Sylvia Stockler-Ipsiroglu

List of Publications by Year
in descending order

70 papers	2,469 citations	331259 21 h-index	223531 46 g-index
71 all docs	71 docs citations	71 times ranked	3653 citing authors

#	Article	IF	Citations
1	Family Experiences with Care for Children with Inherited Metabolic Diseases in Canada: A Cross-Sectional Survey. Patient, 2022, 15, 171-185.	1.1	1
2	Determining ideal balance among branched-chain amino acids in medical formula for Propionic Acidemia: A proof of concept study in healthy children. Molecular Genetics and Metabolism, 2022, 135, 56-62.	0.5	1
3	Familiesâ€™ healthcare experiences for children with inherited metabolic diseases: protocol for a mixed methods cohort study. BMJ Open, 2022, 12, e055664.	0.8	0
4	Sleep as an outcome measure in ADHD randomized controlled trials: A scoping review. Sleep Medicine Reviews, 2022, 63, 101613.	3.8	10
5	Development of minimally invasive 13C-glucose breath test to examine different exogenous carbohydrate sources in patients with glycogen storage disease type Ia. Molecular Genetics and Metabolism Reports, 2022, 31, 100880.	0.4	2
6	JARID2 haploinsufficiency is associated with a clinically distinct neurodevelopmental syndrome. Genetics in Medicine, 2021, 23, 374-383.	1.1	13
7	Consensus guidelines for the diagnosis and management of pyridoxineâ€dependent epilepsy due to Î±â€aminoadipic semialdehyde dehydrogenase deficiency. Journal of Inherited Metabolic Disease, 2021, 44, 178-192.	1.7	47
8	De novo stop-loss variants in CLDN11 cause hypomyelinating leukodystrophy. Brain, 2021, 144, 411-419.	3.7	12
9	Developments in evidence creation for treatments of inborn errors of metabolism. Journal of Inherited Metabolic Disease, 2021, 44, 88-98.	1.7	13
10	Idiopathic splenomegaly in childhood and the spectrum of RAS-associated lymphoproliferative disease: a case report. BMC Pediatrics, 2021, 21, 45.	0.7	1
11	Morquioâ€like dysostosis multiplex presenting with neuronopathic features is a distinct <scp>GLB1</scp>â€related phenotype. JIMD Reports, 2021, 60, 23-31.	0.7	4
12	Treatable inherited metabolic disorders causing intellectual disability: 2021 review and digital app. Orphanet Journal of Rare Diseases, 2021, 16, 170.	1.2	52
13	Dietary management and growth outcomes in children with propionic acidemia: A natural history study. JIMD Reports, 2021, 61, 67-75.	0.7	1
14	Patient and family engagement in the development of core outcome sets for two rare chronic diseases in children. Research Involvement and Engagement, 2021, 7, 66.	1.1	11
15	Diagnostic yield from routine metabolic screening tests in evaluation of global developmental delay and intellectual disability. Paediatrics and Child Health, 2021, 26, 344-348.	0.3	4
16	Establishing a core outcome set for mucopolysaccharidoses (MPS) in children: study protocol for a rapid literature review, candidate outcomes survey, and Delphi surveys. Trials, 2021, 22, 816.	0.7	3
17	Determining Ideal Balance Among Branched-Chain Amino Acids (BCAA) as a Proof of Concept Study in Healthy Children. Current Developments in Nutrition, 2020, 4, nzaa054_140.	0.1	0
18	Development of Minimally Invasive 13C-Glucose Breath Test to Examine Different Dietary Therapies in Patients with Glycogen Storage Disorders. Current Developments in Nutrition, 2020, 4, nzaa055_034.	0.1	0

