



İLYAS OKUR

PROF.

Email : iokur@gazi.edu.tr

Office Phone : [+90 312 202 4000](tel:+903122024000) Extension: 6019

International Researcher IDs

ScholarID: HZMmZeoAAAAJ

ORCID: 0000-0002-8772-0689

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

ScopusID: 6602370050

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, Eğitimcilerin 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-Asosiye 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., EKMEKCİ ERTEK İ., ÖKTEM R. M., BİBEROĞLU G., Project Supported by Higher Education Institutions, Psikoz 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 güvenliliğini 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, Fenilketonüriili erişkin gönüllülerde insan fenilalanin hidrosilazın adeno ilişkili viral vektör aracılı gen transferi olan SAR444836'nın güvenliliğini, 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 İdüsülfaza 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 güvenliliğini 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, Fenilketonüride 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, Venglustatin 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 SOBI003 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., TUBITAK Project, Metilmalonil Koenzim A Mutaz Enziminin Konformasyonel Bozukluğuna Bağlı Olarak Gelişen Metilmalonik Asidemi Hastalığınınxxda Farmakolojik Şaparon 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'xye 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, Hipofosfatazalı 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., Biberoğlu G., Phase 1 Clinical Research Center, January 2018

Published journal articles indexed by SCI, SSCI, and AHCI

- 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, 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, 2024 (SCI-Expanded)
- 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)
4. **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)
 5. **A possibly new autoinflammatory disease due to compound heterozygous phosphomevalonate kinase gene mutation**
Yıldız Ç., Gezin Yıldırım D., İnci A., Tümer L., Ergin F. B., Sunar Yayla E. N. S., Esmeray Şenol P., Karaçayır N., Eğritiş Gürkan Ö., Okur İ., et al.
Joint Bone Spine, vol.90, no.1, 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. **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)
 8. **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)
 9. **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., Eğritiş Gürkan Ö., Okur İ., Biberoglu 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)
 10. **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)
 11. **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)
 12. **First successful concomitant therapy of immune tolerance induction therapy and desensitization in a CRIM-negative infantile Pompe patient**
Sanli M. E., ERTÖY 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)
 13. **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)
 14. **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., Biberoglu G., Öktem M., Ezgü F. S.
ORPHANET JOURNAL OF RARE DISEASES, vol.16, no.1, 2021 (SCI-Expanded)
 15. **Tralesenidase alfa (AX 250) Enzyme Replacement Therapy for Sanfilippo Syndrome Type**
Maricich S., Okur İ., Ezgu F. S., Lopez d. C. M., Couce L. M., Harmatz P., Batzios S., Cleary M., Solano M., Lin S., et al.

ANNALS OF NEUROLOGY, vol.90, 2021 (SCI-Expanded)

16. **Natural History of Sanfilippo Syndrome Type B in Young Patients: Ongoing Results from Two Large, Prospective Studies**
Maricich S., Amartino H., Giugliani R., Muschol N., Harmatz P., Lopez d. C. M., Couce L. M., Lin S., Batzios S., Cleary M., et al.
ANNALS OF NEUROLOGY, vol.90, 2021 (SCI-Expanded)
17. **Congenital defects of glycosylation: Novel presentations with mainly neurological involvement and variable dysmorphic features**
İNCİ A., Cengiz B., BİBEROĞLU G., OKUR İ., ARHAN E., ÖNER A. Y., KASAPKARA Ç. S., Kucukcongar A., TÜMER L., Ezgu F. S.
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, vol.185, no.9, pp.2739-2747, 2021 (SCI-Expanded)
18. **Hypophosphatasia: is it an underdiagnosed disease even by expert physicians?**
İnci A., Ergin F. B., Yüce B. T., Çiftçi B., Demir E., Buyan N., Okur İ., Biberoglu G., Öktem R. M., Tümer L., et al.
JOURNAL OF BONE AND MINERAL METABOLISM, vol.39, no.4, pp.598-605, 2021 (SCI-Expanded)
19. **Ultra-Rare Disorder in a Young Girl with Lipodystrophy: Analbuminemia**
İNCİ A., Arslan B., OKUR İ., BİBEROĞLU G., ŞANLI M. E., ÖZSAYDI AKTAŞOĞLU E., KILIÇ A., TÜMER L., EZGÜ F. S.
INDIAN JOURNAL OF PEDIATRICS, vol.88, pp.723-0, 2021 (SCI-Expanded)
20. **A CASE OF GLYCOGEN STORAGE DISEASE TYPE 1a MIMICKING FAMILIAL CHYLOMICRONEMIA SYNDROME**
Olgac A., OKUR İ., BİBEROĞLU G., EZGÜ F. S., TÜMER L.
BALKAN JOURNAL OF MEDICAL GENETICS, vol.24, no.1, pp.103-105, 2021 (SCI-Expanded)
21. **Autism: Screening of inborn errors of metabolism and unexpected results**
İnci A., Özaskan A., Okur İ., Biberoglu G., Güney E., Ezgu F. S., Tümer L., İşeri E.
AUTISM RESEARCH, vol.14, no.5, pp.887-896, 2021 (SCI-Expanded)
22. **CDH13 and LPHN3 Gene Polymorphisms in Attention-Deficit/Hyperactivity Disorder: Their Relation to Clinical Characteristics.**
Özaslan A., Güney E., Ergün M. A., Okur İ., Yapar D.
Journal of molecular neuroscience : MN, vol.71, pp.394-408, 2021 (SCI-Expanded)
23. **Natural history of Sanfilippo syndrome type B in young patients: Ongoing results from two large, prospective studies**
Giugliani R., OKUR İ., Ezgu F. S., Muschol N., Harmatz P., de Castro Lopez M., Luz Couce M., Lin S., Batzios S., Cleary M., et al.
MOLECULAR GENETICS AND METABOLISM, vol.132, no.2, 2021 (SCI-Expanded)
24. **The chemical chaperone 4-phenylbutyrate enhances alpha-galactosidase activity subsequent to stop-codon read-through therapy with triamterene in Fabry R227X fibroblasts**
Dündar H., Biberoglu G., İnci A., Işık Gönül İ., Okur İ., Tümer L., Ezgu F. S.
MOLECULAR GENETICS AND METABOLISM, vol.132, no.2, 2021 (SCI-Expanded)
25. **Tralesinidase alfa (AX 250) enzyme replacement therapy for Sanfilippo syndrome type B**
Muschol N., von Cossel K., OKUR İ., Ezgu F. S., de Castro Lopez M., Luz Couce M., Harmatz P., Batzios S., Cleary M., Solano M., et al.
MOLECULAR GENETICS AND METABOLISM, vol.132, no.2, 2021 (SCI-Expanded)
26. **Familial hyperphosphatemic tumoral calcinosis in an unusual and usual sites and dramatic improvement with the treatment of acetazolamide, sevelamer and topical sodium thiosulfate**
ŞANLI M. E., KILIÇ A., ÖZSAYDI AKTAŞOĞLU E., İNCİ A., OKUR İ., Ezgu F. S., TÜMER L.
JOURNAL OF PEDIATRIC ENDOCRINOLOGY & METABOLISM, vol.34, no.6, pp.813-816, 2021 (SCI-Expanded)
27. **Two patients from Turkey with a novel variant in the GM2A gene and review of the literature**
İNCİ A., ERGİN F. B., BİBEROĞLU G., OKUR İ., EZGÜ F. S., TÜMER L.
JOURNAL OF PEDIATRIC ENDOCRINOLOGY & METABOLISM, vol.34, no.6, pp.805-812, 2021 (SCI-Expanded)
28. **Nutritional Status of Syrian Refugees in Early Adolescence Living in Turkey**
Pehlivan Türk Kızılkın M., Özmert E. N., Derman O., Okur İ., Kaynak M. O., Adiguzel A., Sahan-Seref I., Kurekci F., Bideci A., Hasanoglu E.

JOURNAL OF IMMIGRANT AND MINORITY HEALTH, vol.22, no.6, pp.1149-1154, 2020 (SSCI)

29. **Beneficial Effects of Modified Atkins Diet in Glycogen Storage Disease Type IIIa**
Olgac A., İNCİ A., OKUR İ., BİBEROĞLU G., Oguz D., EZGÜ F. S., Kasapkara C. S., Aktas E., TÜMER L.
ANNALS OF NUTRITION AND METABOLISM, vol.76, no.4, pp.233-241, 2020 (SCI-Expanded)
30. **Vitamin D Levels and Bone Mineral Density in Inborn Errors of Metabolism Requiring Specialised Diets**
Olgac A., İNCİ A., OKUR İ., Ezgu F. S., BİBEROĞLU G., Turner L.
JCPSP-JOURNAL OF THE COLLEGE OF PHYSICIANS AND SURGEONS PAKISTAN, vol.29, no.12, pp.1207-1211, 2019 (SCI-Expanded)
31. **High incidence of co-existing factors significantly modifying the phenotype in patients with Fabry disease.**
Koca S., TÜMER L., OKUR İ., ERTEN Y., Bakkaloglu S. A., BİBEROĞLU G., Kasapkara C., Kucukcongar A., DALGIÇ B., ÖZHAN OKTAR S., et al.
Gene, vol.687, pp.280-288, 2019 (SCI-Expanded)
32. **Epilepsy in Biotinidase Deficiency Is Distinct from Early Myoclonic Encephalopathy**
Guliyeva U., OKUR İ., Dulac O., Khalilov O., Guliyeva S.
NEUROPEDIATRICS, vol.49, no.6, pp.417-418, 2018 (SCI-Expanded)
33. **Hematologic Findings of Inherited Metabolic Disease: They are More Than Expected**
Sal E., Yenicesu I., OKUR İ., KAYA Z., EZGÜ F. S., KOÇAK Ü., TÜMER L., Gursel T., Hasanoglu A.
JOURNAL OF PEDIATRIC HEMATOLOGY ONCOLOGY, vol.40, no.5, pp.355-359, 2018 (SCI-Expanded)
34. **Bi-allelic Mutations in KLHL7 Cause a Crisponi/CISS1-like Phenotype Associated with Early-Onset Retinitis Pigmentosa.**
Angius A., Uva P., Buers I., Oppo M., Puddu A., Onano S., Persico I., Loi A., Marcia L., Höhne W., et al.
American journal of human genetics, vol.102, pp.713, 2018 (SCI-Expanded)
35. **Patient With Niemann-Pick Type C Presenting With a Jaw Mass Characterized With Lymph Node Involvement by Niemann-Pick Cells**
İNCİ A., OKUR İ., ESENDAĞLI G., OKUR A., Olgac A., EZGÜ F. S., TÜMER L.
JOURNAL OF PEDIATRIC HEMATOLOGY ONCOLOGY, vol.40, no.3, pp.243-245, 2018 (SCI-Expanded)
36. **Structure-function analyses of microsomal triglyceride transfer protein missense mutations in abetalipoproteinemia and hypobetalipoproteinemia subjects**
Walsh M. T., Di Leo E., OKUR İ., Tarugi P., Hussain M. M.
BIOCHIMICA ET BIOPHYSICA ACTA-MOLECULAR AND CELL BIOLOGY OF LIPIDS, vol.1861, no.11, pp.1623-1633, 2016 (SCI-Expanded)
37. **Bi-allelic Mutations in KLHL7 Cause a Crisponi/CISS1-like Phenotype Associated with Early-Onset Retinitis Pigmentosa**
Angius A., Uva P., Buers I., Oppo M., Puddu A., Onano S., Persico I., Loi A., Marcia L., Hoehne W., et al.
AMERICAN JOURNAL OF HUMAN GENETICS, vol.99, no.1, pp.236-245, 2016 (SCI-Expanded)
38. **Audiologic evaluations of children with mucopolysaccharidosis**
Gokdogan C., Altınyay Ş., Gokdogan O., Tutar H., Gündüz B., Okur İ., Tümer L., Kemaloğlu Y. K.
BRAZILIAN JOURNAL OF OTORHINOLARYNGOLOGY, vol.82, no.3, pp.281-284, 2016 (SCI-Expanded)
39. **COBALAMIN C DEFICIENCY WITH INFANTILE SPASM AND CUTANEOUS FINDINGS: A UNIQUE CASE**
Ozturk Z., Arhan E., Aydin K., Hirfanoglu T., Tumer L., Okur İ., Serdaroglu A., Akbas Y., Karaoglu B.
GENETIC COUNSELING, vol.27, no.3, pp.399-403, 2016 (SCI-Expanded)
40. **The Janus-faced manifestations of homozygous familial hypobetalipoproteinemia due to apolipoprotein B truncations**
Di Leo E., Eminoglu T., Magnolo L., Bolkent M. G., TÜMER L., OKUR İ., Tarugi P.
JOURNAL OF CLINICAL LIPIDOLOGY, vol.9, no.3, pp.400-405, 2015 (SCI-Expanded)
41. **Neonates with inborn errors of metabolism: spectrum and short-term outcomes at a tertiary care hospital**
Gunduz M., Unal S., OKUR İ., Ayranci-Sucakli I., Guzel F., Koc N.
TURKISH JOURNAL OF PEDIATRICS, vol.57, no.1, pp.45-52, 2015 (SCI-Expanded)

