Benjamin W Darbro

List of Publications by Year
in descending order

62	1,624	304743	330143
papers	citations	22 h-index	37 g-index

65	65	65	3118
all docs	docs citations	times ranked	citing authors

#	Article	IF	Citations
1	Sporadic hypothalamic hamartoma is a ciliopathy with somatic and bi-allelic contributions. Human Molecular Genetics, 2022, 31, 2307-2316.	2.9	8
2	Intron mutations and early transcription termination in Duchenne and Becker muscular dystrophy. Human Mutation, 2022, 43, 511-528.	2.5	16
3	Additive and Interactive Genetically Contextual Effects of HbA1c on cg19693031 Methylation in Type 2 Diabetes. Genes, 2022, 13, 683.	2.4	4
4	Whole-Exome Sequencing of Patients With Posterior Segment Uveitis. American Journal of Ophthalmology, 2021, 221, 246-259.	3.3	10
5	Using an aquatic model, <scp>Xenopus laevis</scp>, to uncover the role of chromodomain 1 in craniofacial disorders. Genesis, 2021, 59, e23394.	1.6	10
6	Genetic Analysis of Multiple Myeloma Identifies Cytogenetic Alterations Implicated in Disease Complexity and Progression. Cancers, 2021, 13, 517.	3.7	12
7	Combination therapies for MPNSTs targeting RABL6A-RB1 signaling. Oncotarget, 2021, 12, 10-14.	1.8	5
8	Development and comparison of novel bioluminescent mouse models of pancreatic neuroendocrine neoplasm metastasis. Scientific Reports, 2021, 11, 10252.	3.3	4
9	RABL6A Promotes Pancreatic Neuroendocrine Tumor Angiogenesis and Progression In Vivo. Biomedicines, 2021, 9, 633.	3.2	4
10	The emerging role of somatic tumor sequencing in the treatment of urothelial cancer. Asian Journal of Urology, 2021, 8, 391-399.	1.2	6
11	Utility of Flow Cytometry and Fluorescence In Situ Hybridization in Follow-up Monitoring of Plasma Cell Myeloma. American Journal of Clinical Pathology, 2021, 156, 198-204.	0.7	2
12	Sequential genetic testing of livingâ€related donors for inherited renal disease to promote informed choice and enhance safety of living donation. Transplant International, 2021, 34, 2696-2705.	1.6	7
13	Novel Intragenic PAX6 Deletion in a Pedigree with Aniridia, Morbid Obesity, and Diabetes. Current Eye Research, 2020, 45, 91-96.	1.5	10
14	Progressive Optic Disc Cupping Over 20 Years in a Patient with TBK1-Associated Glaucoma. Ophthalmology Glaucoma, 2020, 3, 167-168.	1.9	0
15	Candidate modifier genes for immune function in 22q11.2 deletion syndrome. Molecular Genetics & Genomic Medicine, 2020, 8, e1057.	1.2	3
16	Coactivation of NF-ÎºB and Notch signaling is sufficient to induce B-cell transformation and enables B-myeloid conversion. Blood, 2020, 135, 108-120.	1.4	14
17	Expanding the genotypic and phenotypic spectrum of severe serine biosynthesis disorders. Human Mutation, 2020, 41, 1615-1628.	2.5	14
18	A missense mutation in the MLKL brace region promotes lethal neonatal inflammation and hematopoietic dysfunction. Nature Communications, 2020, 11, 3150.	12.8	75

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19	Longitudinal phenotype development in a minipig model of neurofibromatosis type 1. Scientific Reports, 2020, 10, 5046.	3.3	13
20	RABL6A Is an Essential Driver of MPNSTs that Negatively Regulates the RB1 Pathway and Sensitizes Tumor Cells to CDK4/6 Inhibitors. Clinical Cancer Research, 2020, 26, 2997-3011.	7.0	34
21	In trans variant calling reveals enrichment for compound heterozygous variants in genes involved in neuronal development and growth.. Genetical Research, 2019, 101, e8.	0.9	3
22	A case of primary cutaneous Ewing sarcoma in a neutropenic patient. Journal of Cutaneous Pathology, 2019, 46, 238-241.	1.3	1
23	RABL6A inhibits tumor-suppressive PP2A/AKT signaling to drive pancreatic neuroendocrine tumor growth. Journal of Clinical Investigation, 2019, 129, 1641-1653.	8.2	25
24	Uniparental disomy unveils a novel recessive mutation in POMT2. Neuromuscular Disorders, 2018, 28, 592-596.	0.6	20
25	Identification of Isthmin 1 as a Novel Clefting and Craniofacial Patterning Gene in Humans. Genetics, 2018, 208, 283-296.	2.9	18
26	Changes in gene expression in small bowel neuroendocrine tumors associated with progression to metastases. Surgery, 2018, 163, 232-239.	1.9	14
27	A porcine model of neurofibromatosis type 1 that mimics the human disease. JCI Insight, 2018, 3, .	5.0	44
28	A comprehensive evaluation of Hippo pathway silencing in sarcomas. Oncotarget, 2018, 9, 31620-31636.	1.8	19
29	The Eating-Disorder Associated HDAC4 A778T Mutation Alters Feeding Behaviors in Female Mice. Biological Psychiatry, 2017, 81, 770-777.	1.3	15
30	Development of Secondary Acute Myeloid Leukemia in a Pediatric Patient Concurrently Receiving Primary Therapy for Ewing Sarcoma. Journal of Pediatric Hematology/Oncology, 2017, 39, e370-e372.	0.6	4
31	Immunohistochemical Markers for Prospective Studies in Neurofibromatosis-1 Porcine Models. Journal of Histochemistry and Cytochemistry, 2017, 65, 607-618.	2.5	21
32	A Gene Implicated in Activation of Retinoic Acid Receptor Targets Is a Novel Renal Agenesis Gene in Humans. Genetics, 2017, 207, 215-228.	2.9	62
33	Recessive coding and regulatory mutations in FBLIM1 underlie the pathogenesis of chronic recurrent multifocal osteomyelitis (CRMO). PLoS ONE, 2017, 12, e0169687.	2.5	63
34	Uniparental Isodisomy of Chromosome 1 Unmasking an Autosomal Recessive 3-Beta Hydroxysteroid Dehydrogenase Type II-Related Congenital Adrenal Hyperplasia. JCRPE Journal of Clinical Research in Pediatric Endocrinology, 2017, 9, 70-73.	0.9	8
35	Neurogenetics in the Genome Era. , 2017, , 257-267.		3
36	Autism Linked to Increased Oncogene Mutations but Decreased Cancer Rate. PLoS ONE, 2016, 11, e0149041.	2.5	25

