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Eğitim Bilgileri

Tıpta Yandal Uzmanlık, University Of California Los Angeles / Tıpta Uzmanlık Kurulu, Türkiye 2005 - 2011
Tıpta Yandal Uzmanlık, Gazi Üniversitesi, Tıp Fakültesi, Dahili Tıp Bilimleri Bölümü, Türkiye 1999 - 2003
Tıpta Uzmanlık, Gazi Üniversitesi, Tıp Fakültesi, Dahili Tıp Bilimleri Bölümü, Türkiye 1993 - 1998
Lisans, Ankara Üniversitesi, Ankara Tıp Fakültesi, Ankara Tıp Pr., Türkiye 1987 - 1993

Yaptığı Tezler

Tıpta Yandal Uzmanlık, Obez çocuklarda sICAM-1, sVCAM-1, sE-SELEKTİN ve CRP düzeyleri, Gazi Üniversitesi, Tıp Fakültesi, 2003
Tıpta Uzmanlık, Hipoksik iskemik ensefalopatili term yenidoğanlarda beyin omirilik sıvısı ve serum nöron spesifik enolaz düzeyleri, Gazi Üniversitesi, Tıp Fakültesi, 1998

Araştırma Alanları

Pediatrik Endokrinoloji ve Metabolizma, Pediatrik Genetik ve Teratoloji

Akademik Unvanlar / Görevler

Prof.Dr., Gazi Üniversitesi, Tıp Fakültesi, Dahili Tıp Bilimleri, 2011 - Devam Ediyor
Prof.Dr., Gazi Üniversitesi, Tıp Fakültesi, Dahili Tıp Bilimleri, 2011 - Devam Ediyor
Doç.Dr., Gazi Üniversitesi, Tıp Fakültesi, Dahili Tıp Bilimleri, 2006 - 2011
Yrd.Doç.Dr., Gazi Üniversitesi, Tıp Fakültesi, Dahili Tıp Bilimleri, 2005 - 2006
Uzman, Gazi Üniversitesi, Tıp Fakültesi, Dahili Tıp Bilimleri, 2000 - 2005
Araştırma Görevlisi, Gazi Üniversitesi, Tıp Fakültesi, Dahili Tıp Bilimleri, 1993 - 1998

Verdiği Dersler

HEREDIRATY STORAGE DISORDERS, Lisans, 2022 - 2023

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

Yönetilen Tezler

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

Jüri Üyelikleri

Akademik Kadroya Atama-Profesörlük, Akademik Kadroya Atama-Profesörlük, Gazi Üniversitesi, Aralık, 2023
Akademik Kadroya Atama-Profesörlük, Akademik Kadroya Atama-Profesörlük, İstanbul Üniversitesi-Cerrahpaşa, Aralık, 2022

SCI, SSCI ve AHCI İndekslerine Giren Dergilerde Yayınlanan Makaleler

I. **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., Yllmaz H., OKUR İ., EZGÜ F. S., et al.
Molecular Syndromology, 2024 (SCI-Expanded)

II. **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.
RHEUMATOLOGY, 2023 (SCI-Expanded)

III. **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, cilt.375, ss.9-20, 2023 (SCI-Expanded)

IV. **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, cilt.18, sa.1, 2023 (SCI-Expanded)

V. **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, cilt.184, sa.4, ss.370-375, 2023 (SCI-Expanded)

VI. **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, cilt.36, sa.3, ss.331-334, 2023 (SCI-Expanded)

VII. **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, cilt.11, 2023 (SCI-Expanded)

VIII. **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.
The Journal of clinical investigation, cilt.133, 2023 (SCI-Expanded)

IX. **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, cilt.90, sa.1, 2023 (SCI-Expanded)

X. **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, cilt.11, 2023 (SCI-Expanded)

XI. **Study design challenges and strategies in clinical trials for rare diseases: Lessons learned from pantothenate kinase-associated neurodegeneration**
Videnovic A., Pfeiffer H. C. V., Tylki-Szymańska A., Berry-Kravis E., EZGÜ F. S., Ganju J., Jurecka A., Lang A. E.
Frontiers in Neurology, cilt.14, 2023 (SCI-Expanded)

