# Can H Fiicioglu

## List of Publications by Citations



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| 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 | Structural variation of chromosomes in autism spectrum disorder. *American Journal of Human Genetics*, **2008**, 82, 477-88 | 11 | 1413 |
| 84 | Multiple phenotypes in phosphoglucomutase 1 deficiency. *New England Journal of Medicine*, **2014**, 370, 533-42 | 59.2 | 197 |
| 83 | A Phase 3 Trial of Sebelipase Alfa in Lysosomal Acid Lipase Deficiency. *New England Journal of Medicine*, **2015**, 373, 1010-20 | 59.2 | 168 |
| 82 | 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 |
| 81 | Diagnosis and treatment of tyrosinemia type I: a US and Canadian consensus group review and recommendations. *Genetics in Medicine*, **2017**, 19, | 8.1 | 99 |
| 80 | Epimerase-deficiency galactosemia is not a binary condition. *American Journal of Human Genetics*, **2006**, 78, 89-102 | 11 | 65 |
| 79 | 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 |
| 78 | 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 |
| 77 | Review of miglustat for clinical management in Gaucher disease type 1. *Therapeutics and Clinical Risk Management*, **2008**, 4, 425-31 | 2.9 | 48 |
| 76 | APRDX1 mutant allele causes a MMACHC secondary epimutation in cblC patients. *Nature Communications*, **2018**, 9, 67 | 17.4 | 45 |
| 75 | 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 |
| 74 | Consensus guidelines for newborn screening, diagnosis and treatment of infantile Krabbe disease. *Orphanet Journal of Rare Diseases*, **2018**, 13, 30 | 4.2 | 40 |
| 73 | 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 |
| 72 | MRI and MRS in HMG-CoA lyase deficiency. *Pediatric Neurology*, **1999**, 20, 375-80 | 2.9 | 36 |
| 71 | 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 |
| 70 | 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 |
| 69 | Long-term safety and efficacy of sapropterin: the PKUDOS registry experience. *Molecular Genetics and Metabolism*, **2015**, 114, 557-63 | 3.7 | 32 |

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| 68 | 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 |
| 67 | ALG1-CDG: Clinical and Molecular Characterization of 39 Unreported Patients. *Human Mutation*, **2016**, 37, 653-60 | 4.7 | 30 |
| 66 | Brain magnetic resonance imaging findings in 49,XXXXY syndrome. *Pediatric Neurology*, **2008**, 38, 450-3 | 2.9 | 28 |
| 65 | 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 |
| 64 | Isolated neonatal seizures: when to suspect inborn errors of metabolism. *Pediatric Neurology*, **2011**, 45, 283-91 | 2.9 | 27 |
| 63 | Cobalamin C Deficiency Shows a Rapidly Progressing Maculopathy With Severe Photoreceptor and Ganglion Cell Loss **2015**, 56, 7875-87 | | 25 |
| 62 | 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 |
| 61 | 3-Methylcrotonyl-CoA carboxylase deficiency: metabolic decompensation in a noncompliant child detected through newborn screening. *Pediatrics*, **2006**, 118, 2555-6 | 7.4 | 23 |
| 60 | Pathogenesis and treatment of spine disease in the mucopolysaccharidoses. *Molecular Genetics and Metabolism*, **2016**, 118, 232-43 | 3.7 | 22 |
| 59 | ECHS1 Deficiency as a Cause of Severe Neonatal Lactic Acidosis. *JIMD Reports*, **2016**, 30, 33-37 | 1.9 | 20 |
| 58 | 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 |
| 57 | Complex care of individuals with multiple sulfatase deficiency: Clinical cases and consensus statement. *Molecular Genetics and Metabolism*, **2018**, 123, 337-346 | 3.7 | 19 |
| 56 | 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 |
| 55 | 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 |
| 54 | TANGO2: expanding the clinical phenotype and spectrum of pathogenic variants. *Genetics in Medicine*, **2019**, 21, 601-607 | 8.1 | 18 |
| 53 | Failure to thrive: when to suspect inborn errors of metabolism. *Pediatrics*, **2009**, 124, 972-9 | 7.4 | 17 |
| 52 | 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 |
| 51 | Low bone mineral density is a common finding in patients with homocystinuria. *Molecular Genetics and Metabolism*, **2016**, 117, 351-4 | 3.7 | 17 |

