

## Assoc. Prof. YILMAZ YILDIZ

### Personal Information

Office Phone: [+90 312 305 1141](tel:+903123051141)

Email: [yilmaz.yildiz@hacettepe.edu.tr](mailto:yilmaz.yildiz@hacettepe.edu.tr)

Web: <https://avesis.hacettepe.edu.tr/yilmaz.yildiz>

### International Researcher IDs

ORCID: 0000-0001-9076-1388

Publons / Web Of Science ResearcherID: AAJ-8666-2020

ScopusID: 54399559500

Yoksis Researcher ID: 169133

### Education Information

Post Doctorate of Medicine, Hacettepe University, Tıp Fakültesi (Türkçe), Dahili Tıp Bilimleri Bölümü, Turkey 2015 - 2018

Expertise In Medicine, Hacettepe University, Tıp Fakültesi, Çocuk Sağlığı Ve Hastalıkları Anabilim Dalı, Turkey 2009 - 2014

Doctorate, Hacettepe University, Tıp Fakültesi (İngilizce), Temel Tıp Bilimleri Bölümü, Turkey 2007 - 2010

### Foreign Languages

English, C2 Mastery

### Certificates, Courses and Trainings

Health&Medicine, Good Clinical Practice, çevrimiçi, 2018

### Dissertations

Expertise In Medicine, Hiperfenilalaninemili hastaların gebelik sonuçlarının incelenmesi, Hacettepe University, Tıp Fakültesi (Türkçe), Dahili Tıp Bilimleri Bölümü, 2014

Doctorate, Investigation of the role of Klf5 gene in muscle degeneration and differentiation, Hacettepe University, Sağlık Bilimleri Enstitüsü, Tıbbi Biyoloji A.B.D., 2010

### Research Areas

Medicine, Health Sciences, Medical Biology, Internal Medicine Sciences, Child Health and Diseases, Pediatric Endocrinology and Metabolism

### Academic Titles / Tasks

Assistant Professor, Hacettepe University, Tıp Fakültesi (Türkçe), Dahili Tıp Bilimleri Bölümü, 2020 - Continues  
Expert PhD, Pediatric Metabolic Diseases, 2018 - 2020  
Research Assistant, Hacettepe University, Tıp Fakültesi (Türkçe), Dahili Tıp Bilimleri Bölümü, 2015 - 2018  
Research Assistant, Hacettepe University, Tıp Fakültesi (Türkçe), Dahili Tıp Bilimleri Bölümü, 2009 - 2014

## Published journal articles indexed by SCI, SSCI, and AHCI

- I. **Coexistence of Two Rare Conditions Complicating the Other's Management: Propionic Acidemia and Apert Syndrome**  
ENSERT CİHAN C. K., AKAR H. T., Yıldız Y., SOĞUKPINAR M., ÜTİNE G. E., ÇELİK H. T.  
Molecular Syndromology, vol.15, no.1, pp.83-88, 2024 (SCI-Expanded)
- II. **Clinical, biochemical, and molecular insights into Cerebrotendinous Xanthomatosis: A nationwide study of 100 Turkish individuals.**  
Zubarioglu T., Kiykım E., Köse E., Eminoglu F. T., Teke Kısa P., Balci M. C., Özer I., İnci A., Çilesiz K., Canda E., et al.  
Molecular genetics and metabolism, vol.142, no.2, pp.108493, 2024 (SCI-Expanded)
- III. **Long-term clinical evaluation of patients with alpha-mannosidosis – A multicenter study**  
KÖSE E., KASAPKARA Ç. S., İNCİ A., YILDIZ Y., Sürücü Kara İ., Kahraman A. B., TÜMER L., DURSUN A., EMİNOĞLU F. T.  
European Journal of Medical Genetics, vol.68, 2024 (SCI-Expanded)
- IV. **The continuously evolving phenotype of succinic semialdehyde dehydrogenase deficiency.**  
Julia-Palacios N. A., Kuseyri Hübschmann O., Olivella M., Pons R., Horvath G., Lücke T., Fung C., Wong S., Cortès-Saladelafont E., Rovira-Remisa M. M., et al.  
Journal of inherited metabolic disease, 2024 (SCI-Expanded)
- V. **Electro-clinical features and long-term outcomes in guanidinoacetate methyltransferase (GAMT) deficiency**  
YILDIZ Y., Ardiçlı D., GÖÇMEN R., YALNIZOĞLU D., Topçu M., Coşkun T., TOKATLI A., Haliloğlu G.  
European Journal of Paediatric Neurology, vol.49, pp.66-72, 2024 (SCI-Expanded)
- VI. **Impact of citrulline substitution on clinical outcome after liver transplantation in carbamoyl phosphate synthetase 1 and ornithine transcarbamylase deficiency.**  
Aldrian D., Waldner B., Vogel G. F., El-Gharbawy A. H., McKiernan P., Vockley J., Landau Y. E., Al Mutairi F., Stepien K. M., Kwok A. M., et al.  
Journal of inherited metabolic disease, vol.47, no.2, pp.220-229, 2024 (SCI-Expanded)
- VII. **Validity and reliability of the MetabQoL 1.0 and assessment of neuropsychiatric burden in organic acidemias: Reflections from Turkey**  
Ersak A. Ş., Çak H. T., YILDIZ Y., Çavdar M., Tunç S., ÖZER N., Zeltner N. A., Huemer M., TOKATLI A., Haliloğlu G.  
Molecular Genetics and Metabolism, vol.141, no.1, 2024 (SCI-Expanded)
- VIII. **Predictors of eventual requirement of phenylalanine-restricted diet in young infants with phenylalanine hydroxylase deficiency initially managed with sapropterin monotherapy**  
ÇIKI K., YILDIZ Y., KAHRAMAN A. B., ÖZGÜL R. K., COŞKUN T., DURSUN A., TOKATLI A., SİVRİ S.  
Molecular Genetics and Metabolism, vol.140, no.3, 2023 (SCI-Expanded)
- IX. **A novel mutation in ATP13A2 gene in a patient with complicated hereditary spastic paraplegia accompanied by tubulopathy.**  
Inan B., Azman F., Aktas D., Yildiz Y., Saygi S.  
Acta neurologica Belgica, vol.123, no.5, pp.1985-1987, 2023 (SCI-Expanded)
- X. **Preliminary evaluation of potential urinary organic acid biomarkers with mass spectrometry in children with autism spectrum disorder**  
Sertoglu E., Balik A. R., Duman U. G., Mavis M. E., Arslan M., YILDIZ Y., Batu J., Olgac A., Hekim Ö.  
Research in Autism Spectrum Disorders, vol.106, 2023 (SSCI)
- XI. **Levodopa-refractory hyperprolactinemia and pituitary findings in inherited disorders of biogenic amine metabolism.**

