Sarah P Young

List of Publications by Year
in descending order

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36 papers	1,034 citations	471371 17 h-index	434063 31 g-index
36 all docs	36 docs citations	36 times ranked	1436 citing authors

#	Article	IF	Citations
1	MPS VI associated ocular phenotypes in an MPS VI murine model and the therapeutic effects of odiparcil treatment. Molecular Genetics and Metabolism, 2022, 135, 143-153.	0.5	7
2	Cerebrospinal fluid amino acids glycine, serine, and threonine in nonketotic hyperglycinemia. Journal of Inherited Metabolic Disease, 2022, 45, 734-747.	1.7	9
3	Laboratory analysis of acylcarnitines, 2020 update: a technical standard of the American College of Medical Genetics and Genomics (ACMG). Genetics in Medicine, 2021, 23, 249-258.	1.1	19
4	Diurnal variability of glucose tetrasaccharide (Glc 4) excretion in patients with glycogen storage disease type III. JIMD Reports, 2021, 58, 37-43.	0.7	4
5	A glutaminase isoform switch drives therapeutic resistance and disease progression of prostate cancer. Proceedings of the National Academy of Sciences of the United States of America, 2021, 118, .	3.3	34
6	Characterization of liver GSD IX Î³2 pathophysiology in a novel Phkg2/ mouse model. Molecular Genetics and Metabolism, 2021, 133, 269-276.	0.5	4
7	Multidimensional predictors of antidepressant responses: Integrating mitochondrial, genetic, metabolic and environmental factors with clinical outcomes. Neurobiology of Stress, 2021, 15, 100407.	1.9	9
8	A retrospective longitudinal study and comprehensive review of adult patients with glycogen storage disease type III. Molecular Genetics and Metabolism Reports, 2021, 29, 100821.	0.4	7
9	Higher dosing of alglucosidase alfa improves outcomes in children with Pompe disease: a clinical study and review of the literature. Genetics in Medicine, 2020, 22, 898-907.	1.1	40
10	Improved muscle function in a phase I/II clinical trial of albuterol in Pompe disease. Molecular Genetics and Metabolism, 2020, 129, 67-72.	0.5	13
11	Comparisons of Infant and Adult Mice Reveal Age Effects for Liver Depot Gene Therapy in Pompe Disease. Molecular Therapy - Methods and Clinical Development, 2020, 17, 133-142.	1.8	10
12	Fenofibrate rapidly decreases hepatic lipid and glycogen storage in neonatal mice with glycogen storage disease type Ia. Human Molecular Genetics, 2020, 29, 286-294.	1.4	16
13	Combined analysis of plasma or serum glucosylsphingosine and globotriaosylsphingosine by UPLC-MS/MS. Clinica Chimica Acta, 2020, 511, 132-137.	0.5	7
14	Response to Heiner-Fokkema et al.. Genetics in Medicine, 2020, 22, 1917-1918.	1.1	1
15	Urine glucose tetrasaccharide: A good biomarker for glycogenoses type II and III? A study of the French cohort. Molecular Genetics and Metabolism Reports, 2020, 23, 100583.	0.4	17
16	A comprehensive testing algorithm for the diagnosis of Fabry disease in males and females. Molecular Genetics and Metabolism, 2020, 130, 209-214.	0.5	26
17	Comparison of dermatan sulfate and heparan sulfate concentrations in serum, cerebrospinal fluid and urine in patients with mucopolysaccharidosis type I receiving intravenous and intrathecal enzyme replacement therapy. Clinica Chimica Acta, 2020, 508, 179-184.	0.5	6
18	Evaluation of X-Linked Adrenoleukodystrophy Newborn Screening in North Carolina. JAMA Network Open, 2020, 3, e1920356.	2.8	44

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19	Liver fibrosis during clinical ascertainment of glycogen storage disease type III: a need for improved and systematic monitoring. Genetics in Medicine, 2019, 21, 2686-2694.	1.1	28
20	The North Carolina Experience with Mucopolysaccharidosis Type I Newborn Screening. Journal of Pediatrics, 2019, 211, 193-200.e2.	0.9	22
21	Bezafibrate Enhances AAV Vector-Mediated Genome Editing in Glycogen Storage Disease Type Ia. Molecular Therapy - Methods and Clinical Development, 2019, 13, 265-273.	1.8	8
22	Bezafibrate induces autophagy and improves hepatic lipid metabolism in glycogen storage disease type Ia. Human Molecular Genetics, 2019, 28, 143-154.	1.4	43
23	Laboratory analysis of amino acids, 2018 revision: a technical standard of the American College of Medical Genetics and Genomics (ACMG). Genetics in Medicine, 2018, 20, 1499-1507.	1.1	17
24	Acetyl- <scp>l</scp> -carnitine deficiency in patients with major depressive disorder. Proceedings of the National Academy of Sciences of the United States of America, 2018, 115, 8627-8632.	3.3	102
25	Laboratory diagnosis of creatine deficiency syndromes: a technical standard and guideline of the American College of Medical Genetics and Genomics. Genetics in Medicine, 2017, 19, 256-263.	1.1	21
26	Natural Progression of Canine Glycogen Storage Disease Type IIIa. Comparative Medicine, 2016, 66, 41-51.	0.4	13
27	Complex III deficiency due to an in-frame MT-CYB deletion presenting as ketotic hypoglycemia and lactic acidosis. Molecular Genetics and Metabolism Reports, 2015, 4, 39-41.	0.4	16
28	Mitochondrial NADP(H) deficiency due to a mutation in NADK2 causes dienoyl-CoA reductase deficiency with hyperlysinemia. Human Molecular Genetics, 2014, 23, 5009-5016.	1.4	63
29	Assessment of toxicity and biodistribution of recombinant AAV8 vectorâ€“mediated immunomodulatory gene therapy in mice with Pompe disease. Molecular Therapy - Methods and Clinical Development, 2014, 1, 14018.	1.8	37
30	Assessing disease severity in Pompe disease: The roles of a urinary glucose tetrasaccharide biomarker and imaging techniques. American Journal of Medical Genetics, Part C: Seminars in Medical Genetics, 2012, 160C, 50-58.	0.7	60
31	Long-term monitoring of patients with infantile-onset Pompe disease on enzyme replacement therapy using a urinary glucose tetrasaccharide biomarker. Genetics in Medicine, 2009, 11, 536-541.	1.1	79
32	Quantification of Creatine and Guanidinoacetate Using GCâ€MS and LCâ€MS/MS for the Detection of Cerebral Creatine Deficiency Syndromes. Current Protocols in Human Genetics, 2007, 54, Unit 17.3.	3.5	35
33	Glucose tetrasaccharide as a biomarker for monitoring the therapeutic response to enzyme replacement therapy for Pompe disease. Molecular Genetics and Metabolism, 2005, 85, 247-254.	0.5	79
34	Analysis of a glucose tetrasaccharide elevated in Pompe disease by stable isotope dilutionâ€“electrospray ionization tandem mass spectrometry. Analytical Biochemistry, 2003, 316, 175-180.	1.1	63
35	A comparison of in vitro acylcarnitine profiling methods for the diagnosis of classical and variant short chain acyl-CoA dehydrogenase deficiency. Clinica Chimica Acta, 2003, 337, 103-113.	0.5	15
36	Liquid Chromatographic Assay for a Glucose Tetrasaccharide, a Putative Biomarker for the Diagnosis of Pompe Disease. Analytical Biochemistry, 2000, 287, 136-143.	1.1	60