Carlos A Bacino

List of Publications by Year
in descending order

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88 papers	4,062 citations	126907 33 h-index	133252 59 g-index
90 all docs	90 docs citations	90 times ranked	8237 citing authors

#	Article	IF	Citations
1	Pharmacokinetics and Exposureâ€“Response of Vosoritide in Children with Achondroplasia. Clinical Pharmacokinetics, 2022, 61, 263-280.	3.5	15
2	Stankiewicz-Isidor syndrome: expanding the clinical and molecular phenotype. Genetics in Medicine, 2022, 24, 179-191.	2.4	9
3	Delineation of a novel neurodevelopmental syndrome associated with PAX5 haploinsufficiency. Human Mutation, 2022, 43, 461-470.	2.5	5
4	A multidisciplinary approach and consensus statement to establish standards of care for Angelman syndrome. Molecular Genetics & Genomic Medicine, 2022, 10, e1843.	1.2	14
5	PRUNE1 c.<scp>933G</scp>>A synonymous variant induces exon 7 skipping, disrupts the <scp>DHHA2</scp> domain, and leads to an atypical <scp>NMIHBA</scp> syndrome presentation: Case report and review of the literature. American Journal of Medical Genetics, Part A, 2022, 188, 1868-1874.	1.2	2
6	A novel, de novo intronic variant in <scp>POGZ</scp> causes <scp>Whiteâ€“Sutton</scp> syndrome. American Journal of Medical Genetics, Part A, 2022, 188, 2198-2203.	1.2	4
7	Phenotypic and genetic spectrum of ATP6V1A encephalopathy: a disorder of lysosomal homeostasis. Brain, 2022, 145, 2687-2703.	7.6	11
8	UBR7 functions with UBR5 in the Notch signaling pathway and is involved in a neurodevelopmental syndrome with epilepsy, ptosis, and hypothyroidism. American Journal of Human Genetics, 2021, 108, 134-147.	6.2	15
9	Rare deleterious mutations of HNRNP genes result in shared neurodevelopmental disorders. Genome Medicine, 2021, 13, 63.	8.2	50
10	Persistent and Stable Growth Promoting Effects of Vosoritide in Children With Achondroplasia for up to 2 Years: Results From the Ongoing Phase 3 Extension Study. Journal of the Endocrine Society, 2021, 5, A670-A671.	0.2	2
11	Safe and persistent growth-promoting effects of vosoritide in children with achondroplasia: 2-year results from an open-label, phase 3 extension study. Genetics in Medicine, 2021, 23, 2443-2447.	2.4	36
12	Evaluating Sleep Disturbances in Children With Rare Genetic Neurodevelopmental Syndromes. Pediatric Neurology, 2021, 123, 30-37.	2.1	21
13	Evidence of feedback regulation of C-type natriuretic peptide during Vosoritide therapy in Achondroplasia. Scientific Reports, 2021, 11, 24278.	3.3	2
14	Widening of the genetic and clinical spectrum of Lambâ€“Shaffer syndrome, a neurodevelopmental disorder due to SOX5 haploinsufficiency. Genetics in Medicine, 2020, 22, 524-537.	2.4	21
15	Agenesis of the corpus callosum and hepatoblastoma. American Journal of Medical Genetics, Part A, 2020, 182, 224-228.	1.2	1
16	Once-daily, subcutaneous vosoritide therapy in children with achondroplasia: a randomised, double-blind, phase 3, placebo-controlled, multicentre trial. Lancet, The, 2020, 396, 684-692.	13.7	92
17	Sudden infant death with dysgenesis of the testes syndrome in anonâ€Amishinfant: A case report. American Journal of Medical Genetics, Part A, 2020, 182, 2751-2754.	1.2	4
18	De Novo Variants in CDK19 Are Associated with a Syndrome Involving Intellectual Disability and Epileptic Encephalopathy. American Journal of Human Genetics, 2020, 106, 717-725.	6.2	23

