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	The Vertebrate Primary Cilium in Development, Homeostasis, and Disease. Cell, 2009, 137, 32-45.	28.9	653
2	Hypomorphic mutations in syndromic encephalocele genes are associated with Bardet-Biedl syndrome. Nature Genetics, 2008, 40, 443-448.	21.4	367
3	TTC21B contributes both causal and modifying alleles across the ciliopathy spectrum. Nature Genetics, 2011, 43, 189-196.	21.4	326
4	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
5	RNAi-Mediated Allelic trans-Interaction at the Imprinted Rtl1/Peg11 Locus. Current Biology, 2005, 15, 743-749.	3.9	301
6	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
7	Mutations in TMEM216 perturb ciliogenesis and cause Joubert, Meckel and related syndromes. Nature Genetics, 2010, 42, 619-625.	21.4	261
8	BBS10 encodes a vertebrate-specific chaperonin-like protein and is a major BBS locus. Nature Genetics, 2006, 38, 521-524.	21.4	259
9	A common allele in RPGRIP1L is a modifier of retinal degeneration in ciliopathies. Nature Genetics, 2009, 41, 739-745.	21.4	255
10	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
11	An organelle-specific protein landscape identifies novel diseases and molecular mechanisms. Nature Communications, 2016, 7, 11491.	12.8	207
12	KIF7 mutations cause fetal hydrolethalus and acrocallosal syndromes. Nature Genetics, 2011, 43, 601-606.	21.4	203
13	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
14	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
15	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
16	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
17	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
18	Pitchfork Regulates Primary Cilia Disassembly and Left-Right Asymmetry. Developmental Cell, 2010, 19, 66-77.	7.0	133

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19	The Emerging Complexity of the Vertebrate Cilium: New Functional Roles for an Ancient Organelle. Developmental Cell, 2006, 11, 9-19.	7.0	131
20	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
21	Ectopic Expression of DLK1 Protein in Skeletal Muscle of Padumnal Heterozygotes Causes the Callipyge Phenotype. Current Biology, 2004, 14, 1858-1862.	3.9	114
22	Copy-Number Variation Contributes to the Mutational Load of Bardet-Biedl Syndrome. American Journal of Human Genetics, 2016, 99, 318-336.	6.2	112
23	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
24	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
25	Identification of cis-suppression of human disease mutations by comparative genomics. Nature, 2015, 524, 225-229.	27.8	106
26	Gene therapy rescues cilia defects and restores olfactory function in a mammalian ciliopathy model. Nature Medicine, 2012, 18, 1423-1428.	30.7	103
27	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
28	TAF1 Variants Are Associated with Dysmorphic Features, Intellectual Disability, and Neurological Manifestations. American Journal of Human Genetics, 2015, 97, 922-932.	6.2	101
29	TMEM231, mutated in orofaciodigital and Meckel syndromes, organizes the ciliary transition zone. Journal of Cell Biology, 2015, 209, 129-142.	5.2	95
30	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
31	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
32	Zebrafish knockout of Down syndrome gene, DYRK1A, shows social impairments relevant to autism. Molecular Autism, 2017, 8, 50.	4.9	86
33	Evolutionarily Assembled cis-Regulatory Module at a Human Ciliopathy Locus. Science, 2012, 335, 966-969.	12.6	84
34	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
35	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
36	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

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37	The kinetochore protein,CENPF, is mutated in human ciliopathy and microcephaly phenotypes. Journal of Medical Genetics, 2015, 52, 147-156.	3.2	75
38	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
39	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
40	Whole-exome resequencing distinguishes cystic kidney diseases from phenocopies in renal ciliopathies. Kidney International, 2014, 85, 880-887.	5.2	67
41	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
42	SSBP1 mutations cause mtDNA depletion underlying a complex optic atrophy disorder. Journal of Clinical Investigation, 2019, 130, 108-125.	8.2	65
43	Interpreting human genetic variation with in vivo zebrafish assays. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2014, 1842, 1960-1970.	3.8	63
44	Mutations in Either TUBB or MAPRE2 Cause Circumferential Skin Creases Kunze Type. American Journal of Human Genetics, 2015, 97, 790-800.	6.2	63
45	Exome-wide Association Study Identifies GREB1L Mutations in Congenital Kidney Malformations. American Journal of Human Genetics, 2017, 101, 789-802.	6.2	63
46	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
47	OTX2mutations contribute to the otocephaly-dysgnathia complex. Journal of Medical Genetics, 2012, 49, 373-379.	3.2	58
48	Unique among ciliopathies: primary ciliary dyskinesia, a motile cilia disorder. F1000prime Reports, 2015, 7, 36.	5.9	58
49	Joubert Syndrome in French Canadians and Identification of Mutations in CEP104. American Journal of Human Genetics, 2015, 97, 744-753.	6.2	56
50	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
51	Temperature-activated ion channels in neural crest cells confer maternal feverâ€"associated birth defects. Science Signaling, 2017, 10, .	3.6	51
52	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
53	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
54	Small molecule inhibition of RAS/MAPK signaling ameliorates developmental pathologies of Kabuki Syndrome. Scientific Reports, 2018, 8, 10779.	3.3	50

