# 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 | 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 |
| 2 | Mechanistic Investigation of GHS-R Mediated Glucose-Stimulated Insulin Secretion in Pancreatic Islets. Biomolecules, 2022, 12, 407. | 4.0 | 3 |
| 3 | Untargeted Metabolomics of Slc13a5 Deficiency Reveal Critical Liverâ€“Brain Axis for Lipid Homeostasis. Metabolites, 2022, 12, 351. | 2.9 | 7 |
| 4 | 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 |
| 5 | Acute Hyperammonemia, Lactic Acidosis, and Ketoacidosis in a Developmentally Normal Child. Clinical Chemistry, 2021, 67, 1572-1574. | 3.2 | 1 |
| 6 | <scp>MED27</scp> Variants Cause Developmental Delay, Dystonia, and Cerebellar Hypoplasia. Annals of Neurology, 2021, 89, 828-833. | 5.3 | 14 |
| 7 | Association between pre-diagnostic leukocyte mitochondrial DNA copy number and survival among colorectal cancer patients. Cancer Epidemiology, 2020, 68, 101778. | 1.9 | 5 |
| 8 | 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 |
| 9 | 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 |
| 10 | 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 |
| 11 | 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 |
| 12 | 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 |
| 13 | 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 |
| 14 | 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 |
| 15 | 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 |
| 16 | Lysosomal Signaling Promotes Longevity by Adjusting Mitochondrial Activity. Developmental Cell, 2019, 48, 685-696.e5. | 7.0 | 71 |
| 17 | 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 |
| 18 | Biochemical signatures mimicking multiple carboxylase deficiency in children with mutations in MT-ATP6. Mitochondrion, 2019, 44, 58-64. | 3.4 | 19 |

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|---|---|---|---|
| 19 | Pleiotropic neuropathological and biochemical alterations associated with Myo5a mutation in a rat Model. Brain Research, 2018, 1679, 155-170. | 2.2 | 14 |
| 20 | Pathogenic Variants in Fucokinase Cause a Congenital Disorder of Glycosylation. American Journal of Human Genetics, 2018, 103, 1030-1037. | 6.2 | 18 |
| 21 | 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 |
| 22 | 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 |
| 23 | The phenotypic spectrum of Schaaf-Yang syndrome: 18 new affected individuals from 14 families. Genetics in Medicine, 2017, 19, 45-52. | 2.4 | 94 |
| 24 | 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 |
| 25 | 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 |
| 26 | Lessons learned from additional research analyses of unsolved clinical exome cases. Genome Medicine, 2017, 9, 26. | 8.2 | 184 |
| 27 | A Syndromic Neurodevelopmental Disorder Caused by De Novo Variants in EBF3. American Journal of Human Genetics, 2017, 100, 128-137. | 6.2 | 96 |
| 28 | Loss of Nardilysin, a Mitochondrial Co-chaperone for Î±-Ketoglutarate Dehydrogenase, Promotes mTORC1 Activation and Neurodegeneration. Neuron, 2017, 93, 115-131. | 8.1 | 95 |
| 29 | Use of Exome Sequencing for Infants in Intensive Care Units. JAMA Pediatrics, 2017, 171, e173438. | 6.2 | 348 |
| 30 | Computational Prediction of Position Effects of Apparently Balanced Human Chromosomal Rearrangements. American Journal of Human Genetics, 2017, 101, 206-217. | 6.2 | 51 |
| 31 | 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 |
| 32 | Myokine mediated muscle-kidney crosstalk suppresses metabolic reprogramming and fibrosis in damaged kidneys. Nature Communications, 2017, 8, 1493. | 12.8 | 117 |
| 33 | Heterozygous variants inACTL6A, encoding a component of the BAF complex, are associated with intellectual disability. Human Mutation, 2017, 38, 1365-1371. | 2.5 | 27 |
| 34 | Analyses of SLC13A5 -epilepsy patients reveal perturbations of TCA cycle. Molecular Genetics and Metabolism, 2017, 121, 314-319. | 1.1 | 48 |
| 35 | The genomic landscape of balanced cytogenetic abnormalities associated with human congenital anomalies. Nature Genetics, 2017, 49, 36-45. | 21.4 | 251 |
| 36 | The Cognitive and Behavioral Phenotypes of Individuals with CHRNA7 Duplications. Journal of Autism and Developmental Disorders, 2017, 47, 549-562. | 2.7 | 68 |

