

# İLYAS OKUR

## ASSOC. PROF.

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### Learning Knowledge

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Post Doctorate of Medicine 2005 - 2009	Gazi University, Tıp Fakültesi, Dahili Tıp Bilimleri Bölümü, Turkey
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Expertise In Medicine 1999 - 2004	Gazi University, Tıp Fakültesi, Dahili Tıp Bilimleri Bölümü, Turkey
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Undergraduate 1992 - 1998	Istanbul University, İstanbul Tıp Fakültesi, İstanbul Tıp Pr., Turkey
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### Certificates, Courses and Trainings

Vocational Training, Eğiticilerin Eğitimi Kursu, Gazi Üniversitesi, 2014

Data Analysis, Temel Biyoistatistik Kursu, Gazi Üniversitesi, 2010

Health&Medicine, Deney Hayvanları Uygulama ve Etik Kursu, Gazi Üniversitesi, 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

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Associate Professor 2014 - Continues	Gazi University, Tıp Fakültesi, Dahili Tıp Bilimleri
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Expert 2014 - 2014	Gazi University, Tıp Fakültesi, Dahili Tıp Bilimleri
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Lecturer 2009 - 2011	Gazi University, Tıp Fakültesi, Dahili Tıp Bilimleri
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## Supported Projects

1. Okur İ., Şıvgın V., Ezgü F. S. , Tümer L., İnci A., Soysal Acar A. Ş. , Arhan E., Börcek A. Ö. , Project Supported by Private Organizations in Other Countries, BMN250-202--Mukopolisakaridoz Tip IIIB (MPS IIIB, Sanfilippo Sendromu Tip B) hastalarında İntraserebroventriküler BMN 250'nin Uzun Süreli Güvenliğini ve Verimliliğini değerlendirmek için çok merkezli, çok uluslu ek çalışma., 2018 - 2023
2. Okur İ., Ezgü F. S. , Tümer L., İnci A., Şıvgın V., Soysal Acar A. Ş. , Kurtipek Ö., Tutar H., Gündüz B., Other Supported Projects, BMN250-902--Mukopolisakaridoz Tip III B 'ye (MPS IIIB) Yönelik Prospektif doğal öykü çalışması, 2017 - 2023
3. Okur İ., Ezgü F. S. , Dünder H., TUBITAK Project, Metilmalonil Koenzim A Mutaz Enziminin Konformasyonel Bozukluğuna Bağlı Olarak Gelişen Metilmalonik Asidemi Hastalığının'xxda Farmakolojik Şaperon Uygulaması İle Enzim Aktivitesinin Yeniden Kazandırılması, 2017 - 2022
4. 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
5. 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
6. Okur İ., Ezgü F. S. , Tümer L., Soysal Acar A. Ş. , Şıvgın V., Kurtipek Ö., Gündüz B., İnci A., Tutar H., Arhan E., Other Supported Projects, BMN250-901-A Study of Mucopolysaccharidosis Type IIIB MPS IIIB (Mukopolisakaridoz tip IIIB'xye yönelik uluslararası çok merkezli, prospektif gözlemsel çalışma)-(BMN250-901 nolu proje kodu), 2015 - 2020
7. Özaslan A., Güney E., Ergün M. A. , Okur İ., Project Supported by Higher Education Institutions, Dikkat eksikliği hiperaktivite bozukluğunda cdh13 ve lphn3 gen polimorfizmleri: Klinik özellikler ve yönetici işlevlerle ilişkileri, 2018 - 2019
8. 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
9. Okur İ., Ezgü F. S. , Tümer L., Other Supported Projects, 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
10. 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
11. OKUR İ., Project Supported by Other Official Institutions, Kronik böbrek yetmezliği nedeniyle hemodiyaliz ve periton diyalizi uygulanan hastalarda Fabry hastalığını taranması, 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

