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	Î³-Hydroxybutyric Acid. New England Journal of Medicine, 2005, 352, 2721-2732.	27.0	285
2	Clinical spectrum of succinic semialdehyde dehydrogenase deficiency. Neurology, 2003, 60, 1413-1417.	1.1	191
3	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
4	Clinical Use of CSF Neurotransmitters. Pediatric Neurology, 2015, 53, 277-286.	2.1	164
5	Gamma-aminobutyric acid (GABA) metabolism in mammalian neural and nonneural tissues. Comparative Biochemistry and Physiology A, Comparative Physiology, 1995, 112, 247-263.	0.6	163
6	Pharmacologic rescue of lethal seizures in mice deficient in succinate semialdehyde dehydrogenase. Nature Genetics, 2001, 29, 212-216.	21.4	149
7	The Clinical Phenotype of Succinic Semialdehyde Dehydrogenase Deficiency (4-Hydroxybutyric) Tj ETQq1 1 0.784314 rgBT /Overlock 10	2.1	134
8	4-Hydroxybutyric Acid and the Clinical Phenotype of Succinic Semialdehyde Dehydrogenase Deficiency, an Inborn Error of GABA Metabolism. Neuropediatrics, 1998, 29, 14-22.	0.6	128
9	Magnetic resonance spectroscopy of neurotransmitters in human brain. Annals of Neurology, 2003, 54, S25-S31.	5.3	126
10	Stable-Isotope Dilution Analysis of D- and L-2-Hydroxyglutaric Acid: Application to the Detection and Prenatal Diagnosis of D- and L-2-Hydroxyglutaric Acidemias. Pediatric Research, 1993, 34, 277-280.	2.3	117
11	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
12	3-hydroxy-3-methylglutaryl-coenzyme A lyase deficiency: review of 18 reported patients. European Journal of Pediatrics, 1988, 148, 180-186.	2.7	115
13	Succinic semialdehyde dehydrogenase deficiency in children and adults. Annals of Neurology, 2003, 54, S73-S80.	5.3	108
14	Significant behavioral disturbances in succinic semialdehyde dehydrogenase (SSADH) deficiency (Gamma-Hydroxybutyric aciduria). Biological Psychiatry, 2003, 54, 763-768.	1.3	103
15	Succinic semialdehyde dehydrogenase deficiency: Lessons from mice and men. Journal of Inherited Metabolic Disease, 2009, 32, 343-352.	3.6	97
16	New Rhodamine Nitroxide Based Fluorescent Probes for Intracellular Hydroxyl Radical Identification in Living Cells. Organic Letters, 2012, 14, 50-53.	4.6	96
17	2-Methylbutyryl-Coenzyme A Dehydrogenase Deficiency: A New Inborn Error of L-Isoleucine Metabolism. Pediatric Research, 2000, 47, 830-833.	2.3	92
18	Stable isotope dilution analysis of 4-hydroxybutyric acid: An accurate method for quantification in physiological fluids and the prenatal diagnosis of 4-hydroxybutyric aciduria. Biological Mass Spectrometry, 1990, 19, 89-93.	0.5	89

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19	Decreased GABA-A binding on FMZ-PET in succinic semialdehyde dehydrogenase deficiency. Neurology, 2009, 73, 423-429.	1.1	88
20	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
21	??-Hydroxybutyric Acid. Toxicological Reviews, 2004, 23, 3-20.	2.5	84
22	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
23	Inherited disorders of neurotransmitters in children and adults. Clinical Biochemistry, 2005, 38, 1051-1058.	1.9	80
24	Succinic semialdehyde dehydrogenase deficiency: an inborn error of gamma-aminobutyric acid metabolism. Clinica Chimica Acta, 1983, 133, 33-42.	1.1	73
25	Two Exon-Skipping Mutations as the Molecular Basis of Succinic Semialdehyde Dehydrogenase Deficiency (4-Hydroxybutyric Aciduria). American Journal of Human Genetics, 1998, 63, 399-408.	6.2	73
26	3-Methylcrotonyl-coenzyme A carboxylase deficiency in Amish/Mennonite adults identified by detection of increased acylcarnitines in blood spots of their children. Journal of Pediatrics, 1998, 132, 519-523.	1.8	72
27	Molecular Cloning of the Mature NAD+-dependent Succinic Semialdehyde Dehydrogenase from Rat and Human. Journal of Biological Chemistry, 1995, 270, 461-467.	3.4	70
28	Succinic Semialdehyde Dehydrogenase: Biochemicalâ€“Molecularâ€“Clinical Disease Mechanisms, Redox Regulation, and Functional Significance. Antioxidants and Redox Signaling, 2011, 15, 691-718.	5.4	68
29	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
30	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
31	Multiple syndromes of 3-methylglutaconic aciduria. Pediatric Neurology, 1993, 9, 120-123.	2.1	66
32	Highly Stable and Sensitive Fluorescent Probes (LysoProbes) for Lysosomal Labeling and Tracking. Scientific Reports, 2015, 5, 8576.	3.3	66
33	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
34	Clinical aspects of the disorders of GABA metabolism in children. Current Opinion in Neurology, 2004, 17, 107-113.	3.6	64
35	Oxidation of 4-hydroxy-2-nonenal by succinic semialdehyde dehydrogenase (ALDH5A). Journal of Neurochemistry, 2004, 86, 298-305.	3.9	63
36	Disorders of GABA metabolism: SSADH and GABA-transaminase deficiencies. Journal of Pediatric Epilepsy, 2015, 03, 217-227.	0.2	63

