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Education Information

Post Doctorate of Medicine, University Of California Los Angeles / Tıpta Uzmanlık Kurulu, Turkey 2005 - 2011
Post Doctorate of Medicine, Gazi University, Tıp Fakültesi, Dahili Tıp Bilimleri Bölümü, Turkey 1999 - 2003
Expertise In Medicine, Gazi University, Tıp Fakültesi, Dahili Tıp Bilimleri Bölümü, Turkey 1993 - 1998
Undergraduate, Ankara University, Ankara Tıp Fakültesi, Ankara Tıp Pr., Turkey 1987 - 1993

Dissertations

Post Doctorate of Medicine, Obez çocuklarda sICAM-1, sVCAM-1, sE-SELEKTİN ve CRP düzeyleri, Gazi University, Tıp Fakültesi, 2003
Expertise In Medicine, Hipoksik iskemik ensefalopatili term yenidoğanlarda beyin omirilik sıvısı ve serum nöron spesifik enolaz düzeyleri, Gazi University, Tıp Fakültesi, 1998

Research Areas

Pediatric Endocrinology and Metabolism, Pediatric Genetics and Teratology

Academic Titles / Tasks

Professor, Gazi University, Tıp Fakültesi, Dahili Tıp Bilimleri, 2011 - Continues
Professor, Gazi University, Tıp Fakültesi, Dahili Tıp Bilimleri, 2011 - Continues
Associate Professor, Gazi University, Tıp Fakültesi, Dahili Tıp Bilimleri, 2006 - 2011
Assistant Professor, Gazi University, Tıp Fakültesi, Dahili Tıp Bilimleri, 2005 - 2006
Expert, Gazi University, Tıp Fakültesi, Dahili Tıp Bilimleri, 2000 - 2005
Research Assistant, Gazi University, Tıp Fakültesi, Dahili Tıp Bilimleri, 1993 - 1998

Courses

HEREDIRATY STORAGE DISORDERS, Undergraduate, 2022 - 2023
ORGANOMEGALİ İLE SEYREDEN METABOLİK HASTALIKLAR, Undergraduate, 2022 - 2023
PEDİATRİDE GENETİK HASTALIKLARA YAKLAŞIM, Undergraduate, 2022 - 2023
GENETIC APPROACH IN PEDIATRICS, Undergraduate, 2023 - 2024
PEDİATRİDE GENETİK HASTALIKLARA YAKLAŞIM, Undergraduate, 2023 - 2024
ORGANOMEGALİ İLE SEYREDEN METABOLİK HASTALIKLAR, Undergraduate, 2023 - 2024
GENETIC APPROACH IN PEDIATRICS, Undergraduate, 2022 - 2023
INBORN ERRORS OF METABOLISM, Undergraduate, 2022 - 2023
INBORN ERRORS OF METABOLISM, Undergraduate, 2023 - 2024
HEREDIRATY STORAGE DISORDERS, Undergraduate, 2023 - 2024
VİTAMİNS İN HEALTH AND DİSEASE STATES, Undergraduate, 2017 - 2018, 2016 - 2017
PEDİATRİDE GENETİK YAKLAŞIM, Undergraduate, 2017 - 2018, 2016 - 2017
Healthy Nutrition, Undergraduate, 2017 - 2018
GENETİK APPROACH İN PEDİATRİCS, Undergraduate, 2017 - 2018
KALITSAL DEPO HASTALIKLARI, Undergraduate, 2017 - 2018, 2016 - 2017
HEREDİTARY STORAGE DİSORDERS, Undergraduate, 2017 - 2018, 2016 - 2017
INBORN ERRORS OF METABOLİSM, Undergraduate, 2017 - 2018, 2016 - 2017

Advising Theses

Ezgü F. S., Osteogenezis imperfekta tanısı ile takip edilen hastaların klinik, moleküler, laboratuvar ve radyolojik özelliklerinin retrospektif analizi, Expertise In Medicine, I.ÜSTÜNDAĞ(Student), 2023
Ezgü F. S., Nıemann-pıck tip C tanısı ile takip edilen hastaların epidemiyolojik, klinik, moleküler ve laboratuvar özellikleri ile lizosfingolipid düzeylerinin retrospektif analizi, Expertise In Medicine, A.YILMAZ(Student), 2023
Ezgü F. S., Utility of resequencing and reanalysis for unsolved rare diseases, Postgraduate, Ö.FARUK(Student), 2022
EZGÜ F. S., Moleküler ya da enzimatik analiz ile mukopolisakkaridoz tanısı almış olgularda böbrek ve üriner sistem tutulumunun araştırılması, Expertise In Medicine, F.ÖZAK(Student), 2016
EZGÜ F. S., Hiperfenilalaninemili Hastalarda Kan ve Beyin Fenilalanin Düzeylerinin Ölçümü İle Farklı Tedavi Yöntemlerinin Etkinliklerinin Değerlendirilmesi, Expertise In Medicine, A.KÜÇÜKÇONGAR(Student), 2012
EZGÜ F. S., Alfa-galaktosidaz A eksikliğinde genotip fenotip ilişkisi, Expertise In Medicine, S.KOCA(Student), 2010