Yıldız Y., Kuseyri Hübschmann O., Akgöz Karaosmanoğlu A., Manti F., Karaca M., Schwartz I. V. D., Pons R., López-Laso E., Palacios N. A. J., Porta F., et al.

Journal of inherited metabolic disease, 2023 (SCI-Expanded)

- XII. **COVID-19 in inherited metabolic disorders: Clinical features and risk factors for disease severity**  
KAHRAMAN A. B., YILDIZ Y., ÇIKI K., Erdal I., AKAR H. T., DURSUN A., TOKATLI A., SİVRİ S.  
Molecular Genetics and Metabolism, vol.139, no.2, 2023 (SCI-Expanded)
- XIII. **Successful management of rhabdomyolysis with triheptanoin in a child with severe long-chain 3-hydroxyacyl-coenzyme A dehydrogenase (LCHAD) deficiency**  
KAHRAMAN A. B., YILDIZ Y., GÖKMEN ÖZEL H., KADAYIFÇILAR S., SİVRİ S.  
Neuromuscular Disorders, vol.33, no.4, pp.315-318, 2023 (SCI-Expanded)
- XIV. **Splenic Gaucheroma Leading to Incidental Diagnosis of Gaucher Disease in a 46-Year-Old Man with a Rare GBA Mutation: A Case Report**  
Erdal İ., YILDIZ Y., ÖNAL G., AKTEPE O. H., Düzgün S. A., Sağlam A., DÖKMECİ S., SİVRİ H. S.  
Endocrine, Metabolic and Immune Disorders - Drug Targets, vol.23, no.2, pp.230-234, 2023 (SCI-Expanded)
- XV. **The Türkiye-Syria Earthquake: a response from the editors of the Turkish Journal of Pediatrics**  
DÜZÖVA A., AKGÜL S., ÜTİNE G. E., YILDIZ Y.  
Turkish Journal of Pediatrics, vol.65, no.1, pp.1-2, 2023 (SCI-Expanded)
- XVI. **Two tales of LPIN1 deficiency: from fatal rhabdomyolysis to favorable outcome of acute compartment syndrome**  
KAHRAMAN A. B., Karakaya B., YILDIZ Y., KAMACI S., KESİCİ S., ŞİMŞEK KİPER P. Ö., KURT ŞÜKÜR E. D., BAYRAKCI B., Haliloglu G.  
Neuromuscular Disorders, vol.32, no.11-12, pp.931-934, 2022 (SCI-Expanded)
- XVII. **Three-Country Snapshot of Ornithine Transcarbamylase Deficiency**  
Seker Yilmaz B., Baruteau J., ARSLAN N., AYDIN H. İ., Barth M., Bozaci A. E., Brassier A., CANDA E., Cano A., Chronopoulou E., et al.  
LIFE-BASEL, vol.12, no.11, 2022 (SCI-Expanded)
- XVIII. **Novel Cranial Imaging Findings and a Splice-Site Variant in a Patient with Tyrosinemia Type III, and a Summary of Published Cases**  
KAHRAMAN A. B., AKAR H. T., Lafci N. G., YILDIZ Y., Tokatli A.  
MOLECULAR SYNDROMOLOGY, vol.13, no.3, pp.193-199, 2022 (SCI-Expanded)
- XIX. **Comment on the "NASPGHAN Position Paper on the Diagnosis and Management of Pediatric Acute Liver Failure".**  
Yildiz Y., Tokatli A.  
Journal of pediatric gastroenterology and nutrition, vol.74, 2022 (SCI-Expanded)
- XX. **Increased ocular wall thickness and decreased globe volume in children with mucopolysaccharidosis type VI**  
Özkale Yavuz Ö., Ayaz E., Yıldız Y., Karaosmanoğlu A., Bulut E. G., Sivri H. S., Karlı Oğuz H. K.  
DIAGNOSTIC AND INTERVENTIONAL RADIOLOGY, vol.1, pp.1-18, 2022 (SCI-Expanded)
- XXI. **Clinico-radiological features, molecular spectrum, and identification of prognostic factors in developmental and epileptic encephalopathy due to inosine triphosphate pyrophosphatase (ITPase) deficiency**  
Scala M., Wortmann S. B., Kaya N., Stellingwerff M. D., Pistorio A., Glamuzina E., van Karnebeek C. D., Skrypnik C., Iwanicka-Pronicka K., Piekutowska-Abramczuk D., et al.  
HUMAN MUTATION, vol.43, no.3, pp.403-419, 2022 (SCI-Expanded)
- XXII. **Clinical Heterogeneity in MT-ATP6 Pathogenic Variants: Same Genotype-Different Onset**  
Capiou S., Smet J., De Paepe B., YILDIZ Y., Arslan M., Stevens O., Verschoore M., Stepman H., Seneca S., Vanlander A.  
CELLS, vol.11, no.3, 2022 (SCI-Expanded)
- XXIII. **Genetic Counseling for Phenylketonuria Complicated by Undiagnosed Parental Hyperphenylalaninemia in a Single Family Genetische Beratung bei Phenylketonurie, kompliziert durch nicht diagnostizierte elterliche Hyperphenylalaninämie in einer einzigen Familie**  
ÇİKLİ K., ÖZGÜL R. K., YILDIZ Y.

