# Dennis E Bulman

List of Publications by Year
in descending order



| | | | |
|---|---|---|---|
| 66 papers | 6,509 citations | 147726 31 h-index | 123376 61 g-index |
| 70 all docs | 70 docs citations | 70 times ranked | 7222 citing authors |

| # | Article | IF | Citations |
|---|---|---|---|
| 1 | Familial Hemiplegic Migraine and Episodic Ataxia Type-2 Are Caused by Mutations in the Ca2+ Channel Gene CACNL1A4. Cell, 1996, 87, 543-552. | 13.5 | 2,287 |
| 2 | The Duchenne muscular dystrophy gene product is localized in sarcolemma of human skeletal muscle. Nature, 1988, 333, 466-469. | 13.7 | 650 |
| 3 | A full genome search in multiple sclerosis. Nature Genetics, 1996, 13, 472-476. | 9.4 | 638 |
| 4 | A Population-Based Study of Multiple Sclerosis in Twins. New England Journal of Medicine, 1986, 315, 1638-1642. | 13.9 | 579 |
| 5 | FORGE Canada Consortium: Outcomes of a 2-Year National Rare-Disease Gene-Discovery Project. American Journal of Human Genetics, 2014, 94, 809-817. | 2.6 | 219 |
| 6 | Dystrophin expression in the human retina is required for normal function as defined by electroretinography. Nature Genetics, 1993, 4, 82-86. | 9.4 | 151 |
| 7 | Genomic DNA Methylation Signatures Enable Concurrent Diagnosis and Clinical Genetic Variant Classification in Neurodevelopmental Syndromes. American Journal of Human Genetics, 2018, 102, 156-174. | 2.6 | 135 |
| 8 | Mapping the gene for acetazolamide responsive hereditary paryoxysmal cerebellar ataxia to chromosome 19p. Human Molecular Genetics, 1995, 4, 279-284. | 1.4 | 102 |
| 9 | Translated mutation in the Nurr1 gene as a cause for Parkinson's disease. Movement Disorders, 2006, 21, 906-909. | 2.2 | 93 |
| 10 | Characterization of translational frame exception patients in Duchenne/Becker muscular dystrophy. Human Molecular Genetics, 1993, 2, 737-744. | 1.4 | 89 |
| 11 | Next-generation sequencing for diagnosis of rare diseases in the neonatal intensive care unit. Cmaj, 2016, 188, E254-E260. | 0.9 | 86 |
| 12 | BAFopathiesâ€™ DNA methylation epi-signatures demonstrate diagnostic utility and functional continuum of Coffinâ€“Siris and Nicolaidesâ€“Baraitser syndromes. Nature Communications, 2018, 9, 4885. | 5.8 | 83 |
| 13 | An approach to ascertain probands with a non-traditional risk factor for carotid atherosclerosis. Atherosclerosis, 1999, 144, 429-434. | 0.4 | 78 |
| 14 | Point mutation in the human dystrophin gene: Identification through Western blot analysis. Genomics, 1991, 10, 457-460. | 1.3 | 74 |
| 15 | 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. | 1.1 | 65 |
| 16 | Mutations in the enzyme glutathione peroxidase 4 cause Sedaghatian-type spondylometaphyseal dysplasia. Journal of Medical Genetics, 2014, 51, 470-474. | 1.5 | 64 |
| 17 | Dystrophin is localized to the plasma membrane of human skeletal muscle fibers by electron-microscopic cytochemical study. Muscle and Nerve, 1990, 13, 376-380. | 1.0 | 59 |
| 18 | The defining DNA methylation signature of Floating-Harbor Syndrome. Scientific Reports, 2016, 6, 38803. | 1.6 | 55 |

| # | Article | IF | Citations |
|---|---|---|---|
| 19 | Homozygous nonsense mutation in SYNJ1 associated with intractable epilepsy and tau pathology. Neurobiology of Aging, 2015, 36, 1222.e1-1222.e5. | 1.5 | 50 |
| 20 | 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. | 0.9 | 48 |
| 21 | Congenital Visual Impairment and Progressive Microcephaly Due to Lysylâ€“Transfer Ribonucleic Acid (RNA) Synthetase (KARS) Mutations. Journal of Child Neurology, 2015, 30, 1037-1043. | 0.7 | 47 |
| 22 | 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. | 1.4 | 46 |
| 23 | Mandibulofacial Dysostosis with Microcephaly: Mutation and Database Update. Human Mutation, 2016, 37, 148-154. | 1.1 | 45 |
| 24 | 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. | 2.1 | 43 |
| 25 | A novel mutation in the IHH gene causes brachydactyly type A1: a 95-year-old mystery resolved. Human Genetics, 2002, 111, 368-375. | 1.8 | 42 |
| 26 | Refinement of the DYT15 locus in myoclonus dystonia. Movement Disorders, 2007, 22, 888-892. | 2.2 | 41 |
| 27 | Voluntary Running Triggers VGF-Mediated Oligodendrogenesis to Prolong the Lifespan of Snf2h-Null Ataxic Mice. Cell Reports, 2016, 17, 862-875. | 2.9 | 39 |
| 28 | Compound heterozygous mutations in the gene PIGP are associated with early infantile epileptic encephalopathy. Human Molecular Genetics, 2017, 26, 1706-1715. | 1.4 | 39 |
| 29 | Dystrophin is tightly associated with the sarcolemma of mammalian skeletal muscle fibers. Experimental Cell Research, 1991, 192, 278-288. | 1.2 | 37 |
| 30 | Ataxia Telangiectasia Diagnosed on Newborn Screeningâ€“Case Cohort of 5 Years' Experience. Frontiers in Immunology, 2019, 10, 2940. | 2.2 | 37 |
| 31 | 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. | 0.7 | 36 |
| 32 | 17p13.3 microduplications are associated with split-hand/foot malformation and long-bone deficiency (SHFLD). European Journal of Human Genetics, 2011, 19, 1144-1151. | 1.4 | 32 |
| 33 | A family segregating lethal neonatal coenzyme Q<sub>10</sub> deficiency caused by mutations in COQ9. Journal of Inherited Metabolic Disease, 2018, 41, 719-729. | 1.7 | 30 |
| 34 | Large deletions account for an increasing number of mutations in SGCE. Movement Disorders, 2008, 23, 456-460. | 2.2 | 27 |
| 35 | Mutations in the glucocerebrosidase gene are common in patients with Parkinson's disease from Eastern Canada. International Journal of Neuroscience, 2016, 126, 415-421. | 0.8 | 27 |
| 36 | The ONDRISeq panel: custom-designed next-generation sequencing of genes related to neurodegeneration. Npj Genomic Medicine, 2016, 1, 16032. | 1.7 | 26 |

