in Medicine, 2021, 23, 1624-1635.	2.4	7
9	Generation of a Mouse Model to Study the Noonan Syndrome Gene Lztr1 in the Telencephalon. Frontiers in Cell and Developmental Biology, 2021, 9, 673995.	3.7	1
10	Hyperleucinosis during infections in maple syrup urine disease post liver transplantation. Molecular Genetics and Metabolism Reports, 2021, 27, 100763.	1.1	3
11	Neutropenia and intellectual disability are hallmarks of biallelic and de novo CLPB deficiency. Genetics in Medicine, 2021, 23, 1705-1714.	2.4	22
12	A role for sustained MAPK activity in the mouse ventral telencephalon. Developmental Biology, 2021, 476, 137-147.	2.0	6
13	Molecular characterization of mucopolysaccharidosis type <scp>IVA</scp> patients in the Andean region of Colombia. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2021, 187, 388-395.	1.6	7
14	Acute liver dysfunction with delayed peak of serum aminotransferase levels as a presentation of ornithine transcarbamylase deficiency in females. American Journal of Medical Genetics, Part A, 2021, 185, 909-915.	1.2	0
15	A recurrent, de novo pathogenic variant in ARPC4 disrupts actin filament formation and causes a neurodevelopmental disorder with microcephaly and speech delay. Human Genetics and Genomics Advances, 2021, 3, 100072.	1.7	4
16	725. Complete Blood Count Values Vary in Degree of Change with Day of Fever in Children with Dengue Fever. Open Forum Infectious Diseases, 2021, 8, S461-S462.	0.9	1
17	Enhanced MAPK1 Function Causes a Neurodevelopmental Disorder within the RASopathy Clinical Spectrum. American Journal of Human Genetics, 2020, 107, 499-513.	6.2	48
18	Novel progressive acrodysostosisâ€like skeletal dysplasia, cerebellar atrophy, and ichthyosis. American Journal of Medical Genetics, Part A, 2020, 182, 2214-2221.	1.2	1

#	Article	IF	Citations
19	An international telemedicine program for diagnosis of genetic disorders: Partnership of pediatrician and geneticist. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2020, 184, 996-1008.	1.6	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. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2020, 184, 1052-1059.	1.6	4
21	Clinical and molecular analysis of 26 individuals with Noonan syndrome in a reference institution in Colombia. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2020, 184, 1042-1051.	1.6	6
22	Gaucher disease and SARS-CoV-2 infection: Emerging management challenges. Molecular Genetics and Metabolism, 2020, 130, 164-169.	1.1	25
23	Loss- or Gain-of-Function Mutations in ACOX1 Cause Axonal Loss via Different Mechanisms. Neuron, 2020, 106, 589-606.e6.	8.1	71
24	Introduction to the special issue on Clinical Genetics in Latin America. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2020, 184, 873-875.	1.6	0
25	Disruptive variants of CSDE1 associate with autism and interfere with neuronal development and synaptic transmission. Science Advances, 2019, 5, eaax2166.	10.3	35
26	Phenotypic and biochemical analysis of an international cohort of individuals with variants in NAA10 and NAA15. Human Molecular Genetics, 2019, 28, 2900-2919.	2.9	46
27	Expanding and Underscoring the Hepatoâ€Encephalopathic Phenotype of QIL1/MIC13. Hepatology, 2019, 70, 1066-1070.	7.3	17
28	Mutations in the Neuronal Vesicular SNARE VAMP2 Affect Synaptic Membrane Fusion and Impair Human Neurodevelopment. American Journal of Human Genetics, 2019, 104, 721-730.	6.2	88
29	Substantial pain burden in frequency, intensity, interference and chronicity among children and adults with neurofibromatosis Type 1. American Journal of Medical Genetics, Part A, 2019, 179, 602-607.	1.2	20