Klinische Padiatrie, 2022 (SCI-Expanded)

- XXIV. **Hyperinsulinism may be Underreported in Hypoglycemic Patients with Phosphomannomutase 2 Deficiency.**  
Vuralli D., Yildiz Y., Ozon A., Dursun A., Gonc N., Tokatlı A., Sivri H. S., Alikasifoglu A.  
Journal of clinical research in pediatric endocrinology, vol.14, pp.275-286, 2022 (SCI-Expanded)
- XXV. **Single Institutional Experience with GM1 Gangliosidosis: Clinical and Laboratory Results of 14 Patients.**  
Akar H. T., Yıldız Y., Güvenkaya G., Çıkkı K., Kahraman A. B., Erdal İ., Coşkun T., Dursun A., Sivri H. S., Tokatlı A.  
Balkan medical journal, vol.39, pp.345-350, 2022 (SCI-Expanded)
- XXVI. **Assessment of intellectual impairment, health-related quality of life, and behavioral phenotype in patients with neurotransmitter related disorders: Data from the iNTD registry**  
Keller M., Brennenstuhl H., Kuseyri Hübschmann O., Manti F., Julia Palacios N. A., Friedman J., Yıldız Y., Koht J. A., Wong S., Zafeiriou D. I., et al.  
JOURNAL OF INHERITED METABOLIC DISEASE, vol.44, no.6, pp.1489-1502, 2021 (SCI-Expanded)
- XXVII. **Biallelic mutations in ELFN1 gene associated with developmental and epileptic encephalopathy and joint laxity**  
DUR SUN A., YALNIZOĞLU D., Yılmaz D. Y., Oguz K. K., GÜLBAKAN B., KOŞUKCU C., AKAR H. T., Kahraman A. B., Acar N. V., GÜNBEY C., et al.  
EUROPEAN JOURNAL OF MEDICAL GENETICS, vol.64, no.11, 2021 (SCI-Expanded)
- XXVIII. **Perplexing Etiology of Hyperphenylalaninemia in an Infant Referred via Newborn Screening.**  
Çıkkı K., Akar H. T., Özgül R. K., Gülbakan B., Yıldız Y.  
Clinical chemistry, vol.67, no.10, pp.1428-1431, 2021 (SCI-Expanded)
- XXIX. **Insights into the expanding phenotypic spectrum of inherited disorders of biogenic amines**  
Kuseyri Hübschmann O., Horvath G., Cortès-Saladela font E., Yıldız Y., Mastrangelo M., Pons R., Friedman J., Mercimek-Andrews S., Wong S., Pearson T. S., et al.  
NATURE COMMUNICATIONS, vol.12, no.1, 2021 (SCI-Expanded)
- XXX. **COVID-19-related anxiety in phenylketonuria patients**  
Akar H. T., Karaboncuk Y., Çıkkı K., Kahraman A. B., Erdal İ., Coşkun T., Tokatlı A., Dursun A., Yıldız Y., Sivri H. S.  
TURKISH JOURNAL OF PEDIATRICS, vol.63, no.5, pp.790-800, 2021 (SCI-Expanded)
- XXXI. **Homozygous missense VPS16 variant is associated with a novel disease, resembling mucopolysaccharidosis-plus syndrome in two siblings**  
Yıldız Y., Koşukcu C., Aygün D., Akçaboy M., Öztekk Çelebi F. Z., Taşçı Yıldız Y., Şahin G., Aytekin C., Yüksel D., Lay İ., et al.  
CLINICAL GENETICS, vol.100, no.3, pp.308-317, 2021 (SCI-Expanded)
- XXXII. **DNACJ12 deficiency in patients with unexplained hyperphenylalaninemia: two new patients and a novel variant**  
Çıkkı K., Yıldız Y., Yücel Yılmaz D., Pektaş E., Tokatlı A., Özgül R. K., Sivri H. S., Dursun A.  
METABOLIC BRAIN DISEASE, vol.36, no.6, pp.1405-1410, 2021 (SCI-Expanded)
- XXXIII. **Genotypic and phenotypic features in Turkish patients with classic nonketotic hyperglycinemia**  
Bayrak H., Yıldız Y., Olğaç A., Kasapkara Ç. S., Küçükcong ar A., Zenciroğ lu A., Yüksel D., Ceylaner S., Kılıç M.  
METABOLIC BRAIN DISEASE, vol.36, no.6, pp.1213-1222, 2021 (SCI-Expanded)
- XXXIV. **Brain MR patterns in inherited disorders of monoamine neurotransmitters: An analysis of 70 patients**  
Hübschmann O. K., Mohr A., Friedman J., Manti F., Horvath G., Cortès-Saladela font E., Mercimek-Andrews S., Yildiz Y., Pons R., Kulhánek J., et al.  
JOURNAL OF INHERITED METABOLIC DISEASE, vol.44, no.4, pp.1070-1082, 2021 (SCI-Expanded)
- XXXV. **Recurrent Postinfectious Rhabdomyolysis in a 5-Year-Old Girl**  
Torun E., Yildiz Y., Yazici M., Çolak F., Kasapkara C.  
KLINISCHE PADIATRIE, vol.233, no.04, pp.203-205, 2021 (SCI-Expanded)
- XXXVI. **Complicated peripartum course in a patient with very long-chain acyl-coenzyme A dehydrogenase (VLCAD) deficiency**

