

Michael F Wangler

List of Publications by Year in descending order

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Version: 2024-02-01

88
papers

4,876
citations

125106

35
h-index

124990

64
g-index

96
all docs

96
docs citations

96
times ranked

8635
citing authors

#	ARTICLE	IF	CITATIONS
1	Advances in Next-Generation Sequencing Technologies and Functional Investigation of Candidate Variants in Neurological and Behavioral Disorders. , 2022, , 390-404.		0
2	Drosophila functional screening of de novo variants in autism uncovers damaging variants and facilitates discovery of rare neurodevelopmental diseases. Cell Reports, 2022, 38, 110517.	2.9	24
3	ModelMatcher: A scientistâ€centric online platform to facilitate collaborations between stakeholders of rare and undiagnosed disease research. Human Mutation, 2022, , .	1.1	5
4	An Integrated Phenotypic and Genotypic Approach Reveals a Highâ€Risk Subtype Association for <sc><i>EBF3</i></sc> Missense Variants Affecting the Zinc Finger Domain. Annals of Neurology, 2022, 92, 138-153.	2.8	5
5	Novel <i>CIC</i> variants identified in individuals with neurodevelopmental phenotypes. Human Mutation, 2022, 43, 889-899.	1.1	1
6	The microRNA processor<i>DROSHA</i> is a candidate gene for a severe progressive neurological disorder. Human Molecular Genetics, 2022, 31, 2934-2950.	1.4	6
7	Clinical diagnosis of metabolic disorders using untargeted metabolomic profiling and disease-specific networks learned from profiling data. Scientific Reports, 2022, 12, 6556.	1.6	15
8	Complex effects on CaV2.1 channel gating caused by a CACNA1A variant associated with a severe neurodevelopmental disorder. Scientific Reports, 2022, 12, .	1.6	10
9	Evidence for an association between <sc>Coffinâ€Siris</sc> syndrome and congenital diaphragmatic hernia. American Journal of Medical Genetics, Part A, 2022, 188, 2718-2723.	0.7	3
10	Germline mutation in POLR2A: a heterogeneous, multi-systemic developmental disorder characterized by transcriptional dysregulation. Human Genetics and Genomics Advances, 2021, 2, 100014.	1.0	10
11	Clinical sites of the Undiagnosed Diseases Network: unique contributions to genomic medicine and science. Genetics in Medicine, 2021, 23, 259-271.	1.1	18
12	Retrospective Diagnosis of Ataxia-Telangiectasia in an Adolescent Patient With a Remote History of T-Cell Leukemia. Journal of Pediatric Hematology/Oncology, 2021, 43, e138-e140.	0.3	2
13	<sc><i>MED27</i></sc> Variants Cause Developmental Delay, Dystonia, and Cerebellar Hypoplasia. Annals of Neurology, 2021, 89, 828-833.	2.8	14
14	Phenotypic and protein localization heterogeneity associated with <i>AHDC1</i> pathogenic proteinâ€truncating alleles in Xiaâ€Gibbs syndrome. Human Mutation, 2021, 42, 577-591.	1.1	14
15	Rare deleterious <i>de novo</i> missense variants in <i>Rnf2/Ring2</i> are associated with a neurodevelopmental disorder with unique clinical features. Human Molecular Genetics, 2021, 30, 1283-1292.	1.4	17
16	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
17	Heterozygous loss-of-function variants significantly expand the phenotypes associated with loss of GDF11. Genetics in Medicine, 2021, 23, 1889-1900.	1.1	13
18	TNPO2 variants associate with human developmental delays, neurologic deficits, and dysmorphic features and alter TNPO2 activity in Drosophila. American Journal of Human Genetics, 2021, 108, 1669-1691.	2.6	23

