Can H Fiicioglu

List of Publications by Year in Descending Order

85 papers	3,277 citations	23 h-index	56 g-index
95 ext. papers	3,841 ext. citations	5.2 avg, IF	4.58 L-index

#	Paper	IF	Citations
85	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	3.7	0
84	Epimutations in both the TESK2 and MMACHC promoters in the Epi-cblC inherited disorder of intracellular metabolism of vitamin B.. *Clinical Epigenetics*, **2022**, 14, 52	7.7	1
83	Provider Perspectives on the Impact of the COVID-19 Pandemic on Newborn Screening. *International Journal of Neonatal Screening*, **2021**, 7,	2.6	2
82	Galactokinase deficiency: lessons from the GalNet registry. *Genetics in Medicine*, **2021**, 23, 202-210	8.1	7
81	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	8.1	3
80	Should patients with Phosphomannomutase 2-CDG (PMM2-CDG) be screened for adrenal insufficiency?. *Molecular Genetics and Metabolism*, **2021**, 133, 397-399	3.7	1
79	Diagnostic journey and impact of enzyme replacement therapy for mucopolysaccharidosis IVA: a sibling control study. *Orphanet Journal of Rare Diseases*, **2020**, 15, 336	4.2	1
78	The Importance of Succinylacetone: Tyrosinemia Type I Presenting with Hyperinsulinism and Multiorgan Failure Following Normal Newborn Screening. *International Journal of Neonatal Screening*, **2020**, 6,	2.6	3
77	Persistent dyslipidemia in treatment of lysosomal acid lipase deficiency. *Orphanet Journal of Rare Diseases*, **2020**, 15, 58	4.2	5
76	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	2.7	3
75	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	2.2	11
74	Serial Magnetic Resonance Imaging (MRI) in Pyruvate Dehydrogenase Complex Deficiency. *Journal of Child Neurology*, **2020**, 35, 137-145	2.5	3
73	Newborn Screening for Pompe Disease: Pennsylvania Experience. *International Journal of Neonatal Screening*, **2020**, 6,	2.6	6
72	Response to Neeleman et al. *Genetics in Medicine*, **2020**, 22, 439-440	8.1	
71	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	4.2	5
70	Imaging of non-neuronopathic Gaucher disease: recent advances in quantitative imaging and comprehensive assessment of disease involvement. *Insights Into Imaging*, **2019**, 10, 70	5.6	6
69	Failure to Thrive: An Expanded Differential Diagnosis. *Journal of Pediatric Genetics*, **2019**, 8, 27-32	0.7	2

68	Biomarkers of oxidative stress, inflammation, and vascular dysfunction in inherited cystathionine β-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	5.4	3
67	Increased Clinical Sensitivity and Specificity of Plasma Protein -Glycan Profiling for Diagnosing Congenital Disorders of Glycosylation by Use of Flow Injection-Electrospray Ionization-Quadrupole Time-of-Flight Mass Spectrometry. *Clinical Chemistry*, **2019**, 65, 653-663	5.5	18
66	Phenotype, treatment practice and outcome in the cobalamin-dependent remethylation disorders and MTHFR deficiency: Data from the E-HOD registry. *Journal of Inherited Metabolic Disease*, **2019**, 42, 333-352	5.4	28
65	Characteristics and outcomes of patients with formiminoglutamic aciduria detected through newborn screening. *Journal of Inherited Metabolic Disease*, **2019**, 42, 140-146	5.4	7
64	Early Indicators of Creatine Transporter Deficiency. *Journal of Pediatrics*, **2019**, 206, 283-285	3.6	3
63	TANGO2: expanding the clinical phenotype and spectrum of pathogenic variants. *Genetics in Medicine*, **2019**, 21, 601-607	8.1	18
62	Complex care of individuals with multiple sulfatase deficiency: Clinical cases and consensus statement. *Molecular Genetics and Metabolism*, **2018**, 123, 337-346	3.7	19
61	Treatment outcome of creatine transporter deficiency: international retrospective cohort study. *Metabolic Brain Disease*, **2018**, 33, 875-884	3.9	16
60	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	2.5	11
59	APRDX1 mutant allele causes a MMACHC secondary epimutation in cblC patients. *Nature Communications*, **2018**, 9, 67	17.4	45
58	Neuropsychological implications of Cobalamin C (CblC) disease in Hispanic children detected through newborn screening. *Applied Neuropsychology: Child*, **2018**, 7, 143-149	1.4	1
57	Consensus guidelines for newborn screening, diagnosis and treatment of infantile Krabbe disease. *Orphanet Journal of Rare Diseases*, **2018**, 13, 30	4.2	40
56	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	3.3	5
55	Pharmacokinetics of glycerol phenylbutyrate in pediatric patients 2 months to 2 years of age with urea cycle disorders. *Molecular Genetics and Metabolism*, **2018**, 125, 251-257	3.7	4
54	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	8.1	12
53	New tools and approaches to newborn screening: ready to open Pandora's box?. *Journal of Physical Education and Sports Management*, **2017**, 3, a001842	2.8	8
52	Diagnosis and treatment of tyrosinemia type I: a US and Canadian consensus group review and recommendations. *Genetics in Medicine*, **2017**, 19,	8.1	99
51	Safety and efficacy of glycerol phenylbutyrate for management of urea cycle disorders in patients aged 2months to 2years. *Molecular Genetics and Metabolism*, **2017**, 122, 46-53	3.7	11

