Brett H Graham

List of Publications by Year
in descending order

104 papers	8,460 citations	50276 46 h-index	48315 88 g-index
110 all docs	110 docs citations	110 times ranked	15388 citing authors

#	Article	IF	Citations
1	Glial Lipid Droplets and ROS Induced by Mitochondrial Defects Promote Neurodegeneration. Cell, 2015, 160, 177-190.	28.9	617
2	Recurrent reciprocal 1q21.1 deletions and duplications associated with microcephaly or macrocephaly and developmental and behavioral abnormalities. Nature Genetics, 2008, 40, 1466-1471.	21.4	535
3	A mouse model for mitochondrial myopathy and cardiomyopathy resulting from a deficiency in the heart/muscle isoform of the adenine nucleotide translocator. Nature Genetics, 1997, 16, 226-234.	21.4	523
4	Recurrent reciprocal 16p11.2 rearrangements associated with global developmental delay, behavioural problems, dysmorphism, epilepsy, and abnormal head size. Journal of Medical Genetics, 2010, 47, 332-341.	3.2	447
5	Use of Exome Sequencing for Infants in Intensive Care Units. JAMA Pediatrics, 2017, 171, e173438.	6.2	348
6	Marked Changes in Mitochondrial DNA Deletion Levels in Alzheimer Brains. Genomics, 1994, 23, 471-476.	2.9	290
7	Sox9 and NFIA Coordinate a Transcriptional Regulatory Cascade during the Initiation of Gliogenesis. Neuron, 2012, 74, 79-94.	8.1	287
8	The genomic landscape of balanced cytogenetic abnormalities associated with human congenital anomalies. Nature Genetics, 2017, 49, 36-45.	21.4	251
9	Fatty Acid Oxidation-Driven Src Links Mitochondrial Energy Reprogramming and Oncogenic Properties in Triple-Negative Breast Cancer. Cell Reports, 2016, 14, 2154-2165.	6.4	232
10	Mutations in NGLY1 cause an inherited disorder of the endoplasmic reticulumâ€“associated degradation pathway. Genetics in Medicine, 2014, 16, 751-758.	2.4	191
11	Lessons learned from additional research analyses of unsolved clinical exome cases. Genome Medicine, 2017, 9, 26.	8.2	184
12	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
13	Mutations in the Mitochondrial Methionyl-tRNA Synthetase Cause a Neurodegenerative Phenotype in Flies and a Recessive Ataxia (ARSAL) in Humans. PLoS Biology, 2012, 10, e1001288.	5.6	147
14	Recurrent De Novo and Biallelic Variation of ATAD3A , Encoding a Mitochondrial Membrane Protein, Results in Distinct Neurological Syndromes. American Journal of Human Genetics, 2016, 99, 831-845.	6.2	146
15	Mutations in FBXL4 Cause Mitochondrial Encephalopathy and a Disorder of Mitochondrial DNA Maintenance. American Journal of Human Genetics, 2013, 93, 471-481.	6.2	137
16	Clinical course of sly syndrome (mucopolysaccharidosis type VII). Journal of Medical Genetics, 2016, 53, 403-418.	3.2	133
17	De novo truncating mutations in ASXL3 are associated with a novel clinical phenotype with similarities to Bohring-Opitz syndrome. Genome Medicine, 2013, 5, 11.	8.2	128
18	The GABA Transaminase, ABAT, Is Essential for Mitochondrial Nucleoside Metabolism. Cell Metabolism, 2015, 21, 417-427.	16.2	119

#	Article	IF	Citations
19	Myokine mediated muscle-kidney crosstalk suppresses metabolic reprogramming and fibrosis in damaged kidneys. Nature Communications, 2017, 8, 1493.	12.8	117
20	22q13.3 deletion syndrome: Clinical and molecular analysis using array CGH. American Journal of Medical Genetics, Part A, 2010, 152A, 573-581.	1.2	116
21	Structures and molecular mechanisms for common 15q13.3 microduplications involving CHRNA7: benign or pathological?. Human Mutation, 2010, 31, 840-850.	2.5	111
