

# Michael F Wangler

## List of Publications by Year in descending order

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88  
papers

4,876  
citations

109264

35  
h-index

110317

64  
g-index

96  
all docs

96  
docs citations

96  
times ranked

8021  
citing authors

#	ARTICLE	IF	CITATIONS
1	Use of Exome Sequencing for Infants in Intensive Care Units. <i>JAMA Pediatrics</i> , 2017, 171, e173438.	3.3	348
2	A <i>Drosophila</i> Genetic Resource of Mutants to Study Mechanisms Underlying Human Genetic Diseases. <i>Cell</i> , 2014, 159, 200-214.	13.5	322
3	Peroxisome biogenesis disorders in the Zellweger spectrum: An overview of current diagnosis, clinical manifestations, and treatment guidelines. <i>Molecular Genetics and Metabolism</i> , 2016, 117, 313-321.	0.5	314
4	Effect of Genetic Diagnosis on Patients with Previously Undiagnosed Disease. <i>New England Journal of Medicine</i> , 2018, 379, 2131-2139.	13.9	261
5	Association between Beckwith-Wiedemann syndrome and assisted reproductive technology: A case series of 19 patients. <i>Fertility and Sterility</i> , 2005, 83, 349-354.	0.5	214
6	Lessons learned from additional research analyses of unsolved clinical exome cases. <i>Genome Medicine</i> , 2017, 9, 26.	3.6	184
7	MARRVEL: Integration of Human and Model Organism Genetic Resources to Facilitate Functional Annotation of the Human Genome. <i>American Journal of Human Genetics</i> , 2017, 100, 843-853.	2.6	181
8	Racial disparity in the frequency of recurrence of preterm birth. <i>American Journal of Obstetrics and Gynecology</i> , 2007, 196, 131.e1-131.e6.	0.7	171
9	Model Organisms Facilitate Rare Disease Diagnosis and Therapeutic Research. <i>Genetics</i> , 2017, 207, 9-27.	1.2	165
10	Fruit Flies in Biomedical Research. <i>Genetics</i> , 2015, 199, 639-653.	1.2	149
11	The Undiagnosed Diseases Network: Accelerating Discovery about Health and Disease. <i>American Journal of Human Genetics</i> , 2017, 100, 185-192.	2.6	142
12	Heterozygous De Novo and Inherited Mutations in the Smooth Muscle Actin (ACTG2) Gene Underlie Megacystis-Microcolon-Intestinal Hypoperistalsis Syndrome. <i>PLoS Genetics</i> , 2014, 10, e1004258.	1.5	122
13	A Syndromic Neurodevelopmental Disorder Caused by De Novo Variants in EBF3. <i>American Journal of Human Genetics</i> , 2017, 100, 128-137.	2.6	96
14	Loss of Nardilysin, a Mitochondrial Co-chaperone for $\hat{\alpha}$ -Ketoglutarate Dehydrogenase, Promotes mTORC1 Activation and Neurodegeneration. <i>Neuron</i> , 2017, 93, 115-131.	3.8	95
15	Clinical spectrum of individuals with pathogenic <i>F1</i> missense variants affecting p.Met1149, p.Arg1276, and p.Lys1423: genotype-phenotype study in neurofibromatosis type 1. <i>Human Mutation</i> , 2020, 41, 299-315.	1.1	80
16	Clinically severe CACNA1A alleles affect synaptic function and neurodegeneration differentially. <i>PLoS Genetics</i> , 2017, 13, e1006905.	1.5	80
17	The fruit fly at the interface of diagnosis and pathogenic mechanisms of rare and common human diseases. <i>Human Molecular Genetics</i> , 2019, 28, R207-R214.	1.4	72
18	Loss- or Gain-of-Function Mutations in ACOX1 Cause Axonal Loss via Different Mechanisms. <i>Neuron</i> , 2020, 106, 589-606.e6.	3.8	71