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19	Mutations in the KIF21B kinesin gene cause neurodevelopmental disorders through imbalanced canonical motor activity. Nature Communications, 2020, 11, 2441.	12.8	37
20	Parental somatic mosaicism for CNV deletions â€“ A need for more sensitive and precise detection methods in clinical diagnostics settings. Genomics, 2020, 112, 2937-2941.	2.9	14
21	CNVs cause autosomal recessive genetic diseases with or without involvement of SNV/indels. Genetics in Medicine, 2020, 22, 1633-1641.	2.4	36
22	SAT-LB18 A Randomized Controlled Trial of Vosoritide in Children With Achondroplasia. Journal of the Endocrine Society, 2020, 4, .	0.2	0
23	BAZ2B haploinsufficiency as a cause of developmental delay, intellectual disability, and autism spectrum disorder. Human Mutation, 2020, 41, 921-925.	2.5	11
24	Novel deletion of 6p21.31p21.1 associated with laryngeal cleft, developmental delay, dysmorphic features and vascular anomaly. European Journal of Medical Genetics, 2019, 62, 103531.	1.3	4
25	Biallelic and De Novo Variants in DONSON Reveal a Clinical Spectrum of Cell Cycleâ€opathies with Microcephaly, Dwarfism and Skeletal Abnormalities. American Journal of Medical Genetics, Part A, 2019, 179, 2056-2066.	1.2	15
26	Skin fibroblasts of patients with geleophysic dysplasia due to FBN1 mutations have lysosomal inclusions and losartan improves their microfibril deposition defect. Molecular Genetics & Genomic Medicine, 2019, 7, e844.	1.2	8
27	A placebo-controlled trial of folic acid and betaine in identical twins with Angelman syndrome. Orphanet Journal of Rare Diseases, 2019, 14, 232.	2.7	2
28	C-Type Natriuretic Peptide Analogue Therapy in Children with Achondroplasia. New England Journal of Medicine, 2019, 381, 25-35.	27.0	131
29	Copy number variant and runs of homozygosity detection by microarrays enabled more precise molecular diagnoses in 11,020 clinical exome cases. Genome Medicine, 2019, 11, 30.	8.2	42
30	SLC35A2â€CDG: Functional characterization, expanded molecular, clinical, and biochemical phenotypes of 30 unreported Individuals. Human Mutation, 2019, 40, 908-925.	2.5	39
31	Bi-allelic Variants in TONSL Cause SPONASTRIME Dysplasia and a Spectrum of Skeletal Dysplasia Phenotypes. American Journal of Human Genetics, 2019, 104, 422-438.	6.2	27
32	Microdeletions excluding YWHAE and PAFAH1B1 cause a unique leukoencephalopathy: further delineation of the 17p13.3 microdeletion spectrum. Genetics in Medicine, 2019, 21, 1652-1656.	2.4	8
33	Recurrent mosaic MTOR c.5930C > T (p.Thr1977Ile) variant causing megalencephaly, asymmetric polymicrogyria, and cutaneous pigmentary mosaicism: Case report and review of the literature. American Journal of Medical Genetics, Part A, 2019, 179, 475-479.	1.2	11
34	ZMIZ1 Variants Cause a Syndromic Neurodevelopmental Disorder. American Journal of Human Genetics, 2019, 104, 319-330.	6.2	30
35	The expanding neurological phenotype of DNM1L-related disorders. Brain, 2018, 141, e28-e28.	7.6	7
36	A randomized controlled trial of levodopa in patients with Angelman syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 1099-1107.	1.2	18

