Michael F Wangler

List of Publications by Year
in descending order

88 papers	4,876 citations	109321 35 h-index	110387 64 g-index
96 all docs	96 docs citations	96 times ranked	8021 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.	6.4	24
3	ModelMatcher: A scientistâ€centric online platform to facilitate collaborations between stakeholders of rare and undiagnosed disease research. Human Mutation, 2022, , .	2.5	5
4	An Integrated Phenotypic and Genotypic Approach Reveals a Highâ€Risk Subtype Association for <scp>EBF3</scp> Missense Variants Affecting the Zinc Finger Domain. Annals of Neurology, 2022, 92, 138-153.	5.3	5
5	Novel CIC variants identified in individuals with neurodevelopmental phenotypes. Human Mutation, 2022, 43, 889-899.	2.5	1
6	The microRNA processorDROSHAis a candidate gene for a severe progressive neurological disorder. Human Molecular Genetics, 2022, 31, 2934-2950.	2.9	6
7	Clinical diagnosis of metabolic disorders using untargeted metabolomic profiling and disease-specific networks learned from profiling data. Scientific Reports, 2022, 12, 6556.	3.3	15
8	Complex effects on CaV2.1 channel gating caused by a CACNA1A variant associated with a severe neurodevelopmental disorder. Scientific Reports, 2022, 12, .	3.3	10
9	Evidence for an association between <scp>Coffinâ€Siris</scp> syndrome and congenital diaphragmatic hernia. American Journal of Medical Genetics, Part A, 2022, 188, 2718-2723.	1.2	3
10	Germline mutation in POLR2A: a heterogeneous, multi-systemic developmental disorder characterized by transcriptional dysregulation. Human Genetics and Genomics Advances, 2021, 2, 100014.	1.7	10
11	Clinical sites of the Undiagnosed Diseases Network: unique contributions to genomic medicine and science. Genetics in Medicine, 2021, 23, 259-271.	2.4	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.	0.6	2
13	<scp>MED27</scp> Variants Cause Developmental Delay, Dystonia, and Cerebellar Hypoplasia. Annals of Neurology, 2021, 89, 828-833.	5.3	14
14	Phenotypic and protein localization heterogeneity associated with AHDC1 pathogenic proteinâ€truncating alleles in Xiaâ€“Gibbs syndrome. Human Mutation, 2021, 42, 577-591.	2.5	14
15	Rare deleterious de novo missense variants in Rnf2/Ring2 are associated with a neurodevelopmental disorder with unique clinical features. Human Molecular Genetics, 2021, 30, 1283-1292.	2.9	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.	2.7	53
17	Heterozygous loss-of-function variants significantly expand the phenotypes associated with loss of GDF11. Genetics in Medicine, 2021, 23, 1889-1900.	2.4	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.	6.2	23

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19	AHDC1 missense mutations in Xia-Gibbs syndrome. Human Genetics and Genomics Advances, 2021, 2, 100049.	1.7	5
20	ABCD1 and Xâ€linked adrenoleukodystrophy: A disease with a markedly variable phenotype showing conserved neurobiology in animal models. Journal of Neuroscience Research, 2021, 99, 3170-3181.	2.9	7
21	Missense variants in CTNNB1 can be associated with vitreoretinopathyâ€"Seven new cases of CTNNB1â€associated neurodevelopmental disorder including a previously unreported retinal phenotype. Molecular Genetics & Genomic Medicine, 2021, 9, e1542.	1.2	15
22	Clinical spectrum of individuals with pathogenic N F1 missense variants affecting p.Met1149, p.Arg1276, and p.Lys1423: genotypeâ€"phenotype study in neurofibromatosis type 1. Human Mutation, 2020, 41, 299-315.	2.5	80
23	Recurrent arginine substitutions in the ACTG2 gene are the primary driver of disease burden and severity in visceral myopathy. Human Mutation, 2020, 41, 641-654.	2.5	27
24	A Genetic Screen for Genes That Impact Peroxisomes in Drosophila Identifies Candidate Genes for Human Disease. G3: Genes, Genomes, Genetics, 2020, 10, 69-77.	1.8	6
25	BICRA, a SWI/SNF Complex Member, Is Associated with BAF-Disorder Related Phenotypes in Humans and Model Organisms. American Journal of Human Genetics, 2020, 107, 1096-1112.	6.2	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. Frontiers in Cell and Developmental Biology, 2020, 8, 528742.	3.7	13
27	De novo mutations in TOMM70, a receptor of the mitochondrial import translocase, cause neurological impairment. Human Molecular Genetics, 2020, 29, 1568-1579.	2.9	29
28	De Novo Variants in CDK19 Are Associated with a Syndrome Involving Intellectual Disability and Epileptic Encephalopathy. American Journal of Human Genetics, 2020, 106, 717-725.	6.2	23
29	Biases in arginine codon usage correlate with genetic disease risk. Genetics in Medicine, 2020, 22, 1407-1412.	2.4	7
30	Loss- or Gain-of-Function Mutations in ACOX1 Cause Axonal Loss via Different Mechanisms. Neuron, 2020, 106, 589-606.e6.	8.1	71
31	Genome sequencing analysis of a family with a child displaying severe abdominal distention and recurrent hypoglycemia. Molecular Genetics & Genomic Medicine, 2020, 8, e1130.	1.2	5
32	A comprehensive iterative approach is highly effective in diagnosing individuals who are exome negative. Genetics in Medicine, 2019, 21, 161-172.	2.4	60
33	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.	6.2	43
34	Disruptive mutations in TANC2 define a neurodevelopmental syndrome associated with psychiatric disorders. Nature Communications, 2019, 10, 4679.	12.8	43
35	A Genocentric Approach to Discovery of Mendelian Disorders. American Journal of Human Genetics, 2019, 105, 974-986.	6.2	30
36	In Vivo Functional Study of Disease-associated Rare Human Variants Using Drosophila. Journal of Visualized Experiments, 2019, , .	0.3	34