XII. **Endocrinological, immunological and metabolic features of patients with Fabry disease under therapy**
Emecen Sanli M., Kılıç A., İnci A., Okur İ., Ezgü F. S., Tümer L.
Journal of Pediatric Endocrinology and Metabolism, 2023 (SCI-Expanded)

XIII. **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, cilt.13, sa.5, ss.454-458, 2022 (SCI-Expanded)

XIV. **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, cilt.249, ss.50, 2022 (SCI-Expanded)

XV. **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, cilt.136, sa.4, ss.249-259, 2022 (SCI-Expanded)

XVI. **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, cilt.64, sa.4, ss.658-670, 2022 (SCI-Expanded)

XVII. **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, cilt.13, sa.3, ss.226-234, 2022 (SCI-Expanded)

XVIII. **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, cilt.35, sa.4, ss.497-503, 2022 (SCI-Expanded)

XIX. **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.
JOURNAL OF PEDIATRIC ENDOCRINOLOGY & METABOLISM, cilt.35, sa.4, ss.451-462, 2022 (SCI-Expanded)

XX. **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, cilt.17, sa.1, 2022 (SCI-Expanded)

XXI. **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, cilt.35, sa.3, ss.417-420, 2022 (SCI-Expanded)

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.
ORPHANET JOURNAL OF RARE DISEASES, cilt.17, sa.1, 2022 (SCI-Expanded)

XXIII. **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, cilt.13, sa.1, ss.64-68, 2022 (SCI-Expanded)

XXIV. **Safety of sebelipase alfa for the treatment of lysosomal acid lipase deficiency.**
Ezgu F. S.
Expert opinion on drug safety, cilt.21, sa.2, ss.149-155, 2022 (SCI-Expanded)

XXV. **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, cilt.35, sa.2, ss.273-277, 2022 (SCI-Expanded)

XXVI. **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, cilt.38, sa.6, ss.357-364, 2021 (SCI-Expanded)

XXVII. **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, cilt.45, sa.8, ss.1788-1792, 2021 (SCI-Expanded)

XXVIII. **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, cilt.16, sa.1, 2021 (SCI-Expanded)

XXIX. **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, cilt.185, sa.9, ss.2636-2645, 2021 (SCI-Expanded)

XXX. **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, cilt.90, 2021 (SCI-Expanded)

XXXI. **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, cilt.185, sa.9, ss.2739-2747, 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.
ANNALS OF NEUROLOGY, cilt.90, 2021 (SCI-Expanded)

XXXIII. **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, cilt.88, ss.723-0, 2021 (SCI-Expanded)

XXXIV. **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, cilt.39, sa.4, ss.598-605, 2021 (SCI-Expanded)

XXXV. **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, cilt.24, sa.1, ss.103-105, 2021 (SCI-Expanded)

XXXVI. **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.
AUTISM RESEARCH, cilt.14, sa.5, ss.887-896, 2021 (SCI-Expanded)

XXXVII. **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, cilt.21, sa.1, ss.175, 2021 (SCI-Expanded)

XXXVIII. **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, cilt.11, sa.1, ss.7040, 2021 (SCI-Expanded)

XXXIX. **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, cilt.132, sa.2, 2021 (SCI-Expanded)

XL. **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, cilt.132, sa.2, 2021 (SCI-Expanded)

XLI. **The chemical chaperone 4-phenylbutyrate enhances alpha-galactosidase activity subsequent to stop-codon read-through therapy with triamterene in Fabry R227X fibroblasts**
Dündar H., Biberoğlu G., İnci A., Işık Gönül İ., Okur İ., Tümer L., Ezgü F. S.
MOLECULAR GENETICS AND METABOLISM, cilt.132, sa.2, 2021 (SCI-Expanded)