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19	Morquio B Disease. Disease Characteristics and Treatment Options of a Distinct GLB1-Related Dysostosis Multiplex. International Journal of Molecular Sciences, 2020, 21, 9121.	1.8	13
20	Impact of enteral arginine supplementation on lysine metabolism in humans: A proofâ€ofâ€concept for lysineâ€related inborn errors of metabolism. Journal of Inherited Metabolic Disease, 2020, 43, 952-959.	1.7	11
21	Iron deficiency and sleep â€“ A scoping review. Sleep Medicine Reviews, 2020, 51, 101274.	3.8	44
22	Morquioâ€B disease: Clinical and genetic characteristics of a distinct GLB1â€related dysostosis multiplex. JIMD Reports, 2020, 51, 30-44.	0.7	16
23	Outcomes in pediatric studies of medium-chain acyl-coA dehydrogenase (MCAD) deficiency and phenylketonuria (PKU): a review. Orphanet Journal of Rare Diseases, 2020, 15, 12.	1.2	15
24	Evaluation of the quality of clinical data collection for a pan-Canadian cohort of children affected by inherited metabolic diseases: lessons learned from the Canadian Inherited Metabolic Diseases Research Network. Orphanet Journal of Rare Diseases, 2020, 15, 89.	1.2	11
25	Haploinsufficiency of the Notch Ligand DLL1 Causes Variable Neurodevelopmental Disorders. American Journal of Human Genetics, 2019, 105, 631-639.	2.6	42
26	Congenital lactic acidosis, cerebral cysts and pulmonary hypertension in an infant with FOXRED1 related complex 1 deficiency. Molecular Genetics and Metabolism Reports, 2019, 19, 100472.	0.4	1
27	Congenital lactic acidosis, cerebral cysts and pulmonary hypertension in an infant with FOXRED1 related complex I deficiency. Molecular Genetics and Metabolism Reports, 2019, 18, 32-38.	0.4	8
28	Prenatal alcohol exposure and sleep-wake behaviors: exploratory and naturalistic observations in the clinical setting and in an animal model. Sleep Medicine, 2019, 54, 101-112.	0.8	22
29	Nutritional management of phenylalanine hydroxylase (PAH) deficiency in pediatric patients in Canada: a survey of dietitiansâ€™ current practices. Orphanet Journal of Rare Diseases, 2019, 14, 7.	1.2	8
30	Secondary Abnormal CSF Neurotransmitter Metabolite Profiles in a Pediatric Tertiary Care Centre. Canadian Journal of Neurological Sciences, 2018, 45, 206-213.	0.3	5
31	Prolonged granulocyte colony stimulating factor use in glycogen storage disease type 1b associated with acute myeloid leukemia and with shortened telomere length. Pediatric Hematology and Oncology, 2018, 35, 45-51.	0.3	31
32	Challenging sleep-wake behaviors reported in informal, conversational interviews of caregivers of children with fetal alcohol spectrum disorder. International Journal of Developmental Disabilities, 2018, 64, 65-74.	1.3	7
33	Pyruvate Carboxylase Deficiency Type C: A Rare Cause of Acute Transient Flaccid Paralysis with Ketoacidosis. Neuropediatrics, 2018, 49, 369-372.	0.3	7
34	Using a meta-narrative literature review and focus groups with key stakeholders to identify perceived challenges and solutions for generating robust evidence on the effectiveness of treatments for rare diseases. Orphanet Journal of Rare Diseases, 2018, 13, 104.	1.2	16
35	Morquio B patient/caregiver survey: First insight into the natural course of a rare GLB1 related condition. Molecular Genetics and Metabolism Reports, 2018, 16, 57-63.	0.4	12
36	The Indicator Amino Acid Oxidation Method with the Use of L-[1-13C]Leucine Suggests a Higher than Currently Recommended Protein Requirement in Children with Phenylketonuria. Journal of Nutrition, 2017, 147, 211-217.	1.3	5

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37	High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. American Journal of Human Genetics, 2017, 101, 664-685.	2.6	337
38	ACTB Loss-of-Function Mutations Result in a Pleiotropic Developmental Disorder. American Journal of Human Genetics, 2017, 101, 1021-1033.	2.6	83
39	Establishing core outcome sets for phenylketonuria (PKU) and medium-chain Acyl-CoA dehydrogenase (MCAD) deficiency in children: study protocol for systematic reviews and Delphi surveys. Trials, 2017, 18, 603.	0.7	9
40	â€œEmplotted Narrativesâ€•and Structured â€œBehavioral Observationsâ€•Supporting the Diagnosis of Willisâ€Ekbom Disease/Restless Legs Syndrome in Children with Neurodevelopmental Conditions. CNS Neuroscience and Therapeutics, 2016, 22, 894-905.	1.9	18
41	Experiences of caregivers of children with inherited metabolic diseases: a qualitative study. Orphanet Journal of Rare Diseases, 2016, 11, 168.	1.2	38
42	A three-tier algorithm for guanidinoacetate methyltransferase (GAMT) deficiency newborn screening. Molecular Genetics and Metabolism, 2016, 118, 173-177.	0.5	19
43	Pyridoxine-Dependent Epilepsy: An Expanding Clinical Spectrum. Pediatric Neurology, 2016, 59, 6-12.	1.0	136
44	Secondary neurotransmitter deficiencies in epilepsy caused by voltage-gated sodium channelopathies: A potential treatment target?. Molecular Genetics and Metabolism, 2016, 117, 42-48.	0.5	40
45	ISDN2014_0424: Challenging/disruptive sleep/wake behaviours in adolescents with fetal alcohol spectrum disorders: Iatrogenic effects of prescription medications. International Journal of Developmental Neuroscience, 2015, 47, 128-128.	0.7	0
46	Treatment of Creatine Transporter (SLC6A8) Deficiency With Oral S-Adenosyl Methionine as Adjunct to L-arginine, Glycine, and Creatine Supplements. Pediatric Neurology, 2015, 53, 360-363.e2.	1.0	20
47	Arginine:glycine amidinotransferase (AGAT) deficiency: Clinical features and long term outcomes in 16 patients diagnosed worldwide. Molecular Genetics and Metabolism, 2015, 116, 252-259.	0.5	55
48	Individualized long-term outcomes in blood phenylalanine concentrations and dietary phenylalanine tolerance in 11 patients with primary phenylalanine hydroxylase (PAH) deficiency treated with Sapropterin-dihydrochloride. Molecular Genetics and Metabolism, 2015, 114, 409-414.	0.5	5
49	Scoping review of patient- and family-oriented outcomes and measures for chronic pediatric disease. BMC Pediatrics, 2015, 15, 7.	0.7	20
50	Ã¢â‚¬Å“Diagnosis by Behavioral ObservationÃ¢â‚¬Â•Home-Videosomnography Ã¢â‚¬â€œ A Rigorous Ethnographic Approach to Sleep of Children with Neurodevelopmental Conditions. Frontiers in Psychiatry, 2015, 6, 39.	1.3	26
51	Minimally invasive 13C-breath test to examine phenylalanine metabolism in children with phenylketonuria. Molecular Genetics and Metabolism, 2015, 115, 78-83.	0.5	8
52	Retrospective analysis supports algorithm as efficient diagnostic approach to treatable intellectual developmental disabilities. Molecular Genetics and Metabolism, 2015, 115, 1-9.	0.5	13
53	Health economic evaluation of plasma oxysterol screening in the diagnosis of Niemannâ€“Pick Type C disease among intellectually disabled using discrete event simulation. Molecular Genetics and Metabolism, 2015, 114, 226-232.	0.5	11
54	ISDN2014_0220: Personalized Evaluation Model for making informed decisions in treatments for individuals with intellectual disability. International Journal of Developmental Neuroscience, 2015, 47, 66-66.	0.7	2

