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Personal Information

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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
PEDIATRİDE GENETİK HASTALIKLARA YAKLAŞIM, Undergraduate, 2022 - 2023
GENETIC APPROACH IN PEDIATRICS, Undergraduate, 2023 - 2024
PEDIATRİDE 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
PEDIATRİDE GENETİK YAKLAŞIM, Undergraduate, 2017 - 2018, 2016 - 2017
Healthy Nutrition, Undergraduate, 2017 - 2018
GENETİK APPROACH İN PEDIATRİCS, 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., Osteogenesis imperfecta tanısı ile takip edilen hastaların klinik, moleküler, laboratuvar ve radyolojik özelliklerinin retrospektif analizi, Expertise In Medicine, I.ÜSTÜNDAĞ(Student), 2023
Ezgü F. S., Niemann-pick 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. **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**
Özsaydı Aktaşoğlu E., Kılıç A., Emecen Sanlı M., İnci A., Aktaş E., AKDULUM İ., Yaylı N., OKUR İ., EZGÜ F. S., TÜMER L.
Journal of Pediatric Endocrinology and Metabolism, vol.37, no.9, pp.820-824, 2024 (SCI-Expanded)
- II. **A very rare presentation of mitochondrial elongation factor Tu deficiency-TUFM mutation and literature review**
GÖKALP S., İNCİ A., KILIÇ A., Ozsaydi E., ALTUN A. N., DEMİR F., ERGİN F. B., Ozbek M. N., OKUR İ., EZGÜ F. S., et al.
Journal of Pediatric Endocrinology and Metabolism, vol.37, no.6, pp.571-574, 2024 (SCI-Expanded)

- III. **Endocrinological and metabolic profile of Gaucher disease patients treated with enzyme replacement therapy**
KILIÇ A., Emecen Sanli M., Ozsaydi Aktasoglu E., GÖKALP S., BİBEROĞLU G., İnci A., OKUR İ., EZGÜ F. S., TÜMER L.
Journal of Pediatric Endocrinology and Metabolism, vol.37, no.5, pp.413-418, 2024 (SCI-Expanded)
- IV. **Giant thymic cyst overclouding the diagnosis of fibrodysplasia ossificans progressiva: an inconvenient coincidence**
KÜÇÜKALİ B., Tanidir M., Senol P. E., YILDIZ YILDIRIM Ç., KARAYAYIR N., BELDER N., GEZGİN YILDIRIM D., EZGÜ F. S., BAKKALOĞLU EZGÜ S. A.
RHEUMATOLOGY, vol.63, 2024 (SCI-Expanded)
- V. **Efficacy and safety of oral sepiapterin in participants with phenylketonuria on sapropterin dihydrochloride at time of Phase 3 aphenity study entry**
Zori R., Lah M., Offord S., Lund A., Opladen T., Peters H., Guilder L., EZGÜ F. S., Breilyn M. S., Manti F., et al.
MOLECULAR GENETICS AND METABOLISM, no.4, 2024 (SCI-Expanded)
- VI. **Mucopolysaccharidosis type VII (Sly syndrome) - What do we know?**
Grant C. L., Lopez-Valdez J., Marsden D., EZGÜ F. S.
MOLECULAR GENETICS AND METABOLISM, vol.141, no.3, 2024 (SCI-Expanded)
- VII. **Pterin Profiling in Serum, Dried Blood Spot, and Urine Samples Using LC-MS/MS in Patients with Inherited Hyperphenylalaninemia**
Öktem R. M., İnci A., BAYRAK H., DEMİR F., BİBEROĞLU G., Maviş M. E., Gürsu G. G., Yılmaz H., OKUR İ., EZGÜ F. S., et al.
Molecular Syndromology, vol.15, no.3, pp.185-193, 2024 (SCI-Expanded)
- VIII. **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, no.7, pp.650-658, 2023 (SCI-Expanded)
- IX. **Clinical characteristics of adult and paediatric patients with familial hypercholesterolemia: A real-life cross-sectional study from the Turkish National Database**
Sonmez A., Demirci I., Haymana C., Tasci I., Ayvalı M. O., Ata N., EZGÜ F. S., BAYRAM F., Barcin C., Caglayan M., et al.
Atherosclerosis, vol.375, pp.9-20, 2023 (SCI-Expanded)
- X. **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.
ORPHANET JOURNAL OF RARE DISEASES, vol.18, no.1, 2023 (SCI-Expanded)
- XI. **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)
- XII. **Identification of a novel mutation in the ALDOB gene in hereditary fructose intolerance**
Beyzaei Z., EZGÜ F. S., Imanieh M. H., Haghghat M., Dehghani S. M., Honar N., Geramizadeh B.
JOURNAL OF PEDIATRIC ENDOCRINOLOGY & METABOLISM, vol.36, no.3, pp.331-334, 2023 (SCI-Expanded)
- XIII. **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.
Frontiers in Public Health, vol.11, 2023 (SCI-Expanded)
- XIV. **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., Batziros S., et al.
The Journal of clinical investigation, vol.133, 2023 (SCI-Expanded)
- XV. **A possibly new autoinflammatory disease due to compound heterozygous phosphomevalonate kinase gene mutation**