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

1. **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., Biberöglü G., Öktem M., Ezgü F.  
Orphanet journal of rare diseases, vol.16, no.1, pp.438, 2021 (Journal Indexed in SCI Expanded)
2. **Tralesinidase alfa (AX 250) Enzyme Replacement Therapy for Sanfilippo Syndrome Type**  
Maricich S., Okur İ., Ezgu F., 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 (Journal Indexed in SCI)
3. **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 (Journal Indexed in SCI)
4. **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.  
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, vol.185, pp.2739-2747, 2021 (Journal Indexed in SCI)
5. **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 (Journal Indexed in SCI)
6. **Hypophosphatasia: is it an underdiagnosed disease even by expert physicians?**  
İnci A., Ergin F. B. C. , Yüce B. T. , Çiftçi B., Demir E., Buyan N., Okur İ., Biberöglü G., Öktem R. M. , Tümer L., et al.  
Journal of bone and mineral metabolism, vol.39, pp.598-605, 2021 (Journal Indexed in SCI)
7. **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 (Journal Indexed in SCI)
8. **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, 2021 (Journal Indexed in SCI)
9. **The chemical chaperone 4-phenylbutyrate enhances alpha-galactosidase activity subsequent to stop-codon read-through therapy with triamterene in Fabry R227X fibroblasts**  
Dündar H., Biberöglü G., İnci A., Işık Gönül İ., Okur İ., Tümer L., Ezgü F. S.  
MOLECULAR GENETICS AND METABOLISM, vol.132, no.2, 2021 (Journal Indexed in SCI)
10. **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 (Journal Indexed in SCI Expanded)
11. **Tralesinidase alfa (AX 250) enzyme replacement therapy for Sanfilippo syndrome type B**  
Muschol N., von Cossel K., OKUR İ., Ezgu F., 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 (Journal Indexed in SCI)
12. **Natural history of Sanfilippo syndrome type B in young patients: Ongoing results from two large, prospective studies**  
Giugliani R., OKUR İ., Ezgu F., 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 (Journal Indexed in SCI)
13. **Autism: Screening of inborn errors of metabolism and unexpected results**  
İNCİ A., ÖZASLAN A., OKUR İ., BİBEROĞLU G., GÜNEY E., EZGÜ F. S. , TÜMER L., İŞERİ E.  
AUTISM RESEARCH, 2021 (Journal Indexed in SCI)