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55	In Vivo Modeling of the Morbid Human Genome using Danio rerio. Journal of Visualized Experiments, 2013, , e50338.	0.3	49
56	Acoustofluidic rotational tweezing enables high-speed contactless morphological phenotyping of zebrafish larvae. Nature Communications, 2021, 12, 1118.	12.8	49
57	An Essential Role for DYF-11/MIP-T3 in Assembling Functional Intraflagellar Transport Complexes. PLoS Genetics, 2008, 4, e1000044.	3.5	48
58	Loss of Function Mutations in NNT Are Associated With Left Ventricular Noncompaction. Circulation: Cardiovascular Genetics, 2015, 8, 544-552.	5.1	48
59	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
60	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
61	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
62	Mutations in LRRC50 Predispose Zebrafish and Humans to Seminomas. PLoS Genetics, 2013, 9, e1003384.	3.5	38
63	Mutations in FAM50A suggest that Armfield XLID syndrome is a spliceosomopathy. Nature Communications, 2020, 11, 3698.	12.8	38
64	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
65	CFAP45 deficiency causes situs abnormalities and asthenospermia by disrupting an axonemal adenine nucleotide homeostasis module. Nature Communications, 2020, 11, 5520.	12.8	36
66	Germline-Activating RRAS2 Mutations Cause Noonan Syndrome. American Journal of Human Genetics, 2019, 104, 1233-1240.	6.2	35
67	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
68	Oligogenic Effects of 16p11.2 Copy-Number Variation on Craniofacial Development. Cell Reports, 2019, 28, 3320-3328.e4.	6.4	34
69	A Genocentric Approach to Discovery of Mendelian Disorders. American Journal of Human Genetics, 2019, 105, 974-986.	6.2	30
70	Mutations in the Kinesin-2 Motor KIF3B Cause an Autosomal-Dominant Ciliopathy. American Journal of Human Genetics, 2020, 106, 893-904.	6.2	29
71	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
72	Whole Exome Sequencing of a Dominant Retinitis Pigmentosa Family Identifies a Novel Deletion inPRPF31. , 2014, 55, 2121.		26

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73	Mutations in TMEM260 Cause a Pediatric Neurodevelopmental, Cardiac, and Renal Syndrome. American Journal of Human Genetics, 2017, 100, 666-675.	6.2	22
74	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
75	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
76	The complexity of the cilium: spatiotemporal diversity of an ancient organelle. Current Opinion in Cell Biology, 2018, 55, 139-149.	5.4	21
77	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
78	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
79	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
80	Novel CASK mutations in cases with syndromic microcephaly. Human Mutation, 2018, 39, 993-1001.	2.5	16
81	Loss of CBY1 results in a ciliopathy characterized by features of Joubert syndrome. Human Mutation, 2020, 41, 2179-2194.	2.5	16
82	Multidisciplinary approaches for elucidating genetics and molecular pathogenesis of urinary tract malformations. Kidney International, 2022, 101, 473-484.	5.2	16
83	CSGALNACT1â€congenital disorder of glycosylation: A mild skeletal dysplasia with advanced bone age. Human Mutation, 2020, 41, 655-667.	2.5	15
84	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
85	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
86	Ectopic Expression of Retrotransposon-Derived PEG11/RTL1 Contributes to the Callipyge Muscular Hypertrophy. PLoS ONE, 2015, 10, e0140594.	2.5	14
87	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
88	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
89	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
90	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

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91	Delta-Like 1 Homolog (Dlk1): A Marker for Rhabdomyosarcomas Implicated in Skeletal Muscle Regeneration. PLoS ONE, 2013, 8, e60692.	2.5	9
92	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
93	Cell Polarization Defects in Early Heart Development. Circulation Research, 2007, 101, 122-124.	4.5	8
94	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
95	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
96	A case of Bardetâ€'Biedl syndrome caused by a recurrent variant in BBS12: A case report. Biomedical Reports, 2021, 15, 103.	2.0	6
97	Dissecting Intraflagellar Transport, One Molecule at a Time. Developmental Cell, 2014, 31, 263-264.	7.0	3
98	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
99	In Vivo Modeling Of Genetic Mechanisms Associated With Sickle Cell Disease Nephropathy. Blood, 2013, 122, 2224-2224.	1.4	3
100	HEK293T Cells with TFAM Disruption by CRISPR-Cas9 as a Model for Mitochondrial Regulation. Life, 2022, 12, 22.	2.4	3
101	RNAi-Mediated Allelic trans-Interaction at the Imprinted Rtl1/Peg11 Locus. Current Biology, 2005, 15, 884.	3.9	2
102	Zebrafish: A Model System to Study the Architecture of Human Genetic Disease. , 2017, , 651-670.		2
103	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
104	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
105	Oligogenic Effects of 16p11.2 Copy Number Variation on Craniofacial Development. SSRN Electronic Journal, 0, , .	0.4	1
106	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
107	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
108	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

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109	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
110	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