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37	Succinic Semialdehyde Dehydrogenase Deficiency: GABAB receptor-mediated function. Brain Research, 2006, 1090, 15-22.	2.2	62
38	Status epilepticus in mice deficient for succinate semialdehyde dehydrogenase: GABAA receptor-mediated mechanisms. Annals of Neurology, 2006, 59, 42-52.	5.3	61
39	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
40	Placental stem cell correction of murine intermediate maple syrup urine disease. Hepatology, 2013, 57, 1017-1023.	7.3	58
41	Phenotypic heterogeneity in the syndromes of 3-methylglutaconic aciduria. Journal of Pediatrics, 1991, 118, 885-890.	1.8	57
42	A new case of GABA transaminase deficiency facilitated by proton MR spectroscopy. Journal of Inherited Metabolic Disease, 2010, 33, 85-90.	3.6	57
43	Hematological abnormalities and cholestatic liver disease in two patients with mevalonate kinase deficiency. American Journal of Medical Genetics Part A, 1998, 78, 408-412.	2.4	56
44	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
45	Epilepsy in succinic semialdehyde dehydrogenase deficiency, a disorder of GABA metabolism. Brain and Development, 2011, 33, 796-805.	1.1	56
46	Oral D-galactose supplementation in PGM1-CDG. Genetics in Medicine, 2017, 19, 1226-1235.	2.4	55
47	Seizure evolution and amino acid imbalances in murine succinate semialdehyde dehydrogenase (SSADH) deficiency. Neurobiology of Disease, 2004, 16, 556-562.	4.4	54
48	A ketogenic diet rescues the murine succinic semialdehyde dehydrogenase deficient phenotype. Experimental Neurology, 2008, 210, 449-457.	4.1	54
49	Neuropsychiatric Morbidity in Adolescent and Adult Succinic Semialdehyde Dehydrogenase Deficiency Patients. CNS Spectrums, 2008, 13, 598-605.	1.2	54
50	Focal neurometabolic alterations in mice deficient for succinate semialdehyde dehydrogenase. Journal of Neurochemistry, 2002, 81, 71-79.	3.9	53
51	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
52	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
53	4-Hydroxybutyric aciduria: Application of a fluorometric assay to the determination of succinic semialdehyde dehydrogenase activity in extracts of cultured human lymphoblasts. Clinica Chimica Acta, 1991, 196, 219-221.	1.1	52
54	Succinic semialdehyde dehydrogenase from mammalian brain: Subunit analysis using polyclonal antiserum. International Journal of Biochemistry & Cell Biology, 1992, 24, 1493-1499.	0.5	52

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55	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
56	3-Methylglutaconic aciduria associated with Pearson syndrome and respiratory chain defects. Journal of Pediatrics, 1992, 121, 940-942.	1.8	50
57	Vigabatrin therapy in six patients with succinic semialdehyde dehydrogenase deficiency. Journal of Inherited Metabolic Disease, 1995, 18, 143-146.	3.6	50
58	3-Hydroxy-3-methylglutaryl coenzyme A reductase activity in cultured fibroblasts from patients with mevalonate kinase deficiency: differential response to lipid supplied by fetal bovine serum in tissue culture medium. Journal of Lipid Research, 1990, 31, 515-21.	4.2	50
59	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
60	Phenotype of GABA-transaminase deficiency. Neurology, 2017, 88, 1919-1924.	1.1	49
61	3-hydroxy-3-methylglutaryl-coenzyme a lyase deficiency: Report of five new patients. Journal of Inherited Metabolic Disease, 1988, 11, 76-87.	3.6	48
62	Molecular characterization of methylmalonate semialdehyde dehydrogenase deficiency. Journal of Inherited Metabolic Disease, 2000, 23, 497-504.	3.6	48
63	Murine succinate semialdehyde dehydrogenase deficiency. Annals of Neurology, 2003, 54, S81-S90.	5.3	48
64	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
65	Natural history of succinic semialdehyde dehydrogenase deficiency through adulthood. Neurology, 2015, 85, 861-865.	1.1	44
66	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
67	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
68	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
69	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
70	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
71	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
72	Fatal cardiomyopathy associated with 3-hydroxy-3-methylglutaryl-CoA lyase deficiency. Journal of Inherited Metabolic Disease, 1994, 17, 291-294.	3.6	37

