

Sylvia Stockler-Ipsiroglu

List of Publications by Year in descending order

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Version: 2024-02-01

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	High Rate of Recurrent De Novo Mutations in Developmental and Epileptic Encephalopathies. American Journal of Human Genetics, 2017, 101, 664-685.	2.6	337
2	Pyridoxine dependent epilepsy and antiquitin deficiency. Molecular Genetics and Metabolism, 2011, 104, 48-60.	0.5	258
3	Treatable inborn errors of metabolism causing intellectual disability: A systematic literature review. Molecular Genetics and Metabolism, 2012, 105, 368-381.	0.5	172
4	Cerebral Creatine Deficiency Syndromes: Clinical Aspects, Treatment and Pathophysiology. , 2007, 46, 149-166.		145
5	Pyridoxine-Dependent Epilepsy: An Expanding Clinical Spectrum. Pediatric Neurology, 2016, 59, 6-12.	1.0	136
6	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
7	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
8	Lysine restricted diet for pyridoxine-dependent epilepsy: First evidence and future trials. Molecular Genetics and Metabolism, 2012, 107, 335-344.	0.5	97
9	ACTB Loss-of-Function Mutations Result in a Pleiotropic Developmental Disorder. American Journal of Human Genetics, 2017, 101, 1021-1033.	2.6	83
10	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
11	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
12	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
13	Treatable inherited metabolic disorders causing intellectual disability: 2021 review and digital app. Orphanet Journal of Rare Diseases, 2021, 16, 170.	1.2	52
14	Cerebral Creatine Deficiencies: A Group of Treatable Intellectual Developmental Disorders. Seminars in Neurology, 2014, 34, 350-356.	0.5	48
15	Consensus guidelines for the diagnosis and management of pyridoxine-dependent epilepsy due to Î±-amino adipic semialdehyde dehydrogenase deficiency. Journal of Inherited Metabolic Disease, 2021, 44, 178-192.	1.7	47
16	Iron deficiency and sleep " A scoping review. Sleep Medicine Reviews, 2020, 51, 101274.	3.8	44
17	Haploinsufficiency of the Notch Ligand DLL1 Causes Variable Neurodevelopmental Disorders. American Journal of Human Genetics, 2019, 105, 631-639.	2.6	42
18	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

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19	Secondary neurotransmitter deficiencies in epilepsy caused by voltage-gated sodium channelopathies: A potential treatment target?. <i>Molecular Genetics and Metabolism</i> , 2016, 117, 42-48.	0.5	40
20	Experiences of caregivers of children with inherited metabolic diseases: a qualitative study. <i>Orphanet Journal of Rare Diseases</i> , 2016, 11, 168.	1.2	38
21	Prolonged granulocyte colony stimulating factor use in glycogen storage disease type 1b associated with acute myeloid leukemia and with shortened telomere length. <i>Pediatric Hematology and Oncology</i> , 2018, 35, 45-51.	0.3	31
22	“Diagnosis by Behavioral Observation”-Home-Videosomnography - A Rigorous Ethnographic Approach to Sleep of Children with Neurodevelopmental Conditions. <i>Frontiers in Psychiatry</i> , 2015, 6, 39.	1.3	26
23	Cultural aspects in the management of inborn errors of metabolism. <i>Journal of Inherited Metabolic Disease</i> , 2012, 35, 1147-1152.	1.7	22
24	Prenatal alcohol exposure and sleep-wake behaviors: exploratory and naturalistic observations in the clinical setting and in an animal model. <i>Sleep Medicine</i> , 2019, 54, 101-112.	0.8	22
25	Treatment of Creatine Transporter (SLC6A8) Deficiency With Oral S-Adenosyl Methionine as Adjunct to L-arginine, Glycine, and Creatine Supplements. <i>Pediatric Neurology</i> , 2015, 53, 360-363.e2.	1.0	20
26	Scoping review of patient- and family-oriented outcomes and measures for chronic pediatric disease. <i>BMC Pediatrics</i> , 2015, 15, 7.	0.7	20
27	A three-tier algorithm for guanidinoacetate methyltransferase (GAMT) deficiency newborn screening. <i>Molecular Genetics and Metabolism</i> , 2016, 118, 173-177.	0.5	19
28	Long-term developmental progression in infants and young children taking sapropterin for phenylketonuria: a two-year analysis of safety and efficacy. <i>Genetics in Medicine</i> , 2015, 17, 365-373.	1.1	18
29	“Emplotted Narratives” and Structured “Behavioral Observations” Supporting the Diagnosis of Willis-Ekbom Disease/Restless Legs Syndrome in Children with Neurodevelopmental Conditions. <i>CNS Neuroscience and Therapeutics</i> , 2016, 22, 894-905.	1.9	18
30	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. <i>Orphanet Journal of Rare Diseases</i> , 2018, 13, 104.	1.2	16
31	Morquio B disease: Clinical and genetic characteristics of a distinct <i>GLB1</i>-related dysostosis multiplex. <i>JIMD Reports</i> , 2020, 51, 30-44.	0.7	16
32	Outcomes in pediatric studies of medium-chain acyl-coA dehydrogenase (MCAD) deficiency and phenylketonuria (PKU): a review. <i>Orphanet Journal of Rare Diseases</i> , 2020, 15, 12.	1.2	15
33	Retrospective analysis supports algorithm as efficient diagnostic approach to treatable intellectual developmental disabilities. <i>Molecular Genetics and Metabolism</i> , 2015, 115, 1-9.	0.5	13
34	Morquio B Disease. Disease Characteristics and Treatment Options of a Distinct GLB1-Related Dysostosis Multiplex. <i>International Journal of Molecular Sciences</i> , 2020, 21, 9121.	1.8	13
35	JARID2 haploinsufficiency is associated with a clinically distinct neurodevelopmental syndrome. <i>Genetics in Medicine</i> , 2021, 23, 374-383.	1.1	13
36	Developments in evidence creation for treatments of inborn errors of metabolism. <i>Journal of Inherited Metabolic Disease</i> , 2021, 44, 88-98.	1.7	13