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19	Fibrochondrogenesis Results from Mutations in the COL11A1 Type XI Collagen Gene. American Journal of Human Genetics, 2010, 87, 708-712.	2.6	69
20	IRF2BPL Is Associated with Neurological Phenotypes. American Journal of Human Genetics, 2018, 103, 245-260.	2.6	69
21	De Novo Truncating Variants in ASXL2 Are Associated with a Unique and Recognizable Clinical Phenotype. American Journal of Human Genetics, 2016, 99, 991-999.	2.6	68
22	Loss-of-Function Variants in MYLK Cause Recessive Megacystis Microcolon Intestinal Hypoperistalsis Syndrome. American Journal of Human Genetics, 2017, 101, 123-129.	2.6	67
23	Missense variants in the middle domain of <i>DNM1L</i> in cases of infantile encephalopathy alter peroxisomes and mitochondria when assayed in <i>Drosophila</i> . Human Molecular Genetics, 2016, 25, 1846-1856.	1.4	62
24	Expanding the clinical phenotype of individuals with a 3-bp in-frame deletion of the NF1 gene (c.2970_2972del): an update of genotype-phenotype correlation. Genetics in Medicine, 2019, 21, 867-876.	1.1	62
25	A comprehensive iterative approach is highly effective in diagnosing individuals who are exome negative. Genetics in Medicine, 2019, 21, 161-172.	1.1	60
26	Biallelic Mutations in ATP5F1D, which Encodes a Subunit of ATP Synthase, Cause a Metabolic Disorder. American Journal of Human Genetics, 2018, 102, 494-504.	2.6	59
27	Mother's Genome or Maternally-Inherited Genes Acting in the Fetus Influence Gestational Age in Familial Preterm Birth. Human Heredity, 2009, 68, 209-219.	0.4	57
28	De Novo Truncating Mutations in AHDC1 in Individuals with Syndromic Expressive Language Delay, Hypotonia, and Sleep Apnea. American Journal of Human Genetics, 2014, 94, 784-789.	2.6	57
29	Functional variants in TBX2 are associated with a syndromic cardiovascular and skeletal developmental disorder. Human Molecular Genetics, 2018, 27, 2454-2465.	1.4	54
30	Model organisms contribute to diagnosis and discovery in the undiagnosed diseases network: current state and a future vision. Orphanet Journal of Rare Diseases, 2021, 16, 206.	1.2	53
31	Diagnosis of adenylosuccinate lyase deficiency by metabolomic profiling in plasma reveals a phenotypic spectrum. Molecular Genetics and Metabolism Reports, 2016, 8, 61-66.	0.4	48
32	Ari-1 Regulates Myonuclear Organization Together with Parkin and Is Associated with Aortic Aneurysms. Developmental Cell, 2018, 45, 226-244.e8.	3.1	46
33	De novo mutations in MED13, a component of the Mediator complex, are associated with a novel neurodevelopmental disorder. Human Genetics, 2018, 137, 375-388.	1.8	46
34	Factors associated with preterm delivery in mothers of children with Beckwith-Wiedemann syndrome: A case cohort study from the BWS registry. American Journal of Medical Genetics, Part A, 2005, 134A, 187-191.	0.7	44
35	De Novo Variants in WDR37 Are Associated with Epilepsy, Colobomas, Dysmorphism, Developmental Delay, Intellectual Disability, and Cerebellar Hypoplasia. American Journal of Human Genetics, 2019, 105, 413-424.	2.6	43
36	Disruptive mutations in TANC2 define a neurodevelopmental syndrome associated with psychiatric disorders. Nature Communications, 2019, 10, 4679.	5.8	43

