

Carlos A Bacino

List of Publications by Year in descending order

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Version: 2024-02-01

88
papers

4,062
citations

145106

33
h-index

156644

58
g-index

90
all docs

90
docs citations

90
times ranked

8828
citing authors

#	ARTICLE	IF	CITATIONS
1	Pharmacokinetics and Exposureâ€“Response of Vosoritide in Children with Achondroplasia. <i>Clinical Pharmacokinetics</i> , 2022, 61, 263-280.	1.6	15
2	Stankiewicz-Isidor syndrome: expanding the clinical and molecular phenotype. <i>Genetics in Medicine</i> , 2022, 24, 179-191.	1.1	9
3	Delineation of a novel neurodevelopmental syndrome associated with <i>PAX5</i> haploinsufficiency. <i>Human Mutation</i> , 2022, 43, 461-470.	1.1	5
4	A multidisciplinary approach and consensus statement to establish standards of care for Angelman syndrome. <i>Molecular Genetics & Genomic Medicine</i> , 2022, 10, e1843.	0.6	14
5	<i>PRUNE1</i> c.933G>A synonymous variant induces exon 7 skipping, disrupts the <i>DHHA2</i> domain, and leads to an atypical <i>NMIHBA</i> syndrome presentation: Case report and review of the literature. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 1868-1874.	0.7	2
6	A novel, de novo intronic variant in <i>POGZ</i> causes <i>Whiteâ€“Sutton</i> syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 2198-2203.	0.7	4
7	Phenotypic and genetic spectrum of ATP6V1A encephalopathy: a disorder of lysosomal homeostasis. <i>Brain</i> , 2022, 145, 2687-2703.	3.7	11
8	UBR7 functions with UBR5 in the Notch signaling pathway and is involved in a neurodevelopmental syndrome with epilepsy, ptosis, and hypothyroidism. <i>American Journal of Human Genetics</i> , 2021, 108, 134-147.	2.6	15
9	Rare deleterious mutations of HNRNP genes result in shared neurodevelopmental disorders. <i>Genome Medicine</i> , 2021, 13, 63.	3.6	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. <i>Journal of the Endocrine Society</i> , 2021, 5, A670-A671.	0.1	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. <i>Genetics in Medicine</i> , 2021, 23, 2443-2447.	1.1	36
12	Evaluating Sleep Disturbances in Children With Rare Genetic Neurodevelopmental Syndromes. <i>Pediatric Neurology</i> , 2021, 123, 30-37.	1.0	21
13	Evidence of feedback regulation of C-type natriuretic peptide during Vosoritide therapy in Achondroplasia. <i>Scientific Reports</i> , 2021, 11, 24278.	1.6	2
14	Widening of the genetic and clinical spectrum of <i>Lambâ€“Shaffer</i> syndrome, a neurodevelopmental disorder due to <i>SOX5</i> haploinsufficiency. <i>Genetics in Medicine</i> , 2020, 22, 524-537.	1.1	21
15	Agenesis of the corpus callosum and hepatoblastoma. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 224-228.	0.7	1
16	Once-daily, subcutaneous vosoritide therapy in children with achondroplasia: a randomised, double-blind, phase 3, placebo-controlled, multicentre trial. <i>Lancet, The</i> , 2020, 396, 684-692.	6.3	92
17	Sudden infant death with dysgenesis of the testes syndrome in an Amish infant: A case report. <i>American Journal of Medical Genetics, Part A</i> , 2020, 182, 2751-2754.	0.7	4
18	De Novo Variants in <i>CDK19</i> Are Associated with a Syndrome Involving Intellectual Disability and Epileptic Encephalopathy. <i>American Journal of Human Genetics</i> , 2020, 106, 717-725.	2.6	23