42. **Monocarboxylate Transporter 1 Deficiency and Ketone Utilization**
van Hasselt P. M., Ferdinandusse S., Monroe G. R., Ruiter J. P. N., Turkenburg M., Geerlings M. J., Duran K., Harakalova M., van der Zwaag B., Monavari A. A., et al.
NEW ENGLAND JOURNAL OF MEDICINE, vol.371, no.20, pp.1900-1907, 2014 (SCI-Expanded)
43. **Serum dipeptidyl peptidase-IV: A better screening test for early detection of mucopolysaccharidosis?**
Kurt I., Sertoglu E., OKUR İ., Tapan S., Uyanik M., Kayadibi H., EZGÜ F. S., Aydin H. I., Hasanoglu A.
CLINICA CHIMICA ACTA, vol.431, pp.250-254, 2014 (SCI-Expanded)
44. **Screening for Fabry disease in patients undergoing dialysis for chronic renal failure in Turkey: Identification of new case with novel mutation**
OKUR İ., Ezgu F. S., BİBEROĞLU G., Turner L., ERTEN Y., Isitman M., Eminoglu F. T., Hasanoglu A.
GENE, vol.527, no.1, pp.42-47, 2013 (SCI-Expanded)
45. **Quality of life in children treated with restrictive diet for inherited metabolic disease**
Eminoglu T. F., Soysal S. A., TÜMER L., OKUR İ., Hasanoglu A.
PEDIATRICS INTERNATIONAL, vol.55, no.4, pp.428-433, 2013 (SCI-Expanded)
46. **Oxidized low-density lipoprotein levels and carotid intima-media thickness as markers of early atherosclerosis in prepubertal obese children**
OKUR İ., TÜMER L., EZGÜ F. S., Yesilkaya E., Aral A., ÖZHAN OKTAR S., BİDECİ A., Hasanoglu A.
JOURNAL OF PEDIATRIC ENDOCRINOLOGY & METABOLISM, vol.26, pp.657-662, 2013 (SCI-Expanded)
47. **Two novel deletions in hypotonia-cystinuria syndrome**
Regal L., Aydin H. I., Dieltjens A., Van Esch H., Francois I., OKUR İ., Zeybek C., Meulemans S., Van Mol C., Van Bruwaene L., et al.
MOLECULAR GENETICS AND METABOLISM, vol.107, no.3, pp.614-616, 2012 (SCI-Expanded)
48. **ASSOCIATION BETWEEN SOLUBLE CD40 LIGAND AND PROTHROMBOTIC STATE IN CHILDREN WITH HYPERCHOLESTEROLEMIA**
Kucukcongar A., Eminoglu F. T., Okur İ., Aral A., Hasanoglu A., Tumer L.
JOURNAL OF INHERITED METABOLIC DISEASE, vol.35, 2012 (SCI-Expanded)
49. **MUCOPOLYSACCHARIDOSIS: EFFECTS OF ENZYME-REPLACEMENT THERAPY IN 27 CHILDREN WITH MPS I, II AND VI**
Hasanoglu A., Tumer L., Ezgu F. S., Okur İ., Eminoglu F. T., Kasapkara C. S., Kucukcongar A.
JOURNAL OF INHERITED METABOLIC DISEASE, vol.35, 2012 (SCI-Expanded)
50. **GENOTYPIC FEATURES OF 41 PATIENTS WITH GAUCHER DISEASE FROM TURKEY**
Hasanoglu A., Akay G., Ezgu F. S., Biberoglu G., Tumer L., Okur İ., Kucukcongar A., Kasapkara C., Polat M., Ciftci B.
JOURNAL OF INHERITED METABOLIC DISEASE, vol.35, 2012 (SCI-Expanded)
51. **THE VALUE OF THE SURROGATE TESTS (SERUM DIPEPTIDYL PEPTIDASE-IV, ADENOSINE DEAMINASE-1, CHITOTRIOSIDASE) IN THE DIAGNOSIS OF MUCOPOLYSACCHARIDOSIS**
Kurt I., Hasanoglu A., Aydin H., Okur İ., Sertoglu E., Tapan S., Ezgu F. S.
JOURNAL OF INHERITED METABOLIC DISEASE, vol.35, 2012 (SCI-Expanded)
52. **COMPREHENSIVE SEQUENCING OF MITOCHONDRIAL DNA IN PATIENTS WITH SUSPECTED MITOCHONDRIAL DISEASE: IS THERE A NEED FOR A REVISED MOLECULAR DIAGNOSTIC ALGORITHM?**
Ezgu F. S., Kucukcongar A., Ciftci B., Kasapkara C., Hasanoglu A., Tumer L., Okur İ., Gunduz M., Polat M., Bahceci S.
JOURNAL OF INHERITED METABOLIC DISEASE, vol.35, 2012 (SCI-Expanded)
53. **Hypercalcemia in glycogen storage disease type I patients of Turkish origin**
Kasapkara C. S., TÜMER L., OKUR İ., Eminoglu T., EZGÜ F. S., Hasanoglu A.
TURKISH JOURNAL OF PEDIATRICS, vol.54, no.1, pp.35-37, 2012 (SCI-Expanded)
54. **The levels of asymmetric dimethylarginine, homocysteine and carotid intima-media thickness in hypercholesterolemic children**
Hasanoglu A., OKUR İ., Oren A. C., BİBEROĞLU G., ÖZHAN OKTAR S., Eminoglu F. T., TÜMER L.
TURKISH JOURNAL OF PEDIATRICS, vol.53, no.5, pp.522-527, 2011 (SCI-Expanded)
55. **Very long-chain acyl CoA dehydrogenase deficiency which was accepted as infanticide**

- Eminoglu T. F., TÜMER L., OKUR İ., EZGÜ F. S., BİBEROĞLU G., Hasanoglu A.
FORENSIC SCIENCE INTERNATIONAL, vol.210, 2011 (SCI-Expanded)
56. **IDUA Mutational Profiling of a Cohort of 102 European Patients with Mucopolysaccharidosis Type I: Identification and Characterization of 35 Novel alpha-L-iduronidase (IDUA) Alleles**
Bertola F., Filocamo M., Casati G., Mort M., Rosano C., Tytki-Szymanska A., Tuysuz B., Gabrielli O., Grossi S., Scarpa M., et al.
HUMAN MUTATION, vol.32, no.6, 2011 (SCI-Expanded)
 57. **N-carbamylglutamate treatment for acute neonatal hyperammonemia in isovaleric acidemia**
Kasapkara C. S., EZGÜ F. S., OKUR İ., TÜMER L., BİBEROĞLU G., Hasanoglu A.
EUROPEAN JOURNAL OF PEDIATRICS, vol.170, no.6, pp.799-801, 2011 (SCI-Expanded)
 58. **Harderoporphyria due to homozygosity for coproporphyrinogen oxidase missense mutation H327R**
Hasanoglu A., Balwani M., Kasapkara C. S., EZGÜ F. S., OKUR İ., TÜMER L., Cakmak A., Nazarenko I., Yu C., Clavero S., et al.
JOURNAL OF INHERITED METABOLIC DISEASE, vol.34, no.1, pp.225-231, 2011 (SCI-Expanded)
 59. **A NOVEL MUTATION OF THE CLAUDIN 16 GENE IN FAMILIAL HYPOMAGNESEMIA WITH HYPERCALCIURIA AND NEPHROCALCINOSIS MIMICKING RICKETS**
Kasapkara C. S., Tumer L., OKUR İ., Hasanoglu A.
GENETIC COUNSELING, vol.22, no.2, pp.187-192, 2011 (SCI-Expanded)
 60. **A NOVEL MUTATION IN A TURKISH PATIENT WITH DIHYDROPTERIDINE REDUCTASE (DHPR) DEFICIENCY**
Aydin H., Okur İ., Vurucu S., Mxller L. B.
JOURNAL OF INHERITED METABOLIC DISEASE, vol.34, 2011 (SCI-Expanded)
 61. **AN ADULT PATIENT WITH LATE DIAGNOSED LYSINURIC PROTEIN INTOLERANCE**
Aydin H., Okur İ., Cetin T., Kurt I.
JOURNAL OF INHERITED METABOLIC DISEASE, vol.34, 2011 (SCI-Expanded)
 62. **TWO NOVEL MUTATIONS IN TURKISH PATIENTS WITH SJVGREN LARSSON SYNDROME**
Aydin H., Okur İ., Unay B., Nakano H., Wanders R. J.
JOURNAL OF INHERITED METABOLIC DISEASE, vol.34, 2011 (SCI-Expanded)
 63. **NOVEL DELETION IN HYPOTONIA-CYSTINURIA SYNDROME**
Aydin H., Okur İ., Creemers J. W. M., COŞKUN T.
JOURNAL OF INHERITED METABOLIC DISEASE, vol.34, 2011 (SCI-Expanded)
 64. **AN INFANTILE CASE OF ZELLWEGER SYNDROME PRESENTED WITH KABUKI-LIKE PHENOTYPE**
Ezgu F. S., Eminoglu T., OKUR İ., Gunduz M., Tumer L., Hasanoglu A., Dalgic B.
GENETIC COUNSELING, vol.22, no.2, pp.217-220, 2011 (SCI-Expanded)
 65. **A NOVEL MUTATION IN A TURKISH PATIENT WITH THE INFANTILE FORM OF TAY-SACHS DISEASE**
Okur İ., Aydin H., Akin R.
JOURNAL OF INHERITED METABOLIC DISEASE, vol.34, 2011 (SCI-Expanded)
 66. **The role of viral agents in aetiopathogenesis of acute rheumatic fever**
Olgunturk R., OKUR İ., Cirak M. Y., OĞUZ A. D., Akalin N., Turet S., Tunaoglu S.
CLINICAL RHEUMATOLOGY, vol.30, no.1, pp.15-20, 2011 (SCI-Expanded)
 67. **THE SCANNING OF COMMONLY SEEN MUTATIONS OF GLUCOSE-6-PHOSPHATASE AND GLUCOSE-6-PHOSPHATASE TRANSLOCASE GENES IN GLYCOGEN STORAGE TYPE 1A AND TYPE 1B DISEASE PATIENTS BY THE MICROELECTRONIC ARRAY TECHNOLOGY**
Eminoglu F. T., Tumer L., Ezgu F. S., Okur İ., Biberoglu G., Hasanoglu A.
JOURNAL OF INHERITED METABOLIC DISEASE, vol.33, 2010 (SCI-Expanded)
 68. **FOUR CASES OF NIEMANN-PICK TYPE C DISEASE PRESENTED WITH EARLY ONSET CHOLESTASIS**
Kucukcongar A., Okur İ., Ezgu F. S., Turner L., Dalgic B., Hasanoglu A.
JOURNAL OF INHERITED METABOLIC DISEASE, vol.33, 2010 (SCI-Expanded)
 69. **HARDEROPORPHYRIA PHENOTYPE DUE TO A HOMOZYGOUS H237R MISSENSE MUTATION**
Kasapkara C. S., Hasanoglu A., Ezgu F. S., Okur İ., Tumer L., Cakmak A., Balwani M., Nazarenko I., Clavero S., Yu C., et al.