XLII. **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, cilt.14, sa.1, ss.33, 2021 (SCI-Expanded)

XLIII. **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, cilt.34, sa.6, ss.805-812, 2021 (SCI-Expanded)

XLIV. **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, cilt.34, sa.6, ss.813-816, 2021 (SCI-Expanded)

XLV. **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, cilt.76, sa.4, ss.233-241, 2020 (SCI-Expanded)

XLVI. **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, cilt.10, sa.10, 2020 (SCI-Expanded)

XLVII. **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, cilt.182, sa.7, ss.1608-1614, 2020 (SCI-Expanded)

XLVIII. **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, cilt.33, sa.1, ss.165-170, 2020 (SCI-Expanded)

XLIX. **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., Öztunali Ç., Alimli A. G., Boyunaga Ö. L., Uçar M., Ezgü F. S., Tümer L., et al.
TURKISH JOURNAL OF MEDICAL SCIENCES, cilt.50, sa.4, ss.1048-1061, 2020 (SCI-Expanded)

L. **A new NBIA patient from Turkey with homozygous C19ORF12 mutation.**
Kasapkara C. S., TÜMER L., Gregory A., Ezgu F. S., İNCİ A., Derinkuyu B. E., Fox R., Rogers C., Hayflick S.
Acta neurologica Belgica, cilt.119, sa.4, ss.623-625, 2019 (SCI-Expanded)

LI. **Vitamin D Levels and Bone Mineral Density in Inborn Errors of Metabolism Requiring Specialised Diets**
Olgac A., İNCİ A., OKUR İ., Ezgu F. S., BİBEROĞLU G., Turner L.
JCPSP-JOURNAL OF THE COLLEGE OF PHYSICIANS AND SURGEONS PAKISTAN, cilt.29, sa.12, ss.1207-1211, 2019 (SCI-Expanded)

LII. **A Spectrum of Clinical Findings from ALPS to CVID: Several Novel LRBA Defects.**
ÇAĞDAŞ AYVAZ D. N., OSKAY HALAÇLI S., TAN Ç., Lo B., Cetinkaya P. G., Esenboga S., Karaatmaca B., Matthews H., Balci-Hayta B., Arikoglu T., et al.
Journal of clinical immunology, cilt.39, sa.7, ss.726-738, 2019 (SCI-Expanded)

LIII. **A 7-YEAR-OLD BOY WITH HAND TREMORS AND A NOVEL MUTATION FOR L-2-HYDROXYGLUTARIC**

ACIDURIA
Olgac A., Orgun T. L., Ezgu F. S., Biberoglu G., Tumer L.
BALKAN JOURNAL OF MEDICAL GENETICS, cilt.22, sa.2, ss.93-96, 2019 (SCI-Expanded)

LIV. **High incidence of co-existing factors significantly modifying the phenotype in patients with Fabry disease.**
Koca S., TÜMER L., OKUR İ., ERTEN Y., Bakkaloglu S. A., BİBEROĞLU G., Kasapkara C., Kucukcongar A., DALGIÇ B., ÖZHAN OKTAR S., et al.
Gene, cilt.687, ss.280-288, 2019 (SCI-Expanded)

LV. **Multicystic Dysplastic Kidney and Incontinentia Pigmenti: Coexistence of 2 Rare Diseases.**
Sürmeli Döven S., Delibas A., Türsen Ü., Ezgü F. S.
Iranian journal of kidney diseases, cilt.13, sa.1, ss.67-70, 2019 (SCI-Expanded)

LVI. **The role of IFIH1 gene rs1990760 and rs2111485 single-nucleotide polymorphisms in generalized vitiligo predisposition**
Onan D., Yorulmaz A., EZGÜ F. S., HAYRAN K. M., Kulcu S., Artuz R. F., Yalcin B.
TURKISH JOURNAL OF MEDICAL SCIENCES, cilt.49, sa.1, ss.206-211, 2019 (SCI-Expanded)

