Dennis E Bulman

List of Publications by Year
in descending order

66 papers	6,509 citations	147801 31 h-index	123424 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. CMAJ Open, 2021, 9, E802-E809.	2.4	65
2	Time-dependent decline of T-cell receptor excision circle levels in ZAP-70 deficiency. Journal of Allergy and Clinical Immunology: in Practice, 2020, 8, 806-808.e2.	3.8	10
3	A Case Report of Myoclonus-Dystonia with Isolated Myoclonus Phenotype and Novel Mutation Successfully Treated with Deep Brain Stimulation. Neurology and Therapy, 2020, 9, 187-191.	3.2	4
4	Genotypes of chronic progressive external ophthalmoplegia in a large adult-onset cohort. Mitochondrion, 2019, 49, 227-231.	3.4	20
5	Genetic Variation in the Ontario Neurodegenerative Disease Research Initiative. Canadian Journal of Neurological Sciences, 2019, 46, 491-498.	0.5	7
6	Ataxia Telangiectasia Diagnosed on Newborn Screeningâ€“Case Cohort of 5 Years' Experience. Frontiers in Immunology, 2019, 10, 2940.	4.8	37
7	An evaluation of genetic causes and environmental risks for bilateral optic atrophy. PLoS ONE, 2019, 14, e0225656.	2.5	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. Nutrition, 2019, 61, 208-212.	2.4	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. Journal of Visualized Experiments, 2018, , .	0.3	17
14	T-cell receptor excision circle levels and safety of paediatric immunization: A population-based self-controlled case series analysis. Human Vaccines and Immunotherapeutics, 2018, 14, 1378-1391.	3.3	3
15	Genomic DNA Methylation Signatures Enable Concurrent Diagnosis and Clinical Genetic Variant Classification in Neurodevelopmental Syndromes. American Journal of Human Genetics, 2018, 102, 156-174.	6.2	135
16	A family segregating lethal neonatal coenzyme Q₁₀ deficiency caused by mutations in COQ9. Journal of Inherited Metabolic Disease, 2018, 41, 719-729.	3.6	30
17	BAFopathiesâ€™ DNA methylation epi-signatures demonstrate diagnostic utility and functional continuum of Coffinâ€“Siris and Nicolaidesâ€“Baraitser syndromes. Nature Communications, 2018, 9, 4885.	12.8	83
18	Compound heterozygous mutations in the gene PIGP are associated with early infantile epileptic encephalopathy. Human Molecular Genetics, 2017, 26, 1706-1715.	2.9	39

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19	KMT2D p.Gln3575His segregating in a family with autosomal dominant choanal atresia strengthens the Kabuki/CHARGE connection. American Journal of Medical Genetics, Part A, 2017, 173, 183-189.	1.2	15
20	A novel multisystem disease associated with recessive mutations in the tyrosylâ€tRNA synthetase (YARS) gene. American Journal of Medical Genetics, Part A, 2017, 173, 126-134.	1.2	36
21	Mandibulofacial Dysostosis with Microcephaly: Mutation and Database Update. Human Mutation, 2016, 37, 148-154.	2.5	45
22	The defining DNA methylation signature of Floating-Harbor Syndrome. Scientific Reports, 2016, 6, 38803.	3.3	55
23	The ONDRISeq panel: custom-designed next-generation sequencing of genes related to neurodegeneration. Npj Genomic Medicine, 2016, 1, 16032.	3.8	26
24	Voluntary Running Triggers VGF-Mediated Oligodendrogenesis to Prolong the Lifespan of Snf2h-Null Ataxic Mice. Cell Reports, 2016, 17, 862-875.	6.4	39
25	Severe Neonatal Presentation of Mitochondrial Citrate Carrier (SLC25A1) Deficiency. JIMD Reports, 2016, 30, 73-79.	1.5	21
26	Next-generation sequencing for diagnosis of rare diseases in the neonatal intensive care unit. Cmaj, 2016, 188, E254-E260.	2.0	86
27	Severe connective tissue laxity including aortic dilatation in Sotos syndrome. American Journal of Medical Genetics, Part A, 2016, 170, 531-535.	1.2	9
28	Syndrome disintegration: Exome sequencing reveals that Fitzsimmons syndrome is a coâ€occurrence of multiple events. American Journal of Medical Genetics, Part A, 2016, 170, 1820-1825.	1.2	19
29	Mutations in the glucocerebrosidase gene are common in patients with Parkinson's disease from Eastern Canada. International Journal of Neuroscience, 2016, 126, 415-421.	1.6	27
30	Resolution of refractory hypotension and anuria in a premature newborn with lossâ€ofâ€function of ACE. American Journal of Medical Genetics, Part A, 2015, 167, 1654-1658.	1.2	10
31	Meconium ileus in a Lebanese family secondary to mutations in the GUCY2C gene. European Journal of Human Genetics, 2015, 23, 990-992.	2.8	24
32	Congenital Visual Impairment and Progressive Microcephaly Due to Lysylâ€“Transfer Ribonucleic Acid (RNA) Synthetase (KARS) Mutations. Journal of Child Neurology, 2015, 30, 1037-1043.	1.4	47
33	Two novel disease-causing variants in BMPR1B are associated with brachydactyly type A1. European Journal of Human Genetics, 2015, 23, 1640-1645.	2.8	17
34	Autosomal recessive axonal polyneuropathy in a sibling pair due to a novel homozygous mutation in IGHMBP2. Neuromuscular Disorders, 2015, 25, 794-799.	0.6	16
35	Homozygous nonsense mutation in SYNJ1 associated with intractable epilepsy and tau pathology. Neurobiology of Aging, 2015, 36, 1222.e1-1222.e5.	3.1	50
36	Mutations in the enzyme glutathione peroxidase 4 cause Sedaghatian-type spondylometaphyseal dysplasia. Journal of Medical Genetics, 2014, 51, 470-474.	3.2	64

