# K Michael Gibson

List of Publications by Year
in descending order



| 188 | 7,294 | 36303 | 79698 |
|---|---|---|---|
| papers | citations | 51 h-index | 73 g-index |

| 192 | 192 | 192 | 6736 |
|---|---|---|---|
| all docs | docs citations | times ranked | citing authors |

| # | Article | IF | Citations |
|---|---|---|---|
| 1 | Farnesol induces protection against murine CNS inflammatory demyelination and modifies gut microbiome. Clinical Immunology, 2022, 235, 108766. | 3.2 | 13 |
| 2 | Intestinal Dysbiosis as a component of pathophysiology in succinic semialdehyde dehydrogenase deficiency (SSADHD). Molecular Genetics and Metabolism, 2022, 135, 42-46. | 1.1 | 2 |
| 3 | Discovery and Optimization of 5-Hydroxy-Diclofenac toward a New Class of Ligands with Nanomolar Affinity for the CaMKIIÎ± Hub Domain. Journal of Medicinal Chemistry, 2022, 65, 6656-6676. | 6.4 | 3 |
| 4 | Dysbiosis of the intestinal microbiome as a component of pathophysiology in the inborn errors of metabolism. Molecular Genetics and Metabolism, 2021, 132, 1-10. | 1.1 | 11 |
| 5 | Succinic Semialdehyde Dehydrogenase Deficiency: Review of the Natural History Study. Journal of Child Neurology, 2021, 36, 1153-1161. | 1.4 | 15 |
| 6 | Postmortem Analyses in a Patient With Succinic Semialdehyde Dehydrogenase Deficiency (SSADHD): II. Histological, Lipid, and Gene Expression Outcomes in Regional Brain Tissue. Journal of Child Neurology, 2021, 36, 1177-1188. | 1.4 | 7 |
| 7 | Preferential accumulation of the active S-(+) isomer in murine retina highlights novel mechanisms of vigabatrin-associated retinal toxicity. Epilepsy Research, 2021, 170, 106536. | 1.6 | 3 |
| 8 | A Randomized Controlled Trial of SGS-742, a Î³-aminobutyric acid B (GABA-B) Receptor Antagonist, for Succinic Semialdehyde Dehydrogenase Deficiency. Journal of Child Neurology, 2021, 36, 1189-1199. | 1.4 | 6 |
| 9 | Author Response: Novel ALDH5A1 Variants and Genotype: Phenotype Correlation in SSADH Deficiency. Neurology, 2021, 96, 1060-1060. | 1.1 | 0 |
| 10 | Development of a Quality-of-Life Survey for Patients With Succinic Semialdehyde Dehydrogenase Deficiency, a Rare Disorder of GABA Metabolism. Journal of Child Neurology, 2021, 36, 1223-1230. | 1.4 | 2 |
| 11 | Novel Dual-Organelle-Targeting Probe (RCPP) for Simultaneous Measurement of Organellar Acidity and Alkalinity in Living Cells. ACS Omega, 2021, 6, 31447-31456. | 3.5 | 9 |
| 12 | Transcriptome analysis in mice treated with vigabatrin identifies dysregulation of genes associated with retinal signaling circuitry. Epilepsy Research, 2020, 166, 106395. | 1.6 | 2 |
| 13 | Novel ALDH5A1 variants and genotype. Neurology, 2020, 95, e2675-e2682. | 1.1 | 14 |
| 14 | Novel biomarkers and age-related metabolite correlations in plasma and dried blood spots from patients with succinic semialdehyde dehydrogenase deficiency. Orphanet Journal of Rare Diseases, 2020, 15, 261. | 2.7 | 3 |
| 15 | Cellular and molecular outcomes of glutamine supplementation in the brain of succinic semialdehyde dehydrogenase<scp>â€deficient</scp> mice. JIMD Reports, 2020, 56, 58-69. | 1.5 | 3 |
| 16 | A Missense Variant in ALDH5A1 Associated with Canine Succinic Semialdehyde Dehydrogenase Deficiency (SSADHD) in the Saluki Dog. Genes, 2020, 11, 1033. | 2.4 | 3 |
| 17 | Functional analysis of thirty-four suspected pathogenic missense variants in ALDH5A1 gene associated with succinic semialdehyde dehydrogenase deficiency. Molecular Genetics and Metabolism, 2020, 130, 172-178. | 1.1 | 8 |