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37	A metabolomic map of Zellweger spectrum disorders reveals novel disease biomarkers. <i>Genetics in Medicine</i> , 2018, 20, 1274-1283.	1.1	40
38	Peroxisomes Are Required for Lipid Metabolism and Muscle Function in <i>Drosophila melanogaster</i> . <i>PLoS ONE</i> , 2014, 9, e100213.	1.1	38
39	<i>Drosophila</i> and genome-wide association studies: a review and resource for the functional dissection of human complex traits. <i>DMM Disease Models and Mechanisms</i> , 2017, 10, 77-88.	1.2	37
40	A Recurrent De Novo Variant in <i>NACC1</i> Causes a Syndrome Characterized by Infantile Epilepsy, Cataracts, and Profound Developmental Delay. <i>American Journal of Human Genetics</i> , 2017, 100, 343-351.	2.6	35
41	The phenotypic spectrum of Xia-Gibbs syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 1315-1326.	0.7	34
42	In Vivo Functional Study of Disease-associated Rare Human Variants Using <i>Drosophila</i> . <i>Journal of Visualized Experiments</i> , 2019, , .	0.2	34
43	<i>BICRA</i> , a SWI/SNF Complex Member, Is Associated with BAF-Disorder Related Phenotypes in Humans and Model Organisms. <i>American Journal of Human Genetics</i> , 2020, 107, 1096-1112.	2.6	32
44	Peroxisomal biogenesis is genetically and biochemically linked to carbohydrate metabolism in <i>Drosophila</i> and mouse. <i>PLoS Genetics</i> , 2017, 13, e1006825.	1.5	31
45	A Genocentric Approach to Discovery of Mendelian Disorders. <i>American Journal of Human Genetics</i> , 2019, 105, 974-986.	2.6	30
46	Diagnosis of a mild peroxisomal phenotype with next-generation sequencing. <i>Molecular Genetics and Metabolism Reports</i> , 2016, 9, 75-78.	0.4	29
47	De novo mutations in <i>TOMM70</i> , a receptor of the mitochondrial import translocase, cause neurological impairment. <i>Human Molecular Genetics</i> , 2020, 29, 1568-1579.	1.4	29
48	Recurrent arginine substitutions in the <i>ACTG2</i> gene are the primary driver of disease burden and severity in visceral myopathy. <i>Human Mutation</i> , 2020, 41, 641-654.	1.1	27
49	Atypical presentation of Leigh syndrome associated with a Leber hereditary optic neuropathy primary mitochondrial DNA mutation. <i>Molecular Genetics and Metabolism</i> , 2011, 103, 153-160.	0.5	24
50	De novo missense variant in the GTPase effector domain (GED) of <i>DNM1L</i> leads to static encephalopathy and seizures. <i>Journal of Physical Education and Sports Management</i> , 2019, 5, a003673.	0.5	24
51	<i>Drosophila</i> functional screening of de novo variants in autism uncovers damaging variants and facilitates discovery of rare neurodevelopmental diseases. <i>Cell Reports</i> , 2022, 38, 110517.	2.9	24
52	De Novo Variants in <i>CDK19</i> Are Associated with a Syndrome Involving Intellectual Disability and Epileptic Encephalopathy. <i>American Journal of Human Genetics</i> , 2020, 106, 717-725.	2.6	23
53	<i>TNPO2</i> variants associate with human developmental delays, neurologic deficits, and dysmorphic features and alter <i>TNPO2</i> activity in <i>Drosophila</i> . <i>American Journal of Human Genetics</i> , 2021, 108, 1669-1691.	2.6	23
54	Inborn Errors of Metabolism Involving Complex Molecules. <i>Pediatric Clinics of North America</i> , 2018, 65, 353-373.	0.9	22

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55	Clinical sites of the Undiagnosed Diseases Network: unique contributions to genomic medicine and science. <i>Genetics in Medicine</i> , 2021, 23, 259-271.	1.1	18
56	Rare deleterious <i>de novo</i> missense variants in <i>Rnf2/Ring2</i> are associated with a neurodevelopmental disorder with unique clinical features. <i>Human Molecular Genetics</i> , 2021, 30, 1283-1292.	1.4	17
57	A homozygous mutation in <i>PEX16</i> identified by whole-exome sequencing ending a diagnostic odyssey. <i>Molecular Genetics and Metabolism Reports</i> , 2015, 5, 15-18.	0.4	16
58	Xia's "Gibbs syndrome in adulthood: a case report with insight into the natural history of the condition. <i>Journal of Physical Education and Sports Management</i> , 2019, 5, a003608.	0.5	15
59	Missense variants in <i>CTNNB1</i> can be associated with vitreoretinopathy—Seven new cases of <i>CTNNB1</i> -associated neurodevelopmental disorder including a previously unreported retinal phenotype. <i>Molecular Genetics &amp; Genomic Medicine</i> , 2021, 9, e1542.	0.6	15
60	Clinical diagnosis of metabolic disorders using untargeted metabolomic profiling and disease-specific networks learned from profiling data. <i>Scientific Reports</i> , 2022, 12, 6556.	1.6	15
61	<i>MED27</i> Variants Cause Developmental Delay, Dystonia, and Cerebellar Hypoplasia. <i>Annals of Neurology</i> , 2021, 89, 828-833.	2.8	14
62	Phenotypic and protein localization heterogeneity associated with <i>AHDC1</i> pathogenic protein-truncating alleles in Xia's "Gibbs syndrome. <i>Human Mutation</i> , 2021, 42, 577-591.	1.1	14
63	An <i>SCN1B</i> Variant Affects Both Cardiac-Type (NaV1.5) and Brain-Type (NaV1.1) Sodium Currents and Contributes to Complex Concomitant Brain and Cardiac Disorders. <i>Frontiers in Cell and Developmental Biology</i> , 2020, 8, 528742.	1.8	13
64	Heterozygous loss-of-function variants significantly expand the phenotypes associated with loss of <i>GDF11</i> . <i>Genetics in Medicine</i> , 2021, 23, 1889-1900.	1.1	13
65	Unusually Early Presentation of Small-Bowel Adenocarcinoma in a Patient With Peutz-Jeghers Syndrome. <i>Journal of Pediatric Hematology/Oncology</i> , 2013, 35, 323-328.	0.3	12
66	Germline mutation in <i>POLR2A</i> : a heterogeneous, multi-systemic developmental disorder characterized by transcriptional dysregulation. <i>Human Genetics and Genomics Advances</i> , 2021, 2, 100014.	1.0	10
67	Complex effects on <i>CaV2.1</i> channel gating caused by a <i>CACNA1A</i> variant associated with a severe neurodevelopmental disorder. <i>Scientific Reports</i> , 2022, 12, .	1.6	10
68	Inheritance pattern of Beckwith-Wiedemann syndrome is heterogeneous in 291 families with an affected proband. <i>American Journal of Medical Genetics, Part A</i> , 2005, 137A, 16-21.	0.7	8
69	The expanding neurological phenotype of <i>DNM1L</i> -related disorders. <i>Brain</i> , 2018, 141, e28-e28.	3.7	7
70	Biases in arginine codon usage correlate with genetic disease risk. <i>Genetics in Medicine</i> , 2020, 22, 1407-1412.	1.1	7
71	<i>ABCD1</i> and X-linked adrenoleukodystrophy: A disease with a markedly variable phenotype showing conserved neurobiology in animal models. <i>Journal of Neuroscience Research</i> , 2021, 99, 3170-3181.	1.3	7
72	A Genetic Screen for Genes That Impact Peroxisomes in <i>Drosophila</i> Identifies Candidate Genes for Human Disease. <i>G3: Genes, Genomes, Genetics</i> , 2020, 10, 69-77.	0.8	6