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37	The fruit fly at the interface of diagnosis and pathogenic mechanisms of rare and common human diseases. Human Molecular Genetics, 2019, 28, R207-R214.	2.9	72
38	De novo missense variant in the GTPase effector domain (GED) of DNM1L leads to static encephalopathy and seizures. Journal of Physical Education and Sports Management, 2019, 5, a003673.	1.2	24
39	Digital necrosis in an infant with severe spinal muscular atrophy. Neurology: Genetics, 2019, 5, e361.	1.9	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 genotypeâ€“phenotype correlation. Genetics in Medicine, 2019, 21, 867-876.	2.4	62
41	Xiaâ€“Gibbs syndrome in adulthood: a case report with insight into the natural history of the condition. Journal of Physical Education and Sports Management, 2019, 5, a003608.	1.2	15
42	Biallelic Mutations in ATP5F1D, which Encodes a Subunit of ATP Synthase, Cause a Metabolic Disorder. American Journal of Human Genetics, 2018, 102, 494-504.	6.2	59
43	Ari-1 Regulates Myonuclear Organization Together with Parkin and Is Associated with Aortic Aneurysms. Developmental Cell, 2018, 45, 226-244.e8.	7.0	46
44	A metabolomic map of Zellweger spectrum disorders reveals novel disease biomarkers. Genetics in Medicine, 2018, 20, 1274-1283.	2.4	40
45	The phenotypic spectrum of Xiaâ€Gibbs syndrome. American Journal of Medical Genetics, Part A, 2018, 176, 1315-1326.	1.2	34
46	Inborn Errors of Metabolism Involving Complex Molecules. Pediatric Clinics of North America, 2018, 65, 353-373.	1.8	22
47	The expanding neurological phenotype of DNM1L-related disorders. Brain, 2018, 141, e28-e28.	7.6	7
48	Effect of Genetic Diagnosis on Patients with Previously Undiagnosed Disease. New England Journal of Medicine, 2018, 379, 2131-2139.	27.0	261
49	IRF2BPL Is Associated with Neurological Phenotypes. American Journal of Human Genetics, 2018, 103, 245-260.	6.2	69
50	De novo mutations in MED13, a component of the Mediator complex, are associated with a novel neurodevelopmental disorder. Human Genetics, 2018, 137, 375-388.	3.8	46
51	Functional variants in TBX2 are associated with a syndromic cardiovascular and skeletal developmental disorder. Human Molecular Genetics, 2018, 27, 2454-2465.	2.9	54
52	The Undiagnosed Diseases Network: Accelerating Discovery about Health and Disease. American Journal of Human Genetics, 2017, 100, 185-192.	6.2	142
53	A Recurrent De Novo Variant in NACC1 Causes a Syndrome Characterized by Infantile Epilepsy, Cataracts, and Profound Developmental Delay. American Journal of Human Genetics, 2017, 100, 343-351.	6.2	35
54	Drosophilaand genome-wide association studies: a review and resource for the functional dissection of human complex traits. DMM Disease Models and Mechanisms, 2017, 10, 77-88.	2.4	37