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19	Mutations in the KIF21B kinesin gene cause neurodevelopmental disorders through imbalanced canonical motor activity. <i>Nature Communications</i> , 2020, 11, 2441.	5.8	37
20	Parental somatic mosaicism for CNV deletions – A need for more sensitive and precise detection methods in clinical diagnostics settings. <i>Genomics</i> , 2020, 112, 2937-2941.	1.3	14
21	CNVs cause autosomal recessive genetic diseases with or without involvement of SNV/indels. <i>Genetics in Medicine</i> , 2020, 22, 1633-1641.	1.1	36
22	SAT-LB18 A Randomized Controlled Trial of Vosoritide in Children With Achondroplasia. <i>Journal of the Endocrine Society</i> , 2020, 4, .	0.1	0
23	<i>BAZ2B</i> haploinsufficiency as a cause of developmental delay, intellectual disability, and autism spectrum disorder. <i>Human Mutation</i> , 2020, 41, 921-925.	1.1	11
24	Novel deletion of 6p21.31p21.1 associated with laryngeal cleft, developmental delay, dysmorphic features and vascular anomaly. <i>European Journal of Medical Genetics</i> , 2019, 62, 103531.	0.7	4
25	Biallelic and <i>De Novo</i> Variants in <i>DONSON</i> Reveal a Clinical Spectrum of Cell Cycleopathies with Microcephaly, Dwarfism and Skeletal Abnormalities. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 2056-2066.	0.7	15
26	Skin fibroblasts of patients with geleophysic dysplasia due to <i>FBN1</i> mutations have lysosomal inclusions and losartan improves their microfibril deposition defect. <i>Molecular Genetics & Genomic Medicine</i> , 2019, 7, e844.	0.6	8
27	A placebo-controlled trial of folic acid and betaine in identical twins with Angelman syndrome. <i>Orphanet Journal of Rare Diseases</i> , 2019, 14, 232.	1.2	2
28	C-Type Natriuretic Peptide Analogue Therapy in Children with Achondroplasia. <i>New England Journal of Medicine</i> , 2019, 381, 25-35.	13.9	131
29	Copy number variant and runs of homozygosity detection by microarrays enabled more precise molecular diagnoses in 11,020 clinical exome cases. <i>Genome Medicine</i> , 2019, 11, 30.	3.6	42
30	SLC35A2 ^{CDG} : Functional characterization, expanded molecular, clinical, and biochemical phenotypes of 30 unreported Individuals. <i>Human Mutation</i> , 2019, 40, 908-925.	1.1	39
31	Bi-allelic Variants in TONSL Cause SPONASTRIME Dysplasia and a Spectrum of Skeletal Dysplasia Phenotypes. <i>American Journal of Human Genetics</i> , 2019, 104, 422-438.	2.6	27
32	Microdeletions excluding YWHAE and PFAFH1B1 cause a unique leukoencephalopathy: further delineation of the 17p13.3 microdeletion spectrum. <i>Genetics in Medicine</i> , 2019, 21, 1652-1656.	1.1	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. <i>American Journal of Medical Genetics, Part A</i> , 2019, 179, 475-479.	0.7	11
34	ZMIZ1 Variants Cause a Syndromic Neurodevelopmental Disorder. <i>American Journal of Human Genetics</i> , 2019, 104, 319-330.	2.6	30
35	The expanding neurological phenotype of DNMI1-related disorders. <i>Brain</i> , 2018, 141, e28-e28.	3.7	7
36	A randomized controlled trial of levodopa in patients with Angelman syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 1099-1107.	0.7	18