JOURNAL OF INHERITED METABOLIC DISEASE, vol.33, 2010 (SCI-Expanded)

70. **N-CARBAMYLGLUTAMATE TREATMENT FOR ACUTE NEONATAL HYPERAMMONAEMIA IN ISOVALERIC ACIDAEMIA**

Kasapkara C. S., Ezgu F. S., Tumer L., Biberoglu G., Okur İ., Hasanoglu A.

JOURNAL OF INHERITED METABOLIC DISEASE, vol.33, 2010 (SCI-Expanded)

71. **HYPERCALCAEMIA IN GLYCOGEN STORAGE DISEASE TYPE 1 PATIENTS OF TURKISH ORIGIN**

Kasapkara C. S., Tumer L., Okur İ., Eminoglu F. T., Ezgu F. S., Hasanoglu A.

JOURNAL OF INHERITED METABOLIC DISEASE, vol.33, 2010 (SCI-Expanded)

72. **GENOTYPE-PHENOTYPE CORRELATIONS IN TURKISH PATIENTS WITH ALPHA GALACTOSIDASE A DEFICIENCY**

Koca S., Ezgu F. S., Okur İ., Biberoglu G., Tumer L., Hasanoglu A.

JOURNAL OF INHERITED METABOLIC DISEASE, vol.33, 2010 (SCI-Expanded)

73. **Screening for isolated sulfite oxidase/molibden cofactor deficiencies among the pediatric patients with encephalopathy and mental-motor retardation**

Keles E., Ezgu F. S., Tosun M. S., Okur İ., Eminoglu F. T., Tumer L., Hasanoglu A.

EUROPEAN JOURNAL OF MEDICAL RESEARCH, vol.14, pp.84, 2009 (SCI-Expanded)

74. **3-Methylcrotonyl-CoA Carboxylase Deficiency: Phenotypic Variability in a Family**

Eminoglu F. T., Ozelik A. A., OKUR İ., TÜMER L., BİBEROĞLU G., DEMİR E., Hasanoglu A., Baumgartner M. R.

JOURNAL OF CHILD NEUROLOGY, vol.24, no.4, pp.478-481, 2009 (SCI-Expanded)

75. **Lipid apheresis applications in childhood: Experience in the University Hospital of Gazi**

Eminoglu T. F., Yenicesu I., TÜMER L., OKUR İ., Dilsiz G., Hasanoglu A.

TRANSFUSION AND APHERESIS SCIENCE, vol.39, no.3, pp.235-240, 2008 (SCI-Expanded)

76. **Crisponi Syndrome: A New Case With Additional Features and New Mutation in CRLF1**

OKUR İ., TÜMER L., Crisponi L., Eminoglu F. T., Chiappe F., CİNAZ P., Yenicesu I., Hasanoglu A.

AMERICAN JOURNAL OF MEDICAL GENETICS PART A, no.24, pp.3237-3239, 2008 (SCI-Expanded)

77. **Multisystem involvement in a patient due to accumulation of amylopectin-like material with diminished branching enzyme activity**

Eminoglu T. F., Tumer L., OKUR İ., Olgunturk R., Hasanoglu A., Gonul I. I., Dalgic B.

JOURNAL OF INHERITED METABOLIC DISEASE, vol.31, 2008 (SCI-Expanded)

78. **The impact of inherited metabolic diseases on quality of life: A pilot study**

Tumer L., Eminoglu F. T., Soysal A. S., Okur İ., Hasanoglu A.

JOURNAL OF INHERITED METABOLIC DISEASE, vol.31, pp.147, 2008 (SCI-Expanded)

79. **Crisponi syndrome due to a novel mutation on the cytokine receptor-like factor I (CRLF1) gene**

Okur İ., Tumer L., Eminoglu F. T., Crisponi L., Cinaz P., Yenicesu I., Hasanoglu A.

JOURNAL OF INHERITED METABOLIC DISEASE, vol.31, pp.153, 2008 (SCI-Expanded)

80. **The neonatal case diagnosed with nonketotic hyperglycinemia with a preliminary diagnosis of the chloralhydrate intoxication**

Okur İ., Eminoglu F. T., Tumer L., Ezgu F. S., Biberoglu G., Hasanoglu A.

JOURNAL OF INHERITED METABOLIC DISEASE, vol.31, pp.3, 2008 (SCI-Expanded)

81. **The same novel mutation determined in 2 Hurler-Scheie patients who are the children of different families**

Hasanoglu A., Okur İ., Eminoglu F. T., Tumer L., Biberoglu G., Bertola F., Ezgu F. S.

JOURNAL OF INHERITED METABOLIC DISEASE, vol.31, pp.112, 2008 (SCI-Expanded)

82. **Very long-chain acyl CoA dehydrogenase deficiency which was accepted as infanticide**

Eminoglu F. T., Tumer L., Okur İ., Goekmen Z., Ezgu F. S., Biberoglu G., Hasanoglu A.

JOURNAL OF INHERITED METABOLIC DISEASE, vol.31, pp.33, 2008 (SCI-Expanded)

83. **Molecular analysis of 82 mucopolysaccharidosis type I patients: Mutational spectrum in the European population and identification of 28 novel mutations**

Bertola F., Parini R., Casati G., Tylki-Szymanska A., Okur İ., TÜYSÜZ B., Dalmau J., Gonzales M. A., Antuzzi D., Barone R., et al.

JOURNAL OF INHERITED METABOLIC DISEASE, vol.31, pp.108, 2008 (SCI-Expanded)

84. **Rapid screening of 10 common mutations in Turkish Gaucher patients using electronic DNA microarray**
Ezgu F. S., Hasanoglu A., OKUR İ., Biberoglu G., Tumer L., Eminoglu T., Dogan H.
BLOOD CELLS MOLECULES AND DISEASES, vol.40, no.2, pp.246-247, 2008 (SCI-Expanded)
85. **3-Methylcrotonylglycinuria in a family: Late and different clinical presentation**
Eminoglu F. T., Turner L., Okur İ., Biberoglu G., Derin B., Cinasal Demir G., Hasanoglu A., Ozcelik A., Demir E.
JOURNAL OF INHERITED METABOLIC DISEASE, vol.30, pp.40, 2007 (SCI-Expanded)
86. **Long-term effect of low-density lipoprotein apheresis: Experience in four children with familial homozygous hypercholesterolemia**
Hasanoglu A., Yenicesu I., Eminoglu F. T., Okur İ., Tumer L.
JOURNAL OF INHERITED METABOLIC DISEASE, vol.30, pp.127, 2007 (SCI-Expanded)
87. **Multisystem involvement: A rare and unusual presentation of GSD type IV**
Tumer L., Eminoglu F. T., Okur İ., Hasanoglu A., Olgunturk R.
JOURNAL OF INHERITED METABOLIC DISEASE, vol.30, pp.60, 2007 (SCI-Expanded)
88. **Investigation of 10 common mutations in Turkish Gaucher patients by use of the nanochip microelectronic array technology**
Hasanoglu A., Ezgu F. S., Okur İ., Eminoglu F. T., Biberoglu G., Tumer L.
JOURNAL OF INHERITED METABOLIC DISEASE, vol.30, pp.108, 2007 (SCI-Expanded)
89. **The first results of 18 months experience with lysosomal storage disease**
Biberoglu G., Hasanoglu A., Ezgu F. S., Tumer L., Okur İ., Eminoglu F. T., Yalcinkaya D.
JOURNAL OF INHERITED METABOLIC DISEASE, vol.30, pp.95, 2007 (SCI-Expanded)
90. **Cryptic exon activation by disruption of an exon splice enhancer: A novel mechanism causing 3-methylcrotonyl-CoA carboxylase deficiency**
Eminoglu F. T., Turner L., Okur İ., Biberoglu G., Derin B., Cinasal D. G., Hasanoglu A., Ozcelik A., Demir E.
JOURNAL OF INHERITED METABOLIC DISEASE, vol.30, pp.40, 2007 (SCI-Expanded)
91. **Incidence of osteoporosis in a metabolic unit**
Hasanoglu A., Turner L., Ezgu F. S., Eminoglu F. T., Okur İ.
JOURNAL OF INHERITED METABOLIC DISEASE, vol.29, pp.159, 2006 (SCI-Expanded)
92. **The effects of laronidase treatment in a patient with Hurler syndrome: Results of one year therapy**
Hasanoglu A., Tumer L., Ezgu F. S., Gunduz M., Okur İ., Eminoglu T.
JOURNAL OF INHERITED METABOLIC DISEASE, vol.29, pp.149, 2006 (SCI-Expanded)
93. **Vitamin D intoxication and hypercalcaemia in an infant treated with pamidronate infusions**
Ezgu F. S., Buyan N., Gunduz M., Tumer L., OKUR İ., Hasanoglu A.
EUROPEAN JOURNAL OF PEDIATRICS, vol.163, no.3, pp.163-165, 2004 (SCI-Expanded)