50	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,	2.6	4
49	Missed Newborn Screening Case of Carnitine Palmitoyltransferase-II Deficiency. *JIMD Reports*, **2017** , 33, 93-97	1.9	12
48	Argininosuccinic Acid Lyase Deficiency Missed by Newborn Screen. *JIMD Reports*, **2017**, 34, 43-47	1.9	3
47	Response to van Rijt et al. *Genetics in Medicine*, **2016**, 18, 1324	8.1	
46	Pathogenesis and treatment of spine disease in the mucopolysaccharidoses. *Molecular Genetics and Metabolism*, **2016**, 118, 232-43	3.7	22
45	ECHS1 Deficiency as a Cause of Severe Neonatal Lactic Acidosis. *JIMD Reports*, **2016**, 30, 33-37	1.9	20
44	ALG1-CDG: Clinical and Molecular Characterization of 39 Unreported Patients. *Human Mutation*, **2016**, 37, 653-60	4.7	30
43	Morbidity and mortality among exclusively breastfed neonates with medium-chain acyl-CoA dehydrogenase deficiency. *Genetics in Medicine*, **2016**, 18, 1315-1319	8.1	8
42	Low bone mineral density is a common finding in patients with homocystinuria. *Molecular Genetics and Metabolism*, **2016**, 117, 351-4	3.7	17
41	Long-term safety and efficacy of sapropterin: the PKUDOS registry experience. *Molecular Genetics and Metabolism*, **2015**, 114, 557-63	3.7	32
40	Adolescent presentations of inborn errors of metabolism. *Journal of Adolescent Health*, **2015**, 56, 477-82	5.8	12
39	Cobalamin C Disease Missed by Newborn Screening in a Patient with Low Carnitine Level. *JIMD Reports*, **2015**, 23, 71-5	1.9	7
38	Retinal Structure in Cobalamin C Disease: Mechanistic and Therapeutic Implications. *Ophthalmic Genetics*, **2015**, 36, 339-48	1.2	13
37	A Phase 3 Trial of Sebelipase Alfa in Lysosomal Acid Lipase Deficiency. *New England Journal of Medicine*, **2015**, 373, 1010-20	59.2	168
36	Mudd's disease (MAT I/III deficiency): a survey of data for MAT1A homozygotes and compound heterozygotes. *Orphanet Journal of Rare Diseases*, **2015**, 10, 99	4.2	33
35	An 8-year-old girl with abdominal pain and mental status changes. *Pediatric Emergency Care*, **2015**, 31, 459-62	1.4	1
34	Cobalamin C Deficiency Shows a Rapidly Progressing Maculopathy With Severe Photoreceptor and Ganglion Cell Loss **2015**, 56, 7875-87		25
33	Multiple phenotypes in phosphoglucomutase 1 deficiency. *New England Journal of Medicine*, **2014**, 370, 533-42	59.2	197