22	Mitochondrial fusion but not fission regulates larval growth and synaptic development through steroid hormone production. ELife, 2014, 3, .	6.0	109
23	Comprehensive next-generation sequence analyses of the entire mitochondrial genome reveal new insights into the molecular diagnosis of mitochondrial DNA disorders. Genetics in Medicine, 2013, 15, 388-394.	2.4	106
24	Mitochondrial biology, degenerative diseases and aging. BioFactors, 1998, 7, 187-190.	5.4	105
25	Recurrent Muscle Weakness with Rhabdomyolysis, Metabolic Crises, and Cardiac Arrhythmia Due to Bi-allelic TANGO2 Mutations. American Journal of Human Genetics, 2016, 98, 347-357.	6.2	98
26	A Syndromic Neurodevelopmental Disorder Caused by De Novo Variants in EBF3. American Journal of Human Genetics, 2017, 100, 128-137.	6.2	96
27	Loss of Nardilysin, a Mitochondrial Co-chaperone for Î±-Ketoglutarate Dehydrogenase, Promotes mTORC1 Activation and Neurodegeneration. Neuron, 2017, 93, 115-131.	8.1	95
28	The phenotypic spectrum of Schaaf-Yang syndrome: 18 new affected individuals from 14 families. Genetics in Medicine, 2017, 19, 45-52.	2.4	94
29	Maternal systemic primary carnitine deficiency uncovered by newborn screening: Clinical, biochemical, and molecular aspects. Genetics in Medicine, 2010, 12, 19-24.	2.4	91
30	Expression and sequence analysis of the mouse adenine nucleotide translocase 1 and 2 genes. Gene, 2000, 254, 57-66.	2.2	89
31	Mitochondrial DNA sequence analysis of four Alzheimer's and Parkinson's disease patients. American Journal of Medical Genetics Part A, 1996, 61, 283-289.	2.4	83
32	Transition to Next Generation Analysis of the Whole Mitochondrial Genome: A Summary of Molecular Defects. Human Mutation, 2013, 34, 882-893.	2.5	79
33	Voltage-dependant anion channels: Novel insights into isoform function through genetic models. Biochimica Et Biophysica Acta - Biomembranes, 2012, 1818, 1477-1485.	2.6	74
34	Loss of Frataxin induces iron toxicity, sphingolipid synthesis, and Pdk1/Mef2 activation, leading to neurodegeneration. ELife, 2016, 5, .	6.0	74
35	Lysosomal Signaling Promotes Longevity by Adjusting Mitochondrial Activity. Developmental Cell, 2019, 48, 685-696.e5.	7.0	71
36	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

#	Article	IF	Citations
37	The Cognitive and Behavioral Phenotypes of Individuals with CHRNA7 Duplications. Journal of Autism and Developmental Disorders, 2017, 47, 549-562.	2.7	68
38	Current molecular diagnostic algorithm for mitochondrial disorders. Molecular Genetics and Metabolism, 2010, 100, 111-117.	1.1	66
39	Phenotypic expansion in <scp>DDX</scp>3X â€“ a common cause of intellectual disability in females. Annals of Clinical and Translational Neurology, 2018, 5, 1277-1285.	3.7	66
40	Recurrent ACADVL molecular findings in individuals with a positive newborn screen for very long chain acyl-coA dehydrogenase (VLCAD) deficiency in the United States. Molecular Genetics and Metabolism, 2015, 116, 139-145.	1.1	65
41	Mitochondrial myopathy, lactic acidosis, and sideroblastic anemia (MLASA) plus associated with a novel de novo mutation (m.8969G>A) in the mitochondrial encoded ATP6 gene. Molecular Genetics and Metabolism, 2014, 113, 207-212.	1.1	63
42	MÃ©nage-Ã-Trois 1 Is Critical for the Transcriptional Function of PPARÎ³ Coactivator 1. Cell Metabolism, 2007, 5, 129-142.	16.2	56
43	Measurement of Mitochondrial Oxygen Consumption Using a Clark Electrode. Methods in Molecular Biology, 2012, 837, 63-72.	0.9	56
