Erica E Davis

List of Publications by Year
in descending order

110 papers	8,603 citations	41344 49 h-index	48315 88 g-index
122 all docs	122 docs citations	122 times ranked	12194 citing authors

#	Article	IF	Citations
1	Multidisciplinary approaches for elucidating genetics and molecular pathogenesis of urinary tract malformations. Kidney International, 2022, 101, 473-484.	5.2	16
2	HEK293T Cells with TFAM Disruption by CRISPR-Cas9 as a Model for Mitochondrial Regulation. Life, 2022, 12, 22.	2.4	3
3	Testing for rare genetic causes of obesity: findings and experiences from a pediatric weight management program. International Journal of Obesity, 2022, 46, 1493-1501.	3.4	7
4	A <scp>BBS1 SVA</scp> F retrotransposon insertion is a frequent cause of <scp>Bardetâ€Biedl</scp> syndrome. Clinical Genetics, 2021, 99, 318-324.	2.0	21
5	De novo TRIM8 variants impair its protein localization to nuclear bodies and cause developmental delay, epilepsy, and focal segmental glomerulosclerosis. American Journal of Human Genetics, 2021, 108, 357-367.	6.2	14
6	Acoustofluidic rotational tweezing enables high-speed contactless morphological phenotyping of zebrafish larvae. Nature Communications, 2021, 12, 1118.	12.8	49
7	Haploinsufficiency of the Sin3/HDAC corepressor complex member SIN3B causes a syndromic intellectual disability/autism spectrum disorder. American Journal of Human Genetics, 2021, 108, 929-941.	6.2	15
8	A recessive variant in TFAM causes mtDNA depletion associated with primary ovarian insufficiency, seizures, intellectual disability and hearing loss. Human Genetics, 2021, 140, 1733-1751.	3.8	15
9	A case of Bardetâ€Biedl syndrome caused by a recurrent variant in BBS12: A case report. Biomedical Reports, 2021, 15, 103.	2.0	6
10	A recurrent rare intronic variant in CAPN3 alters mRNA splicing and causes autosomal recessive limbâ€girdle muscular dystrophyâ€1 in three Pakistani pedigrees. American Journal of Medical Genetics, Part A, 2021, , .	1.2	0
11	CSGALNACT1â€congenital disorder of glycosylation: A mild skeletal dysplasia with advanced bone age. Human Mutation, 2020, 41, 655-667.	2.5	15
12	Evidence for secondary-variant genetic burden and non-random distribution across biological modules in a recessive ciliopathy. Nature Genetics, 2020, 52, 1145-1150.	21.4	22
13	Mutations in FAM50A suggest that Armfield XLID syndrome is a spliceosomopathy. Nature Communications, 2020, 11, 3698.	12.8	38
14	Loss of CBY1 results in a ciliopathy characterized by features of Joubert syndrome. Human Mutation, 2020, 41, 2179-2194.	2.5	16
15	CFAP45 deficiency causes situs abnormalities and asthenospermia by disrupting an axonemal adenine nucleotide homeostasis module. Nature Communications, 2020, 11, 5520.	12.8	36
16	Mutations in the Kinesin-2 Motor KIF3B Cause an Autosomal-Dominant Ciliopathy. American Journal of Human Genetics, 2020, 106, 893-904.	6.2	29
17	TCF12 haploinsufficiency causes autosomal dominant Kallmann syndrome and reveals network-level interactions between causal loci. Human Molecular Genetics, 2020, 29, 2435-2450.	2.9	10
18	A Genocentric Approach to Discovery of Mendelian Disorders. American Journal of Human Genetics, 2019, 105, 974-986.	6.2	30