- AKAR H. T., Çağan M., YILDIZ Y., SİVRİ H. S.  
NEUROMUSCULAR DISORDERS, vol.31, no.6, pp.566-569, 2021 (SCI-Expanded)
- XXXVII. **Safety and Efficacy of Liver-Directed Gene Therapy in Patients with Mucopolysaccharidosis Type VI**  
Brunetti-Pierri N., Ferla R., Ginocchio V. M., Rossi A., Fecarotta S., Romano R., Parenti G., YILDIZ Y., Zancan S., Pecorella V., et al.  
MOLECULAR THERAPY, vol.29, no.4, pp.114, 2021 (SCI-Expanded)
- XXXVIII. **Oral health status of children and young adults with maple syrup urine disease in Turkey.**  
Ballıkaya E., Yildiz Y., Koç N., Tokatlı A., Uzamis Tekcicek M., Sivri H. S.  
BMC oral health, vol.21, pp.8, 2021 (SCI-Expanded)
- XXXIX. **Invisible burden of COVID-19: enzyme replacement therapy disruptions**  
Kahraman A. B., Yıldız Y., Çıkkı K., Akar H. T., Erdal İ., Dursun A., Tokatlı A., Sivri H. S.  
JOURNAL OF PEDIATRIC ENDOCRINOLOGY & METABOLISM, vol.34, no.5, pp.539-545, 2021 (SCI-Expanded)
- XL. **Creatine Transporter Deficiency Presenting as Autism Spectrum Disorder**  
Yıldız Y., Göçmen R., Yaramış A., Coşkun T., Haliloğlu G.  
PEDIATRICS, vol.146, no.5, 2020 (SCI-Expanded)
- XLI. **Predictors of acute metabolic decompensation in children with maple syrup urine disease at the emergency department**  
YILDIZ Y., Akcan Yildiz L., DURSUN A., TOKATLI A., COŞKUN T., TEKŞAM Ö., SİVRİ H. S.  
EUROPEAN JOURNAL OF PEDIATRICS, vol.179, no.7, pp.1107-1114, 2020 (SCI-Expanded)
- XLII. **Retrospective evaluation of 85 patients with urea cycle disorders: one center experience, three new mutations**  
Nakip O. S., YILDIZ Y., TOKATLI A.  
JOURNAL OF PEDIATRIC ENDOCRINOLOGY & METABOLISM, vol.33, no.6, pp.721-728, 2020 (SCI-Expanded)
- XLIII. **Successful management of acute pancreatitis due to apolipoprotein C-II deficiency in a 37-day-old infant**  
Yıldız Y., Uysal Y., Çınar H., Özbay H., Kurt Ç., Kılıç M.  
PANCREATOLOGY, vol.20, no.4, pp.644-646, 2020 (SCI-Expanded)
- XLIV. **Two cases of Vici syndrome presenting with corpus callosum agenesis, albinism, and severe developmental delay**  
Hızal M., Yeke B., YILDIZ Y., Öztürk A., Gürbüz B. B., COŞKUN T.  
TURKISH JOURNAL OF PEDIATRICS, vol.62, no.3, pp.474-478, 2020 (SCI-Expanded)
- XLV. **Consensus guideline for the diagnosis and treatment of tetrahydrobiopterin (BH4) deficiencies**  
Opladen T., Lopez-Laso E., Cortes-Saladelafont E., Pearson T. S., SİVRİ H. S., YILDIZ Y., Assmann B., Kurian M. A., Leuzzi V., Heales S., et al.  
ORPHANET JOURNAL OF RARE DISEASES, vol.15, no.1, 2020 (SCI-Expanded)
- XLVI. **Genotypes and estimated prevalence of phosphomannomutase 2 deficiency in Turkey differ significantly from those in Europe**  
YILDIZ Y., Arslan M., Celik G., Kasapkara C. S., Ceylaner S., DURSUN A., SİVRİ H. S., COŞKUN T., TOKATLI A.  
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, vol.182, no.4, pp.705-712, 2020 (SCI-Expanded)
- XLVII. **Comment on: "Multiple acyl-CoA dehydrogenase deficiency in elderly carriers"**  
YILDIZ Y., TOKATLI A.  
JOURNAL OF NEUROLOGY, vol.267, no.4, pp.1209-1210, 2020 (SCI-Expanded)
- XLVIII. **Oral health status of children with phenylketonuria**  
BALLIKAYA E., YILDIZ Y., SİVRİ H. S., TOKATLI A., DURSUN A., Olmez S., COŞKUN T., Tekcicek M. U.  
JOURNAL OF PEDIATRIC ENDOCRINOLOGY & METABOLISM, vol.33, no.3, pp.361-365, 2020 (SCI-Expanded)
- XLIX. **When Pancreatitis Stinks A Rare Cause of Acute Recurrent Pancreatitis in a Child**  
YILDIZ Y., Sahin G., Hosnut F. O.  
PANCREAS, vol.49, no.1, 2020 (SCI-Expanded)
- L. **Inborn errors of metabolism in the differential diagnosis of fatty liver disease**  
YILDIZ Y., SİVRİ H. S.  
TURKISH JOURNAL OF GASTROENTEROLOGY, vol.31, no.1, pp.3-16, 2020 (SCI-Expanded)

