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International Researcher IDs

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Publons / Web Of Science ResearcherID: AAM-2081-2021

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Yoksis Researcher ID: 122033



Learning Knowledge

Post Doctorate of Medicine
2005 - 2010

Gazi University, Tıp Fakültesi, Dahili Tıp Bilimleri, Turkey

Expertise In Medicine
1999 - 2004

Gazi University, Tıp Fakültesi, Dahili Tıp Bilimleri, Turkey

Undergraduate
1992 - 1998

Istanbul University, Istanbul Medical Faculty, İstanbul Tıp Pr., Turkey

Certificates, Courses and Trainings

Health&Medicine, Yeni Nesil Dizi Analiz Kursu, Pediatric Genetic Diseases Association, 2023

Vocational Training, Eğiticilerin Eğitimi Kursu (Training Course for Trainers), Gazi University, 2014

Data Analysis, Temel Biyoistatistik Kursu (Basic Biostatistics Course), Gazi University, 2010

Health&Medicine, Deney Hayvanları Uygulama ve Etik Kursu (Experimental Animals and Ethics Course), Gazi University, 2007

Dissertations

Post Doctorate of Medicine, İdiopatik juvenil osteoporozu olan hastalarda COL1A1, ESR1, VDR, TGFB1, LRP5 ve LRP6 genlerindeki polimorfizmlerin araştırılması, Gazi University, Tıp Fakültesi, Dahili Tıp Bilimleri, 2009

Expertise In Medicine, Akut romatizmal ateşin etiyopatogenezinde viral etkenlerin rolü, Gazi University, Tıp Fakültesi, Dahili Tıp Bilimleri, 2004

Academic Titles / Tasks

Professor
2021 - Continues

Gazi University, Tıp Fakültesi, Dahili Tıp Bilimleri

Associate Professor
2014 - 2021

Gazi University, Tıp Fakültesi, Dahili Tıp Bilimleri

Expert
2014 - 2014

Gazi University, Tıp Fakültesi, Dahili Tıp Bilimleri

Lecturer
2009 - 2011

Gazi University, Tıp Fakültesi, Dahili Tıp Bilimleri

Supported Projects

1. EZGÜ F. S., OKUR İ., TÜMER L., BİBEROĞLU G., ÖKTEM R. M., BAKKALOĞLU EZGÜ S. A., İNCİ A., ERGİN F. B., İNAL T., Project Supported by Higher Education Institutions, PMVK geni fonksiyonunun araştırılması ve aynı gen üzerindeki DNA varyantlarının etkilerinin belirlenmesi, 2023 - Continues
2. EZGÜ F. S., İNAL T., TÜMER L., ARHAN E., KILIÇ A., DEMİREL S., ÖKTEM R. M., İNCİ A., SERDAROĞLU E., ERGİN F. B., et al., Project Supported by Higher Education Institutions, GALNT3 Gen Mutasyonları Sonucu Kaybedilmiş Olan Protein Fonksiyonların Adeno-Asosiyel Virus Aracılı Gen Transferi ile İn Vitro Olarak Yeniden Kazandırılması, 2023 - Continues
3. TÜMER L., OKUR İ., EZGÜ F. S., ALTUN A. N., İNCİ A., EKMEKÇİ ERTEK İ., ÖKTEM R. M., BİBEROĞLU G., Project Supported by Higher Education Institutions, Psikozyoz kliniğiyle başvuran hastaların kalıtsal metabolik hastalıklar açısından taranması, 2023 - Continues
4. Ezgü F. S., Tümer L., İnci A., Okur İ., Industrial Organizations of Other Countries Supported Project, Enzim replasman Tedavisi (ERT) ile terapötik hedeflere ulaşmış Gaucher Tip 3 (GD3) hastalığı olan erişkin ve pediatrik hastalarda venglustatin etkililiğini ve güvenilirliğini değerlendirmek için faz 3, çok merkezli, çok uluslu, randomize, çift kör, çift sağır, çift plasebolu, aktif karşılaştırmalı çalışma (LEAP2MONO), 2024 - 2027
5. Okur İ., Ezgü F. S., Tümer L., İnci A., Şıvgın V., Arhan E., Project Supported by Private Organizations in Other Countries, A Phase 3B/4 Open-Label Multicenter Study Extension Study to Further Evaluate Safety, Tolerability and Efficacy of Intracerebroventricular AX 250 Treatment in Mucopolysaccharidosis Type IIIB (MPS IIIB, Sanfilippo Syndrome Type B) Patients-AX250-401 (ClinicalTrials.gov ID NCT05492799), 2023 - 2027
6. Ezgü F. S., Tümer L., Okur İ., İnci A., Industrial Organizations of Other Countries Supported Project, Fenilketonüri erişkin gönüllülerde insan fenilalanin hidrokisilazın adeno ilişkili viral vektör aracılı gen transferi olan SAR444836'nın güvenilirliğini, tolerabilitesini ve etkililiğini değerlendirmek için Faz 1/2, açık etiketli, doz artırma ve doz genişletme çalışması, 2023 - 2027
7. Ezgü F. S., Tümer L., Okur İ., İnci A., Industrial Organizations of Other Countries Supported Project, Nöronopatik ve Nöronopatik Olmayan Mukopolisakkaridoz Tip II Pediatrik Katılımcılarda DNL310'un İdrosülfaza Karşı Etkililiğini ve Güvenliliğini Belirlemek İçin Çok merkezli, çift-Kör, Randomize Bir Faz 2/3 Çalışma, 2023 - 2027
8. Ezgü F. S., Tümer L., Okur İ., İnci A., Industrial Organizations of Other Countries Supported Project, Osteogenesis Imperfecta'lı gönüllülerde Setrusumab'ın etkililiğini ve güvenilirliğini değerlendiren bir faz 2 tek kör doz değerlendirme fazı ile bir faz 3 çift kör plasebo kontrollü fazdan oluşan operasyonel olarak kesintisiz, randomize faz 2/3 çalışma, 2023 - 2026
9. Ezgü F. S., Tümer L., Okur İ., İnci A., Industrial Organizations of Other Countries Supported Project, Uzun zincirli yağ asidi oksidasyon bozukluğu (LC-FAOD) olan pediatrik hastalardaki majör klinik olaylarda çift zincirli orta zincirli trigliseridlerle karşılaştırıldığında triheptanoin'in etkisinin belirlendiği çok merkezli çift kör randomize çalışma, 2023 - 2026