Yıldız Ç., Gezgin Yıldırım D., İnci A., Tümer L., Ergin F. B., Sunar Yayla E. N. S., Esmeray Şenol P., Karaçayır N., Eğritaş Gürkan Ö., Okur İ., et al.

Joint Bone Spine, vol.90, no.1, 2023 (SCI-Expanded)

- XVI. **Expert opinion on patient journey, diagnosis and clinical monitoring in acid sphingomyelinase deficiency in Turkey: a pediatric metabolic disease specialist's perspective**
ARSLAN N., Coker M., GÖKÇAY G. F., KIYKIM E., Mungan H. N. O., EZGÜ F. S.
FRONTIERS IN PEDIATRICS, vol.11, 2023 (SCI-Expanded)
- XVII. **Study design challenges and strategies in clinical trials for rare diseases: Lessons learned from pantothenate kinase-associated neurodegeneration**
Videnovic A., Pfeiffer H. C. V., Tyłki-Szymańska A., Berry-Kravis E., EZGÜ F. S., Ganju J., Jurecka A., Lang A. E.
Frontiers in Neurology, vol.14, 2023 (SCI-Expanded)
- XVIII. **Co-Occurring Atypical Galactosemia and Wilson Disease**
Doğulu N., Köse E., Tuna Kırsaçlıoğlu C., Ezgü F. S., Kuloğlu Z., Kansu Tanca A., Eminoğlu F. T.
Molecular Syndromology, vol.13, no.5, pp.454-458, 2022 (SCI-Expanded)
- XIX. **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)
- XX. **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)
- XXI. **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)
- XXII. **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)
- XXIII. **Expected or unexpected clinical findings in liver glycogen storage disease type IX: distinct clinical and molecular variability**
İnci A., Kiliç Yıldırım G., Cengiz Ergin F. B., Sarı S., Eğritaş Gürkan Ö., Okur İ., Biberöğlu 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)
- XXIV. **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)
- XXV. **Identification of a novel mutation in the PHKA2 gene in a child with liver cirrhosis**
Beyzaei Z., Ezgu F. S., Imanieh M. H., Geramizadeh B.
JOURNAL OF PEDIATRIC ENDOCRINOLOGY & METABOLISM, vol.35, no.3, pp.417-420, 2022 (SCI-Expanded)
- XXVI. **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.
ORPHANET JOURNAL OF RARE DISEASES, vol.17, no.1, 2022 (SCI-Expanded)
- XXVII. **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.

- ORPHANET JOURNAL OF RARE DISEASES, vol.17, no.1, 2022 (SCI-Expanded)
- XXVIII. **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)
- XXIX. **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)
- XXX. **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.
MOLECULAR SYNDROMOLOGY, vol.13, no.1, pp.64-68, 2022 (SCI-Expanded)
- XXXI. **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)
- XXXII. **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)
- XXXIII. **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)
- XXXIV. **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)
- XXXV. **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)
- XXXVI. **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.
American journal of medical genetics. Part A, vol.185, no.9, pp.2636-2645, 2021 (SCI-Expanded)
- XXXVII. **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)
- XXXVIII. **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)
- XXXIX. **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.
JOURNAL OF BONE AND MINERAL METABOLISM, vol.39, no.4, pp.598-605, 2021 (SCI-Expanded)
- XL. **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)