- LI. **Rare cause of high anion gap metabolic acidosis in an infant: Succinyl-CoA:3-ketoacid transferase deficiency**  
YILDIZ Y., Azapagasi E.  
JOURNAL OF PAEDIATRICS AND CHILD HEALTH, vol.55, no.11, pp.1395-1396, 2019 (SCI-Expanded)
- LII. **Determinants of Riboflavin Responsiveness in Multiple Acyl-CoA Dehydrogenase Deficiency**  
YILDIZ Y., TALİM B., Haliloglu G., Topaloglu H., AKÇÖREN Z., DURSUN A., SİVRİ H. S., COŞKUN T., TOKATLI A.  
PEDIATRIC NEUROLOGY, vol.99, pp.69-75, 2019 (SCI-Expanded)
- LIII. **Maternal phenylketonuria in Turkey: outcomes of 71 pregnancies and issues in management**  
YILDIZ Y., SİVRİ H. S.  
EUROPEAN JOURNAL OF PEDIATRICS, vol.178, no.7, pp.1005-1011, 2019 (SCI-Expanded)
- LIV. **Cognitive and behavioral impairment in mild hyperphenylalaninemia**  
Evinç S., Pektaş E., Foto-Özdemir D., Yıldız Y., Karaboncuk Y., Bilginer-Gürbüz B., Dursun A., Tokatlı A., Coskun T., Öktem F., et al.  
TURKISH JOURNAL OF PEDIATRICS, vol.60, no.6, pp.617-624, 2018 (SCI-Expanded)
- LVI. **Post-mortem detection of FLAD1 mutations in 2 Turkish siblings with hypotonia in early infancy**  
YILDIZ Y., Olsen R. K. J., SİVRİ H. S., AKÇÖREN Z., Nygaard H. H., TOKATLI A.  
NEUROMUSCULAR DISORDERS, vol.28, no.9, pp.787-790, 2018 (SCI-Expanded)
- LVI. **Oral health status in patients with mucopolysaccharidoses**  
BALLIKAYA E., Eymirli P. S., YILDIZ Y., AVCU N., SİVRİ H. S., Uzamis-Tekcicek M.  
TURKISH JOURNAL OF PEDIATRICS, vol.60, no.4, pp.400-406, 2018 (SCI-Expanded)
- LVII. **Hyperinsulinemic Hypoglycemia in Congenital Disorder of Glycosylation Type-1a (CDG-1a)**  
Vuralli D., YILDIZ Y., SİVRİ H. S., ALİKAŞİFOĞLU A.  
HORMONE RESEARCH IN PAEDIATRICS, vol.90, pp.362-363, 2018 (SCI-Expanded)
- LVIII. **Hereditary Dopamine Transporter Deficiency Syndrome: Challenges in Diagnosis and Treatment**  
YILDIZ Y., Pektaş E., TOKATLI A., Haliloglu G.  
NEUROPEDIATRICS, vol.48, no.1, pp.49-52, 2017 (SCI-Expanded)
- LIX. **Partial hydatidiform mole in a phenylketonuria patient treated with sapropterin dihydrochloride**  
YILDIZ Y., DURSUN A., TOKATLI A., COŞKUN T., Sivri S.  
GYNECOLOGICAL ENDOCRINOLOGY, vol.33, no.1, pp.19-20, 2017 (SCI-Expanded)
- LX. **Late-diagnosed phenylketonuria in an eight-year-old boy with dyslexia and attention-deficit hyperactivity disorder**  
YILDIZ Y., DURSUN A., TOKATLI A., COŞKUN T., SİVRİ H. S.  
TURKISH JOURNAL OF PEDIATRICS, vol.58, no.1, pp.94-96, 2016 (SCI-Expanded)
- LXI. **Periostin is temporally expressed as an extracellular matrix component in skeletal muscle regeneration and differentiation**  
OZDEMIR C., Akpulat U., SHARAFI P., YILDIZ Y., ONBAŞILAR İ., Kocaefe C.  
GENE, vol.553, no.2, pp.130-139, 2014 (SCI-Expanded)
- LXII. **Localized acute generalized exanthematous pustulosis with amoxicillin and clavulanic acid**  
Ozkaya-Parlakay A., Azkur D., KARA A., YILDIZ Y., ORHAN D., CENGİZ A. B., Ersoy-Evans S.  
TURKISH JOURNAL OF PEDIATRICS, vol.53, no.2, pp.229-232, 2011 (SCI-Expanded)

