

# Erica E Davis

## List of Publications by Year in descending order

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110  
papers

8,603  
citations

41258

49  
h-index

48187

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. <i>Kidney International</i> , 2022, 101, 473-484.	2.6	16
2	HEK293T Cells with TFAM Disruption by CRISPR-Cas9 as a Model for Mitochondrial Regulation. <i>Life</i> , 2022, 12, 22.	1.1	3
3	Testing for rare genetic causes of obesity: findings and experiences from a pediatric weight management program. <i>International Journal of Obesity</i> , 2022, 46, 1493-1501.	1.6	7
4	A <i>BBS1</i> SVA retrotransposon insertion is a frequent cause of Bardet-Biedl syndrome. <i>Clinical Genetics</i> , 2021, 99, 318-324.	1.0	21
5	De novo TRIM8 variants impair its protein localization to nuclear bodies and cause developmental delay, epilepsy, and focal segmental glomerulosclerosis. <i>American Journal of Human Genetics</i> , 2021, 108, 357-367.	2.6	14
6	Acoustofluidic rotational tweezing enables high-speed contactless morphological phenotyping of zebrafish larvae. <i>Nature Communications</i> , 2021, 12, 1118.	5.8	49
7	Haploinsufficiency of the Sin3/HDAC corepressor complex member SIN3B causes a syndromic intellectual disability/autism spectrum disorder. <i>American Journal of Human Genetics</i> , 2021, 108, 929-941.	2.6	15
8	A recessive variant in TFAM causes mtDNA depletion associated with primary ovarian insufficiency, seizures, intellectual disability and hearing loss. <i>Human Genetics</i> , 2021, 140, 1733-1751.	1.8	15
9	A case of Bardet-Biedl syndrome caused by a recurrent variant in BBS12: A case report. <i>Biomedical Reports</i> , 2021, 15, 103.	0.9	6
10	A recurrent rare intronic variant in CAPN3 alters mRNA splicing and causes autosomal recessive limb-girdle muscular dystrophy in three Pakistani pedigrees. <i>American Journal of Medical Genetics, Part A</i> , 2021, , .	0.7	0
11	CSGALNACT1 congenital disorder of glycosylation: A mild skeletal dysplasia with advanced bone age. <i>Human Mutation</i> , 2020, 41, 655-667.	1.1	15
12	Evidence for secondary-variant genetic burden and non-random distribution across biological modules in a recessive ciliopathy. <i>Nature Genetics</i> , 2020, 52, 1145-1150.	9.4	22
13	Mutations in FAM50A suggest that Armfield XLID syndrome is a spliceosomopathy. <i>Nature Communications</i> , 2020, 11, 3698.	5.8	38
14	Loss of CBY1 results in a ciliopathy characterized by features of Joubert syndrome. <i>Human Mutation</i> , 2020, 41, 2179-2194.	1.1	16
15	CFAP45 deficiency causes situs abnormalities and asthenospermia by disrupting an axonemal adenine nucleotide homeostasis module. <i>Nature Communications</i> , 2020, 11, 5520.	5.8	36
16	Mutations in the Kinesin-2 Motor KIF3B Cause an Autosomal-Dominant Ciliopathy. <i>American Journal of Human Genetics</i> , 2020, 106, 893-904.	2.6	29
17	TCF12 haploinsufficiency causes autosomal dominant Kallmann syndrome and reveals network-level interactions between causal loci. <i>Human Molecular Genetics</i> , 2020, 29, 2435-2450.	1.4	10
18	A Genocentric Approach to Discovery of Mendelian Disorders. <i>American Journal of Human Genetics</i> , 2019, 105, 974-986.	2.6	30