| 18 | Longitudinal metabolomics in dried bloodspots yields profiles informing newborn screening for succinic semialdehyde dehydrogenase deficiency. JIMD Reports, 2020, 53, 29-38. | 1.5 | 3 |

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| 19 | Post-mortem tissue analyses in a patient with succinic semialdehyde dehydrogenase deficiency (SSADHD). I. Metabolomic outcomes. Metabolic Brain Disease, 2020, 35, 601-614. | 2.9 | 16 |
| 20 | Vigabatrin-Induced Retinal Functional Alterations and Second-Order Neuron Plasticity in C57BL/6J Mice. , 2020, 61, 17. | | 11 |
| 21 | Zellweger spectrum disorder: A cross-sectional study of symptom prevalence using input from family caregivers. Molecular Genetics and Metabolism Reports, 2020, 25, 100694. | 1.1 | 4 |
| 22 | Microbiota Manipulation as a Metagenomic Therapeutic Approach for Rare Inherited Metabolic Disorders. Clinical Pharmacology and Therapeutics, 2019, 106, 505-507. | 4.7 | 5 |
| 23 | Gamma-Hydroxybutyrate content in dried bloodspots facilitates newborn detection of succinic semialdehyde dehydrogenase deficiency. Molecular Genetics and Metabolism, 2019, 128, 109-112. | 1.1 | 10 |
| 24 | Temporal metabolomics in dried bloodspots suggests multipathway disruptions in aldh5a1 mice, a model of succinic semialdehyde dehydrogenase deficiency. Molecular Genetics and Metabolism, 2019, 128, 397-408. | 1.1 | 6 |
| 25 | Cardioprotection Effects of LPTC-5 Involve Mitochondrial Protection and Dynamics. ACS Omega, 2019, 4, 9868-9877. | 3.5 | 1 |
| 26 | Maternal glutamine supplementation in murine succinic semialdehyde dehydrogenase deficiency, a disorder of Î³â€aminobutyric acid metabolism. Journal of Inherited Metabolic Disease, 2019, 42, 1030-1039. | 3.6 | 4 |
| 27 | Dâ€2â€hydroxyglutaric aciduria Type I: Functional analysis of D2HGDH missense variants. Human Mutation, 2019, 40, 975-982. | 2.5 | 8 |
| 28 | Rett syndrome ( MECP2 ) and succinic semialdehyde dehydrogenase ( ALDH5A1 ) deficiency in a developmentally delayed female. Molecular Genetics & Genomic Medicine, 2019, 7, e629. | 1.2 | 2 |
| 29 | Emotional experience in parents of children with Zellweger spectrum disorders: A qualitative study. Molecular Genetics and Metabolism Reports, 2019, 19, 100459. | 1.1 | 14 |
| 30 | Ageâ€related phenotype and biomarker changes in <scp>SSADH</scp> deficiency. Annals of Clinical and Translational Neurology, 2019, 6, 114-120. | 3.7 | 27 |
| 31 | Preclinical tissue distribution and metabolic correlations of vigabatrin, an antiepileptic drug associated with potential useâ€limiting visual field defects. Pharmacology Research and Perspectives, 2019, 7, e00456. | 2.4 | 10 |
| 32 | Metabolomic analyses of vigabatrin (VGB)-treated mice: GABA-transaminase inhibition significantly alters amino acid profiles in murine neural and non-neural tissues. Neurochemistry International, 2019, 125, 151-162. | 3.8 | 13 |
| 33 | Membraneâ€Facilitated Allosteric Modulation of GABA A Receptor by Farnesol: An In Silico Modeling and Simulation Study. FASEB Journal, 2019, 33, 809.8. | 0.5 | 0 |
| 34 | Succinic semialdehyde dehydrogenase deficiency, a disorder of GABA metabolism: an update on pharmacological and enzymeâ€replacement therapeutic strategies. Journal of Inherited Metabolic Disease, 2018, 41, 699-708. | 3.6 | 30 |
| 35 | Toxicologic/transport properties of NCS-382, a Î³-hydroxybutyrate (GHB) receptor ligand, in neuronal and epithelial cells: Therapeutic implications for SSADH deficiency, a GABA metabolic disorder. Toxicology in Vitro, 2018, 46, 203-212. | 2.4 | 10 |