10. Ezgü F. S., Tümer L., Okur İ., İnci A., Börcek A. Ö., Şıvgın V., Özger İlhan S., Industrial Organizations of Other Countries Supported Project, Study of Safety, Tolerability and Efficacy of PBGM01 in Pediatric Subjects With GM1 Gangliosidosis (Imagine-1) (Phase 1 and 2) (ClinicalTrials.gov Identifier: NCT04713475), 2022 - 2026
11. Tümer L., Ezgü F. S., Okur İ., İnci A., Industrial Organizations of Other Countries Supported Project, Fabry Hastalığı olan gönüllülerde Migalastat hidroklorür monoterapisinin uzun süreli güvenliliği ve etliliğini değerlendiren açık etiketli uzatma çalışması, 2015 - 2026
12. İnci A., Tümer L., Okur İ., Ezgü F. S., Atalay H. T., Other International Funding Programs, Fabry hastalığı ve sol ventrikülerhipertrofisi olan gönüllülerde normal bakım standardıyla karşılaştırılmalı olarak venglustat'ın solventriküler kitle indeksi üzerindeki etkisini değerlendirmek için randomize, açık etiketli, paralel gruplu, 18 aylık bir Faz 3 çalışma, 2023 - 2025
13. Okur İ., Tümer L., İnci A., Ezgü F. S., Industrial Organizations of Other Countries Supported Project, Fenilketonürde PTC923-MD-PKU Faz 3 Açık Etiketli Uzatma Çalışması, 2022 - 2025
14. İnci A., Ezgü F. S., Tümer L., Okur İ., Other International Funding Programs, Aromatik L aminoacid dekarboksilaz eksikliği tanısı olan hastaların gerçek yaşam verilerini içeren uluslararası gözlemsel bir çalışma, 2022 - 2025
15. Ezgü F. S., Okur İ., İnci A., Tümer L., Arhan E., Soysal Acar A. Ş., Industrial Organizations of Other Countries Supported Project, Venglustatın geç başlangıçlı GM2 gangliosidoz (Tay-Sachs hastalığı ve Sandhoff hastalığı) ile aynı bir kolda juvenil/adolesan geç başlangıçlı GM2 gangliosidozda ve, aynı ve benzer glukozilseramid bazlı sfingolipid yolağı içindeki ultra-nadir hastalıklardaki etkinlik, farmakodinamik, farmakokinetik, güvenlilik ve tolere edilebilirliğini değerlendirmek için çok merkezli, uluslararası, randomize, çift-kör, plasebo kontrollü bir çalışma, EFC15299, 2021 - 2025
16. Okur İ., Şıvgın V., Tümer L., İnci A., Soysal Acar A. Ş., Arhan E., Börcek A. Ö., Yıldırım Gökay N., Gündüz B., Ezgü F. S., Project Supported by Private Organizations in Other Countries, BMN (AX) 250-202--A Multicenter, Multinational, Extension Study to Evaluate the Long Term Safety and Efficacy of Intracerebroventricular AX 250 in Patients With Mucopolysaccharidosis Type IIIB (MPS IIIB, Sanfilippo Syndrome Type B) (Phase 2) (ClinicalTrials.gov Identifier: NCT03784287), 2018 - 2025
17. Okur İ., Arhan E., İnci A., Tümer L., Ezgü F. S., Other International Funding Programs, En az 6 Aydır Tedavi Almamış veya Hiç Tedavi Edilmemiş Fabry Hastalığı Bulunan Erkek ve Kadın Yetişkinlerde Venglustat'ın Nöropatik Ağrı ve Karın Ağrısı üzerindeki Etkisini Değerlendiren Randomize, Çift Kör, Plasebo Kontrollü, 12 Aylık Faz 3 Çalışma - PERIDOT, 2023 - 2024
18. Okur İ., Ezgü F. S., Tümer L., Şıvgın V., Kurtipek Ö., Soysal Acar A. Ş., İnci A., Arhan E., Universities of Other Countries Supported Project, Natural History Study of Infantile and Juvenile GM1 Gangliosidosis (GM1) Patients (ClinicalTrials.gov Identifier: NCT04041102), 2021 - 2023
19. Tümer L., Ezgü F. S., İnci A., Okur İ., Industrial Organizations of Other Countries Supported Project, AT-NIS-001 A study to describe the experience of both patients and their clinicians in the treatment of Fabry Disease with Enzyme, 2021 - 2022
20. Özger İlhan S., Ezgü F. S., Okur İ., Tümer L., Industrial Organizations of Other Countries Supported Project, An Open-label, Ascending Multiple-dose Study to Evaluate Safety, Tolerability, Pharmacokinetics, and Pharmacodynamics of Romosozumab in Children and Adolescents With Osteogenesis Imperfecta, 2020 - 2022
21. Özger İlhan S., Ezgü F. S., Okur İ., Tümer L., Project Supported by Private Organizations in Other Countries, PEDIATRİK MPS IIIA HASTALARDA UZUN SÜRELİ SOBIO03 TEDAVİSİNİN GÜVENLİLİĞİNİ, TOLERABİLİTESİNİ VE ETKİLİLİĞİNİ DEĞERLENDİREN AÇIK, TEK KOLLU, ÇOK MERKEZLİ BİR ÇALIŞMA , 2019 - 2022
22. Okur İ., Ezgü F. S., İnci A., Tümer L., Industrial Organizations of Other Countries Supported Project, An Efficacy and Safety Study of Alirocumab in Children and Adolescents With Heterozygous Familial Hypercholesterolemia, 2018 - 2022
23. Okur İ., Ezgü F. S., Tümer L., İnci A., Şıvgın V., Soysal Acar A. Ş., Kurtipek Ö., Tutar H., Gündüz B., Industrial Organizations of Other Countries Supported Project, BMN (AX) 250-902-A Prospective Natural History Study of Mucopolysaccharidosis Type IIIB (MPS IIIB), 2017 - 2022
24. Ezgü F. S., Tümer L., İnci A., Okur İ., Industrial Organizations of Other Countries Supported Project, A Phase 2b/3 Prospective, Randomized, Double-Blind, Sham-Controlled 3-Part Trial of VTS-270 (2-hydroxypropyl-β-cyclodextrin) in Subjects With Neurologic Manifestations of Niemann-Pick Type C1 (NPC1) Disease (ClinicalTrials.gov Identifier: NCT02534844), 2016 - 2022