LVII. **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.
PEDIATRIC NEPHROLOGY, cilt.33, sa.10, ss.1972, 2018 (SCI-Expanded)

LVIII. **COMPLEMENT FACTOR B MUTATION IN ATYPICAL HEMOLYTICUREMIC SYNDROME; A CASE PRESENTATION**
Tayfur A. C., Yilmaz A. C., Inan Y., Karaokur E., Buyukkaragoz B., Kocak M., EZGÜ F. S.
PEDIATRIC NEPHROLOGY, cilt.33, sa.10, ss.1836-1837, 2018 (SCI-Expanded)

LIX. **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, cilt.40, sa.5, ss.355-359, 2018 (SCI-Expanded)

LX. **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, cilt.40, sa.3, ss.243-245, 2018 (SCI-Expanded)

LXI. **A Myopathy, Lactic Acidosis, Sideroblastic Anemia (MLASA) Case Due to a Novel PUS1 Mutation.**
Kasapkara C. S., TÜMER L., Zanetti N., Ezgu F. S., Lamantea E., Zeviani M.
Turkish journal of haematology : official journal of Turkish Society of Haematology, cilt.34, sa.4, ss.376-377, 2017 (SCI-Expanded)

LXII. **Survival in infants treated with sebelipase Alfa for lysosomal acid lipase deficiency: an open-label, multicenter, dose-escalation study.**
Jones S. A., Rojas-Caro S., Quinn A. G., Friedman M., Marulkar S., Ezgu F. S., Zaki O., Gargus J. J., Hughes J., Plantaz D., et al.
Orphanet journal of rare diseases, cilt.12, sa.1, ss.25, 2017 (SCI-Expanded)

LXIII. **Effect of Cytokine Signaling 3 Gene Polymorphisms in Childhood Obesity.**
Boyraz M., Yesilkaya E., Ezgu F. S., BİDECİ A., Dogan H., ULUCAN K., CİNAZ P.
Journal of clinical research in pediatric endocrinology, cilt.8, sa.4, ss.452-460, 2016 (SCI-Expanded)

LXIV. **Molecular analysis of the AGXT gene in patients suspected with hyperoxaluria type 1 and three novel mutations from Turkey**
Isiyel E., BAKKALOĞLU EZGÜ S. A., ÇALIŞKAN S., AKMAN S., AKİL İ., TABEL Y., Akinci N., BAHAT ÖZDOĞAN E., Ozel A., Eroglu F. K., et al.
MOLECULAR GENETICS AND METABOLISM, cilt.119, sa.4, ss.311-316, 2016 (SCI-Expanded)

LXV. **Early severe anemia as the first sign of cystic fibrosis**
Şişmanlar Eyüboğlu T., Aslan A. T., Köse M., Pekcan S., Ezgü F. S., Budakoğlu I. İ., Yenicesu I.
EUROPEAN JOURNAL OF PEDIATRICS, cilt.175, sa.9, ss.1157-1163, 2016 (SCI-Expanded)

LXVI. **Treatment of Fabry's Disease with the Pharmacologic Chaperone Migalastat.**

Germain D. P., Hughes D. A., Nicholls K., Bichet D. G., Giugliani R., Wilcox W. R., Feliciani C., Shankar S. P., Ezgu F. S., Amartino H., et al.
The New England journal of medicine, cilt.375, sa.6, ss.545-55, 2016 (SCI-Expanded)

LXVII. **Clinical course of sly syndrome (mucopolysaccharidosis type VII).**
Montano A. M., Lock-Hock N., Steiner R. D., Graham B. H., Szlago M., Greenstein R., Pineda M., Gonzalez-Meneses A., ÇOKER M., Bartholomew D., et al.
Journal of medical genetics, cilt.53, sa.6, ss.403-18, 2016 (SCI-Expanded)