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19	Analysis of Single Nucleotide Variants in CRISPR-Cas9 Edited Zebrafish Exomes Shows No Evidence of Off-Target Inflation. Frontiers in Genetics, 2019, 10, 949.	2.3	7
20	Oligogenic Effects of 16p11.2 Copy-Number Variation on Craniofacial Development. Cell Reports, 2019, 28, 3320-3328.e4.	6.4	34
21	RNA sequencing of isolated cell populations expressing human APOL1 G2 risk variant reveals molecular correlates of sickle cell nephropathy in zebrafish podocytes. PLoS ONE, 2019, 14, e0217042.	2.5	3
22	Germline-Activating RRAS2 Mutations Cause Noonan Syndrome. American Journal of Human Genetics, 2019, 104, 1233-1240.	6.2	35
23	Bi-allelic Variants in DYNC1I2 Cause Syndromic Microcephaly with Intellectual Disability, Cerebral Malformations, and Dysmorphic Facial Features. American Journal of Human Genetics, 2019, 104, 1073-1087.	6.2	19
24	Recessive variants in ZNF142 cause a complex neurodevelopmental disorder with intellectual disability, speech impairment, seizures, and dystonia. Genetics in Medicine, 2019, 21, 2532-2542.	2.4	17
25	Mutations in NCAPG2 Cause a Severe Neurodevelopmental Syndrome that Expands the Phenotypic Spectrum of Condensinopathies. American Journal of Human Genetics, 2019, 104, 94-111.	6.2	27
26	SSBP1 mutations cause mtDNA depletion underlying a complex optic atrophy disorder. Journal of Clinical Investigation, 2019, 130, 108-125.	8.2	65
27	SAT-LB071 Loss of Function (LoF) mutations in TCF12 Cause Autosomal Dominant Kallmann Syndrome and Reveal Network-level Interactions Between Causal Loci. Journal of the Endocrine Society, 2019, 3, .	0.2	0
28	Novel CASK mutations in cases with syndromic microcephaly. Human Mutation, 2018, 39, 993-1001.	2.5	16
29	Dual Molecular Effects of Dominant RORA Mutations Cause Two Variants of Syndromic Intellectual Disability with Either Autism or Cerebellar Ataxia. American Journal of Human Genetics, 2018, 102, 744-759.	6.2	51
30	De Novo Pathogenic Variants in CACNA1E Cause Developmental and Epileptic Encephalopathy with Contractures, Macrocephaly, and Dyskinesias. American Journal of Human Genetics, 2018, 103, 666-678.	6.2	87
31	The Human FSGS-Causing ANLN R431C Mutation Induces Dysregulated PI3K/AKT/mTOR/Rac1 Signaling in Podocytes. Journal of the American Society of Nephrology: JASN, 2018, 29, 2110-2122.	6.1	51
32	Small molecule inhibition of RAS/MAPK signaling ameliorates developmental pathologies of Kabuki Syndrome. Scientific Reports, 2018, 8, 10779.	3.3	50
33	The complexity of the cilium: spatiotemporal diversity of an ancient organelle. Current Opinion in Cell Biology, 2018, 55, 139-149.	5.4	21
34	Participant-Partners in Genetic Research: An Exome Study with Families of Children with Unexplained Medical Conditions. Journal of Participatory Medicine, 2018, 10, e2.	1.3	2
35	SMCHD1 mutations associated with a rare muscular dystrophy can also cause isolated arhinia and Bosma arhinia microphthalmia syndrome. Nature Genetics, 2017, 49, 238-248.	21.4	131
36	A truncating mutation in CEP55 is the likely cause of MARCH, a novel syndrome affecting neuronal mitosis. Journal of Medical Genetics, 2017, 54, 490-501.	3.2	45