25. Okur İ., Ezgü F. S., Tümer L., İnci A., Industrial Organizations of Other Countries Supported Project, K020-218--- Standart Bakımda Yeterince Kontrol Altına Alınmayan Bir Üre Döngüsü Bozukluğu Olan Gönüllülerde KB195'in Etkinliğinin ve Güvenliğinin Değerlendirildiği Faz 2, Açık Etiketli Çalışma, 2021 - 2021
26. Okur İ., Ezgü F. S., DüNDAR H., Tümer L., TÜBİTAK Project, Metilmalonil Koenzim A Mutaz Enziminin Konformasyonel Bozukluğuna Bağlı Olarak Gelişen Metilmalonik Asidemi Hastalığının Farmakolojik Şaperon Uygulaması İle Enzim Aktivitesinin Yeniden Kazandırılması, 2017 - 2021
27. Okur İ., Ezgü F. S., Tümer L., Özger İlhan S., Şıvgın V., Soysal Acar A. Ş., İnci A., Project Supported by Private Organizations in Other Countries, An Open, Non-controlled, Parallel, Ascending Multiple-dose, Multicenter Study to Assess Safety and Tolerability, Pharmacokinetics and Pharmacodynamics of SOBI003 in Pediatric MPS IIIA Patients (SOBI003-001) , 2018 - 2020
28. Okur İ., Börcek A. Ö., Ezgü F. S., Tümer L., İnci A., Soysal Acar A. Ş., Arhan E., Tutar H., Gündüz B., Project Supported by Private Organizations in Other Countries, A Phase 1/2 Open-Label Dose-Escalation Study to Evaluate the Safety, Tolerability, Pharmacokinetics and Efficacy of Intracerebroventricular BMN 250 in Patients with Mucopolysaccharidosis Type IIIB (MPS IIIB, Sanfilippo Syndrome Type B) (BMN250-201) <https://clinicaltrials.gov/ct2/show/NCT02754076?term=BMN250draw=2rank=2>, 2017 - 2020
29. Okur İ., Ezgü F. S., Tümer L., Soysal Acar A. Ş., Şıvgın V., Kurtipek Ö., Gündüz B., İnci A., Tutar H., Arhan E., Other International Funding Programs, BMN250-901-A Study of Mucopolysaccharidosis Type IIIB MPS IIIB (Mukopolisakkaridoz tip IIIB'xxye yönelik uluslararası çok merkezli, prospektif gözlemsel çalışma)-(BMN250-901 nolu proje kodu), 2015 - 2020
30. Özaslan A., Güney E., Ergün M. A., Okur İ., Project Supported by Higher Education Institutions, CDH13 and LPHN3 Gene Polymorphisms in Attention Deficit Hyperactivity Disorder: Their Relation With Clinical Characteristics and Executive Functions, 2018 - 2019
31. Ezgü F. S., Tümer L., İnci A., Okur İ., Industrial Organizations of Other Countries Supported Project, Hipofosfatazyalı Hastaların Gözlemsel, Boylamsal ve İleriye Dönük, Uzun Süreli Kayıt Çalışması, 2018 - 2019
32. Okur İ., Ezgü F. S., Tümer L., İnci A., Project Supported by Private Organizations in Other Countries, HAUSER-OLE--- Heterozigot Ailevi Hiperkolesterolemisi (HeAH) veya Homozigot Ailevi Hiperkolesterolemisi (HoFH) Olan 10 ve 17 yaşları arasındaki pediatrik Gönüllülerde LDK-K ' nin azaltılmasında diyet ve lipid düşürücü tedaviye ek olarak Evolocumabın güvenliği tolere edilebilirliği ve etkinliğini değerlendiren açık etiketli, tek kollu, çok merkezli çalışma (20120214 protokol numaralı çalışma), 2016 - 2019
33. Okur İ., Ezgü F. S., Tümer L., Other International Funding Programs, HAUSER-RCT Heterozigot Ailevi Hiperkolesterolemisi (HeAH) Olan 10 ve 17 yaşlarındaki pediatrik Gönüllülerde Düşük Yoğunluklu Lipoprotein-Kolesterol (LDL-C)'ün azaltılmasında diyet ve lipid düşürücü tedaviye ek olarak 24 haftalık Evolocumabın uygulamasının etkililiği, güvenliliği ve tolere edilebilirliği belirlemek amaçlı çift kör, randomize, çok merkezli, plasebo kontrollü, paralel grup çalışması (20120123 protokol numaralı), 2016 - 2019
34. OKUR İ., TÜMER L., Project Supported by Higher Education Institutions, MOLEKÜLER YADA ENZİMATİK ANALİZ İLE MUKOPOLİSAKKARİDOZ TANISI ALMIŞ OLGULARDA BÖBREK VE ÜRİNER İSTEM TUTULUMUNUN ARAŞTIRILMASI, 2015 - 2016
35. Okur İ., Project Supported by Other Official Institutions, Screening for Fabry disease in patients undergoing hemodialysis and peritoneal dialysis due to chronic renal failure., 2012 - 2013

