

Dennis E Bulman

List of Publications by Year in descending order

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66
papers

6,509
citations

147566

31
h-index

123241

61
g-index

70
all docs

70
docs citations

70
times ranked

7222
citing authors

#	ARTICLE	IF	CITATIONS
1	Estimate of the contemporary live-birth prevalence of recurrent 22q11.2 deletions: a cross-sectional analysis from population-based newborn screening. <i>CMAJ Open</i> , 2021, 9, E802-E809.	1.1	65
2	Time-dependent decline of T-cell receptor excision circle levels in ZAP-70 deficiency. <i>Journal of Allergy and Clinical Immunology: in Practice</i> , 2020, 8, 806-808.e2.	2.0	10
3	A Case Report of Myoclonus-Dystonia with Isolated Myoclonus Phenotype and Novel Mutation Successfully Treated with Deep Brain Stimulation. <i>Neurology and Therapy</i> , 2020, 9, 187-191.	1.4	4
4	Genotypes of chronic progressive external ophthalmoplegia in a large adult-onset cohort. <i>Mitochondrion</i> , 2019, 49, 227-231.	1.6	20
5	Genetic Variation in the Ontario Neurodegenerative Disease Research Initiative. <i>Canadian Journal of Neurological Sciences</i> , 2019, 46, 491-498.	0.3	7
6	Ataxia Telangiectasia Diagnosed on Newborn Screening—Case Cohort of 5 Years' Experience. <i>Frontiers in Immunology</i> , 2019, 10, 2940.	2.2	37
7	An evaluation of genetic causes and environmental risks for bilateral optic atrophy. <i>PLoS ONE</i> , 2019, 14, e0225656.	1.1	9
8	Effects of fat mass and obesity-associated (FTO) gene polymorphisms on binge eating in women with binge-eating disorder: The moderating influence of attachment style. <i>Nutrition</i> , 2019, 61, 208-212.	1.1	12
9	An evaluation of genetic causes and environmental risks for bilateral optic atrophy. , 2019, 14, e0225656.		0
10	An evaluation of genetic causes and environmental risks for bilateral optic atrophy. , 2019, 14, e0225656.		0
11	An evaluation of genetic causes and environmental risks for bilateral optic atrophy. , 2019, 14, e0225656.		0
12	An evaluation of genetic causes and environmental risks for bilateral optic atrophy. , 2019, 14, e0225656.		0
13	Targeted Next-generation Sequencing and Bioinformatics Pipeline to Evaluate Genetic Determinants of Constitutional Disease. <i>Journal of Visualized Experiments</i> , 2018, , .	0.2	17
14	T-cell receptor excision circle levels and safety of paediatric immunization: A population-based self-controlled case series analysis. <i>Human Vaccines and Immunotherapeutics</i> , 2018, 14, 1378-1391.	1.4	3
15	Genomic DNA Methylation Signatures Enable Concurrent Diagnosis and Clinical Genetic Variant Classification in Neurodevelopmental Syndromes. <i>American Journal of Human Genetics</i> , 2018, 102, 156-174.	2.6	135
16	A family segregating lethal neonatal coenzyme Q ₁₀ deficiency caused by mutations in COQ9. <i>Journal of Inherited Metabolic Disease</i> , 2018, 41, 719-729.	1.7	30
17	BAFopathies™ DNA methylation epi-signatures demonstrate diagnostic utility and functional continuum of Coffin–Siris and Nicolaides–Baraitser syndromes. <i>Nature Communications</i> , 2018, 9, 4885.	5.8	83
18	Compound heterozygous mutations in the gene PIGP are associated with early infantile epileptic encephalopathy. <i>Human Molecular Genetics</i> , 2017, 26, 1706-1715.	1.4	39