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| 50 | Treatment outcome of creatine transporter deficiency: international retrospective cohort study. *Metabolic Brain Disease*, **2018**, 33, 875-884 | 3.9 | 16 |
| 49 | Infant with cardiomyopathy: When to suspect inborn errors of metabolism?. *World Journal of Cardiology*, **2014**, 6, 1149-55 | 2.1 | 15 |
| 48 | 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 |
| 47 | 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 |
| 46 | Retinal Structure in Cobalamin C Disease: Mechanistic and Therapeutic Implications. *Ophthalmic Genetics*, **2015**, 36, 339-48 | 1.2 | 13 |
| 45 | 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 |
| 44 | Long-term follow-up of four patients affected by HHH syndrome. *Clinica Chimica Acta*, **2012**, 413, 1151-5 | 6.2 | 13 |
| 43 | Ovarian function in Duarte galactosemia. *Fertility and Sterility*, **2011**, 96, 469-473.e1 | 4.8 | 13 |
| 42 | 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 |
| 41 | Adolescent presentations of inborn errors of metabolism. *Journal of Adolescent Health*, **2015**, 56, 477-82 | 5.8 | 12 |
| 40 | Missed Newborn Screening Case of Carnitine Palmitoyltransferase-II Deficiency. *JIMD Reports*, **2017**, 33, 93-97 | 1.9 | 12 |
| 39 | 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 |
| 38 | 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 |
| 37 | 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 |
| 36 | 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 |
| 35 | 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 |
| 34 | 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 |
| 33 | Liver pathology in infantile mitochondrial DNA depletion syndrome. *Pediatric and Developmental Pathology*, **2013**, 16, 415-24 | 2.2 | 8 |

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| 32 | Morbidity and mortality among exclusively breastfed neonates with medium-chain acyl-CoA dehydrogenase deficiency. *Genetics in Medicine*, **2016**, 18, 1315-1319 | 8.1 | 8 |
| 31 | Cobalamin C Disease Missed by Newborn Screening in a Patient with Low Carnitine Level. *JIMD Reports*, **2015**, 23, 71-5 | 1.9 | 7 |
| 30 | Characteristics and outcomes of patients with formiminoglutamic aciduria detected through newborn screening. *Journal of Inherited Metabolic Disease*, **2019**, 42, 140-146 | 5.4 | 7 |
| 29 | Galactokinase deficiency: lessons from the GalNet registry. *Genetics in Medicine*, **2021**, 23, 202-210 | 8.1 | 7 |
| 28 | 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 |
| 27 | 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 |
| 26 | Newborn Screening for Pompe Disease: Pennsylvania Experience. *International Journal of Neonatal Screening*, **2020**, 6, | 2.6 | 6 |
| 25 | 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 |
| 24 | Persistent dyslipidemia in treatment of lysosomal acid lipase deficiency. *Orphanet Journal of Rare Diseases*, **2020**, 15, 58 | 4.2 | 5 |
| 23 | 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 |
| 22 | 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 |
| 21 | 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 |
| 20 | A false-positive newborn screening result: goat's milk acidopathy. *Pediatrics*, **2008**, 122, 210-1; author reply 211 | 7.4 | 4 |
| 19 | 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 |
| 18 | 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 |
| 17 | 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 |
| 16 | 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 |
| 15 | Serial Magnetic Resonance Imaging (MRI) in Pyruvate Dehydrogenase Complex Deficiency. *Journal of Child Neurology*, **2020**, 35, 137-145 | 2.5 | 3 |

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| 14 | Argininosuccinic Acid Lyase Deficiency Missed by Newborn Screen. *JIMD Reports*, **2017**, 34, 43-47 | 1.9 | 3 |
| 13 | Early Indicators of Creatine Transporter Deficiency. *Journal of Pediatrics*, **2019**, 206, 283-285 | 3.6 | 3 |
| 12 | 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 |
| 11 | Failure to Thrive: An Expanded Differential Diagnosis. *Journal of Pediatric Genetics*, **2019**, 8, 27-32 | 0.7 | 2 |
| 10 | Provider Perspectives on the Impact of the COVID-19 Pandemic on Newborn Screening. *International Journal of Neonatal Screening*, **2021**, 7, | 2.6 | 2 |
| 9 | 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 |
| 8 | Neuropsychological implications of Cobalamin C (CblC) disease in Hispanic children detected through newborn screening. *Applied Neuropsychology: Child*, **2018**, 7, 143-149 | 1.4 | 1 |
| 7 | An 8-year-old girl with abdominal pain and mental status changes. *Pediatric Emergency Care*, **2015**, 31, 459-62 | 1.4 | 1 |
| 6 | Should patients with Phosphomannomutase 2-CDG (PMM2-CDG) be screened for adrenal insufficiency?. *Molecular Genetics and Metabolism*, **2021**, 133, 397-399 | 3.7 | 1 |
| 5 | 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 |
| 4 | 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 |
| 3 | Response to van Rijt et al. *Genetics in Medicine*, **2016**, 18, 1324 | 8.1 | |
| 2 | Severe metabolic acidosis in a newborn with an abnormal newborn screen. *Clinical Pediatrics*, **2012**, 51, 518-20 | 1.2 | |
| 1 | Response to Neeleman et al. *Genetics in Medicine*, **2020**, 22, 439-440 | 8.1 | |