LXVIII. **A Rare Cause of Lower Extremity Ulcers: Prolidase Deficiency**
ADIŞEN E., Erduran F. B., EZGÜ F. S., Kasapkara C. S., Besio R., Forlino A., Gurer M. A.
INTERNATIONAL JOURNAL OF LOWER EXTREMITY WOUNDS, cilt.15, sa.1, ss.86-91, 2016 (SCI-Expanded)

LXIX. **Inborn Errors of Metabolism**
Ezgu F. S.
ADVANCES IN CLINICAL CHEMISTRY, VOL 73, cilt.73, ss.195-250, 2016 (SCI-Expanded)

LXX. **A Phase 3 Trial of Sebelipase Alfa in Lysosomal Acid Lipase Deficiency**
Burton B. K., Balwani M., Feillet F., Baric I., Burrow T. A., Camarena Grande C., Coker M., Consuelo-Sanchez A., Deegan P., Di Rocco M., et al.
NEW ENGLAND JOURNAL OF MEDICINE, cilt.373, sa.11, ss.1010-1020, 2015 (SCI-Expanded)

LXXI. **Secondary Hemophagocytosis in Propionic Acidemia**
Kasapkara C. S., Kangin M., Ozmen B. O., Ozbek M. N., Demir R., Karatas M., TÜMER L., EZGÜ F. S., Hasanoglu A.
IRANIAN JOURNAL OF PEDIATRICS, cilt.25, sa.3, 2015 (SCI-Expanded)

LXXII. **A Novel Mutation in the Mitochondrial DNA Cytochrome b Gene (MTCYB) in a Patient with Prader Willi Syndrome**
YİŞ U., EZGÜ F. S., Karakaya P., Polat I., ARSLAN N., ÇANKAYA T., GİRAY BOZKAYA Ö., Kurul S. H.
JOURNAL OF CHILD NEUROLOGY, cilt.30, sa.3, ss.378-381, 2015 (SCI-Expanded)

LXXIII. **Situs inversus of the fibula: medialized fibula**
ATALAR H., TURANLI S., Atik O. S., Kaptan A. Y., Ezgu F. S.
EKLEM HASTALIKLARI VE CERRAHISI-JOINT DISEASES AND RELATED SURGERY, cilt.26, sa.1, ss.60-62, 2015 (SCI-Expanded)

LXXIV. **A CASE WITH RARE TYPE OF CONGENITAL DISORDER OF GLYCOSYLATION: PGM1-CDG**
Kucukcongar A., Tumer L., EZGÜ F. S., Kasapkara C. S., Jaeken J., Matthijs G., Rymen D., Dalgic B., Bideci A., Hasanoglu A.
GENETIC COUNSELING, cilt.26, sa.1, ss.87-90, 2015 (SCI-Expanded)

LXXV. **Results of a Global Phase 3, Randomized, Double-Blind, Placebo-Controlled Trial Evaluating the Efficacy and Safety of Sebelipase Alfa as an Enzyme Replacement Therapy in Children and Adults with Lysosomal Acid Lipase Deficiency**
Balwani M., Burton B., Baric I., Bialer M., Burrow T. A., Camarena Grande C., ÇOKER M., Consuelo Sanchez A., Deegan P., Di Rocco M., et al.
HEPATOLOGY, cilt.60, sa.6, 2014 (SCI-Expanded)

LXXVI. **Radiologic findings of Patterson-Lowry rhizomelic dysplasia in two sisters**
Damar C., Boyunaga Ö. L., Derinkuyu B. E., Battaloglu N., EZGÜ F. S.
SKELETAL RADIOLOGY, cilt.43, sa.11, ss.1651-1654, 2014 (SCI-Expanded)

LXXVII. **Mutation Analysis Of Primary Hyperoxaluria Type 1**
Isiyel E., BAKKALOĞLU EZGÜ S. A., EZGÜ F. S., ÇALIŞKAN S., AKMAN S., AKİL İ., TABEL Y., Akinci N., BAHAT ÖZDOĞAN E.
PEDIATRIC NEPHROLOGY, cilt.29, sa.9, ss.1828, 2014 (SCI-Expanded)

