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	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
2	Development and translational imaging of a TP53 porcine tumorigenesis model. Journal of Clinical Investigation, 2014, 124, 4052-4066.	8.2	92
3	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
4	The heritability of hemolysis in stored human red blood cells. Transfusion, 2015, 55, 1178-1185.	1.6	77
5	A missense mutation in the MLKL brace region promotes lethal neonatal inflammation and hematopoietic dysfunction. Nature Communications, 2020, 11, 3150.	12.8	75
6	Seizures Are Regulated by Ubiquitin-specific Peptidase 9 X-linked (USP9X), a De-Ubiquitinase. PLoS Genetics, 2015, 11, e1005022.	3.5	66
7	Eating disorder predisposition is associated with ESRRA and HDAC4 mutations. Journal of Clinical Investigation, 2013, 123, 4706-4713.	8.2	64
8	Heritability of glutathione and related metabolites in stored red blood cells. Free Radical Biology and Medicine, 2014, 76, 107-113.	2.9	63
9	Recessive coding and regulatory mutations in FBLIM1 underlie the pathogenesis of chronic recurrent multifocal osteomyelitis (CRMO). PLoS ONE, 2017, 12, e0169687.	2.5	63
10	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
11	The heritability of metabolite concentrations in stored human red blood cells. Transfusion, 2014, 54, 2055-2063.	1.6	59
12	Mutations of the Sonic Hedgehog Pathway Underlie Hypothalamic Hamartoma with Gelastic Epilepsy. American Journal of Human Genetics, 2016, 99, 423-429.	6.2	59
13	A porcine model of neurofibromatosis type 1 that mimics the human disease. JCI Insight, 2018, 3, .	5.0	44
14	Lactobacillus delbrueckii as the Cause of Urinary Tract Infection. Journal of Clinical Microbiology, 2009, 47, 275-277.	3.9	41
15	PRICKLE1 Interaction with SYNAPSIN I Reveals a Role in Autism Spectrum Disorders. PLoS ONE, 2013, 8, e80737.	2.5	39
16	Mutations in Extracellular Matrix GenesNID1andLAMC1Cause Autosomal Dominant Dandy-Walker Malformation and Occipital Cephaloceles. Human Mutation, 2013, 34, 1075-1079.	2.5	38
17	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
18	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

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19	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
20	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
21	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
22	Autism Linked to Increased Oncogene Mutations but Decreased Cancer Rate. PLoS ONE, 2016, 11, e0149041.	2.5	25
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	G-protein coupled receptor expression patterns delineate medulloblastoma subgroups. Acta Neuropathologica Communications, 2013, 1, 66.	5.2	22
25	Immunohistochemical Markers for Prospective Studies in Neurofibromatosis-1 Porcine Models. Journal of Histochemistry and Cytochemistry, 2017, 65, 607-618.	2.5	21
26	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
27	Uniparental disomy unveils a novel recessive mutation in POMT2. Neuromuscular Disorders, 2018, 28, 592-596.	0.6	20
28	A comprehensive evaluation of Hippo pathway silencing in sarcomas. Oncotarget, 2018, 9, 31620-31636.	1.8	19
29	Identification of Isthmin 1 as a Novel Clefting and Craniofacial Patterning Gene in Humans. Genetics, 2018, 208, 283-296.	2.9	18
30	TBK1and Flanking Genes in Human Retina. Ophthalmic Genetics, 2014, 35, 35-40.	1.2	17
31	Intron mutations and early transcription termination in Duchenne and Becker muscular dystrophy. Human Mutation, 2022, 43, 511-528.	2.5	16
32	The Eating-Disorder Associated HDAC4 A778T Mutation Alters Feeding Behaviors in Female Mice. Biological Psychiatry, 2017, 81, 770-777.	1.3	15
33	Changes in gene expression in small bowel neuroendocrine tumors associated with progression to metastases. Surgery, 2018, 163, 232-239.	1.9	14
34	Coactivation of NF-Î°B and Notch signaling is sufficient to induce B-cell transformation and enables B-myeloid conversion. Blood, 2020, 135, 108-120.	1.4	14
35	Expanding the genotypic and phenotypic spectrum of severe serine biosynthesis disorders. Human Mutation, 2020, 41, 1615-1628.	2.5	14
36	Longitudinal phenotype development in a minipig model of neurofibromatosis type 1. Scientific Reports, 2020, 10, 5046.	3.3	13

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37	Genetic Analysis of Multiple Myeloma Identifies Cytogenetic Alterations Implicated in Disease Complexity and Progression. Cancers, 2021, 13, 517.	3.7	12
38	RABL6A, a Novel RAB-Like Protein, Controls Centrosome Amplification and Chromosome Instability in Primary Fibroblasts. PLoS ONE, 2013, 8, e80228.	2.5	12
39	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
40	Novel Intragenic PAX6 Deletion in a Pedigree with Aniridia, Morbid Obesity, and Diabetes. Current Eye Research, 2020, 45, 91-96.	1.5	10
41	Whole-Exome Sequencing of Patients With Posterior Segment Uveitis. American Journal of Ophthalmology, 2021, 221, 246-259.	3.3	10
42	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
43	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
44	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
45	Sporadic hypothalamic hamartoma is a ciliopathy with somatic and bi-allelic contributions. Human Molecular Genetics, 2022, 31, 2307-2316.	2.9	8
46	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
47	Evaluation of multiple putative risk alleles within the 15q13.3 region for genetic generalized epilepsy. Epilepsy Research, 2015, 117, 70-73.	1.6	6
48	The emerging role of somatic tumor sequencing in the treatment of urothelial cancer. Asian Journal of Urology, 2021, 8, 391-399.	1.2	6
49	Combination therapies for MPNSTs targeting RABL6A-RB1 signaling. Oncotarget, 2021, 12, 10-14.	1.8	5
50	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
51	Development and comparison of novel bioluminescent mouse models of pancreatic neuroendocrine neoplasm metastasis. Scientific Reports, 2021, 11, 10252.	3.3	4
52	RABL6A Promotes Pancreatic Neuroendocrine Tumor Angiogenesis and Progression In Vivo. Biomedicines, 2021, 9, 633.	3.2	4
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	Additive and Interactive Genetically Contextual Effects of HbA1c on cg19693031 Methylation in Type 2 Diabetes. Genes, 2022, 13, 683.	2.4	4

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55	Neurogenetics in the Genome Era. , 2017, , 257-267.		3
56	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
57	Candidate modifier genes for immune function in 22q11.2 deletion syndrome. Molecular Genetics & Genomic Medicine, 2020, 8, e1057.	1.2	3
58	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
59	GE-02 * PARADOXICAL GENETIC AND EPIDEMIOLOGIC RELATIONSHIPS BETWEEN CANCER AND AUTISM. Neuro-Oncology, 2015, 17, iii7-iii7.	1.2	1
60	A case of primary cutaneous Ewing sarcoma in a neutropenic patient. Journal of Cutaneous Pathology, 2019, 46, 238-241.	1.3	1
61	CNV-ROC: A cost effective, computer-aided analytical performance evaluator of chromosomal microarrays. Journal of Biomedical Informatics, 2015, 54, 106-113.	4.3	0
62	Progressive Optic Disc Cupping Over 20 Years in a Patient with TBK1-Associated Glaucoma. Ophthalmology Glaucoma, 2020, 3, 167-168.	1.9	0