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19	Analysis of Single Nucleotide Variants in CRISPR-Cas9 Edited Zebrafish Exomes Shows No Evidence of Off-Target Inflation. <i>Frontiers in Genetics</i> , 2019, 10, 949.	1.1	7
20	Oligogenic Effects of 16p11.2 Copy-Number Variation on Craniofacial Development. <i>Cell Reports</i> , 2019, 28, 3320-3328.e4.	2.9	34
21	RNA sequencing of isolated cell populations expressing human APOL1 G2 risk variant reveals molecular correlates of sickle cell nephropathy in zebrafish podocytes. <i>PLoS ONE</i> , 2019, 14, e0217042.	1.1	3
22	Germline-Activating RAS2 Mutations Cause Noonan Syndrome. <i>American Journal of Human Genetics</i> , 2019, 104, 1233-1240.	2.6	35
23	Bi-allelic Variants in DYNC112 Cause Syndromic Microcephaly with Intellectual Disability, Cerebral Malformations, and Dysmorphic Facial Features. <i>American Journal of Human Genetics</i> , 2019, 104, 1073-1087.	2.6	19
24	Recessive variants in ZNF142 cause a complex neurodevelopmental disorder with intellectual disability, speech impairment, seizures, and dystonia. <i>Genetics in Medicine</i> , 2019, 21, 2532-2542.	1.1	17
25	Mutations in NCAPG2 Cause a Severe Neurodevelopmental Syndrome that Expands the Phenotypic Spectrum of Condensinopathies. <i>American Journal of Human Genetics</i> , 2019, 104, 94-111.	2.6	27
26	SSBP1 mutations cause mtDNA depletion underlying a complex optic atrophy disorder. <i>Journal of Clinical Investigation</i> , 2019, 130, 108-125.	3.9	65
27	SAT-LB071 Loss of Function (LoF) mutations in TCF12 Cause Autosomal Dominant Kallmann Syndrome and Reveal Network-level Interactions Between Causal Loci. <i>Journal of the Endocrine Society</i> , 2019, 3, .	0.1	0
28	Novel CASK mutations in cases with syndromic microcephaly. <i>Human Mutation</i> , 2018, 39, 993-1001.	1.1	16
29	Dual Molecular Effects of Dominant RORA Mutations Cause Two Variants of Syndromic Intellectual Disability with Either Autism or Cerebellar Ataxia. <i>American Journal of Human Genetics</i> , 2018, 102, 744-759.	2.6	51
30	De Novo Pathogenic Variants in CACNA1E Cause Developmental and Epileptic Encephalopathy with Contractures, Macrocephaly, and Dyskinesias. <i>American Journal of Human Genetics</i> , 2018, 103, 666-678.	2.6	87
31	The Human FSGS-Causing ANLN R431C Mutation Induces Dysregulated PI3K/AKT/mTOR/Rac1 Signaling in Podocytes. <i>Journal of the American Society of Nephrology: JASN</i> , 2018, 29, 2110-2122.	3.0	51
32	Small molecule inhibition of RAS/MAPK signaling ameliorates developmental pathologies of Kabuki Syndrome. <i>Scientific Reports</i> , 2018, 8, 10779.	1.6	50
33	The complexity of the cilium: spatiotemporal diversity of an ancient organelle. <i>Current Opinion in Cell Biology</i> , 2018, 55, 139-149.	2.6	21
34	Participant-Partners in Genetic Research: An Exome Study with Families of Children with Unexplained Medical Conditions. <i>Journal of Participatory Medicine</i> , 2018, 10, e2.	0.7	2
35	SMCHD1 mutations associated with a rare muscular dystrophy can also cause isolated arhinia and Bosma arhinia microphthalmia syndrome. <i>Nature Genetics</i> , 2017, 49, 238-248.	9.4	131
36	A truncating mutation in CEP55 is the likely cause of MARCH, a novel syndrome affecting neuronal mitosis. <i>Journal of Medical Genetics</i> , 2017, 54, 490-501.	1.5	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.	2.6	22
38	Exome-wide Association Study Identifies GREB1L Mutations in Congenital Kidney Malformations. American Journal of Human Genetics, 2017, 101, 789-802.	2.6	63
39	Temperature-activated ion channels in neural crest cells confer maternal fever-associated birth defects. Science Signaling, 2017, 10, .	1.6	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.	2.6	61
41	Zebrafish knockout of Down syndrome gene, DYRK1A, shows social impairments relevant to autism. Molecular Autism, 2017, 8, 50.	2.6	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.	1.4	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.	0.5	1
45	Copy-Number Variation Contributes to the Mutational Load of Bardet-Biedl Syndrome. American Journal of Human Genetics, 2016, 99, 318-336.	2.6	112
46	An organelle-specific protein landscape identifies novel diseases and molecular mechanisms. Nature Communications, 2016, 7, 11491.	5.8	207
47	Targeted resequencing identifies <i>PTCH1</i> as a major contributor to ocular developmental anomalies and extends the SOX2 regulatory network. Genome Research, 2016, 26, 474-485.	2.4	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.	1.4	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.	0.6	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.	1.5	45