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73	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
74	Oxidation of [U-14C]Succinic Semialdehyde in Cultured Human Lymphoblasts: Measurement of Residual Succinic Semialdehyde Dehydrogenase Activity in 11 Patients with 4-Hydroxybutyric Aciduria. Pediatric Research, 1988, 24, 455-460.	2.3	36
75	Relationship between Kinetic Properties of Mutant Enzyme and Biochemical and Clinical Responsiveness to Biotin in Holocarboxylase Synthetase Deficiency. Pediatric Research, 1999, 46, 671-671.	2.3	35
76	Seizures in a boy with succinic semialdehyde dehydrogenase deficiency treated with vigabatrin (Î³-vinyl-GABA). Journal of Inherited Metabolic Disease, 1996, 19, 313-318.	3.6	34
77	VIGABATRIN THERAPY IN PATIENT WITH SUCCINIC SEMIALDEHYDE DEHYDROGENASE DEFICIENCY. Lancet, The, 1989, 334, 1105-1106.	13.7	32
78	Photosensitive absence epilepsy with myoclonias and heterozygosity for succinic semialdehyde dehydrogenase (SSADH) deficiency. Clinical Neurophysiology, 2004, 115, 1417-1422.	1.5	32
79	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
80	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
81	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
82	3â€METHYLGLUTACONIC ACIDURIA IN THE IRAQIâ€JEWISH â€œOPTIC ATROPHY PLUSâ€ (COSTEFF) SYNDROME. Developmental Medicine and Child Neurology, 1994, 36, 167-172.	2.1	30
83	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
84	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
85	Taurine trial in succinic semialdehyde dehydrogenase deficiency and elevated CNS GABA. Neurology, 2014, 82, 940-944.	1.1	30
86	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
87	Murine succinate semialdehyde dehydrogenase (SSADH) deficiency, a heritable disorder of GABA metabolism with epileptic phenotype. IUBMB Life, 2005, 57, 639-644.	3.4	29
88	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
89	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
90	Mevalonate Kinase in Lysates of Cultured Human Fibroblasts and Lymphoblasts: Kinetic Properties, Assay Conditions, Carrier Detection and Measurement of Residual Activity in a Patient with Mevalonic Aciduria. Enzyme, 1989, 41, 47-55.	0.7	27

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91	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
92	Neuropathology in Succinic Semialdehyde Dehydrogenase Deficiency. Pediatric Neurology, 2010, 42, 255-258.	2.1	27
93	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
94	Ageâ€related phenotype and biomarker changes in <scp>SSADH</scp> deficiency. Annals of Clinical and Translational Neurology, 2019, 6, 114-120.	3.7	27
95	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
96	Liver-Directed Adenoviral Gene Transfer in Murine Succinate Semialdehyde Dehydrogenase Deficiency. Molecular Therapy, 2004, 9, 527-539.	8.2	25
97	Expression profiling reveals multiple myelin alterations in murine succinate semialdehyde dehydrogenase deficiency. Journal of Inherited Metabolic Disease, 2006, 29, 143-156.	3.6	25
98	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
99	Succinic semialdehyde dehydrogenase deficiency associated with combined 4-hydroxybutyric and dicarboxylic aciduria: Potential for clinical misdiagnosis based on urinary organic acid profiling. Journal of Pediatrics, 1989, 114, 607-610.	1.8	23
100	3-Hydroxy-3-methylglutaryl-CoA lyase deficiency as detected by radiochemical assay in cell extracts by thin-layer chromatography, and identification of three new cases. Clinical Chemistry, 1990, 36, 297-303.	3.2	23
101	Torin 1 partially corrects vigabatrinâ€induced mitochondrial increase in mouse. Annals of Clinical and Translational Neurology, 2015, 2, 699-706.	3.7	23
102	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
103	MRI Findings in Succinic Semialdehyde Dehydrogenase Deficiency. Neuropediatrics, 2000, 31, 45-46.	0.6	22
104	Delayed-onset dystonia associated with 3-oxothiolase deficiency. Movement Disorders, 2001, 16, 372-375.	3.9	22
105	Incidence and Geographic Distribution of Succinic Semialdehyde Dehydrogenase (SSADH) Deficiency. JIMD Reports, 2016, 34, 111-115.	1.5	22
106	Pre- and postnatal diagnosis of succinic semialdehyde dehydrogenase deficiency using enzyme and metabolite assays. Journal of Inherited Metabolic Disease, 1994, 17, 732-737.	3.6	20
107	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
108	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