Awards

1. Okur İ., Ezgü F. S., Biberoglu G., Ankara il merkezinde dializ hastalarında tarama yöntemi ile Fabry hastalığının belirlenmesi ve aile taramasının önemi, 55. Türkiye Milli Pediatri Kongresi (Sözel Bildiri İkincilik Ödülü), Türkiye Milli Pediatri Derneği, October 2011

Jury Memberships

Appointment to Academic Staff-Assistant Professorship, Appointment to Academic Staff-Assistant Professorship,

Çukurova Üniversitesi, March, 2024

Associate Professor Exam, Associate Professor Exam, T.C. Sağlık Bakanlığı, Manisa Şehir Hastanesi, December, 2023

Appointment to Academic Staff-Assistant Professorship, Appointment to Academic Staff-Assistant Professorship, Lokman Hekim Üniversitesi, November, 2023

Associate Professor Exam, Associate Professor Exam, Marmara Üniversitesi, October, 2023

Associate Professor Exam, Associate Professor Exam, Çukurova Üniversitesi, October, 2023

Expertise In Medicine, Expertise In Medicine, Gazi Üniversitesi, June, 2023

Appointment to Academic Staff-Assistant Professorship, Appointment to Academic Staff-Assistant Professorship, Selçuk Üniversitesi, January, 2023

Appointment to Academic Staff-Professorship, Appointment to Academic Staff-Professorship, Sağlık Bilimleri Üniversitesi, January, 2023

Associate Professor Exam, Associate Professor Exam, Eskişehir Osmangazi Üniversitesi, October, 2022

Appointment to Academic Staff-Assistant Professorship, Appointment Academic Staff, Başkent Üniversitesi, May, 2022

Associate Professor Exam, Associate Professor Exam, Hacettepe University, March, 2022

Associate Professor Exam, Associate Professor Exam, Health Sciences University, March, 2022

Taught Courses And Trainings

İlhan M. N., Okur İ., Güzel Tunçcan Ö., Özkan S., Özger İlhan S., Baran Aksakal F. N., Kula S., Erten Y., İnan N., Bozdayı G., et al., Tıp Fakültesi Eğitici Eğitimi, 2023 - 2023

Okur İ., Training of Trainers Course, 2021 - 2022

Okur İ., Training of Trainers Course, 2022 - 2022

Designed Courses And Trainings

Okur İ., Özkan S., Özger İlhan S., Güzel Tunçcan Ö., Bozdayı G., Kula S., Gazi University Faculty of Medicine Training of Trainers, February 2023

Özger İlhan S., Özkan S., Okur İ., Güzel Tunçcan Ö., Kula S., İnan N., Atan A., Ekmekci Ertek İ., Bozdayı G., Baran Aksakal F. N., Faculty of Medicine Training of Trainers, February 2022

Özkan S., Okur İ., Güzel Tunçcan Ö., Uluoğlu C., Özger İlhan S., Baran Aksakal F. N., Erten Y., Ergün M. A., Küçük Biçer B., Ekmekci Ertek İ., Gazi University Faculty of Medicine Training of Trainers, December 2021

Research Infrastructure Information

Okur İ., Ezgü F. S., Tümer L., İnci A., Özger İlhan S., Şıvgın V., Biberoglu G., Phase 1 Clinical Research Center, January 2018