Jury Memberships

Appointment to Academic Staff-Professorship, Appointment to Academic Staff-Professorship, Gazi Üniversitesi, December, 2023
Appointment to Academic Staff-Professorship, Appointment to Academic Staff-Professorship, İstanbul Üniversitesi-Cerrahpaşa, December, 2022

Published journal articles indexed by SCI, SSCI, and AHCI

I. **A very rare presentation of mitochondrial elongation factor Tu deficiency-TUFM mutation and literature review**
GÖKALP S., İNCİ A., KILIÇ A., Ozsaydi E., ALTUN A. N., DEMİR F., ERGİN F. B., Ozbek M. N., OKUR İ., EZGÜ F. S., et al.
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II. **Endocrinological and metabolic profile of Gaucher disease patients treated with enzyme replacement therapy**
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III. **Giant thymic cyst overclouding the diagnosis of fibrodysplasia ossificans progressiva: an inconvenient coincidence**
KÜÇÜKALİ B., Tanidir M. K., Senol P. E., YILDIZ YILDIRIM Ç., KARAÇAYIR N., BELDER N., GEZGİN YILDIRIM D., EZGÜ F. S., BAKKALOĞLU EZGÜ S. A.
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Öktem R. M., İnci A., BAYRAK H., DEMİR F., BİBEROĞLU G., Maviş M. E., Gürsu G. G., Yllmaz H., OKUR İ., EZGÜ F. S., et al.
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V. **Clinical characteristics of adult and paediatric patients with familial hypercholesterolemia: A real-life cross-sectional study from the Turkish National Database**
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VI. **Consensus clinical management guidelines for acid sphingomyelinase deficiency (Niemann-Pick disease types A, B and A/B)**
Geberhiwot T., Wasserstein M., Wanninayake S., Bolton S. C., Dardis A., Lehman A., Lidove O., Dawson C., Giugliani R., Imrie J., et al.
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VII. **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)

VIII. **Identification of a novel mutation in the ALDOB gene in hereditary fructose intolerance**
Beyzaei Z., EZGÜ F. S., Imanieh M. H., Haghighat M., Dehghani S. M., Honar N., Geramizadeh B.
JOURNAL OF PEDIATRIC ENDOCRINOLOGY & METABOLISM, vol.36, no.3, pp.331-334, 2023 (SCI-Expanded)

IX. **Expert-opinion-based guidance for the care of children with lysosomal storage diseases during the COVID-19 pandemic: An experience-based Turkey perspective**
Akgun A., Gokcay G., Mungan N. O., Sivri H. S., TEZER H., Zeybek C. A., EZGÜ F. S.
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X. **A phase 1/2 study on intracerebroventricular tralesinidase 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.
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XI. **Study design challenges and strategies in clinical trials for rare diseases: Lessons learned from pantothenate kinase-associated neurodegeneration**
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XII. **Endocrinological, immunological and metabolic features of patients with Fabry disease under therapy**
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XIII. **A possibly new autoinflammatory disease due to compound heterozygous phosphomevalonate kinase gene mutation**
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XIV. **Expert opinion on patient journey, diagnosis and clinical monitoring in acid sphingomyelinase deficiency in Turkey: a pediatric metabolic disease specialist's perspective**
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XV. **Co-Occurring Atypical Galactosemia and Wilson Disease**
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XVI. **Longitudinal Natural History of Pediatric Subjects Affected with Mucopolysaccharidosis IIIB**
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XVII. **Chemically modified recombinant human sulfamidase (SOBI003) in mucopolysaccharidosis IIIA patients: Results from an open, non-controlled, multicenter study**
Harmatz P., Muenzer J., EZGÜ F. S., Dalen P., Huledal G., Lindqvist D., Gelius S. S., Wiken M., Onnestam K., Broijersen A.
MOLECULAR GENETICS AND METABOLISM, vol.136, no.4, pp.249-259, 2022 (SCI-Expanded)