| 36 | Indole Alkaloid Derivative B, a Novel Bifunctional Agent That Mitigates 5-Fluorouracil-Induced Cardiotoxicity. ACS Omega, 2018, 3, 15850-15864. | 3.5 | 10 |

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| 37 | In vitro toxicological evaluation of NCS-382, a high-affinity antagonist of Î³-hydroxybutyrate (GHB) binding. Toxicology in Vitro, 2017, 40, 196-202. | 2.4 | 5 |
| 38 | Phenotype of GABA-transaminase deficiency. Neurology, 2017, 88, 1919-1924. | 1.1 | 49 |
| 39 | Oral D-galactose supplementation in PGM1-CDG. Genetics in Medicine, 2017, 19, 1226-1235. | 2.4 | 55 |
| 40 | Gamma-Hydroxybutyrate (GHB) Content in Hair Samples Correlates Negatively with Age in Succinic Semialdehyde Dehydrogenase Deficiency. JIMD Reports, 2017, 36, 93-98. | 1.5 | 9 |
| 41 | Targeted screening of succinic semialdehyde dehydrogenase deficiency (SSADHD) employing an enzymatic assay for Î³-hydroxybutyric acid (GHB) in biofluids. Molecular Genetics and Metabolism Reports, 2017, 11, 81-89. | 1.1 | 4 |
| 42 | Indole-TEMPO conjugates alleviate ischemia-reperfusion injury via attenuation of oxidative stress and preservation of mitochondrial function. Bioorganic and Medicinal Chemistry, 2017, 25, 2545-2568. | 3.0 | 11 |
| 43 | Therapeutic relevance of mTOR inhibition in murine succinate semialdehyde dehydrogenase deficiency (SSADHD), a disorder of GABA metabolism. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2017, 1863, 33-42. | 3.8 | 10 |
| 44 | Aberrant mTOR signaling and disrupted autophagy: The missing link in potential vigabatrinâ€associated ocular toxicity?. Clinical Pharmacology and Therapeutics, 2017, 101, 458-461. | 4.7 | 6 |
| 45 | Multicompartment analysis of proteinâ€restricted phenylketonuric mice reveals amino acid imbalances in brain. Journal of Inherited Metabolic Disease, 2017, 40, 227-235. | 3.6 | 10 |
| 46 | mTOR Inhibition Mitigates Molecular and Biochemical Alterations of Vigabatrin-Induced Visual Field Toxicity in Mice. Pediatric Neurology, 2017, 66, 44-52.e1. | 2.1 | 16 |
| 47 | In vitro modeling of experimental succinic semialdehyde dehydrogenase deficiency (SSADHD) using brain-derived neural stem cells. PLoS ONE, 2017, 12, e0186919. | 2.5 | 5 |
| 48 | mTOR, Autophagy, Aminoacidopathies, and Human Genetic Disorders. , 2016, , 143-166. | | 1 |
| 49 | A pharmacokinetic evaluation and metabolite identification of the <scp>GHB</scp> receptor antagonist <scp>NCS</scp> â€382 in mouse informs novel therapeutic strategies for the treatment of <scp>GHB</scp> intoxication. Pharmacology Research and Perspectives, 2016, 4, e00265. | 2.4 | 11 |
| 50 | Correlation of blood biomarkers with age informs pathomechanisms in succinic semialdehyde dehydrogenase deficiency (SSADHD), a disorder of GABA metabolism. Journal of Inherited Metabolic Disease, 2016, 39, 795-800. | 3.6 | 20 |
| 51 | Pharmacological protection of mitochondrial function mitigates acute limb ischemia/reperfusion injury. Bioorganic and Medicinal Chemistry Letters, 2016, 26, 4042-4051. | 2.2 | 12 |
| 52 | Succinic semialdehyde dehydrogenase deficiency (SSADHD): Pathophysiological complexity and multifactorial trait associations in a rare monogenic disorder of GABA metabolism. Neurochemistry International, 2016, 99, 72-84. | 3.8 | 60 |
| 53 | mTOR inhibitors rescue premature lethality and attenuate dysregulation of GABAergic/glutamatergic transcription in murine succinate semialdehyde dehydrogenase deficiency (SSADHD), a disorder of GABA metabolism. Journal of Inherited Metabolic Disease, 2016, 39, 877-886. | 3.6 | 23 |