Articles Published in Other Journals

1. **Simultaneous succinylacetone-nitisinone measurement in tyrosinemia type I patients and evaluation of the nitisinone therapeutic range**
Öktem R. M., İnci A., Biberoglu G., Okur İ., Ezgu F. S., Tumer L.
Biochimica Clinica, vol.47, no.3, pp.340-345, 2023 (Scopus)
2. **Dietary Fiber Supplementation in Type I Glycogen Storage Disease; Could it Contribute to a Better Metabolic Control?**
Emecen Şanlı M., Aktaş E., İnci A., Okur İ., Ezgu F. S., Tumer L.
GUNCEL PEDIATRI, vol.21, 2023 (ESCI)
3. **MİTOKONDRİYAL HASTALIK NEDENİYLE TETKİK EDİLEN HASTALARDA M.16189T>C DEĞİŞİKLİĞİNİN METABOLİK SENDROM AÇISINDAN İNCELENMESİ**
İNCİ A., HASANOĞLU A., OKUR İ., BİBEROĞLU G., TÜMER L., EZGÜ F. S.
Kocatepe Tıp Dergisi, vol.23, no.3, pp.322-325, 2022 (Peer-Reviewed Journal)
4. **m.3010G>A Değişikliğinin Türk Populasyonunda Sıklık Kusma Sendromuna Etkisi**

ERGİN F. B., İNCİ A., OKUR İ., BİBEROĞLU G., TÜMER L., EZGÜ F. S.

Celal Bayar Üniversitesi Sağlık Bilimleri Enstitüsü Dergisi, 2022 (Peer-Reviewed Journal)

5. **Specialty-Training Program During COVID-19 Pandemic: A Single Center Survey on over 300 Trainees and Trainers**
OĞUZÜLGEN İ. K., KALKANCI A., AYHAN M. S., DERİCİ Ü., ŞENKÖYLÜ A., Gonul I. I., GÜLBAHAR Ö., GÜZEL TUNÇCAN Ö., OKUR İ., İLHAN M. N.
GAZI MEDICAL JOURNAL, vol.33, no.4, pp.381-384, 2022 (ESCI)
6. **PROPIONYLCARNITINE AND FREE CARNITINE ARE NEW BIOMARKERS IN THE FOLLOW-UP PERIOD OF MUCOPOLYSACCHARIDOSIS TO SCREEN OXIDATIVE STRESS**
İNCİ A., OLGAC A., GENÇ DERİN B., BİBEROĞLU G., OKUR İ., EZGÜ F. S., TÜMER L.
Süleyman Demirel Üniversitesi Tıp Fakültesi Dergisi, vol.28, no.4, pp.565-571, 2021 (Peer-Reviewed Journal)
7. **Do cytokines play role in the pathogenesis of mucopolysaccharidosis**
İNCİ A., OLGAC KILIÇKAYA M. A. B., YILMAZ DEMİRTAŞ C., OKUR İ., BİBEROĞLU G., EZGÜ F. S., TÜMER L.
Medicine Science, vol.10, no.4, pp.1492-1497, 2021 (Peer-Reviewed Journal)
8. **The Evaluation of Skeletal Manifestations in Patients with Gaucher Disease**
Kasapkar C. S., Olgac A., OKUR İ., EZGÜ F. S., TÜMER L.
JOURNAL OF PEDIATRIC RESEARCH, vol.8, no.3, pp.257-261, 2021 (ESCI)
9. **Enzim Replasman Tedavisi**
KILIÇ A., OKUR İ.
Türkiye Klinikleri Pediatri Dergisi, vol.1, pp.16-23, 2021 (Scopus)
10. **Citrullinemia with an Atypical Presentation: Paroxysmal Hypoventilation Attacks**
Ozturk Z., HIRFANOĞLU T., İNCİ A., OKUR İ., KOÇ E., TÜMER L., ARHAN E., Aydın K., SERDAROĞLU A.
JOURNAL OF PEDIATRIC NEUROSCIENCES, vol.13, no.2, pp.276-278, 2018 (ESCI)
11. **Association Between Soluble CD40 Ligand and Hypercholesterolemia in Children and Adolescents**
Yavas A. K., Eminoglu T. F., OKUR İ., Aral A., Hasanoglu A., TÜMER L.
JOURNAL OF PEDIATRIC RESEARCH, vol.4, no.1, pp.1-5, 2017 (ESCI)
12. **Clinical course and outcome of glycogen-storage disease type 1a and type 1b**
Eminoglu F. T., TÜMER L., OKUR İ., EZGÜ F. S., Hasanoglu A.
TURK PEDIATRI ARSIVI-TURKISH ARCHIVES OF PEDIATRICS, vol.48, no.2, pp.117-122, 2013 (ESCI)
13. **Giant bronchogenic cyst mimicking tension pneumothorax.**
Demircan S., Kurul İ. C., Tokur M., Memis L., Okur İ.
Asian cardiovascular & thoracic annals, vol.14, pp.244-6, 2006 (Scopus)
14. **Karbonmonoksit Zehirlenmesinde Hiperbarik Oksijen Tedavisi İki Olgu Sunumu**
OKUR İ., SERDAROĞLU A., OKUR A., BUYAN N., DÜNDAR K., ARGAN M., ÖZDEMİR B., GÜCÜYENER K.
TÜRKİYE KLİNİKLERİ J PEDIATR, vol.14, pp.220-222, 2005 (Peer-Reviewed Journal)

Books & Book Chapters

1. **Inherited Metabolic Myopathies - 2024**
Özsaydı Aktaşoğlu E., Tümer L., Yar Sağlam A. S., Usta Salımı D. D., Kasapkar Ç. S., Kahraman A. B., Yıldız Y., Sürücü Kara İ., Eminoğlu F. T., Küçükçongar Yavaş A., et al.
TÜRKİYE KLİNİKLERİ, Ankara, 2024
2. **MATERNAL FENİLKETONÜRİ ve BESLENME TEDAVİSİ**
GÖKALP S., OKUR İ.
in: KALITSAL METABOLİK HASTALIKLARDA BESLENME TEDAVİSİ, Doç. Dr. Fatma Tuba Eminoğlu Prof. Dr. Yusuf Kenan Haspolat Prof. Dr. Coşkun Çeltik Kürşat Bora Çarman Ulaş Emre Akbulut Taşkın Taş, Editor, Orient Yayınları, Ankara, pp.202-208, 2021
3. **Tirozin Metabolizması Bozuklukları**
OKUR İ.
in: TEMEL PEDIATRİ, HASANOĞLU ENVER, DÜŞÜNSEL RUHAN, BİDECİ AYSUN, BODUROĞLU KORAY, Editor, GÜNEŞ

TIP KİTABEVLERİ, Ankara, pp.1144-1148, 2020

4. **Keton Cisim Yapım ve Yıkım Bozuklukları**

OKUR İ.

in: TEMEL PEDİATRİ, HASANOĞLU ENVER, DÜŞÜNSEL RUHAN, BİDECİ AYSUN, BODUROĞLU KORAY, Editor, GÜNEŞ TIP KİTABEVLERİ, Ankara, pp.1132-1135, 2020

5. **Yağ Asidi Oksidasyon Bozuklukları**

İNCİ A., OKUR İ.

in: TEMEL PEDİATRİ, HASANOĞLU ENVER, DÜŞÜNSEL RUHAN, BİDECİ AYSUN, BODUROĞLU KORAY, Editor, GÜNEŞ TIP KİTABEVLERİ, Ankara, pp.1126-1130, 2020

6. **Yağ asidi Oksidasyon Bozuklukları**

İNCİ A., OKUR İ.

in: Temel Pediatri, Prof. Dr. Enver Hasanoğlu, Prof. Dr. Ruhan Düşünsel, Prof. Dr. Aysun Bideci, Prof. Dr. Koray Boduroğlu, Editor, Güneş Tıp Kitapevi, Ankara, pp.1126-1131, 2020

7. **Fruktoz Metabolizması Bozuklukları**

HASANOĞLU A., OKUR İ.

in: TEMEL PEDİATRİ, HASANOĞLU ENVER, DÜŞÜNSEL RUHAN, BİDECİ AYSUN, BODUROĞLU KORAY, Editor, GÜNEŞ TIP KİTABEVLERİ, Ankara, pp.1122-1123, 2020

8. **Metionin Metabolizması Bozuklukları**

OKUR İ.

in: TEMEL PEDİATRİ, HASANOĞLU ENVER, DÜŞÜNSEL RUHAN, BİDECİ AYSUN, BODUROĞLU KORAY, Editor, GÜNEŞ TIP KİTABEVLERİ, Ankara, pp.1156-1158, 2020

9. **Vitaminler**

HASANOĞLU A., OKUR İ.

in: TEMEL PEDİATRİ, HASANOĞLU ENVER, DÜŞÜNSEL RUHAN, BİDECİ AYSUN, BODUROĞLU KORAY, Editor, GÜNEŞ TIP KİTABEVLERİ, Ankara, pp.30-44, 2020

10. **Glikojen Depo Hastalıkları**

HASANOĞLU A., OKUR İ.

in: TEMEL PEDİATRİ, HASANOĞLU ENVER, DÜŞÜNSEL RUHAN, BİDECİ AYSUN, BODUROĞLU KORAY, Editor, GÜNEŞ TIP KİTABEVLERİ, Ankara, pp.1107-1111, 2020

11. **Mitokondriyal Hastalıklar**

OKUR İ.

in: TEMEL PEDİATRİ, HASANOĞLU ENVER, DÜŞÜNSEL RUHAN, BİDECİ AYSUN, BODUROĞLU KORAY, Editor, GÜNEŞ TIP KİTABEVLERİ, Ankara, pp.1136-1139, 2020

12. **Pediyatrik Hastalıklara Özel Beslenme ve Diyet Yönetimi**

Güler T., Mısırlı Özdemir E., Ergen Dibeklioglu S., Erdeniz E. H., Kul M., Bağ İ., Kılıç M., Zeybek S., Bulut M., Erolu E., et al.