Published journal articles indexed by SCI, SSCI, and AHCI

- A very rare presentation of mitochondrial elongation factor Tu deficiency-TUFM mutation and literature review**
GÖKALP S., İNCİ A., KILIÇ A., Ozsaydi E., ALTUN A. N., DEMİR F., ERGİN F. B., Ozbek M. N., OKUR İ., EZGÜ F. S., et al.
Journal of Pediatric Endocrinology and Metabolism, vol.37, no.6, pp.571-574, 2024 (SCI-Expanded)
- Endocrinological and metabolic profile of Gaucher disease patients treated with enzyme replacement therapy**
KILIÇ A., Emecen Sanli M., Ozsaydi Aktasoglu E., GÖKALP S., BİBEROĞLU G., İnci A., OKUR İ., EZGÜ F. S., TÜMER L.
Journal of Pediatric Endocrinology and Metabolism, vol.37, no.5, pp.413-418, 2024 (SCI-Expanded)
- Pterin Profiling in Serum, Dried Blood Spot, and Urine Samples Using LC-MS/MS in Patients with Inherited Hyperphenylalaninemia**

Öktem R. M., İnci A., BAYRAK H., DEMİR F., BİBEROĞLU G., Maviş M. E., Gürsu G. G., Yılmaz H., OKUR İ., EZGÜ F. S., et al.

Molecular Syndromology, vol.15, no.3, pp.185-193, 2024 (SCI-Expanded)

4. **Long-Term Experience with Anaphylaxis and Desensitization to Alglucosidase Alfa in Pompe Disease**
Karagol H. I. E., İnci A., Terece S. P., Kılıç A., Demir F., Yapar D., Köken G., Okur İ., Ezgü F. S., Tümer L., et al.
International Archives of Allergy and Immunology, vol.184, no.4, pp.370-375, 2023 (SCI-Expanded)
5. **A phase 1/2 study on intracerebroventricular tralesenidase alfa in patients with Sanfilippo syndrome type B.**
Muschol N., Koehn A., Von Cossel K., Okur İ., Ezgu F. S., Harmatz P., De Castro Lopez M. J., Couce M. L., Lin S., Batzios S., et al.
The Journal of clinical investigation, vol.133, 2023 (SCI-Expanded)
6. **Endocrinological, immunological and metabolic features of patients with Fabry disease under therapy**
Emecen Sanli M., Kılıç A., İnci A., Okur İ., Ezgü F. S., Tümer L.
Journal of Pediatric Endocrinology and Metabolism, 2023 (SCI-Expanded)
7. **A possibly new autoinflammatory disease due to compound heterozygous phosphomevalonate kinase gene mutation**
Yıldız Ç., Gezgin Yıldırım D., İnci A., Tümer L., Ergin F. B., Sunar Yayla E. N. S., Esmeray Şenol P., Karaçayır N., Eğritaş Gürkan Ö., Okur İ., et al.
Joint Bone Spine, vol.90, no.1, 2023 (SCI-Expanded)
8. **Longitudinal Natural History of Pediatric Subjects Affected with Mucopolysaccharidosis IIIB**
Okur İ., Ezgu F. S., Giugliani R., Muschol N., Koehn A., Amartino H., Harmatz P., De Castro Lopez M. J., Couce M. L., Lin S., et al.
Journal of Pediatrics, vol.249, pp.50, 2022 (SCI-Expanded)
9. **Assessment of auditory functions in patients with hepatic glycogen storage diseases**
ŞANLI M. E., YILDIRIM GÖKAY N., TUTAR H., GÜNDÜZ B., ÖZSAYDI AKTAŞOĞLU E., KILIÇ A., İNCİ A., OKUR İ., EZGÜ F. S., TÜMER L.
TURKISH JOURNAL OF PEDIATRICS, vol.64, no.4, pp.658-670, 2022 (SCI-Expanded)
10. **Expected or unexpected clinical findings in liver glycogen storage disease type IX: distinct clinical and molecular variability**
İnci A., Kılıç Yıldırım G., Cengiz Ergin F. B., Sarı S., Eğritaş Gürkan Ö., Okur İ., Biberöğlü G., Bükülmez A., Ezgü F. S., Dalgıç B., et al.
JOURNAL OF PEDIATRIC ENDOCRINOLOGY & METABOLISM, vol.35, no.4, pp.451-462, 2022 (SCI-Expanded)
11. **Fructose 1,6 bisphosphatase deficiency: outcomes of patients in a single center in Turkey and identification of novel splice site and indel mutations in FBP1**
ŞANLI M. E., Cengiz B., Kilic A., Ozsaydi E., Inci A., Okur İ., Tumer L., Lebigot E., Ezgu F. S.
JOURNAL OF PEDIATRIC ENDOCRINOLOGY & METABOLISM, vol.35, no.4, pp.497-503, 2022 (SCI-Expanded)
12. **An ultra-rare cause of severe hypotonia mimicking Pompe disease in an infant: RRM2B related mitochondrial DNA depletion syndrome with a novel mutation**
İNCİ A., OKUR İ., DEMİR E., BİBEROĞLU G., TÜMER L., SERDAROĞLU A., EZGÜ F. S.
NEUROLOGY ASIA, vol.27, no.1, pp.199-202, 2022 (SCI-Expanded)
13. **First successful concomitant therapy of immune tolerance induction therapy and desensitization in a CRIM-negative infantile Pompe patient**
Sanli M. E., ERTOY KARAGÖL H. İ., KILIÇ A., Aktasoglu E., İNCİ A., OKUR İ., Ezgu F. S., TÜMER L.
JOURNAL OF PEDIATRIC ENDOCRINOLOGY & METABOLISM, vol.35, no.2, pp.273-277, 2022 (SCI-Expanded)
14. **The first case with FBXL4 mutation successfully treated with a parenteral ketogenic diet for lactic acidosis**
İNCİ A., Aktas E., Cengiz Ergin F. B., OKUR İ., BİBEROĞLU G., EZGÜ F. S., TÜMER L.
JOURNAL OF PARENTERAL AND ENTERAL NUTRITION, vol.45, no.8, pp.1788-1792, 2021 (SCI-Expanded)
15. **Clinical and event-based outcomes of patients with mucopolysaccharidosis VI receiving enzyme replacement therapy in Turkey: a case series**