| 54 | Incidence and Geographic Distribution of Succinic Semialdehyde Dehydrogenase (SSADH) Deficiency. JIMD Reports, 2016, 34, 111-115. | 1.5 | 22 |

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| 55 | Anti-inflammatory, analgesic and antioxidant activities of novel kyotorphin-nitroxide hybrid molecules. Bioorganic and Medicinal Chemistry Letters, 2016, 26, 2005-2013. | 2.2 | 12 |
| 56 | Disorders of Neurotransmission. , 2016, , 415-427. | | 0 |
| 57 | Disorders of GABA metabolism: SSADH and GABA-transaminase deficiencies. Journal of Pediatric Epilepsy, 2015, 03, 217-227. | 0.2 | 63 |
| 58 | Torin 1 partially corrects vigabatrinâ€induced mitochondrial increase in mouse. Annals of Clinical and Translational Neurology, 2015, 2, 699-706. | 3.7 | 23 |
| 59 | Biomarkers in a Taurine Trial for Succinic Semialdehyde Dehydrogenase Deficiency. JIMD Reports, 2015, 30, 81-87. | 1.5 | 17 |
| 60 | Inherited disorders of gammaâ€aminobutyric acid metabolism and advances in <scp>ALDH</scp>5A1 mutation identification. Developmental Medicine and Child Neurology, 2015, 57, 611-617. | 2.1 | 44 |
| 61 | Highly Stable and Sensitive Fluorescent Probes (LysoProbes) for Lysosomal Labeling and Tracking. Scientific Reports, 2015, 5, 8576. | 3.3 | 66 |
| 62 | Clinical Use of CSF Neurotransmitters. Pediatric Neurology, 2015, 53, 277-286. | 2.1 | 164 |
| 63 | Lysosomal Targeting with Stable and Sensitive Fluorescent Probes (Superior LysoProbes): Applications for Lysosome Labeling and Tracking during Apoptosis. Scientific Reports, 2015, 5, 9004. | 3.3 | 68 |
| 64 | Targeted fluorescent probes for detection of oxidative stress in the mitochondria. Bioorganic and Medicinal Chemistry Letters, 2015, 25, 3476-3480. | 2.2 | 13 |
| 65 | Natural history of succinic semialdehyde dehydrogenase deficiency through adulthood. Neurology, 2015, 85, 861-865. | 1.1 | 44 |
| 66 | Physiological competition of brain phenylalanine accretion: Initial pharmacokinetic analyses of aminoisobutyric and methylaminoisobutyric acids in Pahenu2âˆ'/âˆ' mice. Molecular Genetics and Metabolism Reports, 2015, 3, 80-87. | 1.1 | 2 |
| 67 | Taurine trial in succinic semialdehyde dehydrogenase deficiency and elevated CNS GABA. Neurology, 2014, 82, 940-944. | 1.1 | 30 |
| 68 | Defects in <scp>GABA</scp> metabolism affect selective autophagy pathways and are alleviated by m<scp>TOR</scp> inhibition. EMBO Molecular Medicine, 2014, 6, 551-566. | 6.9 | 67 |
| 69 | Pharmacologic inhibition of Lâ€tyrosine degradation ameliorates cerebral dopamine deficiency in murine phenylketonuria (PKU). Journal of Inherited Metabolic Disease, 2014, 37, 735-743. | 3.6 | 38 |
| 70 | Therapeutic hepatocyte transplant for inherited metabolic disorders: functional considerations, recent outcomes and future prospects. Journal of Inherited Metabolic Disease, 2014, 37, 165-176. | 3.6 | 8 |
| 71 | Brainâ€“blood amino acid correlates following protein restriction in murine maple syrup urine disease. Orphanet Journal of Rare Diseases, 2014, 9, 73. | 2.7 | 17 |
| 72 | Response to Stove and colleagues concerning newborn screening of succinic semialdehyde dehydrogenase (SSADH) deficiency in dried blood spots. Molecular Genetics and Metabolism, 2013, 110, 196. | 1.1 | 1 |

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| 73 | Nonâ€physiological amino acid (NPAA) therapy targeting brain phenylalanine reduction: pilot studies in PAH<sup>ENU2</sup> mice. Journal of Inherited Metabolic Disease, 2013, 36, 513-523. | 3.6 | 17 |