Akademisyen Yayınevi Kitabevi, Ankara, 2019

13. **Dikkat Eksikliği Ve Hiperaktivite Bozukluğunda Nörometabolik Değerlendirme**

OKUR İ.

in: Dikkat Eksikliği Ve Hiperaktivite Bozukluğu, Şebnem Soysal, Editor, Nobel, pp.483-488, 2019

14. **Normal Çocuklukta Beslenme ve Beslenme Bozuklukları**

TÜMER L., İNCİ A., OKUR İ., Kasapkara Ç. S., OLGAC M. A. B.

in: Lange - Current Tanı ve Tedavi Pediatri, Prof.Dr. Enver Hasanoğlu Prof.Dr. Aysun Bideci Prof.Dr. Elif N. Özmert Prof.Dr. Sevcin A. BAKKALOĞLU EZGÜ, Editor, ema tıp kitapevi, pp.281-308, 2018

15. **Peroksizomal Bozukluklar**

OKUR İ.

in: Yurdakök Pediatri, Murat Yurdakök, Editor, Güneş Tıp Kitapevleri, Ankara, pp.1813-1820, 2017

1. **Fenilketonüri Tanısı İle İzlenen Hastalarda Visseral Adipositenin Değerlendirilmesi**
Gökarp S., Bostancı F., Aktaş E., İnci A., Okur İ., Ezgü F. S., Tümer L.
I. Ulusal Çocuk Beslenme Kongresi, Gaziantep, Turkey, 25 October 2023
2. **Glikojen Depo Tip 1 Hastalığında Lif Takviyesinin Metabolik Kontrole Etkisi**
Emecen Şanlı M., Aktaş E., İnci A., Okur İ., Ezgü F. S., Tümer L.
I. Ulusal Çocuk Beslenme Kongresi, Gaziantep, Turkey, 25 October 2023
3. **Bone Turnover in Patients with Lysosomal Storage Disorders**
Gökarp S., İnci A., Okur İ., Ezgü F. S., Tümer L.
Annual Symposium 2023, Jerusalem, Yerushalayim, Israel, 29 August - 01 September 2023
4. **Pterin Profiling in Serum, Dry Blood Spot and Urine using LC-MS/MS in Patients with Hyperphenylalaninemia**
Öktem R. M., İnci A., Bayrak H., Demir F., Biberoğlu G., Mavis M. E., Okur İ., Ezgü F. S., Tümer L.
Annual Symposium 2023, Jerusalem, Yerushalayim, Israel, 29 August 2023
5. **Intestinal Microbiota Composition of Children with Glycogen Storage Disease Type 1**
Gökarp S., Dinleyici E. Ç., Muluk C., İnci A., Aktaş E., Okur İ., Ezgü F. S., Tümer L.
SSIEM 2023, Yerushalayim, Israel, 29 August 2023
6. **An Alternative for Early Detection of Cardiac Involvement in Gaucher Type 1 Disease: Speckle Tracking Echocardiography**
GÖKALP S., ÜNLÜ S., İNCİ A., OKUR İ., EZGÜ F. S., TÜMER L.
Annual Symposium SSIEM 2023, Israel, 29 August - 01 September 2023
7. **EVENT BASED TREATMENT OUTCOMES OF PATIENTS WITH GAUCHER DISEASE: A DIFFERENT PERSPECTIVE**
Kilic A., İnci A., Okur İ., Tümer L., Ezgü F. S.
Annual Symposium 2023, Jerusalem, Yerushalayim, Israel, 29 August 2023
8. **Fosfomevalonatin Enzim Eksikliğine Bağlı Hiperimmünglobulin D Sendromu mu?**
Yıldız Ç., Gezin Yıldırım D., İnci A., Tümer L., Ergin F. B., Sunar Yayla E. N., Esmeray Şenol P., Karaçayır N., Eğritaş Gürkan Ö., Okur İ., et al.
22. Ulusal Romatoloji Kongresi, Antalya, Turkey, 26 - 30 October 2022, pp.29
9. **3-O Metil Dopa ölçümü ile AADC eksikliği taraması**
Öktem R. M., Biberoğlu G., İnci A., Okur İ., Ezgü F. S., Tümer L.
KBUD Kongre, Lab EXPO 2022, Antalya, Turkey, 03 October 2022
10. **Lysosphingolipids in the screening of sphingolipidoses**
Öktem R. M., İnci A., Biberoğlu G., Okur İ., Ezgü F. S., Tümer L.
360 LYSOSOME_FEBS Advanced Lecture Course_2022, İzmir, Turkey, 04 October 2022
11. **Retargeting phenylbutyrate, ursodeoxycholic acid, pyrimethamine and betaine for beta-glucocerebrosidase recovery in gaucher disease fibroblasts resulting from homozygous p.L483P mutation**
Kiliç A., BİBEROĞLU G., ÖKTEM R. M., İNCİ A., Aydogdu S., Udgu Isık B., IŞIK GÖNÜL İ., OKUR İ., TÜMER L., EZGÜ F. S.
SSIEM Annual Symposium, Germany, 30 August - 02 September 2022, no.1418955
12. **Is The Gut Microbiota Affected By The Special Diet Treatments of Inherited Metabolic Diseases?**
Gökarp S., Dinleyici E. Ç., Muluk C., İnci A., Okur İ., Ezgü F. S., Tümer L.
SSIEM Annual Symposium 2022, Freiburg, Germany, 30 August - 02 September 2022, pp.172-173
13. **Evaluation of simultaneous succinylacetone and nitroisone measurements in type-1 tyrosinemia follow-up.**
Öktem R. M., İnci A., Biberoğlu G., Okur İ., Ezgü F. S., Tümer L.
SSIEM Annual Symposium, Freiburg, Germany, 30 August - 02 September 2022, vol.45, no.222288, pp.99
14. **Analytical performance of guanidinoacetate-creatine test with LC-MS/MS**
Öktem R. M., İnci A., Biberoğlu G., Okur İ., Ezgü F. S., Tümer L.
SSIEM Annual Symposium, Freiburg, Germany, 30 August - 02 September 2022, vol.45, no.1418955, pp.687
15. **Evaluation of succinylacetone and nitroisone measurement: analytical performance requirements**
Öktem R. M., İnci A., Biberoğlu G., Okur İ., Ezgü F. S., Tümer L.

SSIEM Annual Symposium, Freiburg, Germany, 30 August - 02 September 2022, pp.414

16. **NEW PERSPECTIVES FOR THE TREATMENT and FOLLOW UP OF GLYCOGEN STORAGE DISEASE TYPE V: DL-3-HYDROXYBUTYRIC ACID WITH MODIFIED ATKINS DIET and QUADRICEPS FEMORIS SHEAR WAVE ELASTOGRAPHY**
Aktaşoğlu E., Kiliç A., Şanlı M. E., İnci A., Aktas E., Akdulum İ., Yaylı ., Okur İ., Ezgü F. S., Tümer L.
SSIEM Annual Symposium 2022, Freiburg, Germany, 30 August - 02 September 2022, pp.338
17. **A Different Approach For The Treatment Of Gastrointestinal Involvement In A Patient With Early Onset Lysosomal Acid Lipase Deficiency**
Altun Duman A. N., İnci A., Bükülmez A., Gökalp S., Kiliç A., Özsaydi Aktasoglu E., Demir F., Çavuşoğlu Y. H., Okur İ., Ezgü F. S., et al.
SSIEM Annual Symposium 2022, Freiburg, Germany, 30 August - 02 September 2022, pp.604
18. **A Different Approach to the Treatment of Type III Multiple Acyl CoA Dehydrogenase Deficiency: Modified Corn Starch**
İnci A., Aktaş E., Okur İ., Biberoğlu G., Tümer L., Ezgü F. S.
SSIEM 2022-, Freiburg, Germany, 30 August - 02 September 2022, pp.200
19. **Clinical and radiological evaluation of normal liver sizes in malnourished children:Gazi-PULSAM liver scale**
ÖZDÖNMEZ G. T., AKDULUM İ., ÖZTÜRK H., GÜRCAN KAYA N., AKSAKAL M., OKUR İ., KARAKAŞ N. M., KULA S., Konuş Boyunağa Ö., EĞRİTAŞ GÜRKAN Ö.
ESPGHAN 54th ANNUAL MEETING, Kopenhag, Denmark, 20 June 2022
20. **İdrarda organik asit taramasının optimizasyonu**
Öktem R. M., İnci A., Biberoğlu G., Okur İ., Ezgü F. S., Tümer L.
XVI. Uluslararası Katılımlı Metabolik Hastalıklar ve Beslenme Kongresi, Hatay, Turkey, 28 May - 01 June 2022, pp.409-410
21. **Krabbe hastalığında önemli bir biyobelirteç: Psikosin ölçümünün analitik performansı**
Öktem R. M., İnci A., Biberoğlu G., Okur İ., Ezgü F. S., Tümer L.
XVI. Uluslararası Katılımlı Metabolik Hastalıklar ve Beslenme Kongresi, Hatay, Turkey, 28 May - 01 June 2022, pp.411-412
22. **HOMOZİGOT p.L483P MUTASYONUNA BAĞLI GLUKOSEREBROSİDAZ EKSİKLİĞİNDE FENİLBÜTİRAT, URSODEOKSİKOLİK ASİT, BETAİN VE PRİMETAMİNİN YENİDEN HEDEFLERENEREK İNVİTRO ENZİM AKTİVİTESİNİ RESTORASYON POTANSİYELLERİNİN İNCELENMESİ**
Kılıç A., Biberoğlu G., Öktem R. M., İnci A., Işık Gönül İ., Okur İ., Tümer L., Ezgü F. S.
XVI. Uluslararası Katılımlı Metabolik Hastalıklar ve Beslenme Kongresi, Hatay, Turkey, 28 May - 01 June 2022, pp.35
23. **Sık düşünülen ancak nadir saptanan hastalığın yeni saptanmış bir mutasyonu: Transaldolaz (TALDO) Eksikliği**
Çilesiz K., Gökalp S., Demir F., Altun Duman A. N., İnci A., Okur İ., Tümer L., Ezgü F. S.
XVI. Uluslararası Katılımlı Metabolik Hastalıklar ve Beslenme Kongresi, Hatay, Turkey, 28 May - 02 June 2022, pp.354
24. **Mukopolisakkaridoz Tip IVA Tanılı Hastalarda Enzim Replasman Tedavisine Bağlı Anafilaksi ve Yönetimi: Tek Merkez Deneyimi**
ERTOY KARAGÖL H. İ., BAKIRTAŞ A., POLAT TERECE S., İNCİ A., EZGÜ F. S., TÜMER L., OKUR İ., Ayse K., KÖKEN G., DEMİR F., et al.
4. Genç Pediatrik Alerjistler Sempozyumu, Turkey, 19 May 2022
25. **A biomarker for glycogen storage diseases: Glucotetrasaccharid (Glc4)**
Öktem R. M., İnci A., Biberoğlu G., Okur İ., Ezgü F. S., Tümer L.
Uluslararası Katılımlı XXII. Ulusal Klinik Biyokimya Kongresi, Antalya, Turkey, 12 - 15 May 2022, pp.303-304
26. **MPS 6 Hastalarında Klinik Bulgular, ERT önce ve Sonrası Olay Bazlı Değerlendirme**
İNCİ A., OKUR İ., TÜMER L., BİBEROĞLU G., ÖKTEM R. M., EZGÜ F. S.
VII. Uluslararası Katılımlı Lizozomal Hastalıklar Kongresi, Turkey, 25 - 27 November 2021
27. **Gaucher Tip I Hastalığında Kardiyak Tutulumun Erken Saptanması için Bir Alternatif: Speckle Tracking Ekokardiyografi**

GÖKALP S., ÜNLÜ S., İNCİ A., OKUR İ., EZGÜ F. S., TAÇOY G., EMİNOĞLU F. T., KASAPKARA Ç. S., TÜMER L.
VII. Uluslararası Katılımlı Lizozomal Hastalıklar Kongresi 25-27 Kasım 2021 Çevrimiçi Kongre
<http://lizozomal2021.org/>, Turkey, 25 - 27 November 2021

28. **İNFANTİL TİP POMPE HASTALIĞI ULUSAL KONSENSUS ÇALIŞMASI**

Aktaşoğlu E., İNCİ A., OKUR İ., BİBEROĞLU G., ÖKTEM R. M., EZGÜ F. S., TÜMER L., KILIÇ M., GÜNEŞ S., KAĞNICI M., et al.