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37	Mutations in TMEM260 Cause a Pediatric Neurodevelopmental, Cardiac, and Renal Syndrome. American Journal of Human Genetics, 2017, 100, 666-675.	6.2	22
38	Exome-wide Association Study Identifies GREB1L Mutations in Congenital Kidney Malformations. American Journal of Human Genetics, 2017, 101, 789-802.	6.2	63
39	Temperature-activated ion channels in neural crest cells confer maternal feverâ€“associated birth defects. Science Signaling, 2017, 10, .	3.6	51
40	Haploinsufficiency of the Chromatin Remodeler BPTF Causes Syndromic Developmental and Speech Delay, Postnatal Microcephaly, and Dysmorphic Features. American Journal of Human Genetics, 2017, 101, 503-515.	6.2	61
41	Zebrafish knockout of Down syndrome gene, DYRK1A, shows social impairments relevant to autism. Molecular Autism, 2017, 8, 50.	4.9	86
42	Partial uniparental isodisomy of chromosome 16 unmasks a deleterious biallelic mutation in IFT140 that causes Mainzer-Saldino syndrome. Human Genomics, 2017, 11, 16.	2.9	22
43	Zebrafish: A Model System to Study the Architecture of Human Genetic Disease. , 2017, , 651-670.		2
44	A t(5;16) translocation is the likely driver of a syndrome with ambiguous genitalia, facial dysmorphism, intellectual disability, and speech delay. Journal of Physical Education and Sports Management, 2016, 2, a000703.	1.2	1
45	Copy-Number Variation Contributes to the Mutational Load of Bardet-Biedl Syndrome. American Journal of Human Genetics, 2016, 99, 318-336.	6.2	112
46	An organelle-specific protein landscape identifies novel diseases and molecular mechanisms. Nature Communications, 2016, 7, 11491.	12.8	207
47	Targeted resequencing identifies PTCH1 as a major contributor to ocular developmental anomalies and extends the SOX2 regulatory network. Genome Research, 2016, 26, 474-485.	5.5	37
48	DNAH11 Localization in the Proximal Region of Respiratory Cilia Defines Distinct Outer Dynein Arm Complexes. American Journal of Respiratory Cell and Molecular Biology, 2016, 55, 213-224.	2.9	107
49	GWAS Meta-Analysis of Glomerular Filtration Rate in Three Cohorts of Sickle Cell Disease Patients and In Vivo Functional Analysis Reveals Potential Nephropathy Candidate Genes. Blood, 2016, 128, 269-269.	1.4	0
50	Unique among ciliopathies: primary ciliary dyskinesia, a motile cilia disorder. F1000prime Reports, 2015, 7, 36.	5.9	58
51	In vivo Modeling Implicates APOL1 in Nephropathy: Evidence for Dominant Negative Effects and Epistasis under Anemic Stress. PLoS Genetics, 2015, 11, e1005349.	3.5	45
52	Ectopic Expression of Retrotransposon-Derived PEG11/RTL1 Contributes to the Callipyge Muscular Hypertrophy. PLoS ONE, 2015, 10, e0140594.	2.5	14
53	Loss of Function Mutations in NNT Are Associated With Left Ventricular Noncompaction. Circulation: Cardiovascular Genetics, 2015, 8, 544-552.	5.1	48
54	De Novo GMNN Mutations Cause Autosomal-Dominant Primordial Dwarfism Associated with Meier-Gorlin Syndrome. American Journal of Human Genetics, 2015, 97, 904-913.	6.2	65

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55	Mutations in Either TUBB or MAPRE2 Cause Circumferential Skin Creases Kunze Type. American Journal of Human Genetics, 2015, 97, 790-800.	6.2	63
56	TAF1 Variants Are Associated with Dysmorphic Features, Intellectual Disability, and Neurological Manifestations. American Journal of Human Genetics, 2015, 97, 922-932.	6.2	101
57	Identification of cis-suppression of human disease mutations by comparative genomics. Nature, 2015, 524, 225-229.	27.8	106
58	Rapid and Efficient Generation of Transgene-Free iPSC from a Small Volume of Cryopreserved Blood. Stem Cell Reviews and Reports, 2015, 11, 652-665.	5.6	34
59	TMEM231, mutated in orofaciodigital and Meckel syndromes, organizes the ciliary transition zone. Journal of Cell Biology, 2015, 209, 129-142.	5.2	95
60	Joubert Syndrome in French Canadians and Identification of Mutations in CEP104. American Journal of Human Genetics, 2015, 97, 744-753.	6.2	56
61	The kinetochore protein,CENPF, is mutated in human ciliopathy and microcephaly phenotypes. Journal of Medical Genetics, 2015, 52, 147-156.	3.2	75
62	A Novel Ribosomopathy Caused by Dysfunction of RPL10 Disrupts Neurodevelopment and Causes X-Linked Microcephaly in Humans. Genetics, 2014, 198, 723-733.	2.9	92
63	Whole-exome resequencing distinguishes cystic kidney diseases from phenocopies in renal ciliopathies. Kidney International, 2014, 85, 880-887.	5.2	67
64	Whole Exome Sequencing of a Dominant Retinitis Pigmentosa Family Identifies a Novel Deletion inPRPF31. , 2014, 55, 2121.		26
65	Mutations in CSPP1, Encoding a Core Centrosomal Protein, Cause a Range of Ciliopathy Phenotypes in Humans. American Journal of Human Genetics, 2014, 94, 73-79.	6.2	77
66	Dissecting Intraflagellar Transport, One Molecule at a Time. Developmental Cell, 2014, 31, 263-264.	7.0	3
67	Recurrent CNVs and SNVs at the NPHP1 Locus Contribute Pathogenic Alleles to Bardet-Biedl Syndrome. American Journal of Human Genetics, 2014, 94, 745-754.	6.2	80
68	Interpreting human genetic variation with in vivo zebrafish assays. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2014, 1842, 1960-1970.	3.8	63
69	Whole exome sequencing and functional studies identify an intronic mutation in <scp>TRAPPC2</scp> that causes <scp>SEDT</scp>. Clinical Genetics, 2014, 85, 359-364.	2.0	9
70	Evidence for a Dominant Negative Effect Conferred By the APOL1 G2 Sickle Cell Nephropathy Risk Allele in an in Vivo Model. Blood, 2014, 124, 1374-1374.	1.4	0
71	ARMC4 Mutations Cause Primary Ciliary Dyskinesia with Randomization of Left/Right Body Asymmetry. American Journal of Human Genetics, 2013, 93, 357-367.	6.2	150
72	TM4SF20 Ancestral Deletion and Susceptibility to a Pediatric Disorder of Early Language Delay and Cerebral White Matter Hyperintensities. American Journal of Human Genetics, 2013, 93, 197-210.	6.2	43