| 74 | Characterization of 2-(methylamino)alkanoic acid capacity to restrict bloodâ€“brain phenylalanine transport in Pahenu2 mice: Preliminary findings. Molecular Genetics and Metabolism, 2013, 110, S71-S78. | 1.1 | 10 |
| 75 | Improved Amino Acid, Bioenergetic Metabolite and Neurotransmitter Profiles following Human Amnion Epithelial Cell Transplant in Intermediate Maple Syrup Urine Disease Mice. Molecular Genetics and Metabolism, 2013, 109, 132-138. | 1.1 | 42 |
| 76 | Quantitation of gamma-hydroxybutyric acid in dried blood spots: Feasibility assessment for newborn screening of succinic semialdehyde dehydrogenase (SSADH) deficiency. Molecular Genetics and Metabolism, 2013, 109, 255-259. | 1.1 | 29 |
| 77 | Thirty years beyond discoveryâ€"Clinical trials in succinic semialdehyde dehydrogenase deficiency, a disorder of GABA metabolism. Journal of Inherited Metabolic Disease, 2013, 36, 401-410. | 3.6 | 53 |
| 78 | Placental stem cell correction of murine intermediate maple syrup urine disease. Hepatology, 2013, 57, 1017-1023. | 7.3 | 58 |
| 79 | Evidence for treatable inborn errors of metabolism in a cohort of 187 Greek patients with autism spectrum disorder (ASD). Frontiers in Human Neuroscience, 2013, 7, 858. | 2.0 | 53 |
| 80 | Therapeutic Efficacy of Magnesium Valproate in Succinic Semialdehyde Dehydrogenase Deficiency. JIMD Reports, 2012, 8, 133-137. | 1.5 | 15 |
| 81 | New Rhodamine Nitroxide Based Fluorescent Probes for Intracellular Hydroxyl Radical Identification in Living Cells. Organic Letters, 2012, 14, 50-53. | 4.6 | 96 |
| 82 | Succinic Semialdehyde Dehydrogenase: Biochemicalâ€“Molecularâ€“Clinical Disease Mechanisms, Redox Regulation, and Functional Significance. Antioxidants and Redox Signaling, 2011, 15, 691-718. | 5.4 | 68 |
| 83 | Epilepsy in succinic semialdehyde dehydrogenase deficiency, a disorder of GABA metabolism. Brain and Development, 2011, 33, 796-805. | 1.1 | 56 |
| 84 | Familial 6p22.2 duplication associates with mild developmental delay and increased SSADH activity. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2011, 156, 448-453. | 1.7 | 5 |
| 85 | A new class of Î²-carboline alkaloid-peptide conjugates with therapeutic efficacy in acute limb ischemia/reperfusion injury. European Journal of Medicinal Chemistry, 2011, 46, 1453-1462. | 5.5 | 20 |
| 86 | Efficacy of Vigabatrin Intervention in a Mild Phenotypic Expression of Succinic Semialdehyde Dehydrogenase Deficiency. JIMD Reports, 2011, 2, 119-123. | 1.5 | 13 |
| 87 | A new case of GABA transaminase deficiency facilitated by proton MR spectroscopy. Journal of Inherited Metabolic Disease, 2010, 33, 85-90. | 3.6 | 57 |
| 88 | Neuropathology in Succinic Semialdehyde Dehydrogenase Deficiency. Pediatric Neurology, 2010, 42, 255-258. | 2.1 | 27 |
| 89 | Synthesis and Characterization of Novel Indole Derivatives Reveal Improved Therapeutic Agents for Treatment of Ischemia/Reperfusion (I/R) Injury. Journal of Medicinal Chemistry, 2010, 53, 6763-6767. | 6.4 | 27 |
| 90 | Plasticity of postsynaptic, but not presynaptic, GABAB receptors inSSADH deficient mice. Experimental Neurology, 2010, 225, 114-122. | 4.1 | 16 |

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| 91 | Cerebellar Atrophy in Human and Murine Succinic Semialdehyde Dehydrogenase Deficiency. Journal of Child Neurology, 2010, 25, 1457-1461. | 1.4 | 11 |
| 92 | Hepatocyte Transplantation Improves Phenotype and Extends Survival in a Murine Model of Intermediate Maple Syrup Urine Disease. Molecular Therapy, 2009, 17, 1266-1273. | 8.2 | 30 |