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19	AHDC1 missense mutations in Xia-Gibbs syndrome. <i>Human Genetics and Genomics Advances</i> , 2021, 2, 100049.	1.0	5
20	ABCD1 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
21	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 & Genomic Medicine</i> , 2021, 9, e1542.	0.6	15
22	Clinical spectrum of individuals with pathogenic <i>NF1</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
23	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
24	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
25	BICRA, 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
26	An SCN1B 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
27	De novo mutations in TOMM70, a receptor of the mitochondrial import translocase, cause neurological impairment. <i>Human Molecular Genetics</i> , 2020, 29, 1568-1579.	1.4	29
28	De Novo Variants in CDK19 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
29	Biases in arginine codon usage correlate with genetic disease risk. <i>Genetics in Medicine</i> , 2020, 22, 1407-1412.	1.1	7
30	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
31	Genome sequencing analysis of a family with a child displaying severe abdominal distention and recurrent hypoglycemia. <i>Molecular Genetics & Genomic Medicine</i> , 2020, 8, e1130.	0.6	5
32	A comprehensive iterative approach is highly effective in diagnosing individuals who are exome negative. <i>Genetics in Medicine</i> , 2019, 21, 161-172.	1.1	60
33	De Novo Variants in WDR37 Are Associated with Epilepsy, Colobomas, Dysmorphism, Developmental Delay, Intellectual Disability, and Cerebellar Hypoplasia. <i>American Journal of Human Genetics</i> , 2019, 105, 413-424.	2.6	43
34	Disruptive mutations in TANC2 define a neurodevelopmental syndrome associated with psychiatric disorders. <i>Nature Communications</i> , 2019, 10, 4679.	5.8	43
35	A Genocentric Approach to Discovery of Mendelian Disorders. <i>American Journal of Human Genetics</i> , 2019, 105, 974-986.	2.6	30
36	In Vivo Functional Study of Disease-associated Rare Human Variants Using <i>Drosophila</i> . <i>Journal of Visualized Experiments</i> , 2019, , .	0.2	34

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37	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
38	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
39	Digital necrosis in an infant with severe spinal muscular atrophy. <i>Neurology: Genetics</i> , 2019, 5, e361.	0.9	3
40	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. <i>Genetics in Medicine</i> , 2019, 21, 867-876.	1.1	62
41	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
42	Biallelic Mutations in ATP5F1D, which Encodes a Subunit of ATP Synthase, Cause a Metabolic Disorder. <i>American Journal of Human Genetics</i> , 2018, 102, 494-504.	2.6	59
43	Ari-1 Regulates Myonuclear Organization Together with Parkin and Is Associated with Aortic Aneurysms. <i>Developmental Cell</i> , 2018, 45, 226-244.e8.	3.1	46
44	A metabolomic map of Zellweger spectrum disorders reveals novel disease biomarkers. <i>Genetics in Medicine</i> , 2018, 20, 1274-1283.	1.1	40
45	The phenotypic spectrum of Xia's Gibbs syndrome. <i>American Journal of Medical Genetics, Part A</i> , 2018, 176, 1315-1326.	0.7	34
46	Inborn Errors of Metabolism Involving Complex Molecules. <i>Pediatric Clinics of North America</i> , 2018, 65, 353-373.	0.9	22
47	The expanding neurological phenotype of DNM1L-related disorders. <i>Brain</i> , 2018, 141, e28-e28.	3.7	7
48	Effect of Genetic Diagnosis on Patients with Previously Undiagnosed Disease. <i>New England Journal of Medicine</i> , 2018, 379, 2131-2139.	13.9	261
49	IRF2BPL Is Associated with Neurological Phenotypes. <i>American Journal of Human Genetics</i> , 2018, 103, 245-260.	2.6	69
50	De novo mutations in MED13, a component of the Mediator complex, are associated with a novel neurodevelopmental disorder. <i>Human Genetics</i> , 2018, 137, 375-388.	1.8	46
51	Functional variants in TBX2 are associated with a syndromic cardiovascular and skeletal developmental disorder. <i>Human Molecular Genetics</i> , 2018, 27, 2454-2465.	1.4	54
52	The Undiagnosed Diseases Network: Accelerating Discovery about Health and Disease. <i>American Journal of Human Genetics</i> , 2017, 100, 185-192.	2.6	142
53	A Recurrent De Novo Variant in NACC1 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
54	<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

