Can H FiÃ§icioÇ§lu

List of Publications by Year
in descending order

90 papers	4,226 citations	249298 26 h-index	134545 62 g-index
95 all docs	95 docs citations	95 times ranked	7018 citing authors

#	Article	IF	Citations
1	Perceptions and use of phenylbutyrate metabolite testing in urea cycle disorders: Results of a clinician survey and analysis of a centralized testing database. Molecular Genetics and Metabolism, 2022, 135, 35-41.	0.5	2
2	The diagnosis and management of Gaucher disease in pediatric patients: Where do we go from here?. Molecular Genetics and Metabolism, 2022, 136, 4-21.	0.5	18
3	Newborn Screening for X-Linked Adrenoleukodystrophy: Review of Data and Outcomes in Pennsylvania. International Journal of Neonatal Screening, 2022, 8, 24.	1.2	11
4	Epimutations in both the TESK2 and MMACHC promoters in the Epi-cblC inherited disorder of intracellular metabolism of vitamin B12. Clinical Epigenetics, 2022, 14, 52.	1.8	10
5	The current state of adult metabolic medicine in the United States: Results of a nationwide survey. Genetics in Medicine, 2022, 24, 1722-1731.	1.1	4
6	Lessons Learned From the Longâ€Term Use of Enzyme Replacement Therapy in the Treatment of Lysosomal Acid Lipase Deficiency. Journal of Pediatric Gastroenterology and Nutrition, 2022, 74, 726-727.	0.9	1
7	Galactokinase deficiency: lessons from the GalNet registry. Genetics in Medicine, 2021, 23, 202-210.	1.1	14
8	Transforming the clinical outcome in CRIM-negative infantile Pompe disease identified via newborn screening: the benefits of early treatment with enzyme replacement therapy and immune tolerance induction. Genetics in Medicine, 2021, 23, 845-855.	1.1	26
9	Provider Perspectives on the Impact of the COVID-19 Pandemic on Newborn Screening. International Journal of Neonatal Screening, 2021, 7, 38.	1.2	4
10	Should patients with Phosphomannomutase 2-CDG (PMM2-CDG) be screened for adrenal insufficiency?. Molecular Genetics and Metabolism, 2021, 133, 397-399.	0.5	3
11	The Editorâ€™s Choice for Issue 2, Volume 7. International Journal of Neonatal Screening, 2021, 7, 61.	1.2	1
12	Response to Neeleman et al.. Genetics in Medicine, 2020, 22, 439-440.	1.1	0
13	Serial Magnetic Resonance Imaging (MRI) in Pyruvate Dehydrogenase Complex Deficiency. Journal of Child Neurology, 2020, 35, 137-145.	0.7	7
14	Newborn Screening for Pompe Disease: Pennsylvania Experience. International Journal of Neonatal Screening, 2020, 6, 89.	1.2	24
15	Diagnostic journey and impact of enzyme replacement therapy for mucopolysaccharidosis IVA: a sibling control study. Orphanet Journal of Rare Diseases, 2020, 15, 336.	1.2	2
16	The Importance of Succinylacetone: Tyrosinemia Type I Presenting with Hyperinsulinism and Multiorgan Failure Following Normal Newborn Screening. International Journal of Neonatal Screening, 2020, 6, 39.	1.2	5
17	Persistent dyslipidemia in treatment of lysosomal acid lipase deficiency. Orphanet Journal of Rare Diseases, 2020, 15, 58.	1.2	8
18	Gaucher disease status and treatment assessment: pilot study using magnetic resonance spectroscopy bone marrow fat fractions in pediatric patients. Clinical Imaging, 2020, 63, 1-6.	0.8	7