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37	FORGE Canada Consortium: Outcomes of a 2-Year National Rare-Disease Gene-Discovery Project. American Journal of Human Genetics, 2014, 94, 809-817.	6.2	219
38	The PARLance of Parkinson disease. Autophagy, 2011, 7, 790-792.	9.1	7
39	17p13.3 microduplications are associated with split-hand/foot malformation and long-bone deficiency (SHFLD). European Journal of Human Genetics, 2011, 19, 1144-1151.	2.8	32
40	Mutations in GDF5 presenting as semidominant brachydactyly A1. Human Mutation, 2010, 31, 1155-1162.	2.5	23
41	Brachydactyly A-1 mutations restricted to the central region of the N-terminal active fragment of Indian Hedgehog. European Journal of Human Genetics, 2009, 17, 1112-1120.	2.8	46
42	Large deletions account for an increasing number of mutations in SGCE. Movement Disorders, 2008, 23, 456-460.	3.9	27
43	A Nurr1 point mutant, implicated in Parkinsonâ€™s disease, uncouples ERK1/2-dependent regulation of tyrosine hydroxylase transcription. Neurobiology of Disease, 2008, 29, 117-122.	4.4	43
44	Refinement of the DYT15 locus in myoclonus dystonia. Movement Disorders, 2007, 22, 888-892.	3.9	41
45	Translated mutation in the Nurr1 gene as a cause for Parkinson's disease. Movement Disorders, 2006, 21, 906-909.	3.9	93
46	A century later Farabee has his mutation. Human Genetics, 2005, 117, 285-7.	3.8	12
47	A novel mutation in the IHH gene causes brachydactyly type A1: a 95-year-old mystery resolved. Human Genetics, 2002, 111, 368-375.	3.8	42
48	Evidence Favoring Genetic Heterogeneity for Febrile Convulsions. Epilepsia, 2000, 41, 132-139.	5.1	17
49	An approach to ascertain probands with a non-traditional risk factor for carotid atherosclerosis. Atherosclerosis, 1999, 144, 429-434.	0.8	78
50	Familial Hemiplegic Migraine and Episodic Ataxia Type-2 Are Caused by Mutations in the Ca2+ Channel Gene CACNL1A4. Cell, 1996, 87, 543-552.	28.9	2,287
51	A full genome search in multiple sclerosis. Nature Genetics, 1996, 13, 472-476.	21.4	638
52	Mapping the gene for acetazolamide responsive hereditary paryoxysmal cerebellar ataxia to chromosome 19p. Human Molecular Genetics, 1995, 4, 279-284.	2.9	102
53	Dystrophin expression in the human retina is required for normal function as defined by electroretinography. Nature Genetics, 1993, 4, 82-86.	21.4	151
54	Sarcolemmal distribution of abnormal dystrophin in Xp21 carriers. Neuromuscular Disorders, 1993, 3, 135-140.	0.6	3

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55	Characterization of translational frame exception patients in Duchenne/Becker muscular dystrophy. Human Molecular Genetics, 1993, 2, 737-744.	2.9	89
56	Genomic organization of exons 22 to 25 of the dystrophin gene. Human Molecular Genetics, 1993, 2, 593-594.	2.9	7
57	Frameshift duplication resulting in truncated dystrophin in a patient with Duchenne muscular dystrophy. Human Mutation, 1992, 1, 172-173.	2.5	1
58	Additional dystrophin fragment in Becker muscular dystrophy patients: Correlation with the pattern of DNA deletion. American Journal of Medical Genetics Part A, 1992, 44, 382-384.	2.4	9
59	Point mutation in the human dystrophin gene: Identification through Western blot analysis. Genomics, 1991, 10, 457-460.	2.9	74
60	Dystrophin is tightly associated with the sarcolemma of mammalian skeletal muscle fibers. Experimental Cell Research, 1991, 192, 278-288.	2.6	37
61	Screening of male patients with autosomal recessive Duchenne dystrophy through dystrophin and DNA studies. American Journal of Medical Genetics Part A, 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. Journal of Neuropathology and Experimental Neurology, 1990, 49, 96-105.	1.7	48
63	Dystrophin is localized to the plasma membrane of human skeletal muscle fibers by electron-microscopic cytochemical study. Muscle and Nerve, 1990, 13, 376-380.	2.2	59
64	The Duchenne muscular dystrophy gene product is localized in sarcolemma of human skeletal muscle. Nature, 1988, 333, 466-469.	27.8	650
65	A Population-Based Study of Multiple Sclerosis in Twins. New England Journal of Medicine, 1986, 315, 1638-1642.	27.0	579
66	Case of 22q11.2 Deletion Syndrome Not Identified by TBX1 Screening with a Positive SCID Newborn Screen. LymphoSign Journal, 0, , .	0.2	0