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37	Effect of Genetic Diagnosis on Patients with Previously Undiagnosed Disease. <i>New England Journal of Medicine</i> , 2018, 379, 2131-2139.	13.9	261
38	Further evidence for the involvement of <i>EFL1</i> in a Shwachmanâ€Diamond-like syndrome and expansion of the phenotypic features. <i>Journal of Physical Education and Sports Management</i> , 2018, 4, a003046.	0.5	29
39	Level of residual enzyme activity modulates the phenotype in phosphoglycerate kinase deficiency. <i>Neurology</i> , 2018, 91, e1077-e1082.	1.5	15
40	The phenotypic spectrum of Schaaf-Yang syndrome: 18 new affected individuals from 14 families. <i>Genetics in Medicine</i> , 2017, 19, 45-52.	1.1	94
41	De Novo Disruption of the Proteasome Regulatory Subunit PSMD12 Causes a Syndromic Neurodevelopmental Disorder. <i>American Journal of Human Genetics</i> , 2017, 100, 352-363.	2.6	86
42	An Organismal CNV Mutator Phenotype Restricted to Early Human Development. <i>Cell</i> , 2017, 168, 830-842.e7.	13.5	66
43	Haploinsufficiency for ANKRD11-flanking genes makes the difference between KBC and 16q24.3 microdeletion syndromes: 12 new cases. <i>European Journal of Human Genetics</i> , 2017, 25, 694-701.	1.4	33
44	Lessons learned from additional research analyses of unsolved clinical exome cases. <i>Genome Medicine</i> , 2017, 9, 26.	3.6	184
45	A Syndromic Neurodevelopmental Disorder Caused by De Novo Variants in EBF3. <i>American Journal of Human Genetics</i> , 2017, 100, 128-137.	2.6	96
46	Use of Exome Sequencing for Infants in Intensive Care Units. <i>JAMA Pediatrics</i> , 2017, 171, e173438.	3.3	348
47	High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. <i>American Journal of Human Genetics</i> , 2017, 101, 664-685.	2.6	337
48	De Novo Mutations in SLC25A24 Cause a Craniosynostosis Syndrome with Hypertrichosis, Progeroid Appearance, and Mitochondrial Dysfunction. <i>American Journal of Human Genetics</i> , 2017, 101, 833-843.	2.6	56
49	Haploinsufficiency of the Chromatin Remodeler BPTF Causes Syndromic Developmental and Speech Delay, Postnatal Microcephaly, and Dysmorphic Features. <i>American Journal of Human Genetics</i> , 2017, 101, 503-515.	2.6	61
50	Mutations in Fibronectin Cause a Subtype of Spondylometaphyseal Dysplasia with â€œCorner Fracturesâ€ American Journal of Human Genetics, 2017, 101, 815-823.	2.6	37
51	Universal Prenatal Chromosomal Microarray Analysis: Additive Value and Clinical Dilemmas in Fetuses with a Normal Karyotype. <i>American Journal of Perinatology</i> , 2017, 34, 340-348.	0.6	21
52	Severe Pancytopenia in a Premature Infant. <i>Clinical Pediatrics</i> , 2017, 56, 795-797.	0.4	0
53	Identification of novel candidate disease genes from de novo exonic copy number variants. <i>Genome Medicine</i> , 2017, 9, 83.	3.6	50
54	Clinically severe CACNA1A alleles affect synaptic function and neurodegeneration differentially. <i>PLoS Genetics</i> , 2017, 13, e1006905.	1.5	80