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37	Overexpression of recombinant human antiquitin in <i>E. coli</i> : Partial enzyme activity in selected ALDH7A1 missense mutations associated with pyridoxine-dependent epilepsy. <i>Molecular Genetics and Metabolism</i> , 2014, 111, 462-466.	0.5	12
38	Morquio B patient/caregiver survey: First insight into the natural course of a rare GLB1 related condition. <i>Molecular Genetics and Metabolism Reports</i> , 2018, 16, 57-63.	0.4	12
39	<i>De novo</i> stop-loss variants in <i>CLDN11</i> cause hypomyelinating leukodystrophy. <i>Brain</i> , 2021, 144, 411-419.	3.7	12
40	Health economic evaluation of plasma oxysterol screening in the diagnosis of Niemann-Pick Type C disease among intellectually disabled using discrete event simulation. <i>Molecular Genetics and Metabolism</i> , 2015, 114, 226-232.	0.5	11
41	Impact of enteral arginine supplementation on lysine metabolism in humans: A proof-of-concept for lysine-related inborn errors of metabolism. <i>Journal of Inherited Metabolic Disease</i> , 2020, 43, 952-959.	1.7	11
42	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. <i>Orphanet Journal of Rare Diseases</i> , 2020, 15, 89.	1.2	11
43	Patient and family engagement in the development of core outcome sets for two rare chronic diseases in children. <i>Research Involvement and Engagement</i> , 2021, 7, 66.	1.1	11
44	Granulomatous Herpes Simplex Encephalitis in an Infant With Multicystic Encephalopathy: A Distinct Clinicopathologic Entity?. <i>Pediatric Neurology</i> , 2014, 50, 392-396.	1.0	10
45	Sleep as an outcome measure in ADHD randomized controlled trials: A scoping review. <i>Sleep Medicine Reviews</i> , 2022, 63, 101613.	3.8	10
46	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. <i>Trials</i> , 2017, 18, 603.	0.7	9
47	Minimally invasive ¹³ C-breath test to examine phenylalanine metabolism in children with phenylketonuria. <i>Molecular Genetics and Metabolism</i> , 2015, 115, 78-83.	0.5	8
48	Congenital lactic acidosis, cerebral cysts and pulmonary hypertension in an infant with FOXRED1 related complex I deficiency. <i>Molecular Genetics and Metabolism Reports</i> , 2019, 18, 32-38.	0.4	8
49	Nutritional management of phenylalanine hydroxylase (PAH) deficiency in pediatric patients in Canada: a survey of dietitians' current practices. <i>Orphanet Journal of Rare Diseases</i> , 2019, 14, 7.	1.2	8
50	Challenging sleep-wake behaviors reported in informal, conversational interviews of caregivers of children with fetal alcohol spectrum disorder. <i>International Journal of Developmental Disabilities</i> , 2018, 64, 65-74.	1.3	7
51	Pyruvate Carboxylase Deficiency Type C: A Rare Cause of Acute Transient Flaccid Paralysis with Ketoacidosis. <i>Neuropediatrics</i> , 2018, 49, 369-372.	0.3	7
52	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. <i>Molecular Genetics and Metabolism</i> , 2015, 114, 409-414.	0.5	5
53	The Indicator Amino Acid Oxidation Method with the Use of L-[1- ¹³ C]Leucine Suggests a Higher than Currently Recommended Protein Requirement in Children with Phenylketonuria. <i>Journal of Nutrition</i> , 2017, 147, 211-217.	1.3	5
54	Secondary Abnormal CSF Neurotransmitter Metabolite Profiles in a Pediatric Tertiary Care Centre. <i>Canadian Journal of Neurological Sciences</i> , 2018, 45, 206-213.	0.3	5