44	The C8ORF38 homologue Sicily is a cytosolic chaperone for a mitochondrial complex I subunit. Journal of Cell Biology, 2013, 200, 807-820.	5.2	56
45	Increased COUP-TFII expression in adult hearts induces mitochondrial dysfunction resulting in heart failure. Nature Communications, 2015, 6, 8245.	12.8	55
46	Bacteria, yeast, worms, and flies: Exploiting simple model organisms to investigate human mitochondrial diseases. Developmental Disabilities Research Reviews, 2010, 16, 200-218.	2.9	52
47	Computational Prediction of Position Effects of Apparently Balanced Human Chromosomal Rearrangements. American Journal of Human Genetics, 2017, 101, 206-217.	6.2	51
48	Analyses of SLC13A5 -epilepsy patients reveal perturbations of TCA cycle. Molecular Genetics and Metabolism, 2017, 121, 314-319.	1.1	48
49	Aberrant Function of the C-Terminal Tail of HIST1H1E Accelerates Cellular Senescence and Causes Premature Aging. American Journal of Human Genetics, 2019, 105, 493-508.	6.2	48
50	Impaired Mitochondrial Energy Production Causes Light-Induced Photoreceptor Degeneration Independent of Oxidative Stress. PLoS Biology, 2015, 13, e1002197.	5.6	48
51	The mitochondrial 13513G>A mutation is associated with Leigh disease phenotypes independent of complex I deficiency in muscle. Molecular Genetics and Metabolism, 2008, 94, 485-490.	1.1	47
52	Untargeted metabolomic profiling reveals multiple pathway perturbations and new clinical biomarkers in urea cycle disorders. Genetics in Medicine, 2019, 21, 1977-1986.	2.4	47
53	Clinical, morphological, biochemical, imaging and outcome parameters in 21 individuals with mitochondrial maintenance defect related to FBXL4 mutations. Journal of Inherited Metabolic Disease, 2015, 38, 905-914.	3.6	45
54	Activin Signaling: Effects on Body Composition and Mitochondrial Energy Metabolism. Endocrinology, 2009, 150, 3521-3529.	2.8	43

#	Article	IF	Citations
55	Delineation of a deletion region critical for corpus callosal abnormalities in chromosome 1q43â€“q44. European Journal of Human Genetics, 2012, 20, 176-179.	2.8	42
56	A syndrome of short stature, microcephaly and speech delay is associated with duplications reciprocal to the common Sotos syndrome deletion. European Journal of Human Genetics, 2010, 18, 258-261.	2.8	41
57	The Physiological Properties of a Novel Family of VDAC-Like Proteins from Drosophila melanogaster. Biophysical Journal, 2004, 86, 152-162.	0.5	39
58	Genetic strategies for dissecting mammalian and Drosophila voltage-dependent anion channel functions. Journal of Bioenergetics and Biomembranes, 2008, 40, 207-212.	2.3	39
59	An integrated approach for classifying mitochondrial DNA variants: one clinical diagnostic laboratoryâ€™s experience. Genetics in Medicine, 2012, 14, 620-626.	2.4	39
60	6q22.1 microdeletion and susceptibility to pediatric epilepsy. European Journal of Human Genetics, 2015, 23, 173-179.	2.8	35
61	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
62	Impaired mitochondrial complex I function as a candidate driver in the biological stress response and a concomitant stress-induced brain metabolic reprogramming in male mice. Translational Psychiatry, 2020, 10, 176.	4.8	33
63	Neurologic Dysfunction and Male Infertility in Drosophila porin Mutants. Journal of Biological Chemistry, 2010, 285, 11143-11153.	3.4	32
64	Functional cellular analyses reveal energy metabolism defect and mitochondrial DNA depletion in a case of mitochondrial aconitase deficiency. Molecular Genetics and Metabolism, 2016, 118, 28-34.	1.1	32