İnci A., Okur İ., Tümer L., Biberöğlü G., Öktem M., Ezgü F. S.

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98. **LAL enzim eksikliği**
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99. **Fabry Hastalarında Subklinik Sol Ventrikül Disfonksiyonunun Speckle Tracking Ekokardiyografi ile Değerlendirilmesi**
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106. **Sol çenede Lenfatik tutulum ile giden Niemann Pick tip C olgusu**

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107. **Lizozomal depo hastalıklarına yaklaşım**

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108. **Importance of family screening in Fabry disease: Reaching the bottom of the iceberg**

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109. **COBALAMIN C DEFICIENCY WITH INFANTILE SPASM AND CUTANEOUS FINDINGS A UNIQUE CASE**

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112. **PREVALENCE OF FABRY DISEASE AMONG HEMODIALYSIS PATIENTS IN TURKEY**

Okur İ., BİBEROĞLU G., Ezgu F. S., TÜMER L., Hasanoglu A., Bicik Z., Akin Y., Mumcuoglu M., Ecder T.

50th European-Renal-Association - European-Dialysis-and-Transplant-Association Congress, İstanbul, Turkey, 18 - 21 May 2013, vol.28, pp.321

113. **Three siblings with ext1 CDG**

EZGÜ F. S., KASAPKARA Ç., OKUR İ., KÜÇÜKÇONGAR A., TÜMER L., OKUR A., SARAÇ A., WUYTS W., HUL E. V., HASANOĞLU A.

Annual Symposium of the Society for the Study of Inborn Errors of Metabolism, Switzerland, 30 August - 02 September 2011

114. **TWO NOVEL MUTATIONS IN TWO PATIENTS WITH MEDIUM-CHAIN ACYL-CoA DEHYDROGENASE DEFICIENCY**

Hasanoglu A., Okur İ., Largiader C., Biberoglu G., Tumer L., Eminoglu F. T., EZGÜ F. S.

11th International Conference of Inborn Errors of Metabolism, California, United States Of America, 29 August - 02 September 2009, vol.98, pp.52

115. **Gazi Üniversitesi Tıp Fakültesi Sağlık Çocuk Polikliniğinde izlenen 4 6 aylık bebeklerde demir eksikliği anemisinin sıklığı**

ŞAHİN DAĞLI F., OKUR A., OKUR İ., DUYAN ÇAMURDAN A., BEYAZOVA U.

Pediatri Günleri-II, Kırıkkale, Turkey, 11 May 2004

Academic and Administrative Experience

2019 - Continues	Ethics Committee Member	Gazi University, Rektörlük
2020 - 2023	Vice Dean	Gazi University, Tıp Fakültesi
2018 - 2021	Faculty Board Member	Gazi University, Tıp Fakültesi
2018 - 2021	Faculty Executive Board Member	Gazi University, Tıp Fakültesi

2019 - 2020	Clinical Research Ethics Committee Member	Gazi University, Tıp Fakültesi, Dahili Tıp Bilimleri
2015 - 2020	Education Coordinator (Year III)	Gazi University, Tıp Fakültesi

Courses

Pediatric Metabolism Minor Specialization Training, Expertise In Medicine, 2023 - 2024, 2022 - 2023, 2021 - 2022, 2020 - 2021, 2019 - 2020, 2018 - 2019, 2017 - 2018, 2016 - 2017, 2015 - 2016, 2014 - 2015

Clinical Applied Practice Training, Undergraduate, 2023 - 2024, 2022 - 2023, 2021 - 2022, 2020 - 2021, 2019 - 2020, 2018 - 2019, 2017 - 2018, 2016 - 2017

When Should We Consider Metabolic Disease?, Undergraduate, 2023 - 2024, 2022 - 2023, 2021 - 2022

Vitamin and Mineral Disorders, Undergraduate, 2023 - 2024, 2022 - 2023, 2021 - 2022

Intoduction to Nutrition, Undergraduate, 2021 - 2022, 2020 - 2021, 2019 - 2020, 2018 - 2019, 2017 - 2018, 2016 - 2017, 2015 - 2016, 2014 - 2015

Introduction to Nutrition, Undergraduate, 2021 - 2022

Vitamin ve Mineral Bozuklukları, Undergraduate, 2020 - 2021, 2019 - 2020, 2018 - 2019, 2017 - 2018, 2016 - 2017, 2015 - 2016, 2014 - 2015

Ne Zaman Metabolik Hastalık Düşünelim?, Undergraduate, 2020 - 2021, 2019 - 2020, 2018 - 2019, 2017 - 2018, 2016 - 2017, 2014 - 2015