XVIII. **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)

XIX. **A Patient with a Novel RARS2 Variant Exhibiting Liver Involvement as a New Clinical Feature and Review of the Literature**
Sevinc S., İNCİ A., EZGÜ F. S., EMİNOĞLU F. T.
MOLECULAR SYNDROMOLOGY, vol.13, no.3, pp.226-234, 2022 (SCI-Expanded)

XX. **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.
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XXI. **Expected or unexpected clinical findings in liver glycogen storage disease type IX: distinct clinical and molecular variability**
İnci A., Kılıç Yıldırım G., Cengiz Ergin F. B., Sarı S., Eğritaş Gürkan Ö., Okur İ., Biberoğlu G., Bükülmez A., Ezgü F. S., Dalgıç B., et al.
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XXII. **Addressing the need for patient-friendly medical communications: adaptation of the 2019 recommendations for the management of MPS VI and MPS IVA**
Bruce I. A., EZGÜ F. S., Kampmann C., Kenis V., Mackenzie W., Stevens B., Walker R., Hendriksz C.
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XXIII. **Expert opinion on the recognition, diagnosis and management of children and adults with Fabry disease: a multidisciplinary Turkey perspective**
EZGÜ F. S., ALPSOY E., Bicik Bahcebasi Z., Kasapcopur O., PALAMAR ONAY M., Onay H., ÖZDEMİR B. H., TOPÇUOĞLU M. A., Tufekcioglu O.
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XXV. **A Patient with Recurrent Severe Hypoglycemic Attacks and Mitochondrial Complex III Deficiency, Nuclear Type 3: a Novel UQCRB Variant**
Yekeduz M. K., Oncul U., Kose E., Ezgu F. S., EMİNOĞLU F. T.
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XXVI. **First successful concomitant therapy of immune tolerance induction therapy and desensitization in a CRIM-negative infantile Pompe patient**
Sanli M. E., ERTOY KARAGÖL H. İ., KILIÇ A., Aktasoglu E., İNCİ A., OKUR İ., Ezgu F. S., TÜMER L.
JOURNAL OF PEDIATRIC ENDOCRINOLOGY & METABOLISM, vol.35, no.2, pp.273-277, 2022 (SCI-Expanded)

XXVII. **Safety of sebelipase alfa for the treatment of lysosomal acid lipase deficiency.**
Ezgu F. S.

Expert opinion on drug safety, vol.21, no.2, pp.149-155, 2022 (SCI-Expanded)

XXVIII. **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)

XXIX. **The Genetic Analysis of Cystic Fibrosis Patients With Seven Novel Mutations in the CFTR Gene in the Central Anatolian Region of Turkey.**
Erdogan M., KÖSE M., PEKCAN S., HANGÜL M., Balta B., Kiraz A., Gonen G. A., Zamani A. G., YILDIRIM M. S., RAMASLI GÜRSOY T., et al.
Balkan medical journal, vol.38, no.6, pp.357-364, 2021 (SCI-Expanded)

XXX. **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., Biberoğlu G., Öktem M., Ezgü F. S.
ORPHANET JOURNAL OF RARE DISEASES, vol.16, no.1, 2021 (SCI-Expanded)

XXXI. **Tralesinidase 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)

XXXII. **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.
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AMERICAN JOURNAL OF MEDICAL GENETICS PART A, vol.185, no.9, pp.2739-2747, 2021 (SCI-Expanded)

XXXIV. **Thinking outside "The Box": Case-based didactics for medical education and the instructional legacy of Dr John M. Graham, Jr.**
Sanchez-Lara P. A., Grand K., Haanpaa M. K., Curry C. J., Wang R., Ezgu F. S., Rose C. M., D'Cunha Burkardt D., Conway R. L., Relan A., et al.
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XXXV. **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)

XXXVI. **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., Biberoğlu G., Öktem R. M., Tümer L., et al.
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XXXVII. **A CASE OF GLYCOGEN STORAGE DISEASE TYPE 1a MIMICKING FAMILIAL CHYLOMICRONEMIA SYNDROME**
Olgac A., OKUR İ., BİBEROĞLU G., EZGÜ F. S., TÜMER L.
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XXXVIII. **Autism: Screening of inborn errors of metabolism and unexpected results**
İnci A., Özaslan A., Okur İ., Biberoğlu G., Güney E., Ezgü F. S., Tümer L., İşeri E.
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XXXIX. **Novel mutations in the PHKB gene in an iranian girl with severe liver involvement and glycogen storage disease type IX: a case report and review of literature.**
Beyzaei Z., Ezgu F. S., Geramizadeh B., Alborzi A., Shojazadeh A.
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XL. **Clinical and genetic spectrum of glycogen storage disease in Iranian population using targeted gene sequencing.**