52	Ectopic Expression of Retrotransposon-Derived PEG11/RTL1 Contributes to the Callipyge Muscular Hypertrophy. PLoS ONE, 2015, 10, e0140594.	1.1	14
53	Loss of Function Mutations in <i>NNT</i> 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.	2.6	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.	2.6	63
56	TAF1 Variants Are Associated with Dysmorphic Features, Intellectual Disability, and Neurological Manifestations. American Journal of Human Genetics, 2015, 97, 922-932.	2.6	101
57	Identification of cis-suppression of human disease mutations by comparative genomics. Nature, 2015, 524, 225-229.	13.7	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 orofacioidigital and Meckel syndromes, organizes the ciliary transition zone. Journal of Cell Biology, 2015, 209, 129-142.	2.3	95
60	Joubert Syndrome in French Canadians and Identification of Mutations in CEP104. American Journal of Human Genetics, 2015, 97, 744-753.	2.6	56
61	The kinetochore protein, <i>CENPF</i> , is mutated in human ciliopathy and microcephaly phenotypes. Journal of Medical Genetics, 2015, 52, 147-156.	1.5	75
62	A Novel Ribosomopathy Caused by Dysfunction of RPL10 Disrupts Neurodevelopment and Causes X-Linked Microcephaly in Humans. Genetics, 2014, 198, 723-733.	1.2	92
63	Whole-exome resequencing distinguishes cystic kidney diseases from phenocopies in renal ciliopathies. Kidney International, 2014, 85, 880-887.	2.6	67
64	Whole Exome Sequencing of a Dominant Retinitis Pigmentosa Family Identifies a Novel Deletion in <i>PRPF31</i> . , 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.	2.6	77
66	Dissecting Intraflagellar Transport, One Molecule at a Time. Developmental Cell, 2014, 31, 263-264.	3.1	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.	2.6	80
68	Interpreting human genetic variation with in vivo zebrafish assays. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2014, 1842, 1960-1970.	1.8	63
69	Whole exome sequencing and functional studies identify an intronic mutation in <i>TRAPPC2</i> that causes <i>SED1</i> . Clinical Genetics, 2014, 85, 359-364.	1.0	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.	0.6	0
71	ARMC4 Mutations Cause Primary Ciliary Dyskinesia with Randomization of Left/Right Body Asymmetry. American Journal of Human Genetics, 2013, 93, 357-367.	2.6	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.	2.6	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.	2.6	196
74	&lt;em&gt;In Vivo&lt;/em&gt; Modeling of the Morbid Human Genome using &lt;em&gt;Danio rerio&lt;/em&gt;. Journal of Visualized Experiments, 2013, , e50338.	0.2	49
75	Mutations in LRRC50 Predispose Zebrafish and Humans to Seminomas. PLoS Genetics, 2013, 9, e1003384.	1.5	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.	1.0	10
77	Delta-Like 1 Homolog (Dlk1): A Marker for Rhabdomyosarcomas Implicated in Skeletal Muscle Regeneration. PLoS ONE, 2013, 8, e60692.	1.1	9
78	In Vivo Modeling Of Genetic Mechanisms Associated With Sickle Cell Disease Nephropathy. Blood, 2013, 122, 2224-2224.	0.6	3
79	Evolutionarily Assembled cis-Regulatory Module at a Human Ciliopathy Locus. Science, 2012, 335, 966-969.	6.0	84
80	<i>OTX2</i> mutations contribute to the otocephaly-dysgnathia complex. Journal of Medical Genetics, 2012, 49, 373-379.	1.5	58
81	Gene therapy rescues cilia defects and restores olfactory function in a mammalian ciliopathy model. Nature Medicine, 2012, 18, 1423-1428.	15.2	103
82	The ciliopathies: a transitional model into systems biology of human genetic disease. Current Opinion in Genetics and Development, 2012, 22, 290-303.	1.5	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.	9.4	302
84	TTC21B contributes both causal and modifying alleles across the ciliopathy spectrum. Nature Genetics, 2011, 43, 189-196.	9.4	326
85	KIF7 mutations cause fetal hydroletharus and acrocallosal syndromes. Nature Genetics, 2011, 43, 601-606.	9.4	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.	2.6	178
87	Mutation analysis in Bardetâ€Biedl syndrome by DNA pooling and massively parallel resequencing in 105 individuals. Human Genetics, 2011, 129, 79-90.	1.8	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.	1.8	109
89	Mutations in TMEM216 perturb ciliogenesis and cause Joubert, Meckel and related syndromes. Nature Genetics, 2010, 42, 619-625.	9.4	261
90	Identification of 11 novel mutations in eight BBS genes by high-resolution homozygosity mapping. Journal of Medical Genetics, 2010, 47, 262-267.	1.5	67