14. **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., TÜMER L.  
JOURNAL OF PEDIATRIC ENDOCRINOLOGY & METABOLISM, vol.34, no.6, pp.813-816, 2021 (Journal Indexed in SCI)
15. **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 (Journal Indexed in SCI)
16. **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 (Journal Indexed in SCI)
17. **Nutritional Status of Syrian Refugees in Early Adolescence Living in Turkey**  
PEHLİVANTÜRK KIZILKAN M., ÖZMERT E. N. , DERMAN O., OKUR İ., KAYNAK M. O. , Adiguzel A., Sahan-Seref I., Kurekci F., BİDECİ A., Hasanoglu E.  
JOURNAL OF IMMIGRANT AND MINORITY HEALTH, vol.22, no.6, pp.1149-1154, 2020 (Journal Indexed in SSCI)
18. **Vitamin D Levels and Bone Mineral Density in Inborn Errors of Metabolism Requiring Specialised Diets**  
Olgac A., İNCİ A., OKUR İ., Ezgu F., BİBEROĞLU G., Turner L.  
JCSP-JOURNAL OF THE COLLEGE OF PHYSICIANS AND SURGEONS PAKISTAN, vol.29, no.12, pp.1207-1211, 2019 (Journal Indexed in SCI)
19. **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., BİBEROĞLU G., Kasapkara C., Kucukcongar A., DALGIÇ B., ÖZHAN OKTAR S., et al.  
GENE, vol.687, pp.280-288, 2019 (Journal Indexed in SCI)
20. **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 (Journal Indexed in SCI)
21. **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 (Journal Indexed in SCI)
22. **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 (Journal Indexed in SCI)
23. **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 (Journal Indexed in SCI)
24. **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 (Journal Indexed in SCI)
25. **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 (Journal Indexed in SCI)
26. **Audiologic evaluations of children with mucopolysaccharidosis**  
Gokdogan C., ALTINYAY 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 (Journal Indexed in SCI)
27. **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 (Journal Indexed in SCI)
28. **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 (Journal Indexed in SCI)
29. **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 (Journal Indexed in SCI)
30. **Monocarboxylate Transporter 1 Deficiency and Ketone Utilization**  
van Hasselt P. M. , Ferdinandusse S., Monroe G. R. , Ruiten 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 (Journal Indexed in SCI)
31. **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 (Journal Indexed in SCI)
32. **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., BİBEROĞLU G., Turner L., ERTEN Y., Isitman M., Eminoglu F. T. , Hasanoglu A.  
GENE, vol.527, no.1, pp.42-47, 2013 (Journal Indexed in SCI)
33. **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 (Journal Indexed in SCI)
34. **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 (Journal Indexed in SCI)
35. **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 (Journal Indexed in SCI)
36. **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 (Journal Indexed in SCI)
37. **GENOTYPIC FEATURES OF 41 PATIENTS WITH GAUCHER DISEASE FROM TURKEY**  
Hasanoglu A., Akay G., Ezgu F., Biberoglu G., Tumer L., Okur İ., Kucukcongar A., Kasapkara C., Polat M., Ciftci B.  
JOURNAL OF INHERITED METABOLIC DISEASE, vol.35, 2012 (Journal Indexed in SCI)
38. **COMPREHENSIVE SEQUENCING OF MITOCHONDRIAL DNA IN PATIENTS WITH SUSPECTED MITOCHONDRIAL DISEASE: IS THERE A NEED FOR A REVISED MOLECULAR DIAGNOSTIC ALGORITHM?**  
Ezgu F., 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 (Journal Indexed in SCI)
39. **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 (Journal Indexed in SCI)

40. **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 (Journal Indexed in SCI)
41. **Hypercalcemia in glycogen storage disease type I patients of Turkish origin**  
Kasapkar 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 (Journal Indexed in SCI)
42. **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 (Journal Indexed in SCI)
43. **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 (Journal Indexed in SCI)
44. **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., Tyłki-Szymanska A., Tuysuz B., Gabrielli O., Grossi S., Scarpa M., et al.  
HUMAN MUTATION, vol.32, no.6, 2011 (Journal Indexed in SCI)
45. **N-carbamylglutamate treatment for acute neonatal hyperammonemia in isovaleric acidemia**  
Kasapkar 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 (Journal Indexed in SCI)
46. **Harderoporphyria due to homozygosity for coproporphyrinogen oxidase missense mutation H327R**  
Hasanoglu A., Balwani M., Kasapkar 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 (Journal Indexed in SCI)
47. **AN INFANTILE CASE OF ZELLWEGER SYNDROME PRESENTED WITH KABUKI-LIKE PHENOTYPE**  
Ezgu F., Eminoglu T., OKUR İ., Gunduz M., Tumer L., Hasanoglu A., Dalgic B.  
GENETIC COUNSELING, vol.22, no.2, pp.217-220, 2011 (Journal Indexed in SCI)
48. **A NOVEL MUTATION OF THE CLAUDIN 16 GENE IN FAMILIAL HYPOMAGNESEMIA WITH HYPERCALCIURIA AND NEPHROCALCINOSIS MIMICKING RICKETS**  
Kasapkar C. S. , Tumer L., OKUR İ., Hasanoglu A.  
GENETIC COUNSELING, vol.22, no.2, pp.187-192, 2011 (Journal Indexed in SCI)
49. **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 (Journal Indexed in SCI)
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28. **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
29. **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
30. **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
31. **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
32. **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
33. **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
34. **Renal İnvolve ment in Fabry Disease**  
İNCİ A., BİBEROĞLU G., OKUR İ., PAŞAOĞLU Ö. T. , TÜMER L., PAŞAOĞLU H., EZGÜ F. S.  
13.International Congress of Inborn Errors of Metabolism., Rio de Janeiro, Brazil, 5 - 08 September 2017
35. **İ nvestigation of LDLR Gene Mutations in Turkish Patients With Familial Hypercholesterolemia**  
OKUR İ., İNCİ A., OLGAC 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
36. **Karbonhidrat Metabolizması Bozuklukları**  
OKUR İ.  
Çocuk Gastroenteroloji, Hepatoloji ve Beslenme Güncelleme Toplantısı, Turkey, 1 - 04 June 2017
37. **Tirozinemi İzlem ve Tedavi**