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19	Person Ability Scores as an Alternative to Norm-Referenced Scores as Outcome Measures in Studies of Neurodevelopmental Disorders. American Journal on Intellectual and Developmental Disabilities, 2020, 125, 475-480.	0.8	30
20	Early diagnosis of infantile-onset lysosomal acid lipase deficiency in the advent of available enzyme replacement therapy. Orphanet Journal of Rare Diseases, 2019, 14, 198.	1.2	8
21	Imaging of non-neuronopathic Gaucher disease: recent advances in quantitative imaging and comprehensive assessment of disease involvement. Insights Into Imaging, 2019, 10, 70.	1.6	13
22	Failure to Thrive: An Expanded Differential Diagnosis. Journal of Pediatric Genetics, 2019, 08, 027-032.	0.3	4
23	Biomarkers of oxidative stress, inflammation, and vascular dysfunction in inherited cystathionine Î²â€synthase deficient homocystinuria and the impact of taurine treatment in a phase 1/2 human clinical trial. Journal of Inherited Metabolic Disease, 2019, 42, 424-437.	1.7	11
24	Increased Clinical Sensitivity and Specificity of Plasma Protein N-Glycan Profiling for Diagnosing Congenital Disorders of Glycosylation by Use of Flow Injectionâ€“Electrospray Ionizationâ€“Quadrupole Time-of-Flight Mass Spectrometry. Clinical Chemistry, 2019, 65, 653-663.	1.5	40
25	Phenotype, treatment practice and outcome in the cobalaminâ€dependent remethylation disorders and MTHFR deficiency: Data from the Eâ€HOD registry. Journal of Inherited Metabolic Disease, 2019, 42, 333-352.	1.7	53
26	Characteristics and outcomes of patients with formiminoglutamic aciduria detected through newborn screening. Journal of Inherited Metabolic Disease, 2019, 42, 140-146.	1.7	7
27	Early Indicators of Creatine Transporter Deficiency. Journal of Pediatrics, 2019, 206, 283-285.	0.9	10
28	TANGO2: expanding the clinical phenotype and spectrum of pathogenic variants. Genetics in Medicine, 2019, 21, 601-607.	1.1	41
29	Complex care of individuals with multiple sulfatase deficiency: Clinical cases and consensus statement. Molecular Genetics and Metabolism, 2018, 123, 337-346.	0.5	31
30	Treatment outcome of creatine transporter deficiency: international retrospective cohort study. Metabolic Brain Disease, 2018, 33, 875-884.	1.4	32
31	Intrafamilial variability in the clinical manifestations of mucopolysaccharidosis type II: Data from the Hunter Outcome Survey (HOS). American Journal of Medical Genetics, Part A, 2018, 176, 301-310.	0.7	15
32	A PRDX1 mutant allele causes a MMACHC secondary epimutation in cblC patients. Nature Communications, 2018, 9, 67.	5.8	64
33	Neuropsychological implications of Cobalamin C (CblC) disease in Hispanic children detected through newborn screening. Applied Neuropsychology: Child, 2018, 7, 143-149.	0.7	2
34	Conducting an investigator-initiated randomized double-blinded intervention trial in acute decompensation of inborn errors of metabolism: Lessons from the N-Carbamylglutamate Consortium. Translational Science of Rare Diseases, 2018, 3, 157-170.	1.6	7
35	Pharmacokinetics of glycerol phenylbutyrate in pediatric patients 2â€ months to 2â€ years of age with urea cycle disorders. Molecular Genetics and Metabolism, 2018, 125, 251-257.	0.5	7
36	Consensus guidelines for newborn screening, diagnosis and treatment of infantile Krabbe disease. Orphanet Journal of Rare Diseases, 2018, 13, 30.	1.2	67

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37	Efficacy of early treatment in patients with cobalamin C disease identified by newborn screening: a 16-year experience. Genetics in Medicine, 2017, 19, 926-935.	1.1	16
38	New tools and approaches to newborn screening: ready to open Pandora's box?. Journal of Physical Education and Sports Management, 2017, 3, a001842.	0.5	12
39	Diagnosis and treatment of tyrosinemia type I: a US and Canadian consensus group review and recommendations. Genetics in Medicine, 2017, 19, 1380-1395.	1.1	152
40	Safety and efficacy of glycerol phenylbutyrate for management of urea cycle disorders in patients aged 2 months to 2 years. Molecular Genetics and Metabolism, 2017, 122, 46-53.	0.5	16
41	Utility of Genetic Testing for Confirmation of Abnormal Newborn Screening in Disorders of Long-Chain Fatty Acids: A Missed Case of Carnitine Palmitoyltransferase 1A (CPT1A) Deficiency. International Journal of Neonatal Screening, 2017, 3, 10.	1.2	6
42	ALG1-CDG: Clinical and Molecular Characterization of 39 Unreported Patients. Human Mutation, 2016, 37, 653-660.	1.1	40
43	Morbidity and mortality among exclusively breastfed neonates with medium-chain acyl-CoA dehydrogenase deficiency. Genetics in Medicine, 2016, 18, 1315-1319.	1.1	11
44	Missed Newborn Screening Case of Carnitine Palmitoyltransferase-II Deficiency. JIMD Reports, 2016, 33, 93-97.	0.7	17
45	Low bone mineral density is a common finding in patients with homocystinuria. Molecular Genetics and Metabolism, 2016, 117, 351-354.	0.5	22
46	Argininosuccinic Acid Lyase Deficiency Missed by Newborn Screen. JIMD Reports, 2016, 34, 43-47.	0.7	4
47	Response to van Rijt et al.. Genetics in Medicine, 2016, 18, 1324.	1.1	0
48	Pathogenesis and treatment of spine disease in the mucopolysaccharidoses. Molecular Genetics and Metabolism, 2016, 118, 232-243.	0.5	28
49	ECHS1 Deficiency as a Cause of Severe Neonatal Lactic Acidosis. JIMD Reports, 2016, 30, 33-37.	0.7	26
50	Muddâ€™s disease (MAT I/III deficiency): a survey of data for MAT1A homozygotes and compound heterozygotes. Orphanet Journal of Rare Diseases, 2015, 10, 99.	1.2	39
51	An 8-Year-Old Girl With Abdominal Pain and Mental Status Changes. Pediatric Emergency Care, 2015, 31, 459-462.	0.5	3
52	Cobalamin C Deficiency Shows a Rapidly Progressing Maculopathy With Severe Photoreceptor and Ganglion Cell Loss. , 2015, 56, 7875.		30
53	Long-term safety and efficacy of sapropterin: The PKUDOS registry experience. Molecular Genetics and Metabolism, 2015, 114, 557-563.	0.5	39
54	Adolescent Presentations of Inborn Errors of Metabolism. Journal of Adolescent Health, 2015, 56, 477-482.	1.2	16