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55	MARRVEL: Integration of Human and Model Organism Genetic Resources to Facilitate Functional Annotation of the Human Genome. American Journal of Human Genetics, 2017, 100, 843-853.	6.2	181
56	Loss-of-Function Variants in MYLK Cause Recessive Megacystis Microcolon Intestinal Hypoperistalsis Syndrome. American Journal of Human Genetics, 2017, 101, 123-129.	6.2	67
57	Lessons learned from additional research analyses of unsolved clinical exome cases. Genome Medicine, 2017, 9, 26.	8.2	184
58	A Syndromic Neurodevelopmental Disorder Caused by De Novo Variants in EBF3. American Journal of Human Genetics, 2017, 100, 128-137.	6.2	96
59	Loss of Nardilysin, a Mitochondrial Co-chaperone for Î±-Ketoglutarate Dehydrogenase, Promotes mTORC1 Activation and Neurodegeneration. Neuron, 2017, 93, 115-131.	8.1	95
60	Use of Exome Sequencing for Infants in Intensive Care Units. JAMA Pediatrics, 2017, 171, e173438.	6.2	348
61	Model Organisms Facilitate Rare Disease Diagnosis and Therapeutic Research. Genetics, 2017, 207, 9-27.	2.9	165
62	In Vivo Animal Modeling. , 2017, , 211-234.		2
63	Peroxisomal biogenesis is genetically and biochemically linked to carbohydrate metabolism in Drosophila and mouse. PLoS Genetics, 2017, 13, e1006825.	3.5	31
64	Clinically severe CACNA1A alleles affect synaptic function and neurodegeneration differentially. PLoS Genetics, 2017, 13, e1006905.	3.5	80
65	De Novo Truncating Variants in ASXL2 Are Associated with a Unique and Recognizable Clinical Phenotype. American Journal of Human Genetics, 2016, 99, 991-999.	6.2	68
66	Diagnosis of adenylosuccinate lyase deficiency by metabolomic profiling in plasma reveals a phenotypic spectrum. Molecular Genetics and Metabolism Reports, 2016, 8, 61-66.	1.1	48
67	Diagnosis of a mild peroxisomal phenotype with next-generation sequencing. Molecular Genetics and Metabolism Reports, 2016, 9, 75-78.	1.1	29
68	Dataset for a case report of a homozygous PEX16 F332del mutation. Data in Brief, 2016, 6, 722-727.	1.0	1
69	Peroxisome biogenesis disorders in the Zellweger spectrum: An overview of current diagnosis, clinical manifestations, and treatment guidelines. Molecular Genetics and Metabolism, 2016, 117, 313-321.	1.1	314
70	Missense variants in the middle domain ofDNM1Lin cases of infantile encephalopathy alter peroxisomes and mitochondria when assayed inDrosophila. Human Molecular Genetics, 2016, 25, 1846-1856.	2.9	62
71	A homozygous mutation in PEX16 identified by whole-exome sequencing ending a diagnostic odyssey. Molecular Genetics and Metabolism Reports, 2015, 5, 15-18.	1.1	16
72	A Mitochondrial Translation Defect Identified by Whole-Exome Sequencing Expands the Phenotypic Spectrum forMARS2. Human Mutation, 2015, 36, iii-iii.	2.5	0

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73	Fruit Flies in Biomedical Research. Genetics, 2015, 199, 639-653.	2.9	149
74	Heterozygous De Novo and Inherited Mutations in the Smooth Muscle Actin (ACTG2) Gene Underlie Megacystis-Microcolon-Intestinal Hypoperistalsis Syndrome. PLoS Genetics, 2014, 10, e1004258.	3.5	122
75	A Drosophila Genetic Resource of Mutants to Study Mechanisms Underlying Human Genetic Diseases. Cell, 2014, 159, 200-214.	28.9	322
76	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.	6.2	57
77	Peroxisomes Are Required for Lipid Metabolism and Muscle Function in Drosophila melanogaster. PLoS ONE, 2014, 9, e100213.	2.5	38
78	Unusually Early Presentation of Small-Bowel Adenocarcinoma in a Patient With Peutz-Jeghers Syndrome. Journal of Pediatric Hematology/Oncology, 2013, 35, 323-328.	0.6	12
79	Atypical presentation of Leigh syndrome associated with a Leber hereditary optic neuropathy primary mitochondrial DNA mutation. Molecular Genetics and Metabolism, 2011, 103, 153-160.	1.1	24
80	Antioxidant proteins TSA and PAG interact synergistically with Presenilin to modulate Notch signaling in Drosophila. Protein and Cell, 2011, 2, 554-563.	11.0	3
81	Fibrochondrogenesis Results from Mutations in the COL11A1 Type XI Collagen Gene. American Journal of Human Genetics, 2010, 87, 708-712.	6.2	69
82	Motherâ€™s Genome or Maternally-Inherited Genes Acting in the Fetus Influence Gestational Age in Familial Preterm Birth. Human Heredity, 2009, 68, 209-219.	0.8	57
83	498: Placental pathology findings in cases of familial spontaneous preterm birth. American Journal of Obstetrics and Gynecology, 2009, 201, S186.	1.3	0
84	Racial disparity in the frequency of recurrence of preterm birth. American Journal of Obstetrics and Gynecology, 2007, 196, 131.e1-131.e6.	1.3	171
85	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.	1.2	44
86	Inheritance pattern of Beckwith-Wiedemann syndrome is heterogeneous in 291 families with an affected proband. American Journal of Medical Genetics, Part A, 2005, 137A, 16-21.	1.2	8
87	Association between Beckwith-Wiedemann syndrome and assisted reproductive technology: A case series of 19 patients. Fertility and Sterility, 2005, 83, 349-354.	1.0	214
88	Evidence For and Against Associations between ART and Congenital Malformation Syndromes. , 2005, , .		0