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91	Pitchfork Regulates Primary Cilia Disassembly and Left-Right Asymmetry. <i>Developmental Cell</i> , 2010, 19, 66-77.	3.1	133
92	Individuals with mutations in XPNPEP3, which encodes a mitochondrial protein, develop a nephronophthisis-like nephropathy. <i>Journal of Clinical Investigation</i> , 2010, 120, 791-802.	3.9	102
93	Individuals with mutations in XPNPEP3, which encodes a mitochondrial protein, develop a nephronophthisis-like nephropathy. <i>Journal of Clinical Investigation</i> , 2010, 120, 1362-1362.	3.9	0
94	Epistasis between RET and BBS mutations modulates enteric innervation and causes syndromic Hirschsprung disease. <i>Proceedings of the National Academy of Sciences of the United States of America</i> , 2009, 106, 13921-13926.	3.3	51
95	Functional interactions between the ciliopathy-associated Meckel syndrome 1 (MKS1) protein and two novel MKS1-related (MKSR) proteins. <i>Journal of Cell Science</i> , 2009, 122, 611-624.	1.2	71
96	A common allele in RPGRIP1L is a modifier of retinal degeneration in ciliopathies. <i>Nature Genetics</i> , 2009, 41, 739-745.	9.4	255
97	The Vertebrate Primary Cilium in Development, Homeostasis, and Disease. <i>Cell</i> , 2009, 137, 32-45.	13.5	653
98	CC2D2A Is Mutated in Joubert Syndrome and Interacts with the Ciliopathy-Associated Basal Body Protein CEP290. <i>American Journal of Human Genetics</i> , 2008, 83, 559-571.	2.6	202
99	Hypomorphic mutations in syndromic encephalocele genes are associated with Bardet-Biedl syndrome. <i>Nature Genetics</i> , 2008, 40, 443-448.	9.4	367
100	An Essential Role for DYF-11/MIP-T3 in Assembling Functional Intraflagellar Transport Complexes. <i>PLoS Genetics</i> , 2008, 4, e1000044.	1.5	48
101	Cell Polarization Defects in Early Heart Development. <i>Circulation Research</i> , 2007, 101, 122-124.	2.0	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. <i>American Journal of Human Genetics</i> , 2007, 80, 1-11.	2.6	219
103	The Emerging Complexity of the Vertebrate Cilium: New Functional Roles for an Ancient Organelle. <i>Developmental Cell</i> , 2006, 11, 9-19.	3.1	131
104	The ciliary proteome database: an integrated community resource for the genetic and functional dissection of cilia. <i>Nature Genetics</i> , 2006, 38, 961-962.	9.4	265
105	BBS10 encodes a vertebrate-specific chaperonin-like protein and is a major BBS locus. <i>Nature Genetics</i> , 2006, 38, 521-524.	9.4	259
106	RNAi-Mediated Allelic trans-Interaction at the Imprinted Rtl1/Peg11 Locus. <i>Current Biology</i> , 2005, 15, 743-749.	1.8	301
107	RNAi-Mediated Allelic trans-Interaction at the Imprinted Rtl1/Peg11 Locus. <i>Current Biology</i> , 2005, 15, 884.	1.8	2
108	Ectopic Expression of DLK1 Protein in Skeletal Muscle of Padumnal Heterozygotes Causes the Callipyge Phenotype. <i>Current Biology</i> , 2004, 14, 1858-1862.	1.8	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.	2.0	11
110	Oligogenic Effects of 16p11.2 Copy Number Variation on Craniofacial Development. SSRN Electronic Journal, 0, , .	0.4	1