32	Severe 5,10-methylenetetrahydrofolate reductase deficiency and two MTHFR variants in an adolescent with progressive myoclonic epilepsy. *Pediatric Neurology*, **2014**, 51, 266-70	2.9	13
31	Infant with cardiomyopathy: When to suspect inborn errors of metabolism?. *World Journal of Cardiology*, **2014**, 6, 1149-55	2.1	15
30	Liver pathology in infantile mitochondrial DNA depletion syndrome. *Pediatric and Developmental Pathology*, **2013**, 16, 415-24	2.2	8
29	A Pilot Study of Fluorodeoxyglucose Positron Emission Tomography Findings in Patients with Phenylketonuria before and during Sapropterin Supplementation. *Journal of Clinical Neurology (Korea*, **2013**, 9, 151-6	1.7	9
28	Long-term follow-up of four patients affected by HHH syndrome. *Clinica Chimica Acta*, **2012**, 413, 1151-5	6.2	13
27	3-methylcrotonyl-CoA carboxylase deficiency: clinical, biochemical, enzymatic and molecular studies in 88 individuals. *Orphanet Journal of Rare Diseases*, **2012**, 7, 31	4.2	40
26	Severe metabolic acidosis in a newborn with an abnormal newborn screen. *Clinical Pediatrics*, **2012**, 51, 518-20	1.2	
25	Ovarian function in Duarte galactosemia. *Fertility and Sterility*, **2011**, 96, 469-473.e1	4.8	13
24	Clinical and molecular characterization of five patients with succinyl-CoA:3-ketoacid CoA transferase (SCOT) deficiency. *Biochimica Et Biophysica Acta - Molecular Basis of Disease*, **2011**, 1812, 619-24	6.9	30
23	Isolated neonatal seizures: when to suspect inborn errors of metabolism. *Pediatric Neurology*, **2011**, 45, 283-91	2.9	27
22	Newborn screening for galactosemia: a review of 5 years of data and audit of a revised reporting approach. *Clinical Chemistry*, **2010**, 56, 437-44	5.5	15
21	Monitoring of biochemical status in children with Duarte galactosemia: utility of galactose, galactitol, galactonate, and galactose 1-phosphate. *Clinical Chemistry*, **2010**, 56, 1177-82	5.5	23
20	Genotype-phenotype correlations: sudden death in an infant with very-long-chain acyl-CoA dehydrogenase deficiency. *Journal of Inherited Metabolic Disease*, **2010**, 33 Suppl 3, S129-31	5.4	20
19	Very long-chain acyl-CoA dehydrogenase deficiency in a patient with normal newborn screening by tandem mass spectrometry. *Journal of Pediatrics*, **2010**, 156, 492-4	3.6	33
18	Failure to thrive: when to suspect inborn errors of metabolism. *Pediatrics*, **2009**, 124, 972-9	7.4	17
17	A Delphi clinical practice protocol for the management of very long chain acyl-CoA dehydrogenase deficiency. *Molecular Genetics and Metabolism*, **2009**, 96, 85-90	3.7	126
16	Very long-chain acyl-CoA dehydrogenase deficiency: the effects of accidental fat loading in a patient detected through newborn screening. *Journal of Inherited Metabolic Disease*, **2009**, 32 Suppl 1, S187-90	5.4	4
15	Brain magnetic resonance imaging findings in 49,XXXXY syndrome. *Pediatric Neurology*, **2008**, 38, 450-3	2.9	28

14	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	3.7	19
13	Clinical outcomes of infants with short-chain acyl-coenzyme A dehydrogenase deficiency (SCADD) detected by newborn screening. *Molecular Genetics and Metabolism*, **2008**, 95, 241-2	3.7	14
12	Duarte (DG) galactosemia: a pilot study of biochemical and neurodevelopmental assessment in children detected by newborn screening. *Molecular Genetics and Metabolism*, **2008**, 95, 206-12	3.7	58
11	A false-positive newborn screening result: goat$ milk acidopathy. *Pediatrics*, **2008**, 122, 210-1; author reply 211	7.4	4
10	Review of miglustat for clinical management in Gaucher disease type 1. *Therapeutics and Clinical Risk Management*, **2008**, 4, 425-31	2.9	48
9	Effect of galactose free formula on galactose-1-phosphate in two infants with classical galactosemia. *European Journal of Pediatrics*, **2008**, 167, 595-6	4.1	6
8	Structural variation of chromosomes in autism spectrum disorder. *American Journal of Human Genetics*, **2008**, 82, 477-88	11	1413
7	Development of a newborn screening follow-up algorithm for the diagnosis of isobutyryl-CoA dehydrogenase deficiency. *Genetics in Medicine*, **2007**, 9, 108-16	8.1	41
6	3-Methylcrotonyl-CoA carboxylase deficiency: metabolic decompensation in a noncompliant child detected through newborn screening. *Pediatrics*, **2006**, 118, 2555-6	7.4	23
5	Epimerase-deficiency galactosemia is not a binary condition. *American Journal of Human Genetics*, **2006**, 78, 89-102	11	65
4	Liver transplantation is not curative for methylmalonic acidopathy caused by methylmalonyl-CoA mutase deficiency. *Molecular Genetics and Metabolism*, **2006**, 88, 322-6	3.7	59
3	Galactitol and galactonate in red blood cells of children with the Duarte/galactosemia genotype. *Molecular Genetics and Metabolism*, **2005**, 84, 152-9	3.7	17
2	Biotinidase deficiency: the importance of adequate follow-up for an inconclusive newborn screening result. *European Journal of Pediatrics*, **2005**, 164, 298-301	4.1	11
1	MRI and MRS in HMG-CoA lyase deficiency. *Pediatric Neurology*, **1999**, 20, 375-80	2.9	36