LXXVIII. **Phenotype-genotype correlations in patients with Marinesco-Sjogren syndrome**
Ezgu F. S., Krejci P., Li S., de Sousa C., Graham J. M., Hansmann I., He W., Porpora K., Wand D., Wertelecki W., et al.
CLINICAL GENETICS, cilt.86, sa.1, ss.74-84, 2014 (SCI-Expanded)

LXXIX. **Desensitisation of the youngest patient with Pompe disease in response to alglucosidase alfa**
Karagol I. H. E., Bakirtas A., Yilmaz O., Topal E., Kucukcongar A., Ezgu F. S., Demirsoy M. S., Turktas I.
ALLERGOLOGIA ET IMMUNOPATHOLOGIA, cilt.42, sa.4, ss.372-375, 2014 (SCI-Expanded)

LXXX. **Neonatal multiple sulfatase deficiency with a novel mutation and review of the literature**
NUR B., MIHÇI E., Pepe S., BİBEROĞLU G., EZGÜ F. S., Ballabio A., Oztekin O., DURSUN O.
TURKISH JOURNAL OF PEDIATRICS, cilt.56, sa.4, ss.418-422, 2014 (SCI-Expanded)

LXXXI. **BCS1L gene mutation causing GRACILE syndrome: case report**
Kasapkara C. S., TÜMER L., EZGÜ F. S., Kucukcongar A., Hasanoglu A.
RENAL FAILURE, cilt.36, sa.6, ss.953-954, 2014 (SCI-Expanded)

LXXXII. **Recent advances in the molecular diagnosis of inborn errors of metabolism**
Ezgu F. S.
CLINICAL BIOCHEMISTRY, cilt.47, sa.9, ss.759-760, 2014 (SCI-Expanded)

LXXXIII. **Crimean-congo haemorrhagic fever in pregnancy and in newborn: A case with a unique clinical course**
Unlusoy-Aksu A., Havali C., Tapisiz A., Aktas F., Ezgu F. S.
JOURNAL OF OBSTETRICS AND GYNAECOLOGY, cilt.34, sa.4, ss.360-361, 2014 (SCI-Expanded)

LXXXIV. **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, cilt.431, ss.250-254, 2014 (SCI-Expanded)

LXXXV. **Home sleep study characteristics in patients with mucopolysaccharidosis**
Kasapkara C. S., TÜMER L., ASLAN A. T., Hasanoglu A., EZGÜ F. S., Kucukcongar A., Tunca Z., KÖKTÜRK O.
SLEEP AND BREATHING, cilt.18, sa.1, ss.143-149, 2014 (SCI-Expanded)

LXXXVI. **Diagnosis of glycine encephalopathy in a pediatric patient by detection of a GLDC mutation during initial next generation DNA sequencing**
Ezgu F. S., Ciftci B., Topcu B., Adiyaman G., Gokmenoglu H., Kucukcongar A., Kasapkara C., Biberoglu G., TÜMER L., Hasanoglu A.
METABOLIC BRAIN DISEASE, cilt.29, sa.1, ss.211-213, 2014 (SCI-Expanded)

LXXXVII. **Rapid Molecular Diagnosis of Genetic Diseases by High Resolution Melting Analysis: Fabry and Glycogen Storage 1A Diseases**
Ezgu F. S., Divanoglu Y., Polat M., Bahceci S., Hasanoglu A., Desnick R. J.
GENETIC TESTING AND MOLECULAR BIOMARKERS, cilt.18, sa.1, ss.3-7, 2014 (SCI-Expanded)

LXXXVIII. **Could GSD type I expand the spectrum of disorders with elevated plasma chitotriosidase activity?**
TÜMER L., Kasapkara C. S., BİBEROĞLU G., Ezgu F. S., Hasanoglu A.
JOURNAL OF PEDIATRIC ENDOCRINOLOGY & METABOLISM, cilt.26, sa.11-12, ss.1149-1152, 2013 (SCI-Expanded)