OKUR İ.

Çocuk Gastroenteroloji, Hepatoloji ve Beslenme Güncelleme Toplantısı, Turkey, 1 - 04 June 2017

38. **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

39. **Lizozomal Depo Hastalıklarında Nörolojik Bulgular ve Tedavileri**

OKUR İ.

14.Ulusal Metabolik Hastalıklar ve Beslenme Kongresi, Turkey, 26 - 30 April 2017

40. **Mukopolisakkaridozlar: Tanı ve Tedavide Güncel Yaklaşımları**

OKUR İ.

39. Pediatri Günleri ve 18. Pediatri Hemşireliği Günleri, Turkey, 2 - 05 April 2017

41. **Yenidoğanda Tarama Testleri**

OKUR İ.

İlk 1000 gün 5. Kongresi, Turkey, 19 - 22 March 2017

42. **Ailevi Hiperkolesterolemi Olan Türk Hastalarda LDLR Gen Mutasyonlarının Araştırılması**

OKUR İ., EZGÜ F. S. , İNCİ A., OLGAC M. A. B. , TÜMER L.

2. Ege Endokrin Hastalıklar ve Genetik Sempozyumu, Turkey, 23 - 25 February 2017

43. **In vitro translational readthrough by gentamicin and geneticin improves GLA activity in Fabry disease**

Dündar H., Biberoglu 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

44. **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

45. **ÇOCUKLARDA VİTAMİN VE MİNERAL DESTEĞİ**

OKUR İ.

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

46. **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

47. **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

48. **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

49. **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

50. **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

51. **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

52. **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
53. **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
54. **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
55. **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
56. **Bone mineral density and vitamin D status in inborn errors of metabolism**  
OLGAÇ 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
57. **Do cytokine levels play a role in pathogenesis of mucopolysaccharidosis patients**  
İNCİ A., TÜMER L., Demirtaş C., KARAOĞLU A., OKUR İ., OLGAÇ 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
58. **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
59. **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
60. **BonemineradensityandvitaminDstatusininbornerrorsofmetabolism**  
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
61. **Identification of a novel mutation in Turkish infant with early onset monocarboxylate transporter 1 MCT1 deficiencyasacauseofrecurrent ketoacidosi**  
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
62. **LAL enzim eksikliği**  
OKUR İ.  
11. Ulusal Çocuk Gastroenteroloji Hepatoloji ve Beslenme Kongresi, Turkey, 4 - 07 May 2016
63. **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
64. **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