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55	Morquio type dysostosis multiplex presenting with neuronopathic features is a distinct <sc><i>GLB1</i></sc>-related phenotype. <i>JIMD Reports</i> , 2021, 60, 23-31.	0.7	4
56	Diagnostic yield from routine metabolic screening tests in evaluation of global developmental delay and intellectual disability. <i>Paediatrics and Child Health</i> , 2021, 26, 344-348.	0.3	4
57	Establishing a core outcome set for mucopolysaccharidoses (MPS) in children: study protocol for a rapid literature review, candidate outcomes survey, and Delphi surveys. <i>Trials</i> , 2021, 22, 816.	0.7	3
58	ISDN2014_0220: Personalized Evaluation Model for making informed decisions in treatments for individuals with intellectual disability. <i>International Journal of Developmental Neuroscience</i> , 2015, 47, 66-66.	0.7	2
59	Development of minimally invasive ¹³ C-glucose breath test to examine different exogenous carbohydrate sources in patients with glycogen storage disease type Ia. <i>Molecular Genetics and Metabolism Reports</i> , 2022, 31, 100880.	0.4	2
60	Congenital lactic acidosis, cerebral cysts and pulmonary hypertension in an infant with FOXRED1 related complex 1 deficiency. <i>Molecular Genetics and Metabolism Reports</i> , 2019, 19, 100472.	0.4	1
61	Idiopathic splenomegaly in childhood and the spectrum of RAS-associated lymphoproliferative disease: a case report. <i>BMC Pediatrics</i> , 2021, 21, 45.	0.7	1
62	Dietary management and growth outcomes in children with propionic acidemia: A natural history study. <i>JIMD Reports</i> , 2021, 61, 67-75.	0.7	1
63	Family Experiences with Care for Children with Inherited Metabolic Diseases in Canada: A Cross-Sectional Survey. <i>Patient</i> , 2022, 15, 171-185.	1.1	1
64	Protein Requirements in Children with Phenylketonuria (PKU). <i>FASEB Journal</i> , 2015, 29, 742.9.	0.2	1
65	Determining ideal balance among branched-chain amino acids in medical formula for Propionic Acidemia: A proof of concept study in healthy children. <i>Molecular Genetics and Metabolism</i> , 2022, 135, 56-62.	0.5	1
66	ISDN2014_0424: Challenging/disruptive sleep/wake behaviours in adolescents with fetal alcohol spectrum disorders: iatrogenic effects of prescription medications. <i>International Journal of Developmental Neuroscience</i> , 2015, 47, 128-128.	0.7	0
67	Determining Ideal Balance Among Branched-Chain Amino Acids (BCAA) as a Proof of Concept Study in Healthy Children. <i>Current Developments in Nutrition</i> , 2020, 4, nzaa054_140.	0.1	0
68	Development of Minimally Invasive ¹³ C-Glucose Breath Test to Examine Different Dietary Therapies in Patients with Glycogen Storage Disorders. <i>Current Developments in Nutrition</i> , 2020, 4, nzaa055_034.	0.1	0
69	Metabolic epilepsies: approaches to a diagnostic challenge. <i>Canadian Journal of Neurological Sciences</i> , 2009, 36 Suppl 2, S67-72.	0.3	0
70	Families' healthcare experiences for children with inherited metabolic diseases: protocol for a mixed methods cohort study. <i>BMJ Open</i> , 2022, 12, e055664.	0.8	0