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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., ÜTİNİ G. E., ÇELİK H. T.
Molecular Syndromology, vol.15, no.1, pp.83-88, 2024 (SCI-Expanded)
- II. **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)
- III. **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)
- IV. **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., Haliloglu G.
European Journal of Paediatric Neurology, vol.49, pp.66-72, 2024 (SCI-Expanded)
- V. **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)
- VI. **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., Haliloglu G.
Molecular Genetics and Metabolism, vol.141, no.1, 2024 (SCI-Expanded)
- VII. **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)
- VIII. **A novel mutation in ATP13A2 gene in a patient with complicated hereditary spastic paraparesis 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)
- IX. **Preliminary evaluation of potential urinary organic acid biomarkers with mass spectrometry in children with autism spectrum disorder**
Sertoglu E., Balık 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)
- X. **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)
- XI. **COVID-19 in inherited metabolic disorders: Clinical features and risk factors for disease severity**

- KAHRAMAN A. B., YILDIZ Y., ÇIKI K., Erdal İ., AKAR H. T., DURSUN A., TOKATLI A., SİVRİ S.
Molecular Genetics and Metabolism, vol.139, no.2, 2023 (SCI-Expanded)
- XII. **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)
- XIII. **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)
- XIV. **The Türkiye-Syria Earthquake: a response from the editors of the Turkish Journal of Pediatrics**
DÜZOVA A., AKGÜL S., ÜTİNE G. E., YILDIZ Y.
Turkish Journal of Pediatrics, vol.65, no.1, pp.1-2, 2023 (SCI-Expanded)
- XV. **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., BAYRAKCİ B., Haliloglu G.
Neuromuscular Disorders, vol.32, no.11-12, pp.931-934, 2022 (SCI-Expanded)
- XVI. **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)
- XVII. **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)
- XVIII. **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)
- XIX. **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., Sıvri H. S., Karlı Oğuz H. K.
DIAGNOSTIC AND INTERVENTIONAL RADIOLOGY, vol.1, pp.1-18, 2022 (SCI-Expanded)
- XX. **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., Skrypnyk C., Iwanicka-Pronicka K., Piekutowska-Abramczuk D., et al.
HUMAN MUTATION, vol.43, no.3, pp.403-419, 2022 (SCI-Expanded)
- XXI. **Clinical Heterogeneity in MT-ATP6 Pathogenic Variants: Same Genotype-Different Onset**
Capiau 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)
- XXII. **Hyperinsulinism may be Underreported in Hypoglycemic Patients with Phosphomannomutase 2 Deficiency.**
Vuralli D., Yıldız Y., Ozon A., Dursun A., Gonc N., Tokatlı A., Sıvri H. S., Alikasifoglu A.
Journal of clinical research in pediatric endocrinology, vol.14, pp.275-286, 2022 (SCI-Expanded)
- XXIII. **Single Institutional Experience with GM1 Gangliosidosis: Clinical and Laboratory Results of 14 Patients.**
Akar H. T., Yıldız Y., Güvenkaya G., Çıklı K., Kahraman A. B., Erdal İ., Coşkun T., Dursun A., Sıvri H. S., Tokatlı A.
Balkan medical journal, vol.39, pp.345-350, 2022 (SCI-Expanded)