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|---|---|---|---|
| 37 | Renal cell carcinoma harboring somatic TSC2 mutations in a child with methylmalonic acidemia. Pediatric Blood and Cancer, 2017, 64, e26286. | 1.5 | 9 |
| 38 | 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 |
| 39 | Peroxisomal biogenesis is genetically and biochemically linked to carbohydrate metabolism in Drosophila and mouse. PLoS Genetics, 2017, 13, e1006825. | 3.5 | 31 |
| 40 | Loss of Frataxin induces iron toxicity, sphingolipid synthesis, and Pdk1/Mef2 activation, leading to neurodegeneration. ELife, 2016, 5, . | 6.0 | 74 |
| 41 | Clinical course of sly syndrome (mucopolysaccharidosis type VII). Journal of Medical Genetics, 2016, 53, 403-418. | 3.2 | 133 |
| 42 | 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 |
| 43 | 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 |
| 44 | 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 |
| 45 | 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 |
| 46 | 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 |
| 47 | 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 |
| 48 | 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 |
| 49 | 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 |
| 50 | Expanding the Molecular and Clinical Phenotype of SSR4-CDG. Human Mutation, 2015, 36, 1048-1051. | 2.5 | 22 |
| 51 | 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 |
| 52 | Glial Lipid Droplets and ROS Induced by Mitochondrial Defects Promote Neurodegeneration. Cell, 2015, 160, 177-190. | 28.9 | 617 |
| 53 | The GABA Transaminase, ABAT, Is Essential for Mitochondrial Nucleoside Metabolism. Cell Metabolism, 2015, 21, 417-427. | 16.2 | 119 |
| 54 | 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 |

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|---|---|---|---|
| 55 | Increased COUP-TFII expression in adult hearts induces mitochondrial dysfunction resulting in heart failure. Nature Communications, 2015, 6, 8245. | 12.8 | 55 |
| 56 | 6q22.1 microdeletion and susceptibility to pediatric epilepsy. European Journal of Human Genetics, 2015, 23, 173-179. | 2.8 | 35 |
| 57 | Impaired Mitochondrial Energy Production Causes Light-Induced Photoreceptor Degeneration Independent of Oxidative Stress. PLoS Biology, 2015, 13, e1002197. | 5.6 | 48 |
| 58 | 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 |
| 59 | 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 |
| 60 | Mutations in NGLY1 cause an inherited disorder of the endoplasmic reticulumâ€“associated degradation pathway. Genetics in Medicine, 2014, 16, 751-758. | 2.4 | 191 |
| 61 | Mitochondrial fusion but not fission regulates larval growth and synaptic development through steroid hormone production. ELife, 2014, 3, . | 6.0 | 109 |
| 62 | Exome sequencing of a patient with suspected mitochondrial disease reveals a likely multigenic etiology. BMC Medical Genetics, 2013, 14, 83. | 2.1 | 10 |
| 63 | Depletion of mtDNA with MMA: SUCLA2 and SUCLG1. , 2013, , 163-169. | | 0 |
| 64 | 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 |
| 65 | 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 |
| 66 | Transition to Next Generation Analysis of the Whole Mitochondrial Genome: A Summary of Molecular Defects. Human Mutation, 2013, 34, 882-893. | 2.5 | 79 |
| 67 | 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 |
| 68 | 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 |
| 69 | Mitochondrial Protein Translation-Related Disease: Mitochondrial Ribosomal Proteins and Translation Factors. , 2013, , 277-285. | | 0 |
| 70 | 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 |
| 71 | Sox9 and NFIA Coordinate a Transcriptional Regulatory Cascade during the Initiation of Gliogenesis. Neuron, 2012, 74, 79-94. | 8.1 | 287 |
| 72 | Voltage-dependant anion channels: Novel insights into isoform function through genetic models. Biochimica Et Biophysica Acta - Biomembranes, 2012, 1818, 1477-1485. | 2.6 | 74 |