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73	Defects in the IFT-B Component IFT172 Cause Jeune and Mainzer-Saldino Syndromes in Humans. American Journal of Human Genetics, 2013, 93, 915-925.	6.2	196
74	In Vivo Modeling of the Morbid Human Genome using Danio rerio. Journal of Visualized Experiments, 2013, , e50338.	0.3	49
75	Mutations in LRRC50 Predispose Zebrafish and Humans to Seminomas. PLoS Genetics, 2013, 9, e1003384.	3.5	38
76	Combining fetal sonography with genetic and allele pathogenicity studies to secure a neonatal diagnosis of Bardetâ€“Biedl syndrome. Clinical Genetics, 2013, 83, 553-559.	2.0	10
77	Delta-Like 1 Homolog (Dlk1): A Marker for Rhabdomyosarcomas Implicated in Skeletal Muscle Regeneration. PLoS ONE, 2013, 8, e60692.	2.5	9
78	In Vivo Modeling Of Genetic Mechanisms Associated With Sickle Cell Disease Nephropathy. Blood, 2013, 122, 2224-2224.	1.4	3
79	Evolutionarily Assembled cis-Regulatory Module at a Human Ciliopathy Locus. Science, 2012, 335, 966-969.	12.6	84
80	OTX2mutations contribute to the otocephaly-dysgnathia complex. Journal of Medical Genetics, 2012, 49, 373-379.	3.2	58
81	Gene therapy rescues cilia defects and restores olfactory function in a mammalian ciliopathy model. Nature Medicine, 2012, 18, 1423-1428.	30.7	103
82	The ciliopathies: a transitional model into systems biology of human genetic disease. Current Opinion in Genetics and Development, 2012, 22, 290-303.	3.3	137
83	CCDC39 is required for assembly of inner dynein arms and the dynein regulatory complex and for normal ciliary motility in humans and dogs. Nature Genetics, 2011, 43, 72-78.	21.4	302
84	TTC21B contributes both causal and modifying alleles across the ciliopathy spectrum. Nature Genetics, 2011, 43, 189-196.	21.4	326
85	KIF7 mutations cause fetal hydrolethalus and acrocallosal syndromes. Nature Genetics, 2011, 43, 601-606.	21.4	203
86	TMEM237 Is Mutated in Individuals with a Joubert Syndrome Related Disorder and Expands the Role of the TMEM Family at the Ciliary Transition Zone. American Journal of Human Genetics, 2011, 89, 713-730.	6.2	178
87	Mutation analysis in Bardetâ€“Biedl syndrome by DNA pooling and massively parallel resequencing in 105 individuals. Human Genetics, 2011, 129, 79-90.	3.8	80
88	Identification of 28 novel mutations in the Bardetâ€“Biedl syndrome genes: the burden of private mutations in an extensively heterogeneous disease. Human Genetics, 2010, 127, 583-593.	3.8	109
89	Mutations in TMEM216 perturb ciliogenesis and cause Joubert, Meckel and related syndromes. Nature Genetics, 2010, 42, 619-625.	21.4	261
90	Identification of 11 novel mutations in eight BBS genes by high-resolution homozygosity mapping. Journal of Medical Genetics, 2010, 47, 262-267.	3.2	67