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19	<i>KMT2D</i> p.Gln3575His segregating in a family with autosomal dominant choanal atresia strengthens the Kabuki/CHARGE connection. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 183-189.	0.7	15
20	A novel multisystem disease associated with recessive mutations in the tyrosyl-tRNA synthetase (<i>YARS</i>) gene. <i>American Journal of Medical Genetics, Part A</i> , 2017, 173, 126-134.	0.7	36
21	Mandibulofacial Dysostosis with Microcephaly: Mutation and Database Update. <i>Human Mutation</i> , 2016, 37, 148-154.	1.1	45
22	The defining DNA methylation signature of Floating-Harbor Syndrome. <i>Scientific Reports</i> , 2016, 6, 38803.	1.6	55
23	The ONDRISeg panel: custom-designed next-generation sequencing of genes related to neurodegeneration. <i>Npj Genomic Medicine</i> , 2016, 1, 16032.	1.7	26
24	Voluntary Running Triggers VGF-Mediated Oligodendrogenesis to Prolong the Lifespan of Snf2h-Null Ataxic Mice. <i>Cell Reports</i> , 2016, 17, 862-875.	2.9	39
25	Severe Neonatal Presentation of Mitochondrial Citrate Carrier (SLC25A1) Deficiency. <i>JIMD Reports</i> , 2016, 30, 73-79.	0.7	21
26	Next-generation sequencing for diagnosis of rare diseases in the neonatal intensive care unit. <i>Cmaj</i> , 2016, 188, E254-E260.	0.9	86
27	Severe connective tissue laxity including aortic dilatation in Sotos syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 531-535.	0.7	9
28	Syndrome disintegration: Exome sequencing reveals that Fitzsimmons syndrome is a co-occurrence of multiple events. <i>American Journal of Medical Genetics, Part A</i> , 2016, 170, 1820-1825.	0.7	19
29	Mutations in the glucocerebrosidase gene are common in patients with Parkinson's disease from Eastern Canada. <i>International Journal of Neuroscience</i> , 2016, 126, 415-421.	0.8	27
30	Resolution of refractory hypotension and anuria in a premature newborn with loss of function of ACE. <i>American Journal of Medical Genetics, Part A</i> , 2015, 167, 1654-1658.	0.7	10
31	Meconium ileus in a Lebanese family secondary to mutations in the <i>GUCY2C</i> gene. <i>European Journal of Human Genetics</i> , 2015, 23, 990-992.	1.4	24
32	Congenital Visual Impairment and Progressive Microcephaly Due to Lysyl-tRNA Synthetase (<i>KARS</i>) Mutations. <i>Journal of Child Neurology</i> , 2015, 30, 1037-1043.	0.7	47
33	Two novel disease-causing variants in <i>BMPRI1B</i> are associated with brachydactyly type A1. <i>European Journal of Human Genetics</i> , 2015, 23, 1640-1645.	1.4	17
34	Autosomal recessive axonal polyneuropathy in a sibling pair due to a novel homozygous mutation in <i>IGHMBP2</i> . <i>Neuromuscular Disorders</i> , 2015, 25, 794-799.	0.3	16
35	Homozygous nonsense mutation in <i>SYNJ1</i> associated with intractable epilepsy and tau pathology. <i>Neurobiology of Aging</i> , 2015, 36, 1222.e1-1222.e5.	1.5	50
36	Mutations in the enzyme glutathione peroxidase 4 cause Sedaghatian-type spondylometaphyseal dysplasia. <i>Journal of Medical Genetics</i> , 2014, 51, 470-474.	1.5	64