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55	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
56	Loss-of-Function Variants in MYLK Cause Recessive Megacystis Microcolon Intestinal Hypoperistalsis Syndrome. <i>American Journal of Human Genetics</i> , 2017, 101, 123-129.	2.6	67
57	Lessons learned from additional research analyses of unsolved clinical exome cases. <i>Genome Medicine</i> , 2017, 9, 26.	3.6	184
58	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
59	Loss of Nardilysin, a Mitochondrial Co-chaperone for Î±-Ketoglutarate Dehydrogenase, Promotes mTORC1 Activation and Neurodegeneration. <i>Neuron</i> , 2017, 93, 115-131.	3.8	95
60	Use of Exome Sequencing for Infants in Intensive Care Units. <i>JAMA Pediatrics</i> , 2017, 171, e173438.	3.3	348
61	Model Organisms Facilitate Rare Disease Diagnosis and Therapeutic Research. <i>Genetics</i> , 2017, 207, 9-27.	1.2	165
62	In Vivo Animal Modeling. , 2017, , 211-234.		2
63	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
64	Clinically severe CACNA1A alleles affect synaptic function and neurodegeneration differentially. <i>PLoS Genetics</i> , 2017, 13, e1006905.	1.5	80
65	De Novo Truncating Variants in ASXL2 Are Associated with a Unique and Recognizable Clinical Phenotype. <i>American Journal of Human Genetics</i> , 2016, 99, 991-999.	2.6	68
66	Diagnosis of adenylosuccinate lyase deficiency by metabolomic profiling in plasma reveals a phenotypic spectrum. <i>Molecular Genetics and Metabolism Reports</i> , 2016, 8, 61-66.	0.4	48
67	Diagnosis of a mild peroxisomal phenotype with next-generation sequencing. <i>Molecular Genetics and Metabolism Reports</i> , 2016, 9, 75-78.	0.4	29
68	Dataset for a case report of a homozygous PEX16 F332del mutation. <i>Data in Brief</i> , 2016, 6, 722-727.	0.5	1
69	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
70	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> . <i>Human Molecular Genetics</i> , 2016, 25, 1846-1856.	1.4	62
71	A homozygous mutation in PEX16 identified by whole-exome sequencing ending a diagnostic odyssey. <i>Molecular Genetics and Metabolism Reports</i> , 2015, 5, 15-18.	0.4	16
72	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

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73	Fruit Flies in Biomedical Research. <i>Genetics</i> , 2015, 199, 639-653.	1.2	149
74	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
75	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
76	De Novo Truncating Mutations in AHDC1 in Individuals with Syndromic Expressive Language Delay, Hypotonia, and Sleep Apnea. <i>American Journal of Human Genetics</i> , 2014, 94, 784-789.	2.6	57
77	Peroxisomes Are Required for Lipid Metabolism and Muscle Function in <i>Drosophila melanogaster</i> . <i>PLoS ONE</i> , 2014, 9, e100213.	1.1	38
78	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
79	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
80	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
81	Fibrochondrogenesis Results from Mutations in the COL11A1 Type XI Collagen Gene. <i>American Journal of Human Genetics</i> , 2010, 87, 708-712.	2.6	69
82	Mother's Genome or Maternally-Inherited Genes Acting in the Fetus Influence Gestational Age in Familial Preterm Birth. <i>Human Heredity</i> , 2009, 68, 209-219.	0.4	57
83	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
84	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
85	Factors associated with preterm delivery in mothers of children with Beckwith-Wiedemann syndrome: A case cohort study from the BWS registry. <i>American Journal of Medical Genetics, Part A</i> , 2005, 134A, 187-191.	0.7	44
86	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
87	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
88	Evidence For and Against Associations between ART and Congenital Malformation Syndromes. , 2005, , .		0