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37	Leukemic Transdifferentiation of Follicular Lymphoma Into an Acute Histiocytic Leukemia in a 52-Year-Old Caucasian Woman. Laboratory Medicine, 2016, 47, 155-157.	1.2	9
38	Mutations of the Sonic Hedgehog Pathway Underlie Hypothalamic Hamartoma with Gelastic Epilepsy. American Journal of Human Genetics, 2016, 99, 423-429.	6.2	59
39	The heritability of hemolysis in stored human red blood cells. Transfusion, 2015, 55, 1178-1185.	1.6	77
40	GE-02 * PARADOXICAL GENETIC AND EPIDEMIOLOGIC RELATIONSHIPS BETWEEN CANCER AND AUTISM. Neuro-Oncology, 2015, 17, iii7-iii7.	1.2	1
41	CNV-ROC: A cost effective, computer-aided analytical performance evaluator of chromosomal microarrays. Journal of Biomedical Informatics, 2015, 54, 106-113.	4.3	0
42	Esophageal cancer in a family with hamartomatous tumors and germline PTEN frameshift and SMAD7 missense mutations. Cancer Genetics, 2015, 208, 41-46.	0.4	10
43	Promoting improved utilization of laboratory testing through changes in an electronic medical record: experience at an academic medical center. BMC Medical Informatics and Decision Making, 2015, 15, 11.	3.0	84
44	Seizures Are Regulated by Ubiquitin-specific Peptidase 9 X-linked (USP9X), a De-Ubiquitinase. PLoS Genetics, 2015, 11, e1005022.	3.5	66
45	Evaluation of multiple putative risk alleles within the 15q13.3 region for genetic generalized epilepsy. Epilepsy Research, 2015, 117, 70-73.	1.6	6
46	A novel porcine model of ataxia telangiectasia reproduces neurological features and motor deficits of human disease. Human Molecular Genetics, 2015, 24, 6473-6484.	2.9	38
47	TBK1and Flanking Genes in Human Retina. Ophthalmic Genetics, 2014, 35, 35-40.	1.2	17
48	The heritability of metabolite concentrations in stored human red blood cells. Transfusion, 2014, 54, 2055-2063.	1.6	59
49	An international effort towards developing standards for best practices in analysis, interpretation and reporting of clinical genome sequencing results in the CLARITY Challenge. Genome Biology, 2014, 15, R53.	9.6	101
50	RABL6A Promotes G1â€“S Phase Progression and Pancreatic Neuroendocrine Tumor Cell Proliferation in an Rb1-Dependent Manner. Cancer Research, 2014, 74, 6661-6670.	0.9	32
51	Heritability of glutathione and related metabolites in stored red blood cells. Free Radical Biology and Medicine, 2014, 76, 107-113.	2.9	63
52	Development and translational imaging of a TP53 porcine tumorigenesis model. Journal of Clinical Investigation, 2014, 124, 4052-4066.	8.2	92
53	Evaluating Familial Essential Tremor with Novel Genetic Approaches: Is it a Genotyping or Phenotyping Issue?. Tremor and Other Hyperkinetic Movements, 2014, 4, 258.	2.0	4
54	Genome-wide copy number variation analysis of a Branchio-oto-renal syndrome cohort identifies a recombination hotspot and implicates new candidate genes. Human Genetics, 2013, 132, 1339-1350.	3.8	27

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55	Mutations in Extracellular Matrix GenesNID1andLAMC1Cause Autosomal Dominant Dandy-Walker Malformation and Occipital Cephaloceles. Human Mutation, 2013, 34, 1075-1079.	2.5	38
56	G-protein coupled receptor expression patterns delineate medulloblastoma subgroups. Acta Neuropathologica Communications, 2013, 1, 66.	5.2	22
57	PRICKLE1 Interaction with SYNAPSIN I Reveals a Role in Autism Spectrum Disorders. PLoS ONE, 2013, 8, e80737.	2.5	39
58	Eating disorder predisposition is associated with ESRRA and HDAC4 mutations. Journal of Clinical Investigation, 2013, 123, 4706-4713.	8.2	64
59	RABL6A, a Novel RAB-Like Protein, Controls Centrosome Amplification and Chromosome Instability in Primary Fibroblasts. PLoS ONE, 2013, 8, e80228.	2.5	12
60	Lactobacillus delbrueckii as the Cause of Urinary Tract Infection. Journal of Clinical Microbiology, 2009, 47, 275-277.	3.9	41
61	Co-Regulation of p16INK4a and Migratory Genes in Culture Conditions that Lead to Premature Senescence in Human Keratinocytes. Journal of Investigative Dermatology, 2005, 125, 499-509.	0.7	33
62	Amplification of the chromosome 20q region is associated with expression of HPV-16 E7 in human airway and anogenital epithelial cells. Virology, 2005, 340, 237-244.	2.4	20