- XLII. **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)
- XLIII. **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.
BMC pediatrics, vol.21, no.1, pp.175, 2021 (SCI-Expanded)
- XLIV. **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.
Scientific reports, vol.11, no.1, pp.7040, 2021 (SCI-Expanded)
- XLV. **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)
- XLVI. **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)
- XLVII. **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)
- XLVIII. **Novel PRKAG2 variant presenting as liver cirrhosis: report of a family with 2 cases and review of literature.**
Beyzaei Z., Ezgu F. S., Geramizadeh B., Alborzi A., Shojazadeh A.
BMC medical genomics, vol.14, no.1, pp.33, 2021 (SCI-Expanded)
- XLIX. **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)
- L. **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)
- L. **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)
- LI. **Early indicators of disease progression in Fabry disease that may indicate the need for disease-specific treatment initiation: findings from the opinion-based PREDICT-FD modified Delphi consensus initiative.**
Hughes D. A., Aguiar P., Deegan P. B., Ezgu F. S., Frustaci A., Lidove O., Linhart A., Lubanda J., Moon J. C., Nicholls K., et al.
BMJ open, vol.10, no.10, 2020 (SCI-Expanded)
- LII. **Expanding the clinical spectrum of mitochondrial 3-hydroxy-3-methylglutaryl-CoA synthase deficiency with Turkish cases harboring novel HMGCS2 gene mutations and literature review**
Kilic M., Dorum S., Topak A., Yazici M., EZGÜ F. S., COŞKUN T.
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, vol.182, no.7, pp.1608-1614, 2020 (SCI-Expanded)

- LIII. **A rare case of primary coenzyme Q10 deficiency due to COQ9 mutation**
Olgac A., Oztoprak U., Kasapkara C. S., Kilic M., Yuksel D., Derinkuyu E. B., Yildiz Y. T., Ceylaner S., EZGÜ F. S.
Journal of Pediatric Endocrinology and Metabolism, vol.33, no.1, pp.165-170, 2020 (SCI-Expanded)
- LIV. **Posterior fossa horns; a new calvarial finding of mucopolysaccharidoses with well-known cranial MRI features**
Damar Ç., Derinkuyu B. E., Olgaç Kiliçkaya M. A. B., Öztürk M., Öztunalı Ç., Alimli A. G., Boyunaga Ö. L., Uçar M., Ezgü F. S., Tümer L., et al.
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- XV. **Gaucher Tip I Hastalığında Kardiyak Tutulumun Erken Saptanması için Bir Alternatif: Speckle Tracking Ekokardiyografi**
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<http://lizozomal2021.org/>, Turkey, 25 - 27 November 2021
- XVI. **MPS 6 Hastalarında Klinik Bulgular, ERT önce ve Sonrası Olay Bazlı Değerlendirme**
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- XVII. İNFANTİL TİP POMPE HASTALIĞI ULUSAL KONSENSUS ÇALIŞMASI**
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- XVIII. Gastrointestinal Involvement at the Junction of Wolman Disease and COVID 19**
ALTUN A., İNCİ A., KILIÇ A., Özsaydı Aktaşoğlu E., GÖKALP S., DEMİR F., ÇAVUŞOĞLU Y. H., Yiğit S., BOZDAYI G., OKUR İ., et al.
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- XIX. A PATIENT WITH ADENOSINE KINASE DEFICIENCY DUE TO A NOVEL MUTATION PRESENTING WITH NOVEL DYSMORPHIC AND CARDIAC FINDINGS**
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- XX. Pompe Hastalarında Enzim Replasman Tedavisine Bağlı Anafilaksi ve Yönetimi: Tek Merkez Deneyim**
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XXVIII. Ulusal Alerji ve Klinik İmmünoloji kongresi, Turkey, 13 - 17 October 2021
- XXI. Triamterene-induced suppression of R227X premature termination codon in Fabry disease**
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- XXII. Anatomy of MPS Patients**
EZGÜ F. S.
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- XXIII. Laboratory diagnosis of Mucopolysaccharidosis disorders**
EZGÜ F. S.
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- XXIV. Diagnostic workshop: 'Solve the mystery case'**
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- XXV. Analysis of hereditary nephrotic diseases through next generation DNA sequencing**
YAZICIOĞLU B., BAKKALOĞLU EZGÜ S. A., BÜYÜKKARAGÖZ B., EZGÜ F. S.
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- XXVI. Case Study: Switch from agalsidase alfa to beta**
EZGÜ F. S.
Fabry Academy South Asia, 18-19th October, 2019, Taipei, Taiwan, 18 - 19 October 2019
- XXVII. Fabry mi? Değil mi?**
EZGÜ F. S.
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- XXVIII. Fabry Hastalığı'nda Kalıtım**
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- XXIX. Fabry Hastalığı'nda Tedavi Seçenekleri, Endikasyonlar ve Sorunlar**
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- XXX. Fabry Hastalığı'nda Doğal Seyir**
EZGÜ F. S.
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- XXXI. Akut Porfiriya Benzeri Periferik Nöropati Gelişen Tirozinemi Tip 1 Olgusu**
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- XXXII. Genetik Hastalıklarda Yeni Tedaviler**

