John J Mitchell

List of Publications by Year
in descending order

62 papers	3,048 citations	185998 28 h-index	161609 54 g-index
63 all docs	63 docs citations	63 times ranked	3941 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	Familiesâ€™ healthcare experiences for children with inherited metabolic diseases: protocol for a mixed methods cohort study. BMJ Open, 2022, 12, e055664.	0.8	0
3	Liver-Directed Adeno-Associated Virusâ€“Mediated Gene Therapy for Mucopolysaccharidosis Type VI. , 2022, 1, .		5
4	Core Outcome Sets for Medium-Chain Acyl-CoA Dehydrogenase Deficiency and Phenylketonuria. Pediatrics, 2021, 148, .	1.0	16
5	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
6	Clinical characteristics of patients from Quebec, Canada, with Morquio A syndrome: a longitudinal observational study. Orphanet Journal of Rare Diseases, 2020, 15, 270.	1.2	6
7	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
8	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
9	Muscle problems in juvenile-onset acid maltase deficiency (Pompe disease). Paediatrics and Child Health, 2019, 24, 270-271.	0.3	2
10	Recommendations for the management of MPS VI: systematic evidence- and consensus-based guidance. Orphanet Journal of Rare Diseases, 2019, 14, 118.	1.2	30
11	Recommendations for the management of MPS IVA: systematic evidence- and consensus-based guidance. Orphanet Journal of Rare Diseases, 2019, 14, 137.	1.2	62
12	Health services use among children diagnosed with medium-chain acyl-CoA dehydrogenase deficiency through newborn screening: a cohort study in Ontario, Canada. Orphanet Journal of Rare Diseases, 2019, 14, 70.	1.2	9
13	Hematopoietic stem cell transplant does not prevent neurological deterioration in infants with Farber disease: Case report and literature review. JIMD Reports, 2019, 46, 46-51.	0.7	1
14	Health-Related Quality of Life for Patients With Genetically Determined Leukoencephalopathy. Pediatric Neurology, 2018, 84, 21-26.	1.0	9
15	Impact of long-term elosulfase alfa on activities of daily living in patients with Morquio A syndrome in an open-label, multi-center, phase 3 extension study. Molecular Genetics and Metabolism, 2018, 123, 127-134.	0.5	25
16	Sleep and quality of life of patients with glycogen storage disease on standard and modified uncooked cornstarch. Molecular Genetics and Metabolism, 2018, 123, 326-330.	0.5	14
17	Online Module for Carrier Screening in Ashkenazi Jewish Individuals Compared with Inâ€Person Genetics Education: A Randomized Controlled Trial. Journal of Genetic Counseling, 2018, 27, 426-438.	0.9	9
18	Understanding the Early Presentation of Mucopolysaccharidoses Disorders. FIRE Forum for International Research in Education, 2018, 6, 232640981880034.	0.7	7

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19	Safety issues associated with dietary management in patients with hepatic glycogen storage disease. Molecular Genetics and Metabolism, 2018, 125, 79-85.	0.5	12
20	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
21	Hypersuccinylacetonaemia and normal liver function in maleylacetoacetate isomerase deficiency. Journal of Medical Genetics, 2017, 54, 241-247.	1.5	22
22	Acid Ceramidase Deficiency is characterized by a unique plasma cytokine and ceramide profile that is altered by therapy. Biochimica Et Biophysica Acta - Molecular Basis of Disease, 2017, 1863, 386-394.	1.8	35
23	Assessments of neurocognitive and behavioral function in the mucopolysaccharidoses. Molecular Genetics and Metabolism, 2017, 122, 8-16.	0.5	44
24	Pathophysiology, evaluation, and management of sleep disorders in the mucopolysaccharidoses. Molecular Genetics and Metabolism, 2017, 122, 49-54.	0.5	22
25	Morbidity and mortality associated with Farber disease and prospects for therapy. Expert Opinion on Orphan Drugs, 2017, 5, 717-726.	0.5	6
26	Cardiopulmonary Exercise Testing Reflects Improved Exercise Capacity in Response to Treatment in Morquio A Patients: Results of a 52-Week Pilot Study of Two Different Doses of Elosulfase Alfa. JIMD Reports, 2017, 42, 9-17.	0.7	4
27	Case report: maternal mosaicism resulting in inheritance of a novel GATA6 mutation causing pancreatic agenesis and neonatal diabetes mellitus. Diagnostic Pathology, 2017, 12, 1.	0.9	33
28	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
29	Impact of Elosulfase Alfa on Pain in Patients with Morquio A Syndrome over 52 Weeks. FIRE Forum for International Research in Education, 2017, 5, 232640981771885.	0.7	5
30	Detection and Quantification of Mosaic Mutations in Disease Genes by Next-Generation Sequencing. Journal of Molecular Diagnostics, 2016, 18, 446-453.	1.2	69
31	Impact of longâ€term elosulfase alfa treatment on respiratory function in patients with Morquio A syndrome. Journal of Inherited Metabolic Disease, 2016, 39, 839-847.	1.7	24
32	Long-term endurance and safety of elosulfase alfa enzyme replacement therapy in patients with Morquio A syndrome. Molecular Genetics and Metabolism, 2016, 119, 131-143.	0.5	47
33	Pregnancy in patients with mucopolysaccharidosis: a case series. Molecular Genetics and Metabolism Reports, 2016, 8, 111-115.	0.4	11
34	UPLC-MS/MS detection of disaccharides derived from glycosaminoglycans as biomarkers of mucopolysaccharidoses. Analytica Chimica Acta, 2016, 936, 139-148.	2.6	53
35	Unique medical issues in adult patients with mucopolysaccharidoses. European Journal of Internal Medicine, 2016, 34, 2-10.	1.0	28
36	Expert Opinions on Managing Fertility and Pregnancy in Patients With Mucopolysaccharidosis. FIRE Forum for International Research in Education, 2016, 4, 232640981666937.	0.7	1