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91	Pitchfork Regulates Primary Cilia Disassembly and Left-Right Asymmetry. Developmental Cell, 2010, 19, 66-77.	7.0	133
92	Individuals with mutations in XPNPEP3, which encodes a mitochondrial protein, develop a nephronophthisis-like nephropathy. Journal of Clinical Investigation, 2010, 120, 791-802.	8.2	102
93	Individuals with mutations in XPNPEP3, which encodes a mitochondrial protein, develop a nephronophthisis-like nephropathy. Journal of Clinical Investigation, 2010, 120, 1362-1362.	8.2	0
94	Epistasis between RET and BBS mutations modulates enteric innervation and causes syndromic Hirschsprung disease. Proceedings of the National Academy of Sciences of the United States of America, 2009, 106, 13921-13926.	7.1	51
95	Functional interactions between the ciliopathy-associated Meckel syndrome 1 (MKS1) protein and two novel MKS1-related (MKSR) proteins. Journal of Cell Science, 2009, 122, 611-624.	2.0	71
96	A common allele in RPGRIP1L is a modifier of retinal degeneration in ciliopathies. Nature Genetics, 2009, 41, 739-745.	21.4	255
97	The Vertebrate Primary Cilium in Development, Homeostasis, and Disease. Cell, 2009, 137, 32-45.	28.9	653
98	CC2D2A Is Mutated in Joubert Syndrome and Interacts with the Ciliopathy-Associated Basal Body Protein CEP290. American Journal of Human Genetics, 2008, 83, 559-571.	6.2	202
99	Hypomorphic mutations in syndromic encephalocele genes are associated with Bardet-Biedl syndrome. Nature Genetics, 2008, 40, 443-448.	21.4	367
100	An Essential Role for DYF-11/MIP-T3 in Assembling Functional Intraflagellar Transport Complexes. PLoS Genetics, 2008, 4, e1000044.	3.5	48
101	Cell Polarization Defects in Early Heart Development. Circulation Research, 2007, 101, 122-124.	4.5	8
102	Identification of a Novel BBS Gene (BBS12) Highlights the Major Role of a Vertebrate-Specific Branch of Chaperonin-Related Proteins in Bardet-Biedl Syndrome. American Journal of Human Genetics, 2007, 80, 1-11.	6.2	219
103	The Emerging Complexity of the Vertebrate Cilium: New Functional Roles for an Ancient Organelle. Developmental Cell, 2006, 11, 9-19.	7.0	131
104	The ciliary proteome database: an integrated community resource for the genetic and functional dissection of cilia. Nature Genetics, 2006, 38, 961-962.	21.4	265
105	BBS10 encodes a vertebrate-specific chaperonin-like protein and is a major BBS locus. Nature Genetics, 2006, 38, 521-524.	21.4	259
106	RNAi-Mediated Allelic trans-Interaction at the Imprinted Rtl1/Peg11 Locus. Current Biology, 2005, 15, 743-749.	3.9	301
107	RNAi-Mediated Allelic trans-Interaction at the Imprinted Rtl1/Peg11 Locus. Current Biology, 2005, 15, 884.	3.9	2
108	Ectopic Expression of DLK1 Protein in Skeletal Muscle of Padumnal Heterozygotes Causes the Callipyge Phenotype. Current Biology, 2004, 14, 1858-1862.	3.9	114

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109	Toward Molecular Understanding of Polar Overdominance at the Ovine Callipyge Locus. Cold Spring Harbor Symposia on Quantitative Biology, 2004, 69, 477-484.	1.1	11
110	Oligogenic Effects of 16p11.2 Copy Number Variation on Craniofacial Development. SSRN Electronic Journal, 0, , .	0.4	1