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55	Cobalamin C Disease Missed by Newborn Screening in a Patient with Low Carnitine Level. JIMD Reports, 2015, 23, 71-75.	0.7	12
56	Retinal Structure in Cobalamin C Disease: Mechanistic and Therapeutic Implications. Ophthalmic Genetics, 2015, 36, 339-348.	0.5	16
57	A Phase 3 Trial of Sebelipase Alfa in Lysosomal Acid Lipase Deficiency. New England Journal of Medicine, 2015, 373, 1010-1020.	13.9	212
58	Infant with cardiomyopathy: When to suspect inborn errors of metabolism?. World Journal of Cardiology, 2014, 6, 1149.	0.5	22
59	Multiple Phenotypes in Phosphoglucomutase 1 Deficiency. New England Journal of Medicine, 2014, 370, 533-542.	13.9	236
60	Severe 5,10-Methylenetetrahydrofolate Reductase Deficiency and Two MTHFR Variants in an Adolescent With Progressive Myoclonic Epilepsy. Pediatric Neurology, 2014, 51, 266-270.	1.0	21
61	Liver Pathology in Infantile Mitochondrial DNA Depletion Syndrome. Pediatric and Developmental Pathology, 2013, 16, 415-424.	0.5	11
62	A Pilot Study of Fluorodeoxyglucose Positron Emission Tomography Findings in Patients with Phenylketonuria before and during Sapropterin Supplementation. Journal of Clinical Neurology		

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73	Failure to Thrive: When to Suspect Inborn Errors of Metabolism. Pediatrics, 2009, 124, 972-979.	1.0	21
74	A Delphi clinical practice protocol for the management of very long chain acyl-CoA dehydrogenase deficiency. Molecular Genetics and Metabolism, 2009, 96, 85-90.	0.5	139
75	Very longâ€chain acylâ€CoA dehydrogenase deficiency: The effects of accidental fat loading in a patient detected through newborn screening. Journal of Inherited Metabolic Disease, 2009, 32, 187-190.	1.7	6
76	Effect of galactose free formula on galactose-1-phosphate in two infants with classical galactosemia. European Journal of Pediatrics, 2008, 167, 595-596.	1.3	9
77	Structural Variation of Chromosomes in Autism Spectrum Disorder. American Journal of Human Genetics, 2008, 82, 477-488.	2.6	1,641
78	Brain Magnetic Resonance Imaging Findings in 49,XXXXY Syndrome. Pediatric Neurology, 2008, 38, 450-453.	1.0	32
79	Complex management of a patient with a contiguous Xp11.4 gene deletion involving ornithine transcarbamylase: A role for detailed molecular analysis in complex presentations of classical diseases. Molecular Genetics and Metabolism, 2008, 94, 498-502.	0.5	25
80	Clinical outcomes of infants with short-chain acyl-coenzyme A dehydrogenase deficiency (SCADD) detected by newborn screening. Molecular Genetics and Metabolism, 2008, 95, 241-242.	0.5	17
81	Duarte (DG) galactosemia: A pilot study of biochemical and neurodevelopmental assessment in children detected by newborn screening. Molecular Genetics and Metabolism, 2008, 95, 206-212.	0.5	66
82	A False-positive Newborn Screening Result: Goat's Milk Acidopathy. Pediatrics, 2008, 122, 210-211.	1.0	5
83	Review of miglustat for clinical management in Gaucher disease type 1. Therapeutics and Clinical Risk Management, 2008, Volume 4, 425-431.	0.9	65
84	Development of a newborn screening follow-up algorithm for the diagnosis of isobutyryl-CoA dehydrogenase deficiency. Genetics in Medicine, 2007, 9, 108-116.	1.1	50
85	Epimerase-Deficiency Galactosemia Is Not a Binary Condition. American Journal of Human Genetics, 2006, 78, 89-102.	2.6	77
86	Liver transplantation is not curative for methylmalonic acidopathy caused by methylmalonyl-CoA mutase deficiency. Molecular Genetics and Metabolism, 2006, 88, 322-326.	0.5	72
87	3-Methylcrotonyl-CoA Carboxylase Deficiency: Metabolic Decompensation in a Noncompliant Child Detected Through Newborn Screening. Pediatrics, 2006, 118, 2555-2556.	1.0	24
88	Biotinidase deficiency: the importance of adequate follow-up for an inconclusive newborn screening result. European Journal of Pediatrics, 2005, 164, 298-301.	1.3	14
89	Galactitol and galactonate in red blood cells of children with the Duarte/galactosemia genotype. Molecular Genetics and Metabolism, 2005, 84, 152-159.	0.5	18
90	MRI and MRS in HMG-CoA lyase deficiency. Pediatric Neurology, 1999, 20, 375-380.	1.0	46