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55	Long-term developmental progression in infants and young children taking sapropterin for phenylketonuria: a two-year analysis of safety and efficacy. Genetics in Medicine, 2015, 17, 365-373.	1.1	18
56	Protein Requirements in Children with Phenylketonuria (PKU). FASEB Journal, 2015, 29, 742.9.	0.2	1
57	Early identification of treatable inborn errors of metabolism in children with intellectual disability: The Treatable Intellectual Disability Endeavor protocol in British Columbia. Paediatrics and Child Health, 2014, 19, 469-471.	0.3	40
58	Cerebral Creatine Deficiencies: A Group of Treatable Intellectual Developmental Disorders. Seminars in Neurology, 2014, 34, 350-356.	0.5	48
59	Granulomatous Herpes Simplex Encephalitis in an Infant With Multicystic Encephalopathy: A Distinct Clinicopathologic Entity?. Pediatric Neurology, 2014, 50, 392-396.	1.0	10
60	Overexpression of recombinant human antiquitin in E. coli: Partial enzyme activity in selected ALDH7A1 missense mutations associated with pyridoxine-dependent epilepsy. Molecular Genetics and Metabolism, 2014, 111, 462-466.	0.5	12
61	The metabolic evaluation of the child with an intellectual developmental disorder: Diagnostic algorithm for identification of treatable causes and new digital resource. Molecular Genetics and Metabolism, 2014, 111, 428-438.	0.5	110
62	Mitochondrial Carbonic Anhydrase VA Deficiency Resulting from CA5A Alterations Presents with Hyperammonemia in Early Childhood. American Journal of Human Genetics, 2014, 94, 453-461.	2.6	82
63	Guanidinoacetate methyltransferase (GAMT) deficiency: Outcomes in 48 individuals and recommendations for diagnosis, treatment and monitoring. Molecular Genetics and Metabolism, 2014, 111, 16-25.	0.5	111
64	Treatment of X-linked creatine transporter (SLC6A8) deficiency: systematic review of the literature and three new cases. Molecular Genetics and Metabolism, 2014, 112, 259-274.	0.5	54
65	Cultural aspects in the management of inborn errors of metabolism. Journal of Inherited Metabolic Disease, 2012, 35, 1147-1152.	1.7	22
66	Lysine restricted diet for pyridoxine-dependent epilepsy: First evidence and future trials. Molecular Genetics and Metabolism, 2012, 107, 335-344.	0.5	97
67	Treatable inborn errors of metabolism causing intellectual disability: A systematic literature review. Molecular Genetics and Metabolism, 2012, 105, 368-381.	0.5	172
68	Pyridoxine dependent epilepsy and antiquitin deficiency. Molecular Genetics and Metabolism, 2011, 104, 48-60.	0.5	258
69	Metabolic epilepsies: approaches to a diagnostic challenge. Canadian Journal of Neurological Sciences, 2009, 36 Suppl 2, S67-72.	0.3	0
70	Cerebral Creatine Deficiency Syndromes: Clinical Aspects, Treatment and Pathophysiology. , 2007, 46, 149-166.		145