| 93 | Decreased GABA-A binding on FMZ-PET in succinic semialdehyde dehydrogenase deficiency. Neurology, 2009, 73, 423-429. | 1.1 | 88 |
| 94 | Succinic semialdehyde dehydrogenase deficiency: Lessons from mice and men. Journal of Inherited Metabolic Disease, 2009, 32, 343-352. | 3.6 | 97 |
| 95 | Hepatocyte transplantation (HTx) corrects selected neurometabolic abnormalities in murine intermediate maple syrup urine disease (iMSUD). Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2009, 1792, 1004-1010. | 3.8 | 25 |
| 96 | The effects of a ketogenic diet on ATP concentrations and the number of hippocampal mitochondria in Aldh5a1â¯/â¯ mice. Biochimica Et Biophysica Acta - General Subjects, 2009, 1790, 208-212. | 2.4 | 56 |
| 97 | Comparative genomics of aldehyde dehydrogenase 5a1 (succinate semialdehyde dehydrogenase) and accumulation of gamma-hydroxybutyrate associated with its deficiency. Human Genomics, 2009, 3, 106. | 2.9 | 30 |
| 98 | SSADH deficiency leads to elevated extracellular GABA levels and increased GABAergic neurotransmission in the mouse cerebral cortex. Journal of Inherited Metabolic Disease, 2008, 31, 662-668. | 3.6 | 18 |
| 99 | Neurotransmitter alterations in embryonic succinate semialdehyde dehydrogenase (SSADH) deficiency suggest a heightened excitatory state during development. BMC Developmental Biology, 2008, 8, 112. | 2.1 | 37 |
| 100 | Circadian distribution of generalized tonicâ€“clonic seizures associated with murine succinic semialdehyde dehydrogenase deficiency, a disorder of GABA metabolism. Epilepsy and Behavior, 2008, 13, 290-294. | 1.7 | 18 |
| 101 | A ketogenic diet rescues the murine succinic semialdehyde dehydrogenase deficient phenotype. Experimental Neurology, 2008, 210, 449-457. | 4.1 | 54 |
| 102 | Neuropsychiatric Morbidity in Adolescent and Adult Succinic Semialdehyde Dehydrogenase Deficiency Patients. CNS Spectrums, 2008, 13, 598-605. | 1.2 | 54 |
| 103 | Lipid abnormalities in succinate semialdehyde dehydrogenase (Aldh5a1â¯/â¯) deficient mouse brain provide additional evidence for myelin alterations. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2007, 1772, 556-562. | 3.8 | 18 |
| 104 | Enzymatic and metabolic evidence for a region specific mitochondrial dysfunction in brains of murine succinic semialdehyde dehydrogenase deficiency (Aldh5a1â¯/â¯ mice). Neurochemistry International, 2007, 50, 653-659. | 3.8 | 39 |
| 105 | Diagnostic challenges in a severely delayed infant with hypersomnolence, failure to thrive and arteriopathy: A unique case of Î³â€hydroxybutyric aciduria and Williams syndrome. American Journal of Medical Genetics Part B: Neuropsychiatric Genetics, 2007, 144B, 946-948. | 1.7 | 4 |
| 106 | Altered cerebral glucose and acetate metabolism in succinic semialdehyde dehydrogenaseâ€deficient mice: evidence for glial dysfunction and reduced glutamate/glutamine cycling. Journal of Neurochemistry, 2007, 103, 2077-2091. | 3.9 | 52 |
| 107 | Therapeutic concepts in succinate semialdehyde dehydrogenase (SSADH; ALDH5a1) deficiency (Î³-hydroxybutyric aciduria). Hypotheses evolved from 25Âyears of patient evaluation, studies in Aldh5a1 â¯/â¯ mice and characterization of Î³-hydroxybutyric acid pharmacology. Journal of Inherited Metabolic Disease, 2007, 30, 279-294. | 3.6 | 39 |
| 108 | Evidence for oxidative stress in tissues derived from succinate semialdehyde dehydrogenaseâ€deficient mice. Journal of Inherited Metabolic Disease, 2007, 30, 800-810. | 3.6 | 31 |