Hekimliğe Giriş Uygulamaları (PDÖ, KBE, İB), Undergraduate, 2019 - 2020

Vitamin and Mineral Disorders, Doctorate, 2018 - 2019, 2017 - 2018

Pediatric Uzmanlık Eğitimi, Expertise In Medicine, 2014 - 2015

Kalıtısal Metabolik Hastalıklara Tanısal ve Acil Tedavi Yaklaşımı, Expertise In Medicine, 2014 - 2015

Advising Theses

Okur İ., ÇOCUK SAĞLIĞI VE HASTALIKLARI ASİSTAN VE UZMAN HEKİMLERİ ARASINDA AKILCI İLAÇ KULLANIMINI DEĞERLENDİREN ÇEVİRİMİÇİ ANKET ÇALIŞMASI, Expertise In Medicine, L.ERKUŞ(Student), 2023

Okur İ., KALITSAL METABOLİK HASTALIKLARDA BAKIMVERENİN SAĞLIK OKURYAZARLIĞININ DEĞERLENDİRİLMESİ , Expertise In Medicine, F.YAŞAR(Student), 2022

OKUR İ., İdiyopatik proteinürisi olan çocuk hastalarda alfa-galaktosidaz a eksikliğinin (fabry hastalığının) taranması, Expertise In Medicine, M.AKGÜL(Student), 2018

Okur İ., 2000-2013 yılları arasında Gazi Üniversitesi Hastanesinde yatan ve total pranteral nutrisyon uygulanan çocuk hastaların değerlendirilmesi, Expertise In Medicine, B.TANRIKULU(Student), 2016

Student Project

Social Project, Investigation of malnutrition due to phenylketonuria and examination of their amino acid profiles (TUBITAK 2209-A University Students Research Projects Support Program 2022), Gazi University, Tıp Fakültesi, Dahili Tıp Bilimleri, Turkey, 2023 - 2024

Activities in Scientific Journals

TURKISH JOURNAL OF PEDIATRICS, Committee Member, 2022 - Continues

Türkiye Çocuk Hastalıkları Dergisi, Committee Member, 2017 - 2023

Memberships / Tasks in Scientific Organizations

Turkish National Pediatric Society, Board Member, 2019 - Continues, Turkey
Çocuk Beslenme ve Metabolizma Derneği, Member, 2018 - Continues, Turkey
Turkish National Pediatric Association, Member, 2005 - Continues, Turkey
Society for the Study of Inborn Errors of Metabolism, Member, 2020 - 2021, England

Scientific Refereeing

GAZI MEDICAL JOURNAL, National Scientific Refreed Journal, December 2022

Scientific Consultations

T.C. Sağlık Bakanlığı Türkiye İlaç ve Tıbbi Cihaz Kurumu, Beşeri İlaçlar Ruhsatlandırma Klinik Komisyonu, Scientific Consultancy, Gazi University, Tıp Fakültesi, Dahili Tıp Bilimleri, Turkey, 2020 - Continues
T.C. Sağlık Bakanlığı Türkiye İlaç ve Tıbbi Cihaz Kurumu, Özel Tıbbi Amaçlı Gıdalar Danışma Komisyonu, Scientific Consultancy, Gazi University, Tıp Fakültesi, Dahili Tıp Bilimleri, Turkey, 2021 - 2023

Tasks In Event Organizations

Okur İ., Bideci A., Karakaş N. M., Boduroğlu O. K., Çetinkaya M., Turkish National Pediatric Society&Egyptian Pediatric Association Joint Meeting, Scientific Congress, Egypt, Kasım 2023
Okur İ., Bideci A., Tezer H., 6th A Day of the Pediatrician-Interactive and with Cases, Scientific Congress, Girne, Cyprus (Kkct), Nisan 2023
Okur İ., Bideci A., Tezer H., 66. Türkiye Milli Pediatri Kongresi, Scientific Congress, Girne, Cyprus (Kkct), Ekim 2022
Okur İ., Türkiye Milli Pediatri Derneği ve Bosna Hersek Pediatri Derneği Ortak Toplantısı, Scientific Congress, Sarajevo, Bosnia And Herzegovina, Eylül 2022
Okur İ., Türkiye Milli Pediatri Derneği & Çek Pediatri Derneği Ortak Toplantısı, Scientific Congress, Praha, Czech Republic, Haziran 2022
Okur İ., 5. Çocuk Hekiminin Bir Günü, Scientific Congress, Girne, Cyprus (Kkct), Mayıs 2022
Okur İ., VII. Congress of Lysosomal Diseases with International Participation, Scientific Congress, Turkey, Kasım 2021
Okur İ., Bideci A., 65. Türkiye Milli Pediatri Kongresi, Scientific Congress, Antalya, Turkey, Kasım 2021
Okur İ., 4. Çocuk Hekiminin Bir Günü- İnteraktif Olgularla, Scientific Congress, Girne, Cyprus (Kkct), Ekim 2021
Okur İ., 64. Milli Pediatri Kongresi, Scientific Congress, Turkey, Aralık 2020
Okur İ., 3. Çocuk Hekiminin Bir Günü - İnteraktif Olgularla, Scientific Congress, Turkey, Ekim 2020

