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	Endothelial dysfunction and preeclampsia: role of oxidative stress. Frontiers in Physiology, 2014, 5, 372.	2.8	306
2	Mutations in SLC25A46, encoding a UGO1-like protein, cause an optic atrophy spectrum disorder. Nature Genetics, 2015, 47, 926-932.	21.4	166
3	Pediatric Plexiform Neurofibromas: Impact on Morbidity and Mortality in Neurofibromatosis Type 1. Journal of Pediatrics, 2012, 160, 461-467.	1.8	122
4	Neurofibroma-associated macrophages play roles in tumor growth and response to pharmacological inhibition. Acta Neuropathologica, 2013, 125, 159-168.	7.7	104
5	TAF1 Variants Are Associated with Dysmorphic Features, Intellectual Disability, and Neurological Manifestations. American Journal of Human Genetics, 2015, 97, 922-932.	6.2	101
6	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
7	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
8	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
9	Loss- or Gain-of-Function Mutations in ACOX1 Cause Axonal Loss via Different Mechanisms. Neuron, 2020, 106, 589-606.e6.	8.1	71
10	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
11	Enhanced MAPK1 Function Causes a Neurodevelopmental Disorder within the RASopathy Clinical Spectrum. American Journal of Human Genetics, 2020, 107, 499-513.	6.2	48
12	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
13	Cardiac Disease in Methylmalonic Acidemia. Journal of Pediatrics, 2011, 159, 862-864.	1.8	40
14	Genetic Causes of Macroglossia: Diagnostic Approach. Pediatrics, 2012, 129, e431-e437.	2.1	38
15	Disruptive variants of CSDE1 associate with autism and interfere with neuronal development and synaptic transmission. Science Advances, 2019, 5, eaax2166.	10.3	35
16	Malonyl Coenzyme A Decarboxylase Deficiency: Early Dietary Restriction and Time Course of Cardiomyopathy. Pediatrics, 2012, 130, e456-e460.	2.1	26
17	Gaucher disease and SARS-CoV-2 infection: Emerging management challenges. Molecular Genetics and Metabolism, 2020, 130, 164-169.	1.1	25
18	Neutropenia and intellectual disability are hallmarks of biallelic and de novo CLPB deficiency. Genetics in Medicine, 2021, 23, 1705-1714.	2.4	22

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19	Outcomes of four patients with homocysteine remethylation disorders detected by newborn screening. Genetics in Medicine, 2016, 18, 162-167.	2.4	21
20	POLRMT mutations impair mitochondrial transcription causing neurological disease. Nature Communications, 2021, 12, 1135.	12.8	21
21	Vitamin D deficiency and pre-eclampsia in Colombia: PREVitD study. Pregnancy Hypertension, 2018, 14, 240-244.	1.4	20
22	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
23	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
24	Lethal presentation of neurofibromatosis and Noonan syndrome. American Journal of Medical Genetics, Part A, 2011, 155, 1360-1366.	1.2	17
25	Expanding and Underscoring the Hepatoâ€Encephalopathic Phenotype of QIL1/MIC13. Hepatology, 2019, 70, 1066-1070.	7.3	17
26	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
27	DLG4-related synaptopathy: a new rare brain disorder. Genetics in Medicine, 2021, 23, 888-899.	2.4	16
28	The role of IQSEC2 in syndromic intellectual disability: Narrowing the diagnostic odyssey. , 2017, 173, 2814-2820.		14
29	Cardiac Rhabdomyomas in Tuberous Sclerosis Complex. Journal of Pediatrics, 2018, 192, 264-264.e1.	1.8	13
30	Neuronopathic lysosomal storage diseases: Clinical and pathologic findings. Developmental Disabilities Research Reviews, 2013, 17, 226-246.	2.9	12
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	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
33	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
34	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
35	UBA2 variants underlie a recognizable syndrome with variable aplasia cutis congenita and ectrodactyly. Genetics in Medicine, 2021, 23, 1624-1635.	2.4	7
36	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

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37	Severe cervical scoliosis in the fetus. Prenatal Diagnosis, 2011, 31, 1198-1202.	2.3	6
38	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
39	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
40	A role for sustained MAPK activity in the mouse ventral telencephalon. Developmental Biology, 2021, 476, 137-147.	2.0	6
41	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
42	Globus pallidus involvement as initial presentation of methylmalonic acidemia. Movement Disorders, 2014, 29, 870-870.	3.9	4
43	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
44	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
45	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
46	Recurrent pancreatitis in ornithine transcarbamylase deficiency. Molecular Genetics and Metabolism, 2012, 106, 482-484.	1.1	3
47	Hyperleucinosis during infections in maple syrup urine disease post liver transplantation. Molecular Genetics and Metabolism Reports, 2021, 27, 100763.	1.1	3
48	Craniosynostosis is a feature of Costello syndrome. American Journal of Medical Genetics, Part A, 2022, 188, 1280-1286.	1.2	3
49	Novel progressive acrodysostosisâ€like skeletal dysplasia, cerebellar atrophy, and ichthyosis. American Journal of Medical Genetics, Part A, 2020, 182, 2214-2221.	1.2	1
50	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
51	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
52	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
53	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
54	Prevalence of Genetic Diagnoses in a Cohort With Valvar Pulmonary Stenosis. Circulation Genomic and Precision Medicine, 0, , .	3.6	0