## Articles Published in Other Journals

- I. **Adult-onset carnitine palmitoyl transferase II (CPT II) deficiency presenting with rhabdomyolysis and acute kidney injury**  
Akar H. T., Yıldız Y., Mutluay R., Tekin E., Tokatlı A.  
CEN CASE REPORTS, vol.2023, 2023 (ESCI)
- II. **A Novel Double Homozygous BTB Gene Mutation in A Case of Profound Biotinidase Deficiency**  
DEVECİ K., AKAR H. T., YILDIZ Y., ÖZGÜL R. K.  
Türkiye Çocuk Hastalıkları Dergisi, 2022 (Peer-Reviewed Journal)

- III. **Liver-Directed Adeno-Associated Virus Mediated Gene Therapy for Mucopolysaccharidosis Type VI**  
Brunetti-Pierri N., Ferla R., Ginocchio V. M., Rossi A., Fecarotta S., Romano R., Parenti G., YILDIZ Y., Zancan S., Pecorella V., et al.  
NEJM Evidence, vol.1, 2022 (Peer-Reviewed Journal)
- IV. **The COVID-19 Pandemic and Enzyme Replacement Therapy in Lysosomal Storage Disorders**  
Olgaci A., KASAPKARA Ç. S., Acikel B., Yildiz Y., Molla G. K., Kilic M.  
JOURNAL OF PEDIATRIC RESEARCH, vol.8, no.4, pp.370-376, 2021 (ESCI)
- V. **Difficulties Associated with Enzyme Replacement Therapy for Mucopolysaccharidoses**  
YILDIZ Y., SİVRİ H. S.  
TURKISH ARCHIVES OF PEDIATRICS, vol.56, no.6, pp.602-609, 2021 (ESCI)
- VI. **Infant Acute Lymphoblastic Leukemia with Atypical Presentation**  
YAMAN BAJİN H. İ., YILDIZ Y., akın Ş., AYTAÇ EYÜPOĞLU Ş. S., ÜNAL CANGÜL Ş., KUŞKONMAZ B. B., Cetin M., SİVRİ H. S., GÜMRÜK F.  
Acta Medica, vol.50, no.4, pp.57-59, 2019 (Peer-Reviewed Journal)
- VII. **Kalıtsal Metabolik Hastalıklarda Dental Bulgular**  
YILDIZ Y., SİVRİ H. S.  
Türkiye Klinikleri Çocuk Diş Hekimliği - Özel Konular, vol.2, no.2, pp.28-33, 2016 (Peer-Reviewed Journal)
- VIII. **Mukopolisakkaridozlarda ortopedik sorunlar**  
YILDIZ Y., SİVRİ H. S.  
TOTBİD Dergisi, vol.15, no.4, pp.303-310, 2016 (Peer-Reviewed Journal)
- IX. **Analysis of risk taking behaviour of individual pension system participants the case of Turkey**  
KARA S., YILDIZ Y., KARAN M. B.  
Pressacademia, vol.2, no.3, pp.375, 2015 (Peer-Reviewed Journal)

## Books & Book Chapters

- I. **AKÇAĞAÇ ŞURUBU İDRAR HASTALIĞI ve BESLENME TEDAVİSİ**  
Yıldız Y., Gökmen Özel H.  
in: KALITSAL METABOLİK HASTALIKLARDA BESLENME TEDAVİSİ, Eminoğlu FT,Haspolat YK,Çeltik C,Çarman KB,Akbulut UE,Taş T, Editor, Orient Yayınları, Ankara, pp.235-266, 2021
- II. **Doğuştan Metabolik Hastalıklara Yaklaşım**  
Yıldız Y.  
in: Nelson Pediatri, Murat Yurdakök, Editor, Güneş Kitabevi, Ankara, pp.688-695, 2021
- III. **Karbonhidrat Metabolizması Bozuklukları**  
Yıldız Y.  
in: Nelson Pediatri, Murat Yurdakök, Editor, Güneş Kitabevi, Ankara, pp.777-806, 2021
- IV. **Yıldız Y., Sivri H. S.**  
in: , Kalbiye Yalaz, Editor, Hipokrat Kitabevi, Ankara, pp.343-351, 2021
- V. **Disorders of Amino Acid Metabolism**  
Yıldız Y., Kılıç M.  
in: Pediatrics, Zülfiyar Akelma, Editor, Nobel Tıp Kitabevi, Ankara, pp.1124-1129, 2021
- VI. **Acute Metabolic Crisis**  
Yıldız Y., Haliloğlu V. G.  
in: Handbook of Clinical Acute Pediatric Neurology, Deniz Yüksel, Editor, Nobel Tıp Kitabevi, Ankara, pp.57-65, 2020
- VII. **Nörotransmitter Bozukluklarında Klinik Yaklaşım**  
Yıldız Y., Sivri H. S.  
in: Kalıtsal Metabolik Hastalıklarda Hareket Bozuklukları, Dursun, Ali, Editor, Türkiye Klinikleri Yayınevi, Ankara, pp.20-29, 2020