LXXXIX. **Severe renal tubulopathy in a newborn due to BCS1L gene mutation: Effects of different treatment modalities on the clinical course**
Ezgu F. S., Senaca S., Gunduz M., Turner L., Hasanoglu A., Tiras U., Unsal R., BAKKALOĞLU EZGÜ S. A.
GENE, cilt.528, sa.2, ss.364-366, 2013 (SCI-Expanded)

XC. **Screening for Fabry disease in patients undergoing dialysis for chronic renal failure in Turkey: Identification of new case with novel mutation**
OKUR İ., Ezgu F. S., BİBEROĞLU G., Turner L., ERTEN Y., Isitman M., Eminoglu F. T., Hasanoglu A.
GENE, cilt.527, sa.1, ss.42-47, 2013 (SCI-Expanded)

XCI. **Sleep study characteristics in patients with mucopolysaccharidosis**
ASLAN A. T., Kasapkara C., TÜMER L., Hasanoglu A., Ezgu F. S.
EUROPEAN RESPIRATORY JOURNAL, cilt.42, 2013 (SCI-Expanded)

XCII. **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, cilt.26, ss.657-662, 2013 (SCI-Expanded)

XCIII. **Mild clinical presentation and prolonged survival of a patient with fumarase deficiency due to the combination of a known and a novel mutation in FH gene**
Ezgu F. S., Krejci P., Wilcox W. R.
GENE, cilt.524, sa.2, ss.403-406, 2013 (SCI-Expanded)

XCIV. **Apheresis-inducible cytokine pattern change in children with homozygous familial hypercholesterolemia**
Kucukcongar A., Yenicesu I., TÜMER L., Kasapkara C. S., EZGÜ F. S., Pasaoglu O., Demirtas C., ÇELİK B., Dilsiz G., Hasanoglu A.
TRANSFUSION AND APHERESIS SCIENCE, cilt.48, sa.3, ss.391-396, 2013 (SCI-Expanded)

XCV. **Cystic Fibrosis Presenting with Neonatal Cholestasis Simulating Biliary Atresia in a Patient with a Novel Mutation**
Eminoglu T. F., Polat E., Gokce S., EZGÜ F. S., Senel S., Apaydin S.
INDIAN JOURNAL OF PEDIATRICS, cilt.80, sa.6, ss.502-504, 2013 (SCI-Expanded)

XCVI. **Rapid screening of 12 common mutations in Turkish GSD 1a patients using electronic DNA microarray**
Eminoglu T. F., Ezgu F. S., Hasanoglu A., Tumer L.
GENE, cilt.518, sa.2, ss.346-350, 2013 (SCI-Expanded)

XCVII. **SRD5A3-CDG: A patient with a novel mutation**
Kasapkara C. S., TÜMER L., EZGÜ F. S., Hasanoglu A., Race V., Matthijs G., Jaeken J.
EUROPEAN JOURNAL OF PAEDIATRIC NEUROLOGY, cilt.16, sa.5, ss.554-556, 2012 (SCI-Expanded)

XCVIII. **A CASE OF PYRUVATE CARBOXYLASE DEFICIENCY WITH NEW NEUROIMAGING FEATURES**
Kasapkara C. S., Aydin K., Hasanoglu A., Kucukcongar A., Ezgu F. S., Tumer L.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.35, 2012 (SCI-Expanded)

XCIX. **THE SUBCLINICAL NEUROLOGICAL FINDINGS IN TYPE 1 GAUCHER DISEASE**
Kucukcongar A., Aydin K., Ezgu F. S., Kasapkara C. S., Tumer L., Hasanoglu A.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.35, 2012 (SCI-Expanded)