VII. Uluslararası Katılımlı Lizozomal Hastalıklar Kongresi, Turkey, 25 - 27 November 2021

29. **Gastrointestinal Involvement at the Junction of Wolman Disease and COVID 19**

ALTUN A., İNCİ A., KILIÇ A., ÖZSAYDI Aktaşoğlu E., GÖKALP S., DEMİR F., ÇAVUŞOĞLU Y. H., YİĞİT S., BOZDAYI G., OKUR İ., et al.

14th International Congress Of Inborn Errors Of Metabolism, Sidney, Australia, 21 - 23 November 2021

30. **A PATIENT WITH ADENOSINE KINASE DEFICIENCY DUE TO A NOVEL MUTATION PRESENTING WITH NOVEL DYSMORPHIC AND CARDIAC FINDINGS**

Aktaşoğlu E., KILIÇ A., Emecan Şanlı M., GÖKALP S., İNCİ A., OKUR İ., EZGÜ F. S., SARI S., DALGIÇ B., CEYLANER S., et al.
14th International Congress of Inborn Errors of Metabolism 2021, Australia, 21 - 24 November 2021

31. **Pompe Hastalarında Enzim Replasman Tedavisine Bağlı Anafilaksi ve Yönetimi:Tek Merkez Deneyim**

ERTOY KARAGÖL H. İ., İNCİ A., Polat Tecere S., KILIÇ A., Demir F., YAPAR D., OKUR İ., EZGÜ F. S., TÜMER L., BAKIRTAŞ A.

XXVIII. Ulusal Alerji ve Klinik İmmünoloji kongresi, Turkey, 13 - 17 October 2021

32. **Akut Porfiria Benzeri Periferik Nöropati Gelişen Tirozinemi Tip 1 Olgusu**

AKKUZZU E., ÖZEN DEMİRCİOĞLU P., İNCİ A., OKUR İ., EZGÜ F. S.

16. Çocuk Acil Tıp ve Yoğun Bakım Kongresi, Antalya, Turkey, 2 - 05 October 2019, pp.327-328

33. **Screening of twelve lysosomal storage diseases with LC-MS/MS in Gazi university hospital in Turkey: The first results of validation**

BİBEROĞLU G., İNCİ A., DERİN B., OKUR İ., EZGÜ F. S., TÜMER L.

SSIEM, 3 - 06 September 2019

34. **Beneficial effects of Modified Atkins Diet in Glycogen Storage Disorder Type IIIa**

OLGAÇ KILIÇKAYA M. A. B., İNCİ A., OKUR İ., KASAPKARA Ç. S., BİBEROĞLU G., OĞUZ A. D., AKTAŞ E., EZGÜ F. S., TÜMER L.

SSIEM Annual Symposium 2019, Rotterdam, Netherlands, 3 - 06 September 2019

35. **Next generation DNA sequencing as an initial diagnostic method for congenital defects of glycosylation**

EZGÜ F. S., İNCİ A., ÇİFTÇİ B., TÜMER L., OKUR İ., TOPÇU B., HASANOĞLU A.

SSIEM 2019, 3-6th September, 2019, Rotterdam-The Netherlands, 3 - 06 September 2019

36. **Beneficial Effects of Modified Atkins Diet in Glycogen Storage Disease Type IIIa**

OLGAÇ M. A. B., İNCİ A., OKUR İ., Kasapkara Ç. S., BİBEROĞLU G., OĞUZ A. D., Aktaş E., EZGÜ F. S., TÜMER L.

SSIEM 2019, 3-6th September, 2019, Rotterdam-The Netherlands, 3 - 06 September 2019

37. **Diyet tedavisine cevap veren HMG-CoA liyaz enzim eksikliği olan iki olgu**

KOÇ N., KUYUCU A., GÜNDÜZ M., OKUR İ., ÖZAYDIN E.

Hacettepe Beslenme ve Diyetetik Günleri IV. Mezuniyet Sonrası Eğitim Kursu, Ankara, Turkey, 27 - 29 June 2019

38. **Cornelia de Lange Syndrome and Glycogen Storage Disease Together in a Patient**

KILIÇ A., EMECAN ŞANLI M., ÖZSAYDI E., İNCİ A., OKUR İ., TÜMER L., EZGÜ F. S.

International Inborn Errors Of Metabolism And Nutrition Congress, İstanbul, Turkey, 10 - 14 April 2019

39. **Could Targeted Next Generation Sequencing Be A First Line Diagnostic Method for Lysosomal Storage Disease?**

CENGİZ F. B., İNCİ A., BİBEROĞLU G., ÇİFTÇİ B., TOPÇU B., TOKGÖZ D., YAZAR Ö. F., GÖKMENOĞLU H., RAJ Y., OKUR İ., et al.

International Inborn Errors of Metabolism and Nutrition Congress, 10 - 14 April 2019

40. **Screening of Twelve Lysosomal Storage Diseases with LC-MS/MS in Gazi University Hospital: The First Results of Validation.**

BİBEROĞLU G., İNCİ A., DERİN B., OKUR İ., EZGÜ F. S., TÜMER L.

INTERNATIONAL INBORN ERRORS OF METABOLISM AND NUTRITION CONGRESS 10 - 14 April 2019 İstanbul-

Turkey, 10 - 14 April 2019

41. **Familial Hyperphosphatemic Tumoral Calcinosis in an Unusual Site**
Emecan Şanlı M., Özsaydı E., Kılıç M., İNCİ A., OKUR İ., EZGÜ F. S., TÜMER L.
International Inborn Errors Of Metabolism And Nutrition Congress 10 - 14 April 2019, Istanbul-Turkey, 10 - 14 April 2019
42. **Hyperinsulinemic Hypoglycemia: Think of GLUD1 Gene Mutation Leading To Hyperinsulinism/Hyperammonemia (HI/HA) Syndrome**
Emecan Şanlı M., Kılıç M., Özsaydı E., İNCİ A., OKUR İ., TÜMER L., EZGÜ F. S.
International Inborn Errors Of Metabolism And Nutrition Congress 10 - 14 April 2019, Istanbul-Turkey, 10 - 14 April 2019
43. **Could Targeted Next Generation Sequencing Be A First Line Diagnostic Method for Lysosomal storage Diseases?**
ERGİN F. B., İNCİ A., BİBEROĞLU G., ÇİFTÇİ B., TOPÇU YÜCE A. B., TOKGÖZ D., YAZAR Ö. F., GÖKMENOĞLU H., RAJ Y., OKUR İ., et al.
INTERNATIONAL INBORN ERRORS OF METABOLISM AND NUTRITION CONGRESS 10 - 14 April 2019 Istanbul-Turkey, Turkey, 10 - 14 April 2019
44. **Hyperinsulinemic Hypoglycemia: Think of GLUD1 dgene mutation leading to Hyperinsulinemic hyperammonemia (HI/HA syndrome)**
EMECAN ŞANLI M., KILIÇ A., AKTAŞOĞLU E., İNCİ A., OKUR İ., TÜMER L., EZGÜ F. S.
INTERNATIONAL INBORN ERRORS OF METABOLISM AND NUTRITION CONGRESS 10 - 14 April 2019 Istanbul-Turkey, Turkey, 10 - 14 April 2019
45. **Novel Mutation in FBP1 Gene Presenting with Recurrent Episodes of Vomiting in A Child**
Emecan Şanlı M., Kılıç M., Özsaydı E., İNCİ A., OKUR İ., EZGÜ F. S., TÜMER L.
International Inborn Errors Of Metabolism And Nutrition Congress 10 - 14 April 2019, Istanbul-Turkey, 10 - 14 April 2019
46. **A Very Rare Disease: Hyperornithinemia-Hyperammonemia-Homocitrullinuria (Hhh) Syndrome**
Özsaydı E., Emecan Şanlı M., Kılıç M., İNCİ A., OKUR İ., TÜMER L., EZGÜ F. S.
International Inborn Errors Of Metabolism And Nutrition Congress 10 - 14 April 2019, Istanbul-Turkey, 10 - 14 April 2019
47. **Novel Mutation in Two Siblings with Normouricemic Lesch Nyhan Syndrome**
Emecan Şanlı M., Özsaydı E., Kılıç M., İNCİ A., OKUR İ., EZGÜ F. S., TÜMER L.
International Inborn Errors Of Metabolism And Nutrition Congress 10 - 14 April 2019, Istanbul-Turkey, 10 - 14 April 2019
48. **Could Targeted Next Generation Sequencing Be A First Line Diagnostic Method for Lysosomal storage Diseases**
İNCİ A., OKUR İ., AKKUZU E., DÖĞER E., BİBEROĞLU G., KALKAN G., TÜMER L., EZGÜ F. S.
INTERNATIONAL INBORN ERRORS OF METABOLISM AND NUTRITION CONGRESS 10 - 14 April 2019 Istanbul-Turkey, 10 - 14 April 2019
49. **Growth Hormone Treatment: Reverses Catabolic Process in Inborn Errors of Metabolism**
İNCİ A., OKUR İ., AKKUZU E., DÖĞER E., BİBEROĞLU G., KALKAN G., TÜMER L., EZGÜ F. S.
International Inborn Errors Of Metabolism And Nutrition Congress 10 - 14 April 2019, Istanbul-Turkey, 10 - 14 April 2019
50. **Natural history data for young subjects with Sanfilippo syndrome type B (MPS IIIB)**
Villarreal M. S., OKUR İ., Cleary M., Lopez M. J. d. C., Harmatz P., Lee J., Lin S., Couce M. L., Muschol N., Peters H., et al.
15th Annual Research Meeting of the WORLD Symposium(TM), Florida, United States Of America, 4 - 07 February 2019, vol.126
51. **ICV-administered tralesenidase alfa (BMN 250 NAGLU-IGF2) is well-tolerated and reduces heparan sulfate accumulation in the CNS of subjects with Sanfilippo syndrome type B (MPS IIIB)**
Cleary M., Muschol N., Luz Couce M., Harmatz P., Lee J., Lin S., OKUR İ., Ezgu F. S., Peters H., Villarreal M. S., et al.
15th Annual Research Meeting of the WORLD Symposium(TM), Florida, United States Of America, 4 - 07 February 2019, vol.126