65. **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
66. **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., OLGAC M. A. B. , KARAOĞLU A., BİBEROĞLU G., TÜMER L., hasanoğlu a.  
SSIEM Annual Symposium, 1 - 04 September 2015
67. **Dihydrolipoamide dehydrogenase deficiency diagnosed by using new generation sequencing technology**  
İNCİ A., TÜMER L., OKUR İ., OLGAC M. A. B. , SARI S., çiftçi b., topçu b., EZGÜ F. S.  
SSIEM Annual Symposium, 1 - 04 September 2015
68. **Cobalamin C disease with hypopigmented cutaneous findings A unique case**  
TÜMER L., ARHAN E., OKUR İ., Aydın K., Hirfanoğlu 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
69. **Sol çenede Lenfatik tutulum ile giden Niemann Pick tip C olgusu**  
İNCİ A., OKUR İ., ESENDAĞLI G., OKUR A., OLGAC M. A. B. , EZGÜ F. S. , TÜMER L.  
XIII.Ulusal Metabolik Hastalıklar ve Beslenme Kongresi, Adana, Turkey, 14 - 18 April 2015
70. **Lizozomal depo hastalıklarına yaklaşım**  
OKUR İ.  
2. Marmara Pediatri Kongresi, Turkey, 12 - 14 February 2015
71. **The results of enzyme studies in the diagnosis of lysosomal diseases: 8 years experience of Gazi University, Ankara, Turkey**  
Hasanoğlu 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
72. **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
73. **Importance of family screening in Fabry disease: Reaching the bottom of the iceberg**  
Ezgu F., Koca S., OKUR İ., BİBEROĞLU G., TÜMER L., Bakkaloglu S., ERTEN Y., Hasanoğlu A.  
11th Annual WORLD Symposium of the Lysosomal-Disease-Network, Florida, United States Of America, 9 - 13 February 2015, vol.114
74. **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
75. **PREVALENCE OF FABRY DISEASE AMONG HEMODIALYSIS PATIENTS IN TURKEY**  
Okur İ., BİBEROĞLU G., Ezgu F., TÜMER L., Hasanoğlu A., Bicik Z., Akin Y., Mumcuoglu M., Ecdet T.  
50th European-Renal-Association - European-Dialysis-and-Transplant-Association Congress, İstanbul, Turkey, 18 - 21 May 2013, vol.28, pp.321
76. **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
77. **TWO NOVEL MUTATIONS IN TWO PATIENTS WITH MEDIUM-CHAIN ACYL-CoA DEHYDROGENASE DEFICIENCY**  
Hasanoğlu 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

78. **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.

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

### Academic and Administrative Experience

2020 - Continues	<b>Vice Dean</b>	Gazi University, Tıp Fakültesi
2019 - Continues	<b>Ethics Committee Member</b>	Gazi University, -
2018 - Continues	<b>Fakülte Kurulu Üyesi</b>	Gazi University, Tıp Fakültesi, Dahili Tıp Bilimleri
2018 - Continues	<b>Fakülte Yönetim Kurulu Üyesi</b>	Gazi University, Tıp Fakültesi, Dahili Tıp Bilimleri
2015 - 2020	<b>Yıl Koordinatörü</b>	Gazi University, Tıp Fakültesi, Dahili Tıp Bilimleri

### Courses

Beslenmeye Giriş, Undergraduate, 2014 - 2015, 2015 - 2016, 2016 - 2017, 2017 - 2018, 2018 - 2019, 2019 - 2020, 2020 - 2021

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

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

Klinik Uygulamalı Pratik Eğitimi, Undergraduate, 2016 - 2017, 2017 - 2018, 2018 - 2019, 2019 - 2020

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

Vitamin ve Mineral Bozuklukları, Doctorate, 2017 - 2018, 2018 - 2019

Çocuk Metabolizması Yandal Uzmanlık Eğitimi, Expertise In Medicine, 2014 - 2015

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., 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

### Activities in Scientific Journals

Türkiye Çocuk Hastalıkları Dergisi, Editor, 2017 - Continues



## Memberships / Tasks in Scientific Organizations

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

## Scientific Consultations

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 - Continues  
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

## Scientific Research / Working Group Memberships

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

## Edit Congress and Symposium Activities

Pandemi Döneminde Çocuk Sağlığı Online Konferans, Invited Speaker, Baku, Azerbaijan, 2020  
16 Annual WORLDSymposium 2020 , Attendee, Florida, United States Of America, 2020

## Citations

Total Citations (WOS):375  
h-index (WOS):11

## Research Areas

Health Sciences

## Non Academic Experience

Ministry