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| 109 | Metabolism of Î³-hydroxybutyrate to d-2-hydroxyglutarate in mammals: further evidence for d-2-hydroxyglutarate transhydrogenase. Metabolism: Clinical and Experimental, 2006, 55, 353-358. | 3.4 | 43 |
| 110 | Increased guanidino species in murine and human succinate semialdehyde dehydrogenase (SSADH) deficiency. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2006, 1762, 494-498. | 3.8 | 31 |
| 111 | Succinate semialdehyde dehydrogenase deficiency does not down-regulate Î³-hydroxybutyric acid binding sites in the mouse brain. Molecular Genetics and Metabolism, 2006, 88, 86-89. | 1.1 | 17 |
| 112 | d-2-Hydroxyglutaric aciduria in three patients with proven SSADH deficiency: Genetic coincidence or a related biochemical epiphenomenon?. Molecular Genetics and Metabolism, 2006, 88, 53-57. | 1.1 | 13 |
| 113 | A novel, quantitative assay for homocarnosine in cerebrospinal fluid using stable-isotope dilution liquid chromatographyâ€“tandem mass spectrometry. Journal of Chromatography B: Analytical Technologies in the Biomedical and Life Sciences, 2006, 830, 196-200. | 2.3 | 27 |
| 114 | Expression profiling reveals multiple myelin alterations in murine succinate semialdehyde dehydrogenase deficiency. Journal of Inherited Metabolic Disease, 2006, 29, 143-156. | 3.6 | 25 |
| 115 | Succinic Semialdehyde Dehydrogenase Deficiency: GABAB receptor-mediated function. Brain Research, 2006, 1090, 15-22. | 2.2 | 62 |
| 116 | Status epilepticus in mice deficient for succinate semialdehyde dehydrogenase: GABAA receptor-mediated mechanisms. Annals of Neurology, 2006, 59, 42-52. | 5.3 | 61 |
| 117 | Murine succinate semialdehyde dehydrogenase (SSADH) deficiency, a heritable disorder of GABA metabolism with epileptic phenotype. IUBMB Life, 2005, 57, 639-644. | 3.4 | 29 |
| 118 | Inherited disorders of neurotransmitters in children and adults. Clinical Biochemistry, 2005, 38, 1051-1058. | 1.9 | 80 |
| 119 | Determination of the GABA analogue succinic semialdehyde in urine and cerebrospinal fluid by dinitrophenylhydrazine derivatization and liquid chromatographyâ€“tandem mass spectrometry: Application to SSADH deficiency. Journal of Inherited Metabolic Disease, 2005, 28, 913-920. | 3.6 | 28 |
| 120 | Gamma-hydroxybutyric aciduria: A biochemistâ€™s education from a heritable disorder of GABA metabolism. Journal of Inherited Metabolic Disease, 2005, 28, 247-265. | 3.6 | 14 |
| 121 | Î³-Hydroxybutyric Acid. New England Journal of Medicine, 2005, 352, 2721-2732. | 27.0 | 285 |
| 122 | Liver-Directed Adenoviral Gene Transfer in Murine Succinate Semialdehyde Dehydrogenase Deficiency. Molecular Therapy, 2004, 9, 527-539. | 8.2 | 25 |
| 123 | Absence seizures in succinic semialdehyde dehydrogenase deficient mice: a model of juvenile absence epilepsy. Pharmacology Biochemistry and Behavior, 2004, 79, 547-553. | 2.9 | 65 |
| 124 | Oxidation of 4-hydroxy-2-nonenal by succinic semialdehyde dehydrogenase (ALDH5A). Journal of Neurochemistry, 2004, 86, 298-305. | 3.9 | 63 |
| 125 | ??-Hydroxybutyric Acid. Toxicological Reviews, 2004, 23, 3-20. | 2.5 | 84 |
| 126 | From the street to the brain: neurobiology of the recreational drug Î³-hydroxybutyric acid. Trends in Pharmacological Sciences, 2004, 25, 29-34. | 8.7 | 175 |

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| 127 | Seizure evolution and amino acid imbalances in murine succinate semialdehyde dehydrogenase (SSADH) deficiency. Neurobiology of Disease, 2004, 16, 556-562. | 4.4 | 54 |