Scientific Research / Working Group Memberships

Gazi Üniversitesi Çocuk Metabolizma B.D. Faz 1 Klinik Araştırmalar Merkezi, Gazi University, Türkiye, <http://faz1-klinik.gazi.edu.tr/>, 2018 - Continues

Metrics

Publication: 238
Citation (WoS): 500
Citation (Scopus): 458
H-Index (WoS): 12

H-Index (Scopus): 12

Congress and Symposium Activities

WORLD Symposium 2024, Attendee, California, United States Of America, 2024
Metabolic Emergencies and Responsibilities of the Pediatrician, Session Moderator, Şanlıurfa, Turkey, 2024
Yenidoğanda Metabolik Karaciğer Hastalıkları, Invited Speaker, Şanlıurfa, Turkey, 2023
6th National Pediatric Genetic Congress, Invited Speaker, Aydın, Turkey, 2023
Turkish National Pediatric Society&Egyptian Pediatric Association Joint Meeting, Session Moderator, Kafr-Ash-Shaykh, Egypt, 2023
Turkish National Pediatric Society&Egyptian Pediatric Association Joing Meeting, Invited Speaker, Kafr-Ash-Shaykh, Egypt, 2023
Turkish National Pediatric Society&Croatian Pediatric Society Joint Meeting, Session Moderator, Zagreb, Croatia, 2023
Turkish National Pediatric Association&Romanian Pediatric Association Joint Meeting, Invited Speaker, Bucuresti, Romania, 2023
A Day of the Pediatrician, Fall Training Meeting, Invited Speaker, Diyarbakır, Turkey, 2023
11th Children's Friends Congress, Invited Speaker, İstanbul, Turkey, 2023
39. Ulusal Gastroenteroloji Haftası- 10. Gastroenteroloji Cerrahisi Kongresi, Invited Speaker, Antalya, Turkey, 2022
Türkiye Milli Pediatri Derneği & Sırbistan Pediatri Derneği Ortak Toplantısı, Invited Speaker, Belgrade, Serbia, 2022
66 Türkiye Milli Pediatri Kongresi, Invited Speaker, Girne, Cyprus (Kkct), 2022
Çocuk Hekiminin Bir Günü Güz Eğitim Toplantısı, Invited Speaker, Gaziantep, Turkey, 2022
Türkiye Milli Pediatri Derneği & Bosna Hersek Pediatri Derneği Ortak Toplantısı, Invited Speaker, Sarajevo, Bosnia And Herzegovina, 2022
Türkiye Milli Pediatri Derneği & Çek Pediatri Derneği Ortak Toplantısı, Invited Speaker, Praha, Czech Republic, 2022
14. Ulusal Çocuk Gastroenteroloji, Hepatoloji ve Beslenme Kongresi, Invited Speaker, Lefke, Cyprus (Kkct), 2022
VII. Congress of Lysosomal Diseases with International Participation, Invited Speaker, Ankara, Turkey, 2021
65th Turkish National Pediatric Congress, Invited Speaker, Antalya, Turkey, 2021
4. Çocuk Hekiminin Bir Günü Sempozyumu, Invited Speaker, Girne, Cyprus (Kkct), 2021
16 Annual WORLDSymposium 2020 , Attendee, Florida, United States Of America, 2020
İlk 1000 gün 5. Kongresi, Invited Speaker, Ankara, Turkey, 2017

Invited Talks

66th Turkish National Pediatric Congress, Conference, 66. Türkiye Milli Pediatri Kongresi, Turkey, October 2022
Workshop on Updating the Purpose and Learning Objectives of Medical Education Programs, Workshop, Gazi Üniversitesi, Turkey, June 2022
65th Turkish National Pediatric Congress, Conference, Turkish National Pediatric Society, Turkey, November 2021
4. Çocuk Hekiminin Bir Günü Sempozyumu, Conference, 4. Çocuk Hekiminin Bir Günü Sempozyumu, Turkey, September 2021
Pandemi Döneminde Çocuk Sağlığı Online Konferans (Türkiye Milli Pediatri Derneği, Azerbaycan Tıp Üniversitesi ve ATA Pediatri Derneği Ortak Toplantısı), Conference, Türkiye Milli Pediatri Derneği, Azerbaycan Tıp Üniversitesi ve ATA Pediatri Derneği, Azerbaijan, July 2020
17. UNPSSTR-Eurasia Congress, Conference, Union of National Pediatric Societies of Turkic Republics (UNPSTR), Azerbaijan, September 2019

Research Areas

Medicine, Child Health and Diseases, Pediatric Endocrinology and Metabolism

Non Academic Experience

Ministry, Republic of Türkiye Ministry of Health, Turkish Medicines and Medical Devices Agency (TİTCK)

Ministry, Ministry of Health, Turkish Medicines and Medical Devices Agency (TİTCK)

Professional Association, Turkish National Pediatric Society, Board Membership

Ministry, Ministry of Health Ankara Child Health and Diseases Hematology and Oncology Education and Research Hospital, Pediatric Metabolism Unit

Ministry, Ministry of Health, Dr. Lütfi Kırdar Kartal Education and Research Hospital, Pediatric Metabolism Unit

Ministry, General Staff, Gülhane Military Medical Academy, Department of Pediatric Metabolism

Ministry, Republic of Türkiye, Ministry of Health, Bayburt Public Hospital, Pediatric Unit

Private Hospital, Sincan Medical Center, Pediatric Unit