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73	The microRNA processor <i>DROSHA</i> is a candidate gene for a severe progressive neurological disorder. <i>Human Molecular Genetics</i> , 2022, 31, 2934-2950.	1.4	6
74	Genome sequencing analysis of a family with a child displaying severe abdominal distention and recurrent hypoglycemia. <i>Molecular Genetics &amp; Genomic Medicine</i> , 2020, 8, e1130.	0.6	5
75	AHDC1 missense mutations in Xia-Gibbs syndrome. <i>Human Genetics and Genomics Advances</i> , 2021, 2, 100049.	1.0	5
76	ModelMatcher: A scientist-centric online platform to facilitate collaborations between stakeholders of rare and undiagnosed disease research. <i>Human Mutation</i> , 2022, , .	1.1	5
77	An Integrated Phenotypic and Genotypic Approach Reveals a High-Risk Subtype Association for <i>EBF3</i> Missense Variants Affecting the Zinc Finger Domain. <i>Annals of Neurology</i> , 2022, 92, 138-153.	2.8	5
78	Antioxidant proteins TSA and PAG interact synergistically with Presenilin to modulate Notch signaling in <i>Drosophila</i> . <i>Protein and Cell</i> , 2011, 2, 554-563.	4.8	3
79	Digital necrosis in an infant with severe spinal muscular atrophy. <i>Neurology: Genetics</i> , 2019, 5, e361.	0.9	3
80	Evidence for an association between Coffin-Siris syndrome and congenital diaphragmatic hernia. <i>American Journal of Medical Genetics, Part A</i> , 2022, 188, 2718-2723.	0.7	3
81	In Vivo Animal Modeling. , 2017, , 211-234.		2
82	Retrospective Diagnosis of Ataxia-Telangiectasia in an Adolescent Patient With a Remote History of T-Cell Leukemia. <i>Journal of Pediatric Hematology/Oncology</i> , 2021, 43, e138-e140.	0.3	2
83	Dataset for a case report of a homozygous PEX16 F332del mutation. <i>Data in Brief</i> , 2016, 6, 722-727.	0.5	1
84	Novel <i>CIC</i> variants identified in individuals with neurodevelopmental phenotypes. <i>Human Mutation</i> , 2022, 43, 889-899.	1.1	1
85	498: Placental pathology findings in cases of familial spontaneous preterm birth. <i>American Journal of Obstetrics and Gynecology</i> , 2009, 201, S186.	0.7	0
86	A Mitochondrial Translation Defect Identified by Whole-Exome Sequencing Expands the Phenotypic Spectrum for MARS2. <i>Human Mutation</i> , 2015, 36, iii-iii.	1.1	0
87	Advances in Next-Generation Sequencing Technologies and Functional Investigation of Candidate Variants in Neurological and Behavioral Disorders. , 2022, , 390-404.		0
88	Evidence For and Against Associations between ART and Congenital Malformation Syndromes. , 2005, , .		0