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37	Effect of Genetic Diagnosis on Patients with Previously Undiagnosed Disease. New England Journal of Medicine, 2018, 379, 2131-2139.	27.0	261
38	Further evidence for the involvement of EFL1 in a Shwachmanâ€“Diamond-like syndrome and expansion of the phenotypic features. Journal of Physical Education and Sports Management, 2018, 4, a003046.	1.2	29
39	Level of residual enzyme activity modulates the phenotype in phosphoglycerate kinase deficiency. Neurology, 2018, 91, e1077-e1082.	1.1	15
40	The phenotypic spectrum of Schaaf-Yang syndrome: 18 new affected individuals from 14 families. Genetics in Medicine, 2017, 19, 45-52.	2.4	94
41	De Novo Disruption of the Proteasome Regulatory Subunit PSMD12 Causes a Syndromic Neurodevelopmental Disorder. American Journal of Human Genetics, 2017, 100, 352-363.	6.2	86
42	An Organismal CNV Mutator Phenotype Restricted to Early Human Development. Cell, 2017, 168, 830-842.e7.	28.9	66
43	Haploinsufficiency for ANKRD11-flanking genes makes the difference between KBG and 16q24.3 microdeletion syndromes: 12 new cases. European Journal of Human Genetics, 2017, 25, 694-701.	2.8	33
44	Lessons learned from additional research analyses of unsolved clinical exome cases. Genome Medicine, 2017, 9, 26.	8.2	184
45	A Syndromic Neurodevelopmental Disorder Caused by De Novo Variants in EBF3. American Journal of Human Genetics, 2017, 100, 128-137.	6.2	96
46	Use of Exome Sequencing for Infants in Intensive Care Units. JAMA Pediatrics, 2017, 171, e173438.	6.2	348
47	High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. American Journal of Human Genetics, 2017, 101, 664-685.	6.2	337
48	De Novo Mutations in SLC25A24 Cause a Craniosynostosis Syndrome with Hypertrichosis, Progeroid Appearance, and Mitochondrial Dysfunction. American Journal of Human Genetics, 2017, 101, 833-843.	6.2	56
49	Haploinsufficiency of the Chromatin Remodeler BPTF Causes Syndromic Developmental and Speech Delay, Postnatal Microcephaly, and Dysmorphic Features. American Journal of Human Genetics, 2017, 101, 503-515.	6.2	61
50	Mutations in Fibronectin Cause a Subtype of Spondylometaphyseal Dysplasia with â€œCorner Fracturesâ€• American Journal of Human Genetics, 2017, 101, 815-823.	6.2	37
51	Universal Prenatal Chromosomal Microarray Analysis: Additive Value and Clinical Dilemmas in Fetuses with a Normal Karyotype. American Journal of Perinatology, 2017, 34, 340-348.	1.4	21
52	Severe Pancytopenia in a Premature Infant. Clinical Pediatrics, 2017, 56, 795-797.	0.8	0
53	Identification of novel candidate disease genes from de novo exonic copy number variants. Genome Medicine, 2017, 9, 83.	8.2	50
54	Clinically severe CACNA1A alleles affect synaptic function and neurodegeneration differentially. PLoS Genetics, 2017, 13, e1006905.	3.5	80

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55	1p13.2 deletion displays clinical features overlapping Noonan syndrome, likely related to NRAS gene haploinsufficiency. Genetics and Molecular Biology, 2016, 39, 349-357.	1.3	5
56	Mechanisms for Complex Chromosomal Insertions. PLoS Genetics, 2016, 12, e1006446.	3.5	45
57	4p16.3 microdeletions and microduplications detected by chromosomal microarray analysis: New insights into mechanisms and critical regions. American Journal of Medical Genetics, Part A, 2016, 170, 2540-2550.	1.2	25
58	5q14.3 deletion neurocutaneous syndrome: Contiguous gene syndrome caused by simultaneous deletion of RASA1 and MEF2C: A progressive disease. American Journal of Medical Genetics, Part A, 2016, 170, 688-693.	1.2	9
59	Pathogenetics of alveolar capillary dysplasia with misalignment of pulmonary veins. Human Genetics, 2016, 135, 569-586.	3.8	85
60	De Novo Truncating Variants in SON Cause Intellectual Disability, Congenital Malformations, and Failure to Thrive. American Journal of Human Genetics, 2016, 99, 720-727.	6.2	45
61	Increased bone turnover, osteoporosis, progressive tibial bowing, fractures, and scoliosis in a patient with a finalâ€exon SATB2 frameshift mutation. American Journal of Medical Genetics, Part A, 2016, 170, 3028-3032.	1.2	16
62	Recurrent Muscle Weakness with Rhabdomyolysis, Metabolic Crises, and Cardiac Arrhythmia Due to Bi-allelic TANGO2 Mutations. American Journal of Human Genetics, 2016, 98, 347-357.	6.2	98
63	A homozygous mutation in PEX16 identified by whole-exome sequencing ending a diagnostic odyssey. Molecular Genetics and Metabolism Reports, 2015, 5, 15-18.	1.1	16
64	Alu-mediated diverse and complex pathogenic copy-number variants within human chromosome 17 at p13.3. Human Molecular Genetics, 2015, 24, 4061-4077.	2.9	83
65	Neurodevelopmental and neurobehavioral characteristics in males and females with CDKL5 duplications. European Journal of Human Genetics, 2015, 23, 915-921.	2.8	32
66	Autosomal-Dominant Multiple Pterygium Syndrome Is Caused by Mutations in MYH3. American Journal of Human Genetics, 2015, 96, 841-849.	6.2	55
67	Delineation of candidate genes responsible for structural brain abnormalities in patients with terminal deletions of chromosome 6q27. European Journal of Human Genetics, 2015, 23, 54-60.	2.8	45
68	Heterozygous De Novo and Inherited Mutations in the Smooth Muscle Actin (ACTG2) Gene Underlie Megacystis-Microcolon-Intestinal Hypoperistalsis Syndrome. PLoS Genetics, 2014, 10, e1004258.	3.5	122
69	Isolated Fetal Macrodactyly: Phenotypic and Genetic Disparities in Mosaic Overgrowth Syndrome. Journal of Ultrasound in Medicine, 2014, 33, 1305-1307.	1.7	8
70	Combined array CGH plus SNP genome analyses in a single assay for optimized clinical testing. European Journal of Human Genetics, 2014, 22, 79-87.	2.8	112
71	Parental Somatic Mosaicism Is Underrecognized and Influences Recurrence Risk of Genomic Disorders. American Journal of Human Genetics, 2014, 95, 173-182.	6.2	219
72	Somatic mosaicism detected by exon-targeted, high-resolution aCGH in 10â€‰362 consecutive cases. European Journal of Human Genetics, 2014, 22, 969-978.	2.8	51

