Asst. Prof. YILMAZ YILDIZ

Personal Information

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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 Üniversitesi, 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

Articles Published in Journals That Entered SCI, SSCI and AHCI Indexes

I. **Homozygous missense VPS16 variant is associated with a novel disease, resembling mucopolysaccharidosis-plus syndrome in two siblings**
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II. **DNACJ12 deficiency in patients with unexplained hyperphenylalaninemia: two new patients and a novel variant**
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METABOLIC BRAIN DISEASE, vol.36, no.6, pp.1405-1410, 2021 (Journal Indexed in SCI)

III. **Assessment of intellectual impairment, health-related quality of life, and behavioral phenotype in patients with neurotransmitter related disorders: Data from the iNTD registry**
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IV. **Complicated peripartum course in a patient with very long-chain acyl-coenzyme A dehydrogenase (VLCAD) deficiency**
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V. **Genotypic and phenotypic features in Turkish patients with classic nonketotic hyperglycinemia**
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VI. **Safety and Efficacy of Liver-Directed Gene Therapy in Patients with Mucopolysaccharidosis Type VI**
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MOLECULAR THERAPY, vol.29, no.4, pp.114, 2021 (Journal Indexed in SCI)

VII. **Oral health status of children and young adults with maple syrup urine disease in Turkey.**
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BMC oral health, vol.21, pp.8, 2021 (Journal Indexed in SCI)

VIII. **Invisible burden of COVID-19: enzyme replacement therapy disruptions**
Kahraman A. B. , Yıldız Y., Çıkı K., Akar H. T. , Erdal İ., Dursun A., Tokatlı A., Sivri H. S.
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IX. **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., Yildiz Y., Pons R., Kulhánek J., et al.
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X. **Creatine Transporter Deficiency Presenting as Autism Spectrum Disorder**
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XI. **Recurrent Postinfectious Rhabdomyolysis in a 5-Year-Old Girl**
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KLINISCHE PADIATRIE, 2020 (Journal Indexed in SCI)

XII. **A Rare Cause of Hyperinsulinemic Hypoglycemia: Kabuki Syndrome**
Mısırlıgil M., Yıldız Y., Akın O., Odabaşı G., Arslan M., Ünay B.
Journal of clinical research in pediatric endocrinology, 2020 (Journal Indexed in SCI Expanded)

XIII. **Predictors of acute metabolic decompensation in children with maple syrup urine disease at the emergency department**
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EUROPEAN JOURNAL OF PEDIATRICS, vol.179, no.7, pp.1107-1114, 2020 (Journal Indexed in SCI)

XIV. **Successful management of acute pancreatitis due to apolipoprotein C-II deficiency in a 37-day-old infant**
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XV. **Retrospective evaluation of 85 patients with urea cycle disorders: one center experience, three new mutations**
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XVI. **Consensus guideline for the diagnosis and treatment of tetrahydrobiopterin (BH4) deficiencies**
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ORPHANET JOURNAL OF RARE DISEASES, vol.15, no.1, 2020 (Journal Indexed in SCI)

XVII. **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.
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XVIII. **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 (Journal Indexed in SCI)

XIX. **Genotypes and estimated prevalence of phosphomannomutase 2 deficiency in Turkey differ significantly from those in Europe**
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AMERICAN JOURNAL OF MEDICAL GENETICS PART A, vol.182, no.4, pp.705-712, 2020 (Journal Indexed in SCI)

XX. **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 (Journal Indexed in SCI)

XXI. **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 (Journal Indexed in SCI)

XXII. **Inborn errors of metabolism in the differential diagnosis of fatty liver disease**
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TURKISH JOURNAL OF GASTROENTEROLOGY, vol.31, no.1, pp.3-16, 2020 (Journal Indexed in SCI)

XXIII. **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 (Journal Indexed in SCI)

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XXV. **Maternal phenylketonuria in Turkey: outcomes of 71 pregnancies and issues in management**
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EUROPEAN JOURNAL OF PEDIATRICS, vol.178, no.7, pp.1005-1011, 2019 (Journal Indexed in SCI)

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TURKISH JOURNAL OF PEDIATRICS, vol.60, no.6, pp.617-624, 2018 (Journal Indexed in SCI)

XXVII. **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 (Journal Indexed in SCI)

XXVIII. **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 (Journal Indexed in SCI)

XXIX. **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 (Journal Indexed in SCI)

XXX. **Hereditary Dopamine Transporter Deficiency Syndrome: Challenges in Diagnosis and Treatment**
YILDIZ Y., Pektas E., TOKATLI A., Haliloglu G.
NEUROPEDIATRICS, vol.48, no.1, pp.49-52, 2017 (Journal Indexed in SCI)

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GYNECOLOGICAL ENDOCRINOLOGY, vol.33, no.1, pp.19-20, 2017 (Journal Indexed in SCI)

XXXII. **Late-diagnosed phenylketonuria in an eight-year-old boy with dyslexia and attention-deficit hyperactivity disorder**
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TURKISH JOURNAL OF PEDIATRICS, vol.58, no.1, pp.94-96, 2016 (Journal Indexed in SCI)

XXXIII. **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 (Journal Indexed in SCI)

XXXIV. **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 (Journal Indexed in SCI)

Articles Published in Other Journals

I. **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 (Other Refereed National Journals)

II. **Mukopolisakkaridozlarda ortopedik sorunlar**
YILDIZ Y., SİVRİ H. S.
TOTBİD Dergisi, vol.15, no.4, pp.303-310, 2016 (Other Refereed National Journals)

III. **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 (Other Refereed National Journals)

Books & Book Chapters

I. Yıldız Y., Sivri H. S.
in: , Kalbiye Yalaz, Editor, Hipokrat Kitabevi, Ankara, pp.343-351, 2021

II. **Disorders of Amino Acid Metabolism**
Yıldız Y., Kılıç M.
in: Pediatrics, Zülfikar Akelma, Editor, Nobel Tıp Kitapevi, Ankara, pp.1124-1129, 2021

III. **Acute Metabolic Crisis**
Yıldız Y., Haliloğlu V. G.
in: Handbook of Clinical Acute Pediatric Neurology, Deniz Yüksel, Editor, Nobel Tıp Kitapevi, Ankara, pp.57-65,

2020

IV. **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

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

VI. **Lyme Hastalığı**
Yıldız Y.
in: Neonatoloji El Kitabı, Yurdakök,Murat, Editor, Güneş Tıp Kitabevleri, Ankara, pp.683-685, 2014

VII. **Çocuğa Kötü Muamele: İhmalden İstismara**
Yıldız Y.
in: Rudolph Pediatri, Yurdakök,Murat, Editor, Güneş Tıp Kitabeleri, Ankara, pp.137-143, 2013

Supported Projects

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 - Continues

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

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, England

Scientific Refereeing

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

Cukurova Medical Journal, Journal Indexed in ESCI, June 2021

TURKISH JOURNAL OF PEDIATRICS, Journal Indexed in SCI-E, June 2021

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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

Scientific Research / Working Group Memberships

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

Citations

Total Citations (WOS):116
h-index (WOS):6