65	Peroxisomal biogenesis is genetically and biochemically linked to carbohydrate metabolism in Drosophila and mouse. PLoS Genetics, 2017, 13, e1006825.	3.5	31
66	Elevations of C14:1 and C14:2 Plasma Acylcarnitines in Fasted Children: AÂDiagnostic Dilemma. Journal of Pediatrics, 2016, 169, 208-213.e2.	1.8	30
67	Mitochondrial voltage-dependent anion channel gene family in Drosophila melanogaster: Complex patterns of evolution, genomic organization, and developmental expression. Molecular Genetics and Metabolism, 2005, 85, 308-317.	1.1	29
68	Two mtDNA mutations 14487T>C (M63V, ND6) and 12297T>C (tRNA Leu) in a Leigh syndrome family. Molecular Genetics and Metabolism, 2009, 96, 59-65.	1.1	28
69	Heterozygous variants inACTL6A, encoding a component of the BAF complex, are associated with intellectual disability. Human Mutation, 2017, 38, 1365-1371.	2.5	27
70	Genetic Approaches to Analyzing Mitochondrial Outer Membrane Permeability. Current Topics in Developmental Biology, 2004, 59, 87-118.	2.2	23
71	Diagnostic Challenges of Mitochondrial Disorders: Complexities of Two Genomes. Methods in Molecular Biology, 2012, 837, 35-46.	0.9	23
72	De novo and inherited TCF20 pathogenic variants are associated with intellectual disability, dysmorphic features, hypotonia, and neurological impairments with similarities to Smithâ€“Magenis syndrome. Genome Medicine, 2019, 11, 12.	8.2	23

#	Article	IF	Citations
73	Expanding the Molecular and Clinical Phenotype of SSR4-CDG. Human Mutation, 2015, 36, 1048-1051.	2.5	22
74	Milder clinical and biochemical phenotypes associated with the c.482G > A (p.Arg161Gln) pathogenic variant in cobalamin C disease: Implications for management and screening. Molecular Genetics and Metabolism, 2017, 122, 60-66.	1.1	20
75	Outcome of infants diagnosed with 3-methyl-crotonyl-CoA-carboxylase deficiency by newborn screening. Molecular Genetics and Metabolism, 2012, 106, 439-441.	1.1	19
76	Screen for abnormal mitochondrial phenotypes in mouse ES cells identifies model for Succinyl-CoA Ligase deficiency and mtDNA depletion. DMM Disease Models and Mechanisms, 2014, 7, 271-80.	2.4	19
77	Biochemical signatures mimicking multiple carboxylase deficiency in children with mutations in MT-ATP6. Mitochondrion, 2019, 44, 58-64.	3.4	19
78	Pathogenic Variants in Fucokinase Cause a Congenital Disorder of Glycosylation. American Journal of Human Genetics, 2018, 103, 1030-1037.	6.2	18
79	Alzheimer disease-related presenilin-1 variants exert distinct effects on monoamine oxidase-A activity in vitro. Journal of Neural Transmission, 2011, 118, 987-995.	2.8	16
80	Mitochondrial Dysfunction Mediated by Poly(ADP-Ribose) Polymerase-1 Activation Contributes to Hippocampal Neuronal Damage Following Status Epilepticus. International Journal of Molecular Sciences, 2017, 18, 1502.	4.1	16
81	Atypical presentation of moyamoya disease in an infant with a de novo RNF213 variant. American Journal of Medical Genetics, Part A, 2015, 167, 2742-2747.	1.2	15
82	UBR7 functions with UBR5 in the Notch signaling pathway and is involved in a neurodevelopmental syndrome with epilepsy, ptosis, and hypothyroidism. American Journal of Human Genetics, 2021, 108, 134-147.	6.2	15
83	Intrauterine growth retardation and placental vacuolization as presenting features in a case of GM1 gangliosidosis. Journal of Inherited Metabolic Disease, 2007, 30, 823-823.	3.6	14
84	Expanding the phenotypic spectrum of Succinyl-CoA ligase deficiency through functional validation of a new SUCLG1 variant. Molecular Genetics and Metabolism, 2016, 119, 68-74.	1.1	14