30	Prevalence of pathogenic and likely pathogenic variants in the RASopathy genes in patients who have had panel testing for cardiomyopathy. American Journal of Medical Genetics, Part A, 2019, 179, 608-614.	1.2	9
31	Correlating liver stiffness with disease severity scoring system (DS3) values in Gaucher disease type 1 (GD1) patients. Molecular Genetics and Metabolism, 2018, 123, 357-363.	1.1	11
32	Vitamin D deficiency and pre-eclampsia in Colombia: PREVitD study. Pregnancy Hypertension, 2018, 14, 240-244.	1.4	20
33	Cardiac Rhabdomyomas in Tuberous Sclerosis Complex. Journal of Pediatrics, 2018, 192, 264-264.e1.	1.8	13
34	The Tatton-Brown-Rahman Syndrome: A clinical study of 55 individuals with de novo constitutive DNMT3A variants. Wellcome Open Research, 2018, 3, 46.	1.8	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. Genetics in Medicine, 2016, 18, 162-167.	2.4	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. JMIR Research Protocols, 2016, 5, e102.	1.0	6
38	TAF1 Variants Are Associated with Dysmorphic Features, Intellectual Disability, and Neurological Manifestations. American Journal of Human Genetics, 2015, 97, 922-932.	6.2	101
39	Neurological and cardiac responses after treatment with miglustat and a ketogenic diet in a patient with Sandhoff disease. European Journal of Medical Genetics, 2015, 58, 180-183.	1.3	16
40	The Use of Magnetic Resonance Imaging Screening for Optic Pathway Gliomas in Children with Neurofibromatosis Type 1. Journal of Pediatrics, 2015, 167, 851-856.e1.	1.8	88
41	Mutations in SLC25A46, encoding a UGO1-like protein, cause an optic atrophy spectrum disorder. Nature Genetics, 2015, 47, 926-932.	21.4	166
42	CNS, lung, and lymph node involvement in Gaucher disease type 3 after 11years of therapy: Clinical, histopathologic, and biochemical findings. Molecular Genetics and Metabolism, 2015, 114, 233-241.	1.1	54
43	Globus pallidus involvement as initial presentation of methylmalonic acidemia. Movement Disorders, 2014, 29, 870-870.	3.9	4
44	Endothelial dysfunction and preeclampsia: role of oxidative stress. Frontiers in Physiology, 2014, 5, 372.	2.8	306
45	Neuronopathic lysosomal storage diseases: Clinical and pathologic findings. Developmental Disabilities Research Reviews, 2013, 17, 226-246.	2.9	12
46	Neurofibroma-associated macrophages play roles in tumor growth and response to pharmacological inhibition. Acta Neuropathologica, 2013, 125, 159-168.	7.7	104
47	Malonyl Coenzyme A Decarboxylase Deficiency: Early Dietary Restriction and Time Course of Cardiomyopathy. Pediatrics, 2012, 130, e456-e460.	2.1	26
48	Genetic Causes of Macroglossia: Diagnostic Approach. Pediatrics, 2012, 129, e431-e437.	2.1	38
49	Recurrent pancreatitis in ornithine transcarbamylase deficiency. Molecular Genetics and Metabolism, 2012, 106, 482-484.	1.1	3
50	Pediatric Plexiform Neurofibromas: Impact on Morbidity and Mortality in Neurofibromatosis Type 1. Journal of Pediatrics, 2012, 160, 461-467.	1.8	122
51	Cardiac Disease in Methylmalonic Acidemia. Journal of Pediatrics, 2011, 159, 862-864.	1.8	40
52	Severe cervical scoliosis in the fetus. Prenatal Diagnosis, 2011, 31, 1198-1202.	2.3	6
53	Lethal presentation of neurofibromatosis and Noonan syndrome. American Journal of Medical Genetics, Part A, 2011, 155, 1360-1366.	1.2	17
54	Prevalence of Genetic Diagnoses in a Cohort With Valvar Pulmonary Stenosis. Circulation Genomic and Precision Medicine, 0, , .	3.6	0