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|---|---|---|---|
| 73 | Measurement of Mitochondrial Oxygen Consumption Using a Clark Electrode. Methods in Molecular Biology, 2012, 837, 63-72. | 0.9 | 56 |
| 74 | An integrated approach for classifying mitochondrial DNA variants: one clinical diagnostic laboratoryâ€™s experience. Genetics in Medicine, 2012, 14, 620-626. | 2.4 | 39 |
| 75 | 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 |
| 76 | Diagnostic Challenges of Mitochondrial Disorders: Complexities of Two Genomes. Methods in Molecular Biology, 2012, 837, 35-46. | 0.9 | 23 |
| 77 | 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 |
| 78 | 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 |
| 79 | Structures and molecular mechanisms for common 15q13.3 microduplications involving CHRNA7: benign or pathological?. Human Mutation, 2010, 31, 840-850. | 2.5 | 111 |
| 80 | 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 |
| 81 | 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 |
| 82 | 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 |
| 83 | 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 |
| 84 | Neurologic Dysfunction and Male Infertility in Drosophila porin Mutants. Journal of Biological Chemistry, 2010, 285, 11143-11153. | 3.4 | 32 |
| 85 | Maternal systemic primary carnitine deficiency uncovered by newborn screening: Clinical, biochemical, and molecular aspects. Genetics in Medicine, 2010, 12, 19-24. | 2.4 | 91 |
| 86 | Current molecular diagnostic algorithm for mitochondrial disorders. Molecular Genetics and Metabolism, 2010, 100, 111-117. | 1.1 | 66 |
| 87 | Activin Signaling: Effects on Body Composition and Mitochondrial Energy Metabolism. Endocrinology, 2009, 150, 3521-3529. | 2.8 | 43 |
| 88 | 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 |
| 89 | Genetic strategies for dissecting mammalian and Drosophila voltage-dependent anion channel functions. Journal of Bioenergetics and Biomembranes, 2008, 40, 207-212. | 2.3 | 39 |
| 90 | 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 |

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|---|---|---|---|
| 91 | 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 |
| 92 | MÃ©nage-Ã-Trois 1 Is Critical for the Transcriptional Function of PPARÎ³ Coactivator 1. Cell Metabolism, 2007, 5, 129-142. | 16.2 | 56 |
| 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 | 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 |
| 95 | 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 |
| 96 | Genetic Approaches to Analyzing Mitochondrial Outer Membrane Permeability. Current Topics in Developmental Biology, 2004, 59, 87-118. | 2.2 | 23 |
| 97 | The Physiological Properties of a Novel Family of VDAC-Like Proteins from Drosophila melanogaster. Biophysical Journal, 2004, 86, 152-162. | 0.5 | 39 |
| 98 | Noninvasive, in vivo approaches to evaluating behavior and exercise physiology in mouse models of mitochondrial disease. Methods, 2002, 26, 364-370. | 3.8 | 10 |
| 99 | Expression and sequence analysis of the mouse adenine nucleotide translocase 1 and 2 genes. Gene, 2000, 254, 57-66. | 2.2 | 89 |
| 100 | Mitochondrial biology, degenerative diseases and aging. BioFactors, 1998, 7, 187-190. | 5.4 | 105 |
| 101 | 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 |
| 102 | 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 |
| 103 | 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 |
| 104 | Marked Changes in Mitochondrial DNA Deletion Levels in Alzheimer Brains. Genomics, 1994, 23, 471-476. | 2.9 | 290 |