## VIII. Lyme Hastalığı

Yıldız Y.

in: Neonatoloji El Kitabı, Yurdakök, Murat, Editor, Güneş Tıp Kitabevleri, Ankara, pp.683-685, 2014

## IX. Syphilis

Yıldız Y.

in: Manual of Neonatal Care, Murat Yurdakök, Editor, Güneş Tıp Kitabevleri, Ankara, pp.664-671, 2014

## X. Çocuğa Kötü Muamele: İhmalden İstismara

Yıldız Y.

in: Rudolph Pediatri, Yurdakök, Murat, Editor, Güneş Tıp Kitabeleri, Ankara, pp.137-143, 2013

## Supported Projects

YALÇIN S. S., YILDIZ Y., SABUNCUOĞLU S., ERKEKOĞLU Ü. P., Erdal İ., Yirün A., Project Supported by Higher Education Institutions, Fenilalanin metabolizması bozukluğu olan hastaların tedavi rejimlerine göre serum bisfenol A, bisfenol F, ftalat ve bazı ağır metaller arasındaki ilişkinin incelenmesi, 2022 - Continues

YILDIZ Y., OĞUZ B., ÖNEN E. F., DÜZOVA A., LAY İ., Project Supported by Higher Education Institutions, Glutarik asidüri tip 1 hastalarında böbrek işlevlerinin incelenmesi, 2022 - Continues

Yıldız Y., Sivri H. S., Project Supported by Private Organizations in Other Countries, iNTD - International Working Group on Neurotransmitter Related Diseases, 2017 - 2025

TOKATLI A., DURSUN A., SİVRİ H. S., LAY İ., ÖZGÜL R. K., YÜCEL YILMAZ D., GÜLBAKAN B., YILDIZ Y., Project Supported by Higher Education Institutions, HACETTEPE ÜNİVERSİTESİ İLERİ METABOLİK TESTLER LABORATUVARI ALTYAPISININ YENİLENMESİ, 2020 - 2024

Önenli Mungan H., Yıldız Y., Kor D., Bulut F. D., Sivri H. S., Öktem F., Gökmen Özel H., Tokatlı A., Çıkkı K., Gökçay G. F., et al., Project Supported by Other Private Institutions, Fenilalanin Hidroksilaz (PAH) Enzim Eksikliğine veya Yokluğuna Bağlı Hiperfenilalaninemili Hastaların Retrospektif ve Prospektif Olarak Takip, Tedavi ve İzlemlerinin Yapıldığı Çok Merkezli Kayıt Çalışması, 2020 - 2023

Yıldız Y., Sivri H. S., Civil Society Organizations of Other Countries Supported Project, A Phase I/II Open Label, Dose Escalation, Safety Study in Subjects with Mucopolysaccharidosis type VI (MPS VI) Using Adeno-Associated Viral Vector 8 to Deliver the human ARSB gene to Liver, 2017 - 2022

Yıldız Y., Sivri H. S., Other International Funding Programs, A Non-Interventional Observational Study of Pegvaliase-Naive Adults with Phenylketonuria (PKU) - Concept Elicitation and Cognitive Interviews (BMN 165-901), 2018 - 2019  
Auricchio A., Sivri H. S., Yıldız Y., FP7 Project, MEUSix Ağır bir lizozomal depo hastalığı olan MPS tip VI lı hastalarda gen tedavisi, 2014 - 2017

## Activities in Scientific Journals

TURKISH JOURNAL OF PEDIATRICS, Publication Committee Member, 2022 - Continues

Sürekli Tıp Eğitimi Dergisi STED, Evaluation Committee Member, 2014 - Continues

## Memberships / Tasks in Scientific Organizations

Çocuk Beslenme ve Metabolizma Derneği, Member, 2019 - Continues, Turkey

Society for the Study of Inborn Errors of Metabolism, Member, 2015 - Continues, United Kingdom

## Scientific Refereeing

TURKISH JOURNAL OF PEDIATRICS, Journal Indexed in SCI-E, May 2024

JOURNAL OF PEDIATRIC ENDOCRINOLOGY AND METABOLISM, Journal Indexed in SCI-E, March 2024