| # | Article | IF | Citations |
|---|---|---|---|
| 37 | Meconium ileus in a Lebanese family secondary to mutations in the GUCY2C gene. European Journal of Human Genetics, 2015, 23, 990-992. | 1.4 | 24 |
| 38 | Mutations in GDF5 presenting as semidominant brachydactyly A1. Human Mutation, 2010, 31, 1155-1162. | 1.1 | 23 |
| 39 | Severe Neonatal Presentation of Mitochondrial Citrate Carrier (SLC25A1) Deficiency. JIMD Reports, 2016, 30, 73-79. | 0.7 | 21 |
| 40 | Genotypes of chronic progressive external ophthalmoplegia in a large adult-onset cohort. Mitochondrion, 2019, 49, 227-231. | 1.6 | 20 |
| 41 | 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. | 0.7 | 19 |
| 42 | Evidence Favoring Genetic Heterogeneity for Febrile Convulsions. Epilepsia, 2000, 41, 132-139. | 2.6 | 17 |
| 43 | Two novel disease-causing variants in BMPR1B are associated with brachydactyly type A1. European Journal of Human Genetics, 2015, 23, 1640-1645. | 1.4 | 17 |
| 44 | Targeted Next-generation Sequencing and Bioinformatics Pipeline to Evaluate Genetic Determinants of Constitutional Disease. Journal of Visualized Experiments, 2018, , . | 0.2 | 17 |
| 45 | Autosomal recessive axonal polyneuropathy in a sibling pair due to a novel homozygous mutation in IGHMBP2. Neuromuscular Disorders, 2015, 25, 794-799. | 0.3 | 16 |
| 46 | 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 |
| 47 | 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. | 0.7 | 15 |
| 48 | A century later Farabee has his mutation. Human Genetics, 2005, 117, 285-7. | 1.8 | 12 |
| 49 | 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. | 1.1 | 12 |
| 50 | 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. | 0.7 | 10 |
| 51 | 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. | 2.0 | 10 |
| 52 | 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 |
| 53 | Severe connective tissue laxity including aortic dilatation in Sotos syndrome. American Journal of Medical Genetics, Part A, 2016, 170, 531-535. | 0.7 | 9 |
| 54 | An evaluation of genetic causes and environmental risks for bilateral optic atrophy. PLoS ONE, 2019, 14, e0225656. | 1.1 | 9 |

| # | Article | IF | Citations |
|---|---|---|---|
| 55 | Genomic organization of exons 22 to 25 of the dystrophin gene. Human Molecular Genetics, 1993, 2, 593-594. | 1.4 | 7 |
| 56 | The PARLance of Parkinson disease. Autophagy, 2011, 7, 790-792. | 4.3 | 7 |
| 57 | Genetic Variation in the Ontario Neurodegenerative Disease Research Initiative. Canadian Journal of Neurological Sciences, 2019, 46, 491-498. | 0.3 | 7 |
| 58 | 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. | 1.4 | 4 |
| 59 | Sarcolemmal distribution of abnormal dystrophin in Xp21 carriers. Neuromuscular Disorders, 1993, 3, 135-140. | 0.3 | 3 |
| 60 | 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. | 1.4 | 3 |
| 61 | Frameshift duplication resulting in truncated dystrophin in a patient with Duchenne muscular dystrophy. Human Mutation, 1992, 1, 172-173. | 1.1 | 1 |
| 62 | Case of 22q11.2 Deletion Syndrome Not Identified by TBX1 Screening with a Positive SCID Newborn Screen. LymphoSign Journal, 0, , . | 0.1 | 0 |
| 63 | An evaluation of genetic causes and environmental risks for bilateral optic atrophy. , 2019, 14, e0225656. | | 0 |
| 64 | An evaluation of genetic causes and environmental risks for bilateral optic atrophy. , 2019, 14, e0225656. | | 0 |
| 65 | An evaluation of genetic causes and environmental risks for bilateral optic atrophy. , 2019, 14, e0225656. | | 0 |
| 66 | An evaluation of genetic causes and environmental risks for bilateral optic atrophy. , 2019, 14, e0225656. | | 0 |