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109	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
110	Variable clinical presentation in three patients with 3-methylglutaconyl-coenzyme A hydratase deficiency. Journal of Inherited Metabolic Disease, 1998, 21, 631-638.	3.6	19
111	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
112	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
113	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
114	Further evaluation of Vigabatrin therapy in 4-hydroxybutyric aciduria. European Journal of Pediatrics, 1992, 151, 466-466.	2.7	17
115	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
116	Nonâ€physiological amino acid (NPAA) therapy targeting brain phenylalanine reduction: pilot studies in PAH^{ENU2} mice. Journal of Inherited Metabolic Disease, 2013, 36, 513-523.	3.6	17
117	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
118	Biomarkers in a Taurine Trial for Succinic Semialdehyde Dehydrogenase Deficiency. JIMD Reports, 2015, 30, 81-87.	1.5	17
119	Plasticity of postsynaptic, but not presynaptic, GABAB receptors inSSADH deficient mice. Experimental Neurology, 2010, 225, 114-122.	4.1	16
120	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
121	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
122	4-Hydroxybutyric aciduria in a patient without ataxia or convulsions. European Journal of Pediatrics, 1988, 147, 529-531.	2.7	15
123	Metabolism of [U-14C]-4-hydroxybutyric acid to intermediates of the tricarboxylic acid cycle in extracts of rat liver and kidney mitochondria. European Journal of Drug Metabolism and Pharmacokinetics, 1989, 14, 61-70.	1.6	15
124	Therapeutic Efficacy of Magnesium Valproate in Succinic Semialdehyde Dehydrogenase Deficiency. JIMD Reports, 2012, 8, 133-137.	1.5	15
125	Succinic Semialdehyde Dehydrogenase Deficiency: Review of the Natural History Study. Journal of Child Neurology, 2021, 36, 1153-1161.	1.4	15
126	Mevalonic aciduria: Family studies in mevalonate kinase deficiency, an inborn error of cholesterol biosynthesis. Journal of Inherited Metabolic Disease, 1987, 10, 282-285.	3.6	14

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127	Enzymatic and immunological demonstration of normal and defective succinic semialdehyde dehydrogenase activity in fetal brain, liver and kidney. Journal of Inherited Metabolic Disease, 1993, 16, 523-526.	3.6	14
128	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
129	Emotional experience in parents of children with Zellweger spectrum disorders: A qualitative study. Molecular Genetics and Metabolism Reports, 2019, 19, 100459.	1.1	14
130	Novel ALDH5A1 variants and genotype. Neurology, 2020, 95, e2675-e2682.	1.1	14
131	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
132	Efficacy of Vigabatrin Intervention in a Mild Phenotypic Expression of Succinic Semialdehyde Dehydrogenase Deficiency. JIMD Reports, 2011, 2, 119-123.	1.5	13
133	Targeted fluorescent probes for detection of oxidative stress in the mitochondria. Bioorganic and Medicinal Chemistry Letters, 2015, 25, 3476-3480.	2.2	13
134	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
135	Farnesol induces protection against murine CNS inflammatory demyelination and modifies gut microbiome. Clinical Immunology, 2022, 235, 108766.	3.2	13
136	Pharmacological protection of mitochondrial function mitigates acute limb ischemia/reperfusion injury. Bioorganic and Medicinal Chemistry Letters, 2016, 26, 4042-4051.	2.2	12
137	Anti-inflammatory, analgesic and antioxidant activities of novel kyotorphin-nitroxide hybrid molecules. Bioorganic and Medicinal Chemistry Letters, 2016, 26, 2005-2013.	2.2	12
138	3-Hydroxy-3-methylglutaryl-CoA lyase deficiency as detected by radiochemical assay in cell extracts by thin-layer chromatography, and identification of three new cases. Clinical Chemistry, 1990, 36, 297-303.	3.2	12
139	Cerebellar Atrophy in Human and Murine Succinic Semialdehyde Dehydrogenase Deficiency. Journal of Child Neurology, 2010, 25, 1457-1461.	1.4	11
140	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
141	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
142	Vigabatrin-Induced Retinal Functional Alterations and Second-Order Neuron Plasticity in C57BL/6J Mice. , 2020, 61, 17.		11
143	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
144	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

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145	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
146	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
147	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
148	Indole Alkaloid Derivative B, a Novel Bifunctional Agent That Mitigates 5-Fluorouracil-Induced Cardiotoxicity. ACS Omega, 2018, 3, 15850-15864.	3.5	10
149	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
150	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
151	Phenotypically mild presentation in a patient with 2-methylacetoacetyl-coenzyme A (Î²-keto)thiolase deficiency. Journal of Inherited Metabolic Disease, 1997, 20, 712-713.	3.6	9
152	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
153	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
154	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
155	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
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