Beyzaei Z., Ezgu F. S., Geramizadeh B., Imanieh M. H., Haghighat M., Dehghani S. M., Honar N., Zahmatkeshan M., Jassbi A., Mahboubifar M., et al.
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XLI. **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.
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XLIII. **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.
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XLVII. **Beneficial Effects of Modified Atkins Diet in Glycogen Storage Disease Type IIIa**
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L. **A rare case of primary coenzyme Q10 deficiency due to COQ9 mutation**
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LIV. **A Spectrum of Clinical Findings from ALPS to CVID: Several Novel LRBA Defects.**
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LV. **A 7-YEAR-OLD BOY WITH HAND TREMORS AND A NOVEL MUTATION FOR L-2-HYDROXYGLUTARIC ACIDURIA**
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XII. **Gaucher Tip I Hastalığında Kardiyak Tutulumun Erken Saptanması için Bir Alternatif: Speckle Tracking Ekokardiyografi**

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XIX. **Diagnostic workshop: 'Solve the mystery case'**

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XX. **Laboratory diagnosis of Mucopolysaccharidosis disorders**

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Okur İ., Ezgü F. S., Tümer L., Other International Funding Programs, HAUSER-RCT Heterezigot Ailevi Hiperkolesterolemisi (HeAH) Olan 10 ve 17 yaşlarındaki pediatrik Gönüllülerde Düşük Yoğunluklu Lipoprotein-Kolesterol (LDL-C)'ün azaltılmasında diyete 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

EZGÜ F. S., Project Supported by Higher Education Institutions, MOLEKÜLER YA DA ENZİMATİK ANALİZ İLE MUKOPOLİSAKKRİDOZ TANISI ALMIŞ OLGULARDA BÖBREK VE ÜRİNER SİSTEM TUTULUMUNUN ARAŞTIRILMASI, 2015 - 2016

EZGÜ F. S., Project Supported by Higher Education Institutions, BİRDEN FAZLA BİREYİNDE "KARDİYOFASİYOKUTANÖZ

SENDROM" FENOTİPİ GÖZLENEN BİR AİLEDE MOLEKÜLER BAĞLANTI ANALİZİ İLE SENDROMA YOL AÇAN GEN/GENLERİN BELİRLENMESİ, 2010 - 2015
EZGÜ F. S., Project Supported by Higher Education Institutions, Hiperfenilalaninemili Hastalarda Kan ve Beyin Fenilalanin Düzeylerinin Değerlendirimesi ve Tedavi Etkinliğinin Saptanması, 2011 - 2014
EZGÜ F. S., Project Supported by Higher Education Institutions, ALFA GALAKTOSİDAZ A EKSİKLİĞİNDE GENOTİP-FENOTİP İLİŞKİSİ, 2010 - 2011
EZGÜ F. S., Project Supported by Higher Education Institutions, Prebubertal Obez Çocuklarda Karotis İntima-Media Kalınlığı ve Kan Okside LDL Düzeyleri, 2004 - 2006

Activities in Scientific Journals

TURKISH JOURNAL OF PEDIATRICS, Committee Member, 2022 - Continues
Turkish Journal Of Medical Sciences, Committee Member, 2018 - Continues
Türkiye Klinikleri Pediatri Dergisi, Publication Committee Member, 2013 - Continues

Memberships / Tasks in Scientific Organizations

Çocuk Genetik Hastalıkları Derneği Yönetim Kurulu, Board Member, 2018 - 2023, Turkey

Scientific Refereeing

ORPHANET JOURNAL OF RARE DISEASES, Journal Indexed in SCI-E, November 2023
TURKISH JOURNAL OF PEDIATRICS, Journal Indexed in SCI-E, November 2023
ORPHANET JOURNAL OF RARE DISEASES, Journal Indexed in SCI-E, June 2023
Horizon Europe Project, Horizon Europe Project, Avrupa Birliği, Netherlands, March 2023
TURKISH JOURNAL OF PEDIATRICS, Journal Indexed in SCI-E, December 2022
ORPHANET JOURNAL OF RARE DISEASES, Journal Indexed in SCI-E, November 2022
TURKISH JOURNAL OF PEDIATRICS, Journal Indexed in SCI-E, September 2022
TURKISH JOURNAL OF PEDIATRICS, Journal Indexed in SCI-E, September 2022
ORPHANET JOURNAL OF RARE DISEASES, Journal Indexed in SCI-E, June 2022
TURKISH JOURNAL OF PEDIATRICS, Journal Indexed in SCI-E, April 2022
ORPHANET JOURNAL OF RARE DISEASES, Journal Indexed in SCI-E, April 2022