- XXIV. **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
 Çlkı K., Özgül R. K., Yıldız Y.
Klinische Padiatrie, 2022 (SCI-Expanded)
- XXV. **Biallelic mutations in ELFN1 gene associated with developmental and epileptic encephalopathy and joint laxity**
 DURSUN A., YALNIZOĞLU D., Yılmaz D. Y., Oguz K. K., GÜLBAKAN B., KOŞUKCU C., AKAR H. T., Kahraman A. B., Acar N. V., GÜNBET C., et al.
EUROPEAN JOURNAL OF MEDICAL GENETICS, vol.64, no.11, 2021 (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. **Perplexing Etiology of Hyperphenylalaninemia in an Infant Referred via Newborn Screening.**
 Çırkı K., Akar H. T., Özgül R. K., Gülbakan B., Yıldız Y.
Clinical chemistry, vol.67, no.10, pp.1428-1431, 2021 (SCI-Expanded)
- XXVIII. **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., Öztek 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)
- XXIX. **COVID-19-related anxiety in phenylketonuria patients**
 Akar H. T., Karaboncuk Y., Çırkı K., Kahraman A. B., Erdal İ., Coşkun T., Tokathlı A., Dursun A., Yıldız Y., Sivri H. S.
TURKISH JOURNAL OF PEDIATRICS, vol.63, no.5, pp.790-800, 2021 (SCI-Expanded)
- XXX. **Insights into the expanding phenotypic spectrum of inherited disorders of biogenic amines**
 Kuseyri Hübschmann O., Horvath G., Cortès-Saladelafont 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)
- XXXI. **DNACJ12 deficiency in patients with unexplained hyperphenylalaninemia: two new patients and a novel variant**
 Çırkı K., Yıldız Y., Yücel Yılmaz D., Pektaş E., Tokathlı A., Özgül R. K., Sivri H. S., Dursun A.
METABOLIC BRAIN DISEASE, vol.36, no.6, pp.1405-1410, 2021 (SCI-Expanded)
- XXXII. **Genotypic and phenotypic features in Turkish patients with classic nonketotic hyperglycinemia**
 Bayrak H., Yıldız Y., Olgaç A., Kasapkara Ç. S., Küçükcongar A., Zenciroğlu A., Yüksel D., Ceylaner S., Kılıç M.
METABOLIC BRAIN DISEASE, vol.36, no.6, pp.1213-1222, 2021 (SCI-Expanded)
- XXXIII. **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-Saladelafont E., Mercimek-Andrews S., Yıldız Y., Pons R., Kulhánek J., et al.
JOURNAL OF INHERITED METABOLIC DISEASE, vol.44, no.4, pp.1070-1082, 2021 (SCI-Expanded)
- XXXIV. **Recurrent Postinfectious Rhabdomyolysis in a 5-Year-Old Girl**
 Torun E., Yıldız Y., Yazıcı M., Çolak F., Kasapkara C.
KLINISCHE PADIATRIE, vol.233, no.04, pp.203-205, 2021 (SCI-Expanded)
- XXXV. **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)
- XXXVI. **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)
- XXXVII. **Oral health status of children and young adults with maple syrup urine disease in Turkey.**
Ballikaya E., Yıldız Y., Koç N., Tokatlı A., Uzamis Tekcicek M., Sıvrı H. S.
BMC oral health, vol.21, pp.8, 2021 (SCI-Expanded)
- XXXVIII. **Invisible burden of COVID-19: enzyme replacement therapy disruptions**
Kahraman A. B., Yıldız Y., Çırkı K., Akar H. T., Erdal İ., Dursun A., Tokatlı A., Sıvrı H. S.
JOURNAL OF PEDIATRIC ENDOCRINOLOGY & METABOLISM, vol.34, no.5, pp.539-545, 2021 (SCI-Expanded)
- XXXIX. **Creatine Transporter Deficiency Presenting as Autism Spectrum Disorder**
Yıldız Y., Göçmen R., Yaramış A., Coşkun T., Haliloglu G.
PEDIATRICS, vol.146, no.5, 2020 (SCI-Expanded)
- XL. **Predictors of acute metabolic decompensation in children with maple syrup urine disease at the emergency department**
YILDIZ Y., Akcan Yıldız 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)
- XLI. **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)
- 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. **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)
- XLIV. **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)
- XLV. **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)
- XLVI. **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)
- XLVII. **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)
- XLVIII. **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)
- 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. **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)

- L.I. **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)
- L.II. **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)
- L.III. **Cognitive and behavioral impairment in mild hyperphenylalaninemia**
Evinç S., Pektaş E., Foto-Özdemir D., Yıldız Y., Karaboncuk Y., Bilginer-Gürbüz B., Dursun A., Tokathlı A., Coskun T., Öktem F., et al.
TURKISH JOURNAL OF PEDIATRICS, vol.60, no.6, pp.617-624, 2018 (SCI-Expanded)
- L.IV. **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)
- L.V. **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)
- L.VI. **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)
- L.VII. **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)
- L.VIII. **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)
- L.IX. **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)
- L.X. **Periostin is temporally expressed as an extracellular matrix component in skeletal muscle regeneration and differentiation**
ÖZDEMİR C., Akpulat U., SHARAFI P., YILDIZ Y., ONBAŞILAR İ., Kocafe C.
GENE, vol.553, no.2, pp.130-139, 2014 (SCI-Expanded)
- LXI. **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., Tokathlı A.
CEN CASE REPORTS, vol.2023, 2023 (ESCI)
- II. **A Novel Double Homozygous BTD Gene Mutation in A Case of Profound Biotinidase Deficiency**
DEVECİ K., AKAR H. T., YILDIZ Y., ÖZGÜL R. K.
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- III. **Liver-Directed Adeno-Associated Virus\u2013Mediated 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., Yıldız Y., Molla G. K., Kılıç 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., akin 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. Kalitsal 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**
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- VIII. Lyme Hastalığı**
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- Yıldız Y., Sivri H. S., Other International Funding Programs, A Non-Interventional Observational Study of Pegvaliase-Naïve 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 li 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 ARCHIVES OF PEDIATRICS, Journal Indexed in ESCI, March 2024
JOURNAL OF PEDIATRIC ENDOCRINOLOGY AND METABOLISM, Journal Indexed in SCI-E, March 2024
PEDIATRICS, Journal Indexed in SCI-E, February 2024
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TURKISH JOURNAL OF PEDIATRICS, Journal Indexed in SCI-E, August 2023
JOURNAL OF PEDIATRIC ENDOCRINOLOGY AND METABOLISM, Journal Indexed in SCI-E, August 2023
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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
NOROPSİKIYATRI ARSIVİ-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, Turkey, <http://intd-online.org/>, 2017 - Continues

Metrics

Publication: 105

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 DR.SAMİ ULUS KADIN DOĞUM ÇOCUK SAĞLIĞI VE HASTALIKLARI EĞİTİM VE ARAŞTIRMA HASTANESİ
Ankara Gülhane Eğitim ve Araştırma Hastanesi