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73	Etiopathogenesis of equinovarus foot malformations. European Journal of Medical Genetics, 2014, 57, 473-479.	1.3	43
74	Lysinuric protein intolerance presenting with multiple fractures. Molecular Genetics and Metabolism Reports, 2014, 1, 176-183.	1.1	20
75	TM4SF20 Ancestral Deletion and Susceptibility to a Pediatric Disorder of Early Language Delay and Cerebral White Matter Hyperintensities. American Journal of Human Genetics, 2013, 93, 197-210.	6.2	43
76	WDR35 mutation in siblings with Sensenbrenner syndrome: A ciliopathy with variable phenotype. American Journal of Medical Genetics, Part A, 2012, 158A, 2917-2924.	1.2	40
77	WDR62 missense mutation in a consanguineous family with primary microcephaly. American Journal of Medical Genetics, Part A, 2012, 158A, 622-625.	1.2	12
78	Introductory comments on special sectionâ€"Genomic microduplications: When adding may equal subtracting. American Journal of Medical Genetics, Part A, 2010, 152A, 1063-1065.	1.2	2
79	ATR-16 Due to a De Novo Complex Rearrangement of Chromosome 16. Hemoglobin, 2005, 29, 141-150.	0.8	7
80	EGR2 mutation R359W causes a spectrum of Dejerine-Sottas neuropathy. Neurogenetics, 2001, 3, 153-157.	1.4	60
81	Identification of Y chromatin directly in gonadal tissue by fluorescence in situ hybridization (FISH): Significance for Ullrich-Turner syndrome screening in the cytogenetics laboratory. , 2000, 91, 377-382.		9
82	Detection of a cryptic translocation in a family with mental retardation using FISH and telomere region-specific probes. American Journal of Medical Genetics Part A, 2000, 92, 250-255.	2.4	38
83	Terminal osseous dysplasia and pigmentary defects: Clinical characterization of a novel male lethal X-linked syndrome. American Journal of Medical Genetics Part A, 2000, 94, 102-112.	2.4	25
84	Trisomy 16q in a female newborn with a de novo X;16 translocation and hypoplastic left heart. , 1999, 82, 128-131.		20
85	Normal expression of the Fanconi anemia proteins FAA and FAC and sensitivity to mitomycin C in two Patients with Seckel syndrome. , 1999, 83, 388-391.		14
86	Caution: Telomere crossing. American Journal of Medical Genetics Part A, 1999, 87, 278-280.	2.4	17
87	The Pointer syndrome: A new syndrome with skeletal abnormalities, camptodactyly, facial anomalies, and feeding difficulties. , 1997, 68, 225-230.		6
88	Severe clinical phenotype due to an interstitial deletion of the short arm of chromosome 1: A brief review. , 1997, 71, 189-193.		14