Scientific Consultations

Ankara Üniversitesi Nadir Hastalıklar Uygulama ve Araştırma Merkezi, Scientific Consultancy, Gazi University, Tıp Fakültesi, Dahili Tıp Bilimleri, Turkey, 2023 - Continues
ACURARE, Scientific Consultancy, Gazi University, Tıp Fakültesi, Dahili Tıp Bilimleri, Turkey, 2021 - Continues
TİTCK, Scientific Consultancy, Gazi University, Tıp Fakültesi, Dahili Tıp Bilimleri, Turkey, 2020 - Continues

Tasks In Event Organizations

Ezgü F. S., EMPOWER MPS Meeting, Scientific Congress, London, England, Kasım 2020

Metrics

Publication: 408
Citation (WoS): 1494
Citation (Scopus): 1597
H-Index (WoS): 17
H-Index (Scopus): 18

Invited Talks

Overview: State of the Field in AADCd , Conference, PTC-Experiental Learning Program, England, December 2021
Overview of Current & Future Therapies of MPSII, Conference, Hong Kong, December 2021
Patient Identification and Referral , Conference, PTC-Experiental Learning program, England, December 2021
Approach to MPSII: Diagnosis from organomegaly & short stature, Conference, takeda-MPS II Tutorial, Hong Kong, November 2021
Niemann-Pick Tip C: Tanı, İzlem ve Tedavide Neler Değişti?, Conference, VII. Lizozomal Hastalıklar Kongresi, Turkey, November 2021
AADC deficiency: a disorder with multiple signs and symptoms, Conference, PTC Therapeutics- Treatment Advances in AADC Deficiency: a rare neurotransmitter disorder, Kuwait, October 2021
What do you do with a floppy infant?„ Conference, 10th Europaediatrics, Croatia, October 2021
Lizozomal depo hastalıklarında yeni tedavi stratejileri, Conference, 5. ulusal Çocuk Genetik Kongresi, Turkey, October 2021
Nadir Genetik Hastalık Araştırmalarında Özellikler, Seminar, Klinik Araştırmalar Derneği-Akademika-İleri Düzey İyi Klinik Uygulamalar (İKU) Eğitimi, Turkey, September 2021
Makale Okurken, Seminar, Klinik araştırmalar Derneği - Akademika- İleri Düzey İyi Klinik Uygulamalar (İKU) Eğitimi, Turkey, September 2021
Current & Future Treatment Options in MPS II, Conference, MPS Academy Digital 2021 “Current & Future Treatment Options in MPS II, Ukraine, September 2021
Viewpoints: Management/Practices Pattern, Conference, Takeda-LSD 21, England, August 2021
Fabry Gastropathy: prevalence and response to ERT, Conference, England, August 2021
Diagnosis of skeletal dysplasias, Conference, Biomarin-Differential diagnosis of skeletal dysplasias, England, July 2021
Case examples of differential diagnoses, Conference, Biomarin-Differential diagnosis of skeletal dysplasias, England, July 2021
Nadir nörotransmitter hastalıklar; Hareket bozuklukları belirti ve bulguları, Conference, Türk Pediatri Kurumu-Nörotransmitter Hastalıklar Toplantıları , Turkey, June 2021
AADC eksikliğinin etyolojisi, genetik ve metabolik temelleri-AADC eksikliğinin prevalans ve temel klinik belirtileri, Conference, Türk Pediatri Kurumu-Nörotransmitter Hastalıklar Toplantıları , Turkey, June 2021
Challenges in management of patients during Covid-19 pandemic, Conference, Sanofi-Practical Solutions for Managing Patients with Gaucher Disease During COVID-19 Pandemic, England, June 2021
Challenges associated with MPS IVA: transition to adult care , Conference, England, June 2021
Guidance for the management of MPS IVA: key takeaways , Conference, ISMPS 2021-Improving patient care in MPS IVA through knowledge and empowerment , England, June 2021
Diagnostic and therapeutic update on Neurotransmitter disorders, Conference, The 1ST MENA Metabolic & Genetic Conference, Qatar, June 2021
Fabry Hastalığı, Conference, 43.Pediatri Günleri, Turkey, May 2021
Bebeklik Dönemi Karaciğer Hastalıklarında Genetik ve Metabolik Testler, Conference, 13. ulusal Çocuk Gastroenteroloji, Hepatoloji ve Beslenme Kongresi, Turkey, May 2021
Neurotransmitter-related disorders – a focus on non-ketotic hyperglycinemia, Conference, PTC-The road less travelled:Finding a path to diagnosis in rare neurotransmitter disorders, England, May 2021
Yeni Normal ile Tecrübeler Güncellemesi, Conference, Sanofi-Yeni Normal ile Tecrübeler, Turkey, May 2021
Fabry Hastalığında Spesifik Tedaviler Ve Güncel Yaklaşımlar, Conference, PARTNERSHIP IN FABRY, Turkey, April 2021
• Fabry disease in general (briefly with Fabry awareness month perspective) • Challenges in the diagnosis of Fabry