TURKISH ARCHIVES OF PEDIATRICS, Journal Indexed in ESCI, March 2024  
PEDIATRICS, Journal Indexed in SCI-E, February 2024  
TURKISH JOURNAL OF PEDIATRICS, Journal Indexed in SCI-E, January 2024  
TURKISH ARCHIVES OF PEDIATRICS, Journal Indexed in ESCI, January 2024  
EUROPEAN JOURNAL OF MEDICAL GENETICS, Journal Indexed in SCI-E, January 2024  
TURKISH ARCHIVES OF PEDIATRICS, Journal Indexed in ESCI, December 2023  
TURKISH JOURNAL OF PEDIATRICS, Journal Indexed in SCI-E, November 2023  
CLINICAL CASE REPORTS, Journal Indexed in ESCI, October 2023  
PEDIATRICS, Journal Indexed in SCI-E, October 2023  
EUROPEAN JOURNAL OF MEDICAL GENETICS, Journal Indexed in SCI-E, September 2023  
TURKISH ARCHIVES OF PEDIATRICS, Journal Indexed in ESCI, September 2023  
TURKISH JOURNAL OF PEDIATRICS, Journal Indexed in SCI-E, August 2023  
JOURNAL OF PEDIATRIC ENDOCRINOLOGY AND METABOLISM, Journal Indexed in SCI-E, August 2023  
HUMAN GENE, Journal Indexed in ESCI, June 2023  
BMC MEDICAL EDUCATION, Journal Indexed in SCI-E, May 2023  
JOURNAL OF PEDIATRIC ENDOCRINOLOGY AND METABOLISM, Journal Indexed in SCI-E, May 2023  
TURKISH ARCHIVES OF PEDIATRICS, Journal Indexed in ESCI, May 2023  
EUROPEAN JOURNAL OF NEUROLOGY, Journal Indexed in SCI-E, April 2023  
HUMAN MOLECULAR GENETICS, Journal Indexed in SCI-E, April 2023  
JOURNAL OF PEDIATRIC ENDOCRINOLOGY AND METABOLISM, Journal Indexed in SCI-E, March 2023  
Türkiye Çocuk Hastalıkları Dergisi, National Scientific Refreed Journal, March 2023  
TURKISH ARCHIVES OF PEDIATRICS, Journal Indexed in ESCI, January 2023  
TURKISH ARCHIVES OF PEDIATRICS, Journal Indexed in ESCI, December 2022  
TURKISH ARCHIVES OF PEDIATRICS, Journal Indexed in ESCI, October 2022  
TURKISH JOURNAL OF PEDIATRICS, Journal Indexed in SCI-E, February 2022  
NOROPSIKIYATRI ARSIVI-ARCHIVES OF NEUROPSYCHIATRY, Journal Indexed in SCI-E, December 2021  
Turkish Archives of Pediatrics, Journal Indexed in ESCI, December 2021  
SÜREKLİ TIP EĞİTİMİ DERGİSİ STED, National Scientific Refreed Journal, September 2021  
TURKISH JOURNAL OF PEDIATRICS, Journal Indexed in SCI-E, August 2021  
SÜREKLİ TIP EĞİTİMİ DERGİSİ STED, National Scientific Refreed Journal, July 2021  
TURKISH JOURNAL OF PEDIATRICS, Journal Indexed in SCI-E, June 2021  
TURKISH JOURNAL OF PEDIATRICS, Journal Indexed in SCI-E, June 2021  
Cukurova Medical Journal, Journal Indexed in ESCI, June 2021  
PLOS ONE, SCI Journal, April 2021  
JOURNAL OF PEDIATRIC ENDOCRINOLOGY & METABOLISM, Journal Indexed in SCI-E, January 2021  
JOURNAL OF INTERNATIONAL MEDICAL RESEARCH, Journal Indexed in SCI-E, August 2020  
TURKISH JOURNAL OF PEDIATRICS, Journal Indexed in SCI-E, August 2020  
JOURNAL OF INTEGRATIVE NEUROSCIENCE, Journal Indexed in SCI-E, June 2020  
Journal Of Pediatric Endocrinology & Metabolism, Journal Indexed in SCI-E, April 2020  
ANNALS OF INDIAN ACADEMY OF NEUROLOGY, Journal Indexed in SCI-E, August 2019  
EUROPEAN JOURNAL OF PEDIATRICS, Journal Indexed in SCI-E, June 2019  
Project Supported by Private Organizations in Other Countries, Telethon Foundation, Italy, June 2019

## **Tasks In Event Organizations**

Opladen T., Stevanovic G., Garcia Cazorla A., Bertoldi M., Kulhanek J., Yıldız Y., Conference on rare neurotransmitter diseases, Scientific Congress, Belgrade, Serbia, Ekim 2022

## **Scientific Research / Working Group Memberships**

International Working Group On Neurotransmitter Related Disorders, Hacettepe University, Türkiye, <http://intd-online.org/>, 2017 - Continues

## **Metrics**

Publication: 106

Citation (WoS): 173

Citation (Scopus): 376

H-Index (WoS): 8

H-Index (Scopus): 11

## **Invited Talks**

Conference on rare neurotransmitter diseases, Conference, International Network on Neurotransmitter-Related Disorders, Serbia, September 2022

## **Non Academic Experience**

Ankara Gülhane Eğitim ve Araştırma Hastanesi

ANKARA DR.SAMİ ULUS KADIN DOĞUM ÇOCUK SAĞLIĞI VE HASTALIKLARI EĞİTİM VE ARAŞTIRMA HASTANESİ