- EZGÜ F. S.
4.Ulusal Çocuk Genetik Kongresi, 25-27 Eylül 2019, Ankara, Turkey, 25 - 27 September 2019
- XXXIII. **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.
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- XXXIV. **Screening of twelve lysosomal storage diseases with LC-MS/MS in Gazi university hospital in Turkey: The first results of validation**
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- XXXV. **Next generation DNA sequencing as an initial diagnostic method for congenital defects of glycosylation**
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- XXXVI. **A Rare Case of Primary Coenzyme Q10 deficiency due to COQ9 gene mutation**
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- XXXVII. **Beneficial Effects of Modified Atkins Diet in Glycogen Storage Disease Type IIIa**
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- XXXVIII. **Mild hyperammonemia due to Antiquitin deficiency**
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- XXXIX. **Natural History in Fabry Disease: Classic vs Late-onset phenotype-Paediatrics**
EZGÜ F. S.
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- XL. **Metabolic and rare diseases: new effective treatments**
EZGÜ F. S.
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- XLI. **Population Genetics and Inborn errors of Metabolism**
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- XLII. **Hafif Renal Varyant ile Karakterize Pierson Sendromu: Olgu Sunumu**
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- XLIII. **Fabry Hastalığı**
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- XLIV. **Screening of Twelve Lysosomal Storage Diseases with LC-MS/MS in Gazi University Hospital: The First Results of Validation.**
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- XLV. **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.
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- XLVI. **Could Targeted Next Generation Sequencing Be A First Line Diagnostic Method for Lysosomal Storage Disease?**
CENGİZ F. B., İNCİ A., BİBEROĞLU G., Çiftçi B., Topçu B., Tokgöz D., Yazar Ö. F., Gökmenoğlu H., Raj Y., OKUR İ., et al.
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- XLVII. **Adenylosuccinate Lyase Deficiency in A Turkish Siblings**

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XLVIII. 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.

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XLIX. 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.

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L. Pyruvate Carboxylase Ceficiency in A Child with an Early Diagnosis of KetolysisDefect

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LI. Marinesco-Sjögren Syndrome: Case Report

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LII. A Very Rare Disease: Hyperornithinemia-Hyperammonemia-Homocitrullinuria (Hhh) Syndrome

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LIII. Familial Hyperphosphatemic Tumoral Calcinosis in an Unusual Site

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LIV. 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.

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LV. 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.

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LVI. 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.

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LVII. Hyperammonemia Secondary to Mitochondrial HMG-Coa Synthase Deficiency

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LVIII. Physiopathology and Diagnostic Methods in CDG

EZGÜ F. S.

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LIX. Novel Mutation in FBP1 Gene Presenting with Recurrent Episodes of Vomiting in A Child