85	Pleiotropic neuropathological and biochemical alterations associated with Myo5a mutation in a rat Model. Brain Research, 2018, 1679, 155-170.	2.2	14
86	<scp>MED27</scp> Variants Cause Developmental Delay, Dystonia, and Cerebellar Hypoplasia. Annals of Neurology, 2021, 89, 828-833.	5.3	14
87	Biallelic variants in COX4I1 associated with a novel phenotype resembling Leigh syndrome with developmental regression, intellectual disability, and seizures. American Journal of Medical Genetics, Part A, 2019, 179, 2138-2143.	1.2	11
88	Rapid quantification of underivatized alloisoleucine and argininosuccinate using mixed-mode chromatography with tandem mass spectrometry. Journal of Chromatography B: Analytical Technologies in the Biomedical and Life Sciences, 2019, 1128, 121786.	2.3	11
89	Noninvasive, in vivo approaches to evaluating behavior and exercise physiology in mouse models of mitochondrial disease. Methods, 2002, 26, 364-370.	3.8	10
90	Exome sequencing of a patient with suspected mitochondrial disease reveals a likely multigenic etiology. BMC Medical Genetics, 2013, 14, 83.	2.1	10

#	Article	IF	Citations
91	Renal cell carcinoma harboring somatic TSC2 mutations in a child with methylmalonic acidemia. Pediatric Blood and Cancer, 2017, 64, e26286.	1.5	9
92	Untargeted Metabolomics of Slc13a5 Deficiency Reveal Critical Liverâ€“Brain Axis for Lipid Homeostasis. Metabolites, 2022, 12, 351.	2.9	7
93	Fineâ€“Lubinsky syndrome: Sibling pair suggests possible autosomal recessive inheritance. American Journal of Medical Genetics, Part A, 2007, 143A, 2576-2580.	1.2	5
94	Megaloblastic Anemia Progressing to Severe Thrombotic Microangiopathy in Patients with Disordered Vitamin B12 Metabolism: Case Reports and Literature Review. Journal of Pediatrics, 2018, 202, 315-319.e2.	1.8	5
95	Association between pre-diagnostic leukocyte mitochondrial DNA copy number and survival among colorectal cancer patients. Cancer Epidemiology, 2020, 68, 101778.	1.9	5
96	Insulinemic Potential of Lifestyle Is Inversely Associated with Leukocyte Mitochondrial DNA Copy Number in US White Adults. Journal of Nutrition, 2020, 150, 2156-2163.	2.9	3
97	Mitochondrial DNA sequence analysis of four Alzheimer's and Parkinson's disease patients. American Journal of Medical Genetics Part A, 1996, 61, 283-289.	2.4	3
98	Mechanistic Investigation of GHS-R Mediated Glucose-Stimulated Insulin Secretion in Pancreatic Islets. Biomolecules, 2022, 12, 407.	4.0	3
99	An unusual cause for Coffinâ€“Lowry syndrome: Three brothers with a novel microduplication in RPS6KA3. American Journal of Medical Genetics, Part A, 2019, 179, 2357-2364.	1.2	1
100	Acute Hyperammonemia, Lactic Acidosis, and Ketoacidosis in a Developmentally Normal Child. Clinical Chemistry, 2021, 67, 1572-1574.	3.2	1
101	Depletion of mtDNA with MMA: SUCLA2 and SUCLG1. , 2013, , 163-169.		0
102	An apparent new syndrome of extreme short stature, microcephaly, dysmorphic faces, intellectual disability, and a bone dysplasia of unknown etiology. American Journal of Medical Genetics, Part A, 2020, 182, 1562-1571.	1.2	0
103	Mitochondrial Protein Translation-Related Disease: Mitochondrial Ribosomal Proteins and Translation Factors. , 2013, , 277-285.		0
104	Girl-Boy Twins with Developmental Delay from 16p11.2 Triplication due to Biparental Inheritance from Two Parents with 16p11.2 Duplication. Cytogenetic and Genome Research, 2022, , 1-6.	1.1	0