52. **Epilepsy in Biotinidase Deficiency Is Distinct from Early Myoclonic Encephalopathy**
Guliyeva U., Dulac O., Okur İ., Khalilov O., Guliyeva S.
13th European Congress on Epileptology, Vienna, Austria, 26 - 30 August 2018, vol.59
53. **Respiratory system involvement of 41 Mucopolysaccharidosis patients with the evaluation of KL-6, SPA and SPD levels**
İNCİ A., OKUR İ., Yılmaz Demirtaş C., BİBEROĞLU G., ASLAN A. T., EZGÜ F. S., TÜMER L.
15 th MEMG, Beirut, 29 November - 02 December 2018
54. **UNIQUE CLINICAL AND MOLECULAR FINDINGS IN LARGE COHORT OF PATIENTS WITH GAUCHER DISEASE FROM TURKEY**
Akay Tayfun G., OKUR İ., BİBEROĞLU G., TÜMER L., İNCİ A., Küçükcongar A., Hasanoğlu A., EZGÜ F. S.
Gaucher Symposium, İstanbul, Turkey, 21 - 22 October 2018
55. **ICV-administered BMN 250 (NAGLU-IGF2) is Well Tolerated and Reduces Heparan Sulfate Accumulation in the CNS of Subjects with Sanfilippo Syndrome Type B (MPS IIIB)**
Muschol N., Cleary M., Couce M., Harmatz P., Lee J., Lin S. P., Okur İ., Ezgu F. S., Peters H., Villarreal M., et al.
47th Annual Meeting of the Child-Neurology-Society (CNS), Illinois, United States Of America, 15 - 18 October 2018, vol.84
56. **Glycogen storage disease type 9: Insidious onset,mild form**
TÜMER L., İNCİ A., OKUR İ., BİBEROĞLU G., EZGÜ F. S.
SSIEM, 4 - 07 September 2018
57. **Respiratory system involvement of mucopolysaccharidosis patients with the evaluation of KL-6, SPA and SPD levels**
İNCİ A., OKUR İ., YILMAZ-DEMİRTAŞ C., BİBEROĞLU G., aslan A. T., EZGÜ F. S., TÜMER L.
SSIEM, 4 - 07 September 2018
58. **Determination of succinylacetone in dried blood spot: preliminary results of our laboratory**
BİBEROĞLU G., TÜMER L., OKUR İ., EZGÜ F. S., İNCİ A.
SSIEM, 4 - 07 September 2018
59. **An early diagnosis cerebretendinous xanthomatosis in a patient at the age of 15 years**
İNCİ A., BİBEROĞLU G., OKUR İ., TÜMER L., EZGÜ F. S.
SSIEM, 4 - 07 September 2018
60. **The clinical evaluation of Fabry patientswith Mainz severity score index and DS3 score**
OKUR İ., İNCİ A., bütün s., BİBEROĞLU G., EZGÜ F. S., TÜMER L.
SSIEM, 4 - 07 September 2018
61. **Natural history data for young subjects with Sanfilippo Syndrome Type B (MPS IIIB)**
OKUR İ., Cleary M., de Castro Lopez M. J., Harmatz P., Lees J., Lin S., Luz Couce M., Muschol N., Peters H., Solano Villarrea M., et al.
40th Annual Meeting of the Society-for-Inherited-Metabolic-Disorders (SIMD), California, United States Of America, 11 - 14 March 2018, vol.123, pp.255-256
62. **Natural history data for young subjects with Sanfilippo syndrome type B (MPS IIIB)**
Okur İ., Cleary M., de Castro Lopez M. J., Harmatz P., Lee J., Lin S., Luz Couce M., Muschol N., Peters H., Solano Villarreal M., et al.
We're Organizing Research for Lysosomal Diseases (WORLD) Symposium, California, United States Of America, 5 - 09 February 2018, vol.123
63. **Preliminary Results of Our Laboratoryfor Bile Acid Metabolism Disorders**
BİBEROĞLU G., DERİN B., İNCİ A., OKUR İ., EZGÜ F. S., TÜMER L.
ICIEM, 5 - 08 September 2017
64. **Short Chain Fatty Acid OxidationDefect in an Adult Patient With RefractorySeizures**
İNCİ A., TÜMER L., OKUR İ., BİBEROĞLU G., EZGÜ F. S.
ICIEM, 5 - 08 September 2017
65. **Carnitine Acyl Carnitine TranslocaseDeficiency With Severe Hyperammonemiaand Hypoglycemia**
İNCİ A., OKUR İ., OLGAC M. A. B., AKKUZU E., BİBEROĞLU G., EZGÜ F. S., TÜMER L.
ICIEM, 5 - 08 September 2017

66. **In Vitro Stopcodon Readthrough ofAlfa-Galactosidase and Alfa-GlucosidasePremature Termination Codons UsingGentamicin, Geneticin, and Ataluren:Therapeutic Potential for Fabry and PompeDiseases**
dundar h., BİBEROĞLU G., OKUR İ., TÜMER L., EZGÜ F. S.
ICIEM, 5 - 08 September 2017
67. **Screening ALPL Gene Differences byNext Generation Sequence Techonology inPatients Having Low ALP Levels**
İNCİ A., EZGÜ F. S., topcu b., çiftci b., OKUR İ., BİBEROĞLU G., TÜMER L.
ICIEM, 5 - 08 September 2017
68. **DiagnosticCapability ofNextGenerationDNA Sequencing With A 450 Gene Panel forInborn Errors of Metabolism**
EZGÜ F. S., çiftci b., topcu b., İNCİ A., OKUR İ., BİBEROĞLU G., hasanoğlu a.
ICIEM, 5 - 08 September 2017
69. **Renal Involvement in Fabry Disease**
İNCİ A., BİBEROĞLU G., OKUR İ., PAŞAOĞLU Ö. T., TÜMER L., PAŞAOĞLU H., EZGÜ F. S.
ICIEM, 5 - 08 September 2017
70. **İnvestigation of LDLR Gene Mutations in Turkish Patients With Familial Hypercholesterolemia**
OKUR İ., İNCİ A., OLGAÇ M. A. B., ÇİFTÇİ B., TOPÇU B., TÜMER L., EZGÜ F. S.
13th International Congress of Inborn Errors of Metabolism - ICIEM 2017, 5 - 08 September 2017, vol.5
71. **THE HEMATOLOGIC FINDINGS OF INHERITED METABOLIC DISEASE; THEY ARE MORE THAN EXPECTED**
Yenicesu I., Sal A. E., Okur İ., Kaya Z., Ezgu F. S., Kocak U., Tumer L.
22nd Congress of the European-Hematology-Association, Madrid, Spain, 22 - 25 June 2017, vol.102, pp.829-830
72. **Karbonhidrat Metabolizması Bozuklukları**
OKUR İ.
Çocuk Gastroenteroloji, Hepatoloji ve Beslenme Güncelleme Toplantısı, Turkey, 1 - 04 June 2017
73. **Tirozinemi İzlem ve Tedavi**
OKUR İ.
Çocuk Gastroenteroloji, Hepatoloji ve Beslenme Güncelleme Toplantısı, Turkey, 1 - 04 June 2017
74. **Ciddi hiperammonemi ve hipoglisemi ile giden karnitin-açıl translokaz olgusu**
İNCİ A., OLGAÇ KILIÇKAYA M. A. B., OKUR İ., AKKUZU E., BİBEROĞLU G., EZGÜ F. S., TÜMER L.
14. Ulusal Metabolik Hastalıklar ve Beslenme Kongresi, Muğla, Turkey, 26 - 30 April 2017
75. **Lizozomal Depo Hastalıklarında Nörolojik Bulgular ve Tedavileri**
OKUR İ.
14.Ulusal Metabolik Hastalıklar ve Beslenme Kongresi, Turkey, 26 - 30 April 2017
76. **Mukopolisakkaridozlar: Tanı ve Tedavide Güncel Yaklaşımları**
OKUR İ.
39. Pediatri Günleri ve 18. Pediatri Hemşireliği Günleri, Turkey, 2 - 05 April 2017
77. **Yenidoğanda Tarama Testleri**
OKUR İ.
İlk 1000 gün 5. Kongresi, Turkey, 19 - 22 March 2017
78. **Ailevi Hiperkolesterolemi Olan Türk Hastalarda LDLR Gen Mutasyonlarının Araştırılması**
OKUR İ., EZGÜ F. S., İNCİ A., OLGAÇ M. A. B., TÜMER L.
2. Ege Endokrin Hastalıklar ve Genetik Sempozyumu, Turkey, 23 - 25 February 2017
79. **In vitro translational readthrough by gentamicin and geneticin improves GLA activity in Fabry disease**
Dündar H., Biberoğlu G., Okur İ., Tümer L., Ezgü F. S.
13th Annual Research Meeting on We're Organizing Research for Lysosomal Diseases (WORLD), California, United States Of America, 13 - 17 February 2017, vol.120
80. **Evaluation of chitotriosidase and high sensitive c reactive protein levels in mucopolysaccharidosis**
İNCİ A., GENÇ B., YILMAZ-DEMİRTAŞ C., UDGU B., KARAOĞLU A., OKUR İ., EZGÜ F. S., BİBEROĞLU G., TÜMER L.

13th Middle East Metabolic Group Meeting/ Amman-Jordan, 28 - 30 October 2016

81. **ÇOCUKLARDA VİTAMİN VE MİNERAL DESTEĞİ**

OKUR İ.