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55	1p13.2 deletion displays clinical features overlapping Noonan syndrome, likely related to NRAS gene haploinsufficiency. <i>Genetics and Molecular Biology</i> , 2016, 39, 349-357.	0.6	5
56	Mechanisms for Complex Chromosomal Insertions. <i>PLoS Genetics</i> , 2016, 12, e1006446.	1.5	45
57	4p16.3 microdeletions and microduplications detected by chromosomal microarray analysis: New insights into mechanisms and critical regions. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 2540-2550.	0.7	25
58	5q14.3 deletion neurocutaneous syndrome: Contiguous gene syndrome caused by simultaneous deletion of <i>RASA1</i> and <i>MEF2C</i> : A progressive disease. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 688-693.	0.7	9
59	Pathogenetics of alveolar capillary dysplasia with misalignment of pulmonary veins. <i>Human Genetics</i> , 2016, 135, 569-586.	1.8	85
60	De Novo Truncating Variants in SON Cause Intellectual Disability, Congenital Malformations, and Failure to Thrive. <i>American Journal of Human Genetics</i> , 2016, 99, 720-727.	2.6	45
61	Increased bone turnover, osteoporosis, progressive tibial bowing, fractures, and scoliosis in a patient with a finalâ€xexon <i>SATB2</i> frameshift mutation. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 3028-3032.	0.7	16
62	Recurrent Muscle Weakness with Rhabdomyolysis, Metabolic Crises, and Cardiac Arrhythmia Due to Bi-allelic TANGO2 Mutations. <i>American Journal of Human Genetics</i> , 2016, 98, 347-357.	2.6	98
63	A homozygous mutation in PEX16 identified by whole-exome sequencing ending a diagnostic odyssey. <i>Molecular Genetics and Metabolism Reports</i> , 2015, 5, 15-18.	0.4	16
64	Alu-mediated diverse and complex pathogenic copy-number variants within human chromosome 17 at p13.3. <i>Human Molecular Genetics</i> , 2015, 24, 4061-4077.	1.4	83
65	Neurodevelopmental and neurobehavioral characteristics in males and females with CDKL5 duplications. <i>European Journal of Human Genetics</i> , 2015, 23, 915-921.	1.4	32
66	Autosomal-Dominant Multiple Pterygium Syndrome Is Caused by Mutations in MYH3. <i>American Journal of Human Genetics</i> , 2015, 96, 841-849.	2.6	55
67	Delineation of candidate genes responsible for structural brain abnormalities in patients with terminal deletions of chromosome 6q27. <i>European Journal of Human Genetics</i> , 2015, 23, 54-60.	1.4	45
68	Heterozygous De Novo and Inherited Mutations in the Smooth Muscle Actin (ACTG2) Gene Underlie Megacystis-Microcolon-Intestinal Hypoperistalsis Syndrome. <i>PLoS Genetics</i> , 2014, 10, e1004258.	1.5	122
69	Isolated Fetal Macrodactyly: Phenotypic and Genetic Disparities in Mosaic Overgrowth Syndrome. <i>Journal of Ultrasound in Medicine</i> , 2014, 33, 1305-1307.	0.8	8
70	Combined array CGH plus SNP genome analyses in a single assay for optimized clinical testing. <i>European Journal of Human Genetics</i> , 2014, 22, 79-87.	1.4	112
71	Parental Somatic Mosaicism Is Underrecognized and Influences Recurrence Risk of Genomic Disorders. <i>American Journal of Human Genetics</i> , 2014, 95, 173-182.	2.6	219
72	Somatic mosaicism detected by exon-targeted, high-resolution aCGH in 10â€%362 consecutive cases. <i>European Journal of Human Genetics</i> , 2014, 22, 969-978.	1.4	51

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73	Etiopathogenesis of equinovarus foot malformations. <i>European Journal of Medical Genetics</i> , 2014, 57, 473-479.	0.7	43
74	Lysinuric protein intolerance presenting with multiple fractures. <i>Molecular Genetics and Metabolism Reports</i> , 2014, 1, 176-183.	0.4	20
75	TM4SF20 Ancestral Deletion and Susceptibility to a Pediatric Disorder of Early Language Delay and Cerebral White Matter Hyperintensities. <i>American Journal of Human Genetics</i> , 2013, 93, 197-210.	2.6	43
76	<i>WDR35</i> mutation in siblings with Sensenbrenner syndrome: A ciliopathy with variable phenotype. <i>American Journal of Medical Genetics, Part A</i> , 2012, 158A, 2917-2924.	0.7	40
77	<i>WDR62</i> missense mutation in a consanguineous family with primary microcephaly. <i>American Journal of Medical Genetics, Part A</i> , 2012, 158A, 622-625.	0.7	12
78	Introductory comments on special section "Genomic microduplications: When adding may equal subtracting." <i>American Journal of Medical Genetics, Part A</i> , 2010, 152A, 1063-1065.	0.7	2
79	ATR-16 Due to a De Novo Complex Rearrangement of Chromosome 16. <i>Hemoglobin</i> , 2005, 29, 141-150.	0.4	7
80	EGR2 mutation R359W causes a spectrum of Dejerine-Sottas neuropathy. <i>Neurogenetics</i> , 2001, 3, 153-157.	0.7	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. <i>American Journal of Medical Genetics Part A</i> , 2000, 92, 250-255.	2.4	38
83	Terminal osseous dysplasia and pigmentary defects: Clinical characterization of a novel male lethal X-linked syndrome. <i>American Journal of Medical Genetics Part A</i> , 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. , 1999, 87, 278-280.		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