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- LX. **A Novel Rars2 Mutation in Two Siblings with Microcephaly, Seizures and Liver Involvement**
EMİNOĞLU F. T., Sevinç S., Karaköse Gök T., EZGÜ F. S., İNCİ A., TÜMER L.
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- LXI. **Insights into Lysosomal Storage Diseases**
EZGÜ F. S.
Gaucher Disease Workshop, 22-23rd March 2019, Riyadh, Saudi Arabia, 22 - 23 March 2019
- LXII. **Case Presentations and Discussion**
EZGÜ F. S.
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- LXIII. **Screening Approaches and Laboratory Diagnosis in Gaucher Disease**
EZGÜ F. S.
Gaucher Disease Workshop, 22-23rd March 2019, Riyadh, Saudi Arabia, 22 - 23 March 2019
- LXIV. **The Practical Aspects of Diagnosis Protocols Implementation**
EZGÜ F. S.
Rare Diseases Annual Forum, 15-16th March, 2019, İstanbul-Turkey, 15 March 2019
- LXV. **Future of Rare Diseases Research - Advances in Lysosomal Storage Disorders Management**
EZGÜ F. S.
Rare Diseases Annual Forum, 15-16th March, 2019, İstanbul-Turkey, 15 - 16 March 2019
- LXVI. **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 WORLDSymposium(TM), Florida, United States Of America, 4 - 07 February 2019, vol.126
- LXVII. **Once every 4 weeks-2 mg/kg of pegunigalsidase alfa for treating Fabry disease Preliminary results of a phase 3 study**
Holida M. D., Bernat J., Longo N., Goker-Alpan O., Wallace E., Schiffmann R., Deegan P., Khan N., Tondel C., Eyskens F., et al.
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- LXVIII. **Safety and tolerability of SOBI003 in pediatric MPS IIIA patients key study design features of the ongoing first-in-human study**
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- LXIX. **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.
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- LXX. **Fabry hastalığının tedavisi ve seçenekler**
EZGÜ F. S.
Korkut Yalıtıkaya XIII. Klinik Nörofizyoloji Sempozyumu, Antalya, Turkey, 21 - 23 December 2018
- LXXI. **Pompe hastalığının tedavisinde güncel gelişmeler**
EZGÜ F. S.
Korkut Yalıtıkaya XIII. Klinik Nörofizyoloji Sempozyumu, Antalya, Turkey, 21 - 23 December 2018
- LXXII. **Gaucher Disease, is it still a diagnostic challenge after 30 years of clinical experience. Best Practices**
EZGÜ F. S.
Lysosomal Storage Disease Expert Meeting, Beirut, 2018, 15 December 2018

- LXXIII. Gaucher Disease: from Diagnosis to Treatment**
EZGÜ F. S.
1st Levant Regional Lysosomal Storage Disease Expert Meeting, Beirut, 14 December 2018
- LXXIV. LSDs Landscape**
EZGÜ F. S.
1st Levant Regional Lysosomal Storage Disease Expert Meeting, Beirut, 14 December 2018
- LXXV. Best Practices from Turkey Focus on Awareness, Access Patient Journey**
EZGÜ F. S.
"Second Step Towards Enhancing LSD Patient's Journey" The second Rare Diseases Out Loud Advisory Board Meeting Lysosomal Storage Disorders, Beirut, 2018, 14 December 2018
- LXXVI. Forum conclusion and Take Home Messages**
EZGÜ F. S.
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- LXXVII. RDs International vs local reimbursement guidelines**
EZGÜ F. S.
"Second Step Towards Enhancing LSD Patient's Journey" The second Rare Diseases Out Loud Advisory Board Meeting Lysosomal Storage Disorders, Beirut, 2018, 14 December 2018
- LXXVIII. LSDs landscape From Diagnosis to treatment with special focus on Gaucher Disease**
EZGÜ F. S.
Jordan Rare Disease Advisory Board Meeting "Enhancing LSD Patient's Journey Lysosomal Storage Disorders, Focusing on Gaucher, Beirut, 2018, 13 December 2018
- LXXIX. Turkish experience (Sharing Best Practices): "Moving from Diagnosis, Awareness to Access"**
EZGÜ F. S.
Jordan Rare Disease Advisory Board Meeting "Enhancing LSD Patient's Journey Lysosomal Storage Disorders, Focusing on Gaucher, Beirut, 2018, 13 December 2018
- LXXX. RAR2 mutation in two siblings with microcephaly, seizures and liver involvement**
EMİNOĞLU F. T., S. S., GÖK T., EZGÜ F. S., İNCİ A., TÜMER L.
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- LXXXI. 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.
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- LXXXII. Wrap Up**
EZGÜ F. S.
4th Gaucher Disease Regional Forum, İstanbul, 2 - 03 November 2018
- LXXXIII. Closing Day 1**
EZGÜ F. S.
4th Gaucher Disease Regional Forum, İstanbul, 2 - 03 November 2018
- LXXXIV. Interview with Gaucher Experts**
EZGÜ F. S.
4th Gaucher Disease Regional Forum, 2 - 03 November 2018
- LXXXV. Gaucher Disease Is it still a diagnostic challenge after 30 years of clinical experience?**
EZGÜ F. S.
4th Gaucher Disease Regional Forum, İstanbul, 2 - 03 November 2018
- LXXXVI. Advances in Gaucher Disease Management: Available Choices and Direction of Future Research**
EZGÜ F. S.
4th Gaucher Disease Regional Forum, İstanbul, 2 - 03 November 2018
- LXXXVII. 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., Küçükcongür A., Hasanoğlu A., EZGÜ F. S.
Gaucher Symposium, İstanbul, Turkey, 21 - 22 October 2018