60. Türkiye Milli Pediatri Kongresi, Antalya, Turkey, 9 - 13 November 2016

82. **Evaluation of chitotriosidase and high sensitivity c reactive protein levels in mucopolysaccharidosis patients**

İNCİ A., DERİN B., YILMAZ C., udgu b., KARAOĞLU A., OKUR İ., EZGÜ F. S., BİBEROĞLU G., TÜMER L.
MEMG, 28 - 30 October 2016

83. **Do cytokine levels play a role in the pathogenesis of mucopolysaccharidosis patients**

İNCİ A., TÜMER L., YILMAZ-DEMİRTAŞ C., KARAOĞLU A., OKUR İ., OLGAC M. A. B., EZGÜ F. S., BİBEROĞLU G.
13th Middle East Metabolic Group Meeting/Amman -Jordan, 28 - 30 October 2016

84. **Evaluation of chitotriosidase and high sensitive c reactive protein levels in mucopolysaccharidosis**

İNCİ A., GENÇ B., YILMAZ-DEMİRTAŞ C., UDGU B., KARAOĞLU A., OKUR İ., EZGÜ F. S., BİBEROĞLU G., TÜMER L.
13th MEMG Meeting, 28 ekim-30kasım 2016, Amman, Jordan, 28 - 30 October 2016

85. **Could propionylcarnitine and free carnitine be used as antioxidative markers in mucopolysaccharidosis**

İNCİ A., BİBEROĞLU G., DERİN B., KARAOĞLU A., OKUR İ., EZGÜ F. S., TÜMER L.
MEMG, 28 - 30 October 2016

86. **Early initiation of investigational enzyme replacement therapy in a nine month old infant with mucopolysaccharidosis type VII**

KARAOĞLU A., İNCİ A., BİBEROĞLU G., OKUR İ., kılıçkaya a., TÜMER L., king b., haller c., EZGÜ F. S.
MEMG, 28 - 30 October 2016

87. **Evaluation of gentamycin for stop codon readthrough therapy in Fabry disease**

halil d., BİBEROĞLU G., çiftci b., topcu b., OKUR İ., TÜMER L., EZGÜ F. S.
MEMG, 28 - 30 October 2016

88. **The specificity and sensitivity of next generation semiconductor DNA sequencing in detecting heteroplasmic mitochondrial**

EZGÜ F. S., topcu b., çiftci b., düNDAR H., BİBEROĞLU G., OKUR İ., TÜMER L.
MEMG, 28 - 30 October 2016

89. **Evaluation of chitotriosidase and high sensitivity c reactive protein levels in mucopolysaccharidosis**

İNCİ A., Genç B., Demirtaş C., Udgu B., KARAOĞLU A., OKUR İ., EZGÜ F. S., BİBEROĞLU G., TÜMER L.
SSIEM 2016: Annual Symposium of the Society for the Study of Inborn Errors of Metabolism, Rome, Italy, 6 - 09 September 2016

90. **Early initiation of investigational enzyme replacement therapy in a 9 month old infant with mucopolysaccharidosis type VII**

KARAOĞLU A., İNCİ A., BİBEROĞLU G., OKUR İ., Kılıçkaya A., KELEŞ E., TÜMER L., King B., Hall C., EZGÜ F. S.
SSIEM 2016: Annual Symposium of the Society for the Study of Inborn Errors of Metabolism, Rome, Italy, 6 - 09 September 2016

91. **Type 1 hypersensitivity reaction and desensitization with Elosulphase alpha**

İNCİ A., Kan A., Topuz B., OKUR İ., EZGÜ F. S., TÜMER L.
SSIEM 2016: Annual Symposium of the Society for the Study of Inborn Errors of Metabolism, Rome, Italy, 6 - 09 September 2016

92. **Do cytokine levels play a role in pathogenesis of mucopolysaccharidosis patients**

İNCİ A., TÜMER L., Demirtaş C., KARAOĞLU A., OKUR İ., OLGAC M. A. B., EZGÜ F. S., BİBEROĞLU G.
SSIEM 2016: Annual Symposium of the Society for the Study of Inborn Errors of Metabolism, Rome, Italy, 6 - 09 September 2016

93. **Bone mineral density and vitamin D status in inborn errors of metabolism**

OLGAC M. A. B., TÜMER L., İNCİ A., KARAOĞLU A., OKUR İ., EZGÜ F. S.
SSIEM 2016: Annual Symposium of the Society for the Study of Inborn Errors of Metabolism, Rome, Italy, 6 - 09 September 2016

94. **Identification of a novel mutation in Turkish infant with early onset monocarboxylate transporter 1**

MCT1 deficiency as a cause of recurrent ketoacidosis

OKUR İ., İNCİ A., KELEŞ E., KARAOĞLU A., CEYLANER S., BİBEROĞLU G., EZGÜ F. S., TÜMER L.

SSIEM 2016: Annual Symposium of the Society for the Study of Inborn Errors of Metabolism, Rome, Italy, 6 - 09 September 2016

95. **The specificity and sensitivity of next generation semiconductor DNA sequencing in detecting mitochondrial DNA heteroplasmy**
EZGÜ F. S., Topçu B., Çiftçi B., OKUR İ., TÜMER L.
SSIEM 2016: Annual Symposium of the Society for the Study of Inborn Errors of Metabolism, Rome, Italy, 6 - 09 September 2016
96. **Identification of a novel mutation in Turkish infant with early onset monocarboxylate transporter 1 MCT1 deficiency as a cause of recurrent ketoacidosis**
OKUR İ., İNCİ A., KELEŞ E., KARAOĞLU A., CEYLANER S., BİBEROĞLU G., EZGÜ F. S., TÜMER L.
SSIEM 2016: Annual Symposium of the Society for the Study of Inborn Errors of Metabolism, Rome, Italy, 6 - 09 September 2016, vol.39, pp.35-284
97. **Bone mineral density and vitamin D status in inborn errors of metabolism**
OLGAÇ M. A. B., TÜMER L., İNCİ A., KARAOĞLU B., OKUR İ., EZGÜ F. S.
SSIEM 2016: Annual Symposium of the Society for the Study of Inborn Errors of Metabolism, Rome, Italy, 6 - 09 September 2016
98. **LAL enzim eksikliği**
OKUR İ.
11. Ulusal Çocuk Gastroenteroloji Hepatoloji ve Beslenme Kongresi, Turkey, 4 - 07 May 2016
99. **Fabry Hastalarında Subklinik Sol Ventrikül Disfonksiyonunun Speckle Tracking Ekokardiyografi ile Değerlendirilmesi**
GÖKALP G., OKUR İ., ÜNLÜ S., İNCİ A., EZGÜ F. S., ŞAHİNARSLAN A., TÜMER L.
V. Uluslararası Katılımlı Lizozomal Hastalıklar Kongresi, Turkey, 14 - 17 April 2016
100. **Plasma acylcarnitine levels Are there New İnflammatory markers in lysosomal storage disease**
BİBEROĞLU G., DERİN B., İNCİ A., udgu b., kurnaz p., OKUR İ., EZGÜ F. S., TÜMER L.
MEMG, 29 October - 01 November 2015
101. **Is there any effect of acylcarnitines on proinflammatory process in obese children**
BİBEROĞLU G., DERİN B., İNCİ A., DÖĞER E., OKUR İ., EZGÜ F. S., TÜMER L.
SSIEM, 1 - 04 September 2015
102. **Patient with Niemann Pick type C presenting with lymphatic involvement with Niemann Pick cells in the left jaw**
İNCİ A., OKUR İ., ESENDAĞLI G., OKUR A., EZGÜ F. S., TÜMER L.
Annual Symposium of the Society for the Study of Inborn Errors of Metabolism, Lyon, France, 1 - 04 September 2015
103. **Cobalamin C disease with hypopigmented cutaneous findings A unique case**
TÜMER L., ARHAN E., OKUR İ., Aydın K., Hirfanoglu T., Karaoğlu A., Öztürk Z.
annual symposium of the society for the study of inborn errors of metabolism, Lyon, France, 1 - 04 September 2015, vol.38, pp.319
104. **A completely new approach to the diagnosis of inborn errors development of a 450 gene all metabolic disorders next generation sequencing panel**
EZGÜ F. S., çiftçi b., topçu b., OKUR İ., İNCİ A., OLGAÇ M. A. B., KARAOĞLU A., BİBEROĞLU G., TÜMER L., hasanoğlu a.
SSIEM Annual Symposium, 1 - 04 September 2015
105. **Dihydrolipoamide dehydrogenase deficiency diagnosed by using new generation sequencing technology**
İNCİ A., TÜMER L., OKUR İ., OLGAÇ M. A. B., SARI S., çiftçi b., topçu b., EZGÜ F. S.
SSIEM Annual Symposium, 1 - 04 September 2015
106. **Sol çenede Lenfatik tutulum ile giden Niemann Pick tip C olgusu**
İNCİ A., OKUR İ., ESENDAĞLI G., OKUR A., OLGAÇ M. A. B., EZGÜ F. S., TÜMER L.
XIII. Ulusal Metabolik Hastalıklar ve Beslenme Kongresi, Adana, Turkey, 14 - 18 April 2015

107. **Lizozomal depo hastalıklarına yaklaşım**
OKUR İ.
2. Marmara Pediatri Kongresi, Turkey, 12 - 14 February 2015
108. **Importance of family screening in Fabry disease: Reaching the bottom of the iceberg**
Ezgu F. S., Koca S., OKUR İ., BİBEROĞLU G., TÜMER L., Bakkaloglu S. A., ERTEN Y., Hasanoglu A.
11th Annual WORLD Symposium of the Lysosomal-Disease-Network, Florida, United States Of America, 9 - 13 February 2015, vol.114
109. **COBALAMIN C DEFICIENCY WITH INFANTILE SPASM AND CUTANEOUS FINDINGS A UNIQUE CASE**
ÖZTÜRK Z., ARHAN E., AYDIN K., OKUR İ., TÜMER L., SERDAROĞLU A., HIRFANOĞLU T., AKBAŞ Y., HAVALI C.
31st International Epilepsy Congress, ISTANBUL, 5 - 09 September 2015
110. **COBALAMIN C DEFICIENCY WITH INFANTILE SPASM AND CUTANEOUS FINDINGS: A UNIQUE CASE**
Ozturk Z., Arhan E., Aydin K., Okur İ., Tumer L., Serdaroglu A., Hirfanoglu T., Akbas Y., Havalı C.
31st International Epilepsy Congress, İstanbul, Turkey, 5 - 09 September 2015, vol.56, pp.175
111. **The results of enzyme studies in the diagnosis of lysosomal diseases: 8 years experience of Gazi University, Ankara, Turkey**
Hasanoglu A., BİBEROĞLU G., OKUR İ., Turner L., EZGÜ F. S., Udgu B., Olgac A.
11th Annual WORLD Symposium of the Lysosomal-Disease-Network, Florida, United States Of America, 9 - 13 February 2015, vol.114
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 Sağlam 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

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

Pediatrici Uzmanlık Eğitimi, Expertise In Medicine, 2014 - 2015

Kalıtsal 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 (Kktc), Nisan 2023
Okur İ., Bideci A., Tezer H., 66. Türkiye Milli Pediatri Kongresi, Scientific Congress, Girne, Cyprus (Kktc), 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 (Kktc), 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 (Kktc), 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: 237
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 Joint 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 (Kktc), 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 (Kktc), 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 (Kktc), 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, Azerbaijan Tıp Üniversitesi ve ATA Pediatri Derneği Ortak Toplantısı), Conference, Türkiye Milli Pediatri Derneği, Azerbaijan 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