disease, Conference, Fabry Tedavisinde Fark Yaratan Bilgiler |20.Yılında Fabrazyme Global Deneyimi , Turkey, April 2021
Kadın Hastaya Tanısal Yaklaşım, Conference, PARTNERSHIP IN FABRY, Turkey, April 2021
Fabry Hastalığında Genetik, Kardiyak Varyantlar ve Temel Pedigri, Conference, PARTNERSHIP IN FABRY, Turkey, March 2021
Overview of Gaucher disease and its current management, Conference, Takeda-ICMEA Engage VPRIV Speaker Program, England, March 2021
Genetik ve Klinik Bağlantısı, Conference, Türk Hematoloji Derneği-Gaucher Hastalığı: Sık Belirtilerin Nadir Sebepleri, Turkey, March 2021
Management Options for MPS II: A Practical "Step by Step" Approach, Conference, Takeda-MPS II Tutorial, Hong Kong, March 2021
Gen Tedavisi İle İlgili Son Gelişmeler, Conference, Prof. dr. Hıfzı Özcan 8. Uluslararası Katılımlı Cerebral Palsy ve Gelişimsel Bozukluklar Kongresi, Turkey, February 2021
Long-term management of Gaucher – what we've learned so far, Conference, 6th Gaucher Disease Regional Forum, England, December 2020
Pitfalls: MPS II can be missed as it is less common than other MPS diseases, Conference, The Yin and Yang of Diagnosing MPS II, England, December 2020
Diagnostic challenges: -Laboratory testing, - Classical and non-classical cases, Conference, EMPOWER MPS Meeting, England, November 2020
Gaucher disease biomarkers – available options and utility in clinical practice, Conference, 6th Gaucher Disease Regional Forum, England, November 2020
Given Its rarity-Are Gaucher Disease Patients under-Recognized?, Conference, 56th Annual Meeting of the Thai Society of Hematology, Thailand, September 2020
Mukopolisakkaridoz ve Oligosakkaridozların Tedavisinde Yeni Ufuklar, Conference, ÇUKUROVA, GÜNEYDOĞU ANADOLU ve DOĞU ANADOLU BÖLGELERİ ÇOCUK METABOLİZMA ONLİNE OLGU SUNUMLARI – II, , Turkey, September 2020
Çocuklarda Büyüme Geriligi ve Organomegalinin Görünmeyen Nedenleri, Conference, İstanbul Üniversitesi, Turkey, September 2020
Challenges in Diagnosis of Gaucher Disease, Conference, Thailand, September 2020
Overview of treatment options for management of MPS II patients, Conference, MPS Academy: "Mucopolysaccharidosis type II: past, present and future", Kazakhstan, August 2020
Practical aspects of ERT applications, Conference, MPS Academy: "Mucopolysaccharidosis type II: past, present and future", , Kazakhstan, August 2020

Non Academic Experience

Ministry, Tıpta Uzmanlık Kurulu, Çocuk Metabolizma Hastalıkları Tıpta Uzmanlık Kurulu Müfredat Oluşturma Komisyonu
GAZİ ÜNİVERSİTESİ TIP FAKÜLTESİ
Ministry, Nadir Hastalıklar Bilimsel Danışma Komisyonu, Nadir Hastalıklar Bilimsel Danışma Komisyonu
UNIVERSITY OF CALIFORNIA LOS ANGELES
GAZİ ÜNİVERSİTESİ TIP FAKÜLTESİ
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