- LXXXVIII. **Diagnosis and Testing**
EZGÜ F. S.
Gaucher Disease symposium 2018, İstanbul, 21 - 22 October 2018
- LXXXIX. **FMF'e genetik yaklaşım**
EZGÜ F. S.
VI. Çocuk genetik hastalıkları sempozyumu, Gaziantep, 2018, Turkey, 19 October 2018
- XC. **Complement factor B mutation in atypical hemolytic uremic syndrome: a case presentation**
ÇELEBİ TAYFUR A., ÇALTIK YILMAZ A., İNAN Y., KARAOKUR E., BÜYÜKKARAGÖZ B., KOÇAK M., EZGÜ F. S.
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- XCI. **Association of hereditary focal and segmental glomerulosclerosis with Bartter syndrome: a case report**
YAZICIOĞLU B., BÜYÜKKARAGÖZ B., ALPMAN B. N., IŞIK GÖNÜL İ., EZGÜ F. S., BUYAN N., BAKKALOĞLU EZGÜ S. A.
51st ESPN Congress, Antalya, 3 - 06 October 2018
- XCII. **Multidisipliner bir hastalık: Deneyimler ışığında Fabry**
EZGÜ F. S.
35. Ulusal nefroloji, hipertansiyon, diyaliz ve transplantasyon kongresi, 2018, Antalya, Turkey, 3 - 07 October 2018
- XCIII. **Natural History Data for Young Subjects with Sanfilippo Syndrome Type B (MPS IIIB)**
Okur I., Cleary M., Lopez d. C. M., Harmatz P., Lee J., Lin S. P., Couce M., Muschol N., 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
- XCIV. **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
- XCV. **Çocuklarda spontan pnömotoraks nedeni olarak marfan sendromu**
RAMASLI GÜRSOY T., ŞİŞMANLAR EYÜBOĞLU T., ASLAN A. T., EZGÜ F. S.
Çocuk Göğüs Hastalıkları 3. Kongresi, Turkey, 26 - 28 September 2018
- XCVI. **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
- XCVII. **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
- XCVIII. **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
- XCIX. **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.
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- C. **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
- CI. **Laboratory Diagnosis of MPS Disorders**
EZGÜ F. S.
MPS Masterclass 2018, Belgrade, 8 - 10 July 2018
- CII. **Next generation sequencing in the diagnosis of metabolic diseases**
EZGÜ F. S.
Organelle disease revisited, 2018, Antwerp, 18 May 2018
- CIII. **Erken Başlangıçlı Lizozomal Asit Lipaz Eksikliğinde Enzim Replasman Tedavisi Sonuçları**

EZGÜ F. S.

VI. Uluslararası katılımlı Lizozomal Hastalıklar Kongresi, Turkey, 11 - 15 April 2018

CIV. Natural history data for young subjects with Sanfilippo Syndrome Type B (MPS IIIB)

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CV. RENAL INVOLMENT IN FABRY DİSEASE

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Okur İ., Ezgü F. S., Tümer L., İnci A., Industrial Organizations of Other Countries Supported Project, K020-218--Standart Bakımda Yeterince Kontrol Altına Alnamayan Bir Üre Döngüsü Bozukluğu Olan Gönüllülerde KB195'in Etkinliğinin ve Güvenliğinin Değerlendirildiği Faz 2, Açık Etiketli Çalışma, 2021 - 2021

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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

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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

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

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, BİR DEN FAZLA BİREYİNDE "KARDİYOFAŞİYOKUTANÖZ 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 Değerlendirilmesi 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 Birliđi, 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

Okur İ., Ezgü F. S., 10. Ulusal Tıp Öğrenci Kongresi, Hücre ve Gen Tedavileri, Scientific Congress, Ankara, Turkey, Mayıs 2024
Ezgü F. S., EMPOWER MPS Meeting, Scientific Congress, London, England, Kasım 2020

Metrics

Publication: 422
Citation (WoS): 1591
Citation (Scopus): 1800
H-Index (WoS): 18
H-Index (Scopus): 20

Invited Talks

Overview: State of the Field in AADCd , Conference, PTC-Experiential Learning Program, England, December 2021
Overview of Current & Future Therapies of MPSII, Conference, Hong Kong, December 2021
Patient Identification and Referral , Conference, PTC-Experiential 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 Deđiřti?, 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

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