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37	FORGE Canada Consortium: Outcomes of a 2-Year National Rare-Disease Gene-Discovery Project. <i>American Journal of Human Genetics</i> , 2014, 94, 809-817.	2.6	219
38	The PARLance of Parkinson disease. <i>Autophagy</i> , 2011, 7, 790-792.	4.3	7
39	17p13.3 microduplications are associated with split-hand/foot malformation and long-bone deficiency (SHFLD). <i>European Journal of Human Genetics</i> , 2011, 19, 1144-1151.	1.4	32
40	Mutations in GDF5 presenting as semidominant brachydactyly A1. <i>Human Mutation</i> , 2010, 31, 1155-1162.	1.1	23
41	Brachydactyly A-1 mutations restricted to the central region of the N-terminal active fragment of Indian Hedgehog. <i>European Journal of Human Genetics</i> , 2009, 17, 1112-1120.	1.4	46
42	Large deletions account for an increasing number of mutations in <i>SGCE</i>. <i>Movement Disorders</i> , 2008, 23, 456-460.	2.2	27
43	A Nurr1 point mutant, implicated in Parkinsonâ€™s disease, uncouples ERK1/2-dependent regulation of tyrosine hydroxylase transcription. <i>Neurobiology of Disease</i> , 2008, 29, 117-122.	2.1	43
44	Refinement of the DYT15 locus in myoclonus dystonia. <i>Movement Disorders</i> , 2007, 22, 888-892.	2.2	41
45	Translated mutation in the Nurr1 gene as a cause for Parkinson's disease. <i>Movement Disorders</i> , 2006, 21, 906-909.	2.2	93
46	A century later Farabee has his mutation. <i>Human Genetics</i> , 2005, 117, 285-7.	1.8	12
47	A novel mutation in the IHH gene causes brachydactyly type A1: a 95-year-old mystery resolved. <i>Human Genetics</i> , 2002, 111, 368-375.	1.8	42
48	Evidence Favoring Genetic Heterogeneity for Febrile Convulsions. <i>Epilepsia</i> , 2000, 41, 132-139.	2.6	17
49	An approach to ascertain probands with a non-traditional risk factor for carotid atherosclerosis. <i>Atherosclerosis</i> , 1999, 144, 429-434.	0.4	78
50	Familial Hemiplegic Migraine and Episodic Ataxia Type-2 Are Caused by Mutations in the Ca ²⁺ Channel Gene CACNL1A4. <i>Cell</i> , 1996, 87, 543-552.	13.5	2,287
51	A full genome search in multiple sclerosis. <i>Nature Genetics</i> , 1996, 13, 472-476.	9.4	638
52	Mapping the gene for acetazolamide responsive hereditary paroxysmal cerebellar ataxia to chromosome 19p. <i>Human Molecular Genetics</i> , 1995, 4, 279-284.	1.4	102
53	Dystrophin expression in the human retina is required for normal function as defined by electroretinography. <i>Nature Genetics</i> , 1993, 4, 82-86.	9.4	151
54	Sarcolemmal distribution of abnormal dystrophin in Xp21 carriers. <i>Neuromuscular Disorders</i> , 1993, 3, 135-140.	0.3	3

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55	Characterization of translational frame exception patients in Duchenne/Becker muscular dystrophy. <i>Human Molecular Genetics</i> , 1993, 2, 737-744.	1.4	89
56	Genomic organization of exons 22 to 25 of the dystrophin gene. <i>Human Molecular Genetics</i> , 1993, 2, 593-594.	1.4	7
57	Frameshift duplication resulting in truncated dystrophin in a patient with Duchenne muscular dystrophy. <i>Human Mutation</i> , 1992, 1, 172-173.	1.1	1
58	Additional dystrophin fragment in Becker muscular dystrophy patients: Correlation with the pattern of DNA deletion. <i>American Journal of Medical Genetics Part A</i> , 1992, 44, 382-384.	2.4	9
59	Point mutation in the human dystrophin gene: Identification through Western blot analysis. <i>Genomics</i> , 1991, 10, 457-460.	1.3	74
60	Dystrophin is tightly associated with the sarcolemma of mammalian skeletal muscle fibers. <i>Experimental Cell Research</i> , 1991, 192, 278-288.	1.2	37
61	Screening of male patients with autosomal recessive Duchenne dystrophy through dystrophin and DNA studies. <i>American Journal of Medical Genetics Part A</i> , 1991, 39, 38-41.	2.4	15
62	Age-Related Conversion of Dystrophin-Negative to -Positive Fiber Segments of Skeletal but not Cardiac Muscle Fibers in Heterozygote mdx Mice. <i>Journal of Neuropathology and Experimental Neurology</i> , 1990, 49, 96-105.	0.9	48
63	Dystrophin is localized to the plasma membrane of human skeletal muscle fibers by electron-microscopic cytochemical study. <i>Muscle and Nerve</i> , 1990, 13, 376-380.	1.0	59
64	The Duchenne muscular dystrophy gene product is localized in sarcolemma of human skeletal muscle. <i>Nature</i> , 1988, 333, 466-469.	13.7	650
65	A Population-Based Study of Multiple Sclerosis in Twins. <i>New England Journal of Medicine</i> , 1986, 315, 1638-1642.	13.9	579
66	Case of 22q11.2 Deletion Syndrome Not Identified by <i>TBX1</i> Screening with a Positive SCID Newborn Screen. <i>LymphoSign Journal</i> , 0, , .	0.1	0