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37	Experiences of caregivers of children with inherited metabolic diseases: a qualitative study. Orphanet Journal of Rare Diseases, 2016, 11, 168.	1.2	38
38	The health system impact of false positive newborn screening results for medium-chain acyl-CoA dehydrogenase deficiency: a cohort study. Orphanet Journal of Rare Diseases, 2016, 11, 12.	1.2	38
39	Child and family experiences with inborn errors of metabolism: a qualitative interview study with representatives of patient groups. Journal of Inherited Metabolic Disease, 2016, 39, 139-147.	1.7	26
40	Safety and physiological effects of two different doses of elosulfase alfa in patients with morquio a syndrome: A randomized, doubleâ€blind, pilot study. American Journal of Medical Genetics, Part A, 2015, 167, 2272-2281.	0.7	33
41	Longitudinal analysis of endurance and respiratory function from a natural history study of Morquio A syndrome. Molecular Genetics and Metabolism, 2015, 114, 186-194.	0.5	33
42	Multi-domain impact of elosulfase alfa in Morquio A syndrome in the pivotal phase III trial. Molecular Genetics and Metabolism, 2015, 114, 178-185.	0.5	65
43	Scoping review of patient- and family-oriented outcomes and measures for chronic pediatric disease. BMC Pediatrics, 2015, 15, 7.	0.7	20
44	Metabolic Clinic Atlas: Organization of Care for Children with Inherited Metabolic Disease in Canada. JIMD Reports, 2014, 21, 15-22.	0.7	3
45	Redefining normal bone and mineral clinical biochemistry reference intervals for healthy infants in Canada. Clinical Biochemistry, 2014, 47, 27-32.	0.8	17
46	Phenylalanine hydroxylase deficiency: diagnosis and management guideline. Genetics in Medicine, 2014, 16, 188-200.	1.1	486
47	The Morquio A Clinical Assessment Program: Baseline results illustrating progressive, multisystemic clinical impairments in Morquio A subjects. Molecular Genetics and Metabolism, 2013, 109, 54-61.	0.5	117
48	Achieving the â€œtriple aimâ€•for inborn errors of metabolism: a review of challenges to outcomes research and presentation of a new practice-based evidence framework. Genetics in Medicine, 2013, 15, 415-422.	1.1	29
49	The Prevalence of Hypoglycemia in Children With Vomiting or Decreased Oral Intake and Irritability. Pediatric Emergency Care, 2012, 28, 333-335.	0.5	3
50	Pregnancy of a patient with multiple Acyl-CoA dehydrogenation deficiency (MADD). Molecular Genetics and Metabolism, 2012, 106, 491-494.	0.5	11
51	Effect of nitisinone (NTBC) treatment on the clinical course of hepatorenal tyrosinemia in QuÃ©bec. Molecular Genetics and Metabolism, 2012, 107, 49-54.	0.5	132
52	Longitudinal observations of serum heparin cofactor II-thrombin complex in treated Mucopolysaccharidosis I and II patients. Journal of Inherited Metabolic Disease, 2012, 35, 355-362.	1.7	17
53	Phenylalanine hydroxylase deficiency. Genetics in Medicine, 2011, 13, 607-617.	1.1	240
54	Maternal riboflavin deficiency, resulting in transient neonatal-onset glutaric aciduria Type 2, is caused by a microdeletion in the riboflavin transporter gene GPR172B. Human Mutation, 2011, 32, E1976-E1984.	1.1	96

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55	Combined malonic and methylmalonic aciduria: exome sequencing reveals mutations in the ACSF3 gene in patients with a non-classic phenotype. Journal of Medical Genetics, 2011, 48, 602-605.	1.5	44
56	Rfx6 directs islet formation and insulin production in mice and humans. Nature, 2010, 463, 775-780.	13.7	300
57	CRTAPandLEPRE1mutations in recessive osteogenesis imperfecta. Human Mutation, 2008, 29, 1435-1442.	1.1	196
58	A 25-year longitudinal analysis of treatment efficacy in inborn errors of metabolism. Molecular Genetics and Metabolism, 2008, 95, 11-16.	0.5	33
59	Tetrahydrobiopterin-responsive phenylketonuria: The New South Wales experience. Molecular Genetics and Metabolism, 2005, 86, 81-85.	0.5	33
60	Neonatal diabetes, with hypoplastic pancreas, intestinal atresia and gall bladder hypoplasia: search for the aetiology of a new autosomal recessive syndrome. Diabetologia, 2004, 47, 2160-2167.	2.9	96
61	U-type exchange in a paracentric inversion as a possible mechanism of origin of an inverted tandem duplication of chromosome 8. American Journal of Medical Genetics Part A, 1994, 49, 384-387.	2.4	31
62	Familial cold urticaria. Clinical and Experimental Dermatology, 1993, 18, 338-341.	0.6	43