| 128 | Photosensitive absence epilepsy with myoclonias and heterozygosity for succinic semialdehyde dehydrogenase (SSADH) deficiency. Clinical Neurophysiology, 2004, 115, 1417-1422. | 1.5 | 32 |
| 129 | Clinical aspects of the disorders of GABA metabolism in children. Current Opinion in Neurology, 2004, 17, 107-113. | 3.6 | 64 |
| 130 | Mutational spectrum of the succinate semialdehyde dehydrogenase (ALDH5A1) gene and functional analysis of 27 novel disease-causing mutations in patients with SSADH deficiency. Human Mutation, 2003, 22, 442-450. | 2.5 | 117 |
| 131 | Murine succinate semialdehyde dehydrogenase deficiency. Annals of Neurology, 2003, 54, S81-S90. | 5.3 | 48 |
| 132 | Succinic semialdehyde dehydrogenase deficiency in children and adults. Annals of Neurology, 2003, 54, S73-S80. | 5.3 | 108 |
| 133 | Magnetic resonance spectroscopy of neurotransmitters in human brain. Annals of Neurology, 2003, 54, S25-S31. | 5.3 | 126 |
| 134 | Monitoring of 4-hydroxybutyric acid levels in body fluids during vigabatrin treatment in succinic semialdehyde dehydrogenase deficiency. Annals of Neurology, 2003, 54, 686-689. | 5.3 | 39 |
| 135 | Significant behavioral disturbances in succinic semialdehyde dehydrogenase (SSADH) deficiency (Gamma-Hydroxybutyric aciduria). Biological Psychiatry, 2003, 54, 763-768. | 1.3 | 103 |
| 136 | Clinical spectrum of succinic semialdehyde dehydrogenase deficiency. Neurology, 2003, 60, 1413-1417. | 1.1 | 191 |
| 137 | Therapeutic Intervention in Mice Deficient for Succinate Semialdehyde Dehydrogenase (Î³-Hydroxybutyric Aciduria). Journal of Pharmacology and Experimental Therapeutics, 2002, 302, 180-187. | 2.5 | 81 |
| 138 | Structure of human succinic semialdehyde dehydrogenase gene: identification of promoter region and alternatively processed isoforms. Molecular Genetics and Metabolism, 2002, 76, 348-362. | 1.1 | 49 |
| 139 | Focal neurometabolic alterations in mice deficient for succinate semialdehyde dehydrogenase. Journal of Neurochemistry, 2002, 81, 71-79. | 3.9 | 53 |
| 140 | Enzymatic and Immunologic Identification of Succinic Semialdehyde Dehydrogenase in Rat and Human Neural and Nonneural Tissues. Journal of Neurochemistry, 2002, 65, 851-855. | 3.9 | 26 |
| 141 | Pharmacologic rescue of lethal seizures in mice deficient in succinate semialdehyde dehydrogenase. Nature Genetics, 2001, 29, 212-216. | 21.4 | 149 |
| 142 | Development of a Stable-Isotope Dilution Assay for Î³-Aminobutyric Acid (GABA) Transaminase in Isolated Leukocytes and Evidence That GABA and Î²-Alanine Transaminases Are Identical. Clinical Chemistry, 2001, 47, 525-531. | 3.2 | 20 |
| 143 | Delayed-onset dystonia associated with 3-oxothiolase deficiency. Movement Disorders, 2001, 16, 372-375. | 3.9 | 22 |
| 144 | Organization of the mevalonate kinase (MVK) gene and identification of novel mutations causing mevalonic aciduria and hyperimmunoglobulinaemia D and periodic fever syndrome. European Journal of Human Genetics, 2001, 9, 253-259. | 2.8 | 85 |

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| 145 | Development of a stable-isotope dilution assay for gamma-aminobutyric acid (GABA) transaminase in isolated leukocytes and evidence that GABA and beta-alanine transaminases are identical. Clinical Chemistry, 2001, 47, 525-31. | 3.2 | 9 |
| 146 | Fetal demise with Smith-Lemli-Opitz syndrome confirmed by tissue sterol analysis and the absence of measurable 7-dehydrocholesterol ?7-reductase activity in chorionic villi. , 2000, 20, 238-240. | | 30 |
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