C. **GENOTYPIC FEATURES OF 41 PATIENTS WITH GAUCHER DISEASE FROM TURKEY**
Hasanoglu A., Akay G., Ezgu F. S., Biberoglu G., Tumer L., Okur İ., Kucukcongar A., Kasapkara C., Polat M., Ciftci B.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.35, 2012 (SCI-Expanded)

CI. **COMPREHENSIVE SEQUENCING OF MITOCHONDRIAL DNA IN PATIENTS WITH SUSPECTED MITOCHONDRIAL DISEASE: IS THERE A NEED FOR A REVISED MOLECULAR DIAGNOSTIC ALGORITHM?**
Ezgu F. S., Kucukcongar A., Ciftci B., Kasapkara C., Hasanoglu A., Tumer L., Okur İ., Gunduz M., Polat M., Bahceci S.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.35, 2012 (SCI-Expanded)

CII. **IS THERE ANY EFFECT OF GROWTH HORMONE THERAPY ON CARNITINE AND ACLYLCARNITINE STATUS IN CHILDREN?**
Gursel B., Alev H., Tuba E. F., Ilyas O., Ediz Y., Leyla T., Suheyl E. F., Aysun B., Betul D.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.35, 2012 (SCI-Expanded)

CIII. **LYSOSOMAL STORAGE DISEASES IN OUR COUNTRY: RESULTS OF LAST SIX YEARS**
Alev H., Gursel B., Suheyl E. F., Leyla T., Seher K. C., Aynur K., Basak U.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.35, 2012 (SCI-Expanded)

CIV. **GOOD OUTCOME OF ERT OF TWO INFANTILE POMPE DISEASE CASES**
Hasanoglu A., Kucukcongar A., Ezgu F. S., Tumer L., Reuser A. J. J.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.35, 2012 (SCI-Expanded)

CV. **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, cilt.35, 2012 (SCI-Expanded)

CVI. **DESENSITIZATION OF AN INFANT WITH POMPE DISEASE AND A HISTORY OF ANAPHYLAXIS TO ALGLUCOSIDASE ALFA**
Kucukcongar A., Karagol E., Ezgu F. S., Tumer L., Kasapkara C. S., Hasanoglu A.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.35, 2012 (SCI-Expanded)

CVII. **HYPERPHENYLALANINAEMIA DUE TO PHENYLALANIN HYDROXYLASE DEFICIENCY: EVALUATION OF TETRAHYDOBIOPTERIN RESPONSIVNESS IN PATIENTS FROM TURKEY**
Kucukcongar A., Ezgu F. S., Tumer L., Hasanoglu A.

JOURNAL OF INHERITED METABOLIC DISEASE, cilt.35, 2012 (SCI-Expanded)

CVIII. **SUCCESSFUL MANAGEMENT OF INFUSION-ASSOCIATED REACTIONS IN A 23 YEARS OLD PATIENT WITH MUCOPOLYSACCHARIDOSIS VI RECEIVING RECOMBINANT HUMAN ARYLSULFATASE B**
Hasanoglu A., Kasapkara C. S., Celik G., Kucukcongar A., Ezgu F. S., Tumer L.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.35, 2012 (SCI-Expanded)

CIX. **HOME SLEEP STUDY CHARACTERISTICS IN PATIENTS WITH MUCOPOLYSACCHARIDOSIS**
Kasapkara C. S., Tumer L., Hasanoglu A., Ezgu F. S., Arslan A., Kokturk O., Kucujcongar A.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.35, 2012 (SCI-Expanded)

CX. **BONE METABOLISM IN PATIENTS AFFECTED BY GAUCHER DISEASE**
Hasanoglu A., Tumer L., Ezgu F. S., Kasapkara C. S., Kucukcongar A.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.35, 2012 (SCI-Expanded)

CXI. **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, cilt.35, 2012 (SCI-Expanded)

CXII. **Hypercalcemia in glycogen storage disease type I patients of Turkish origin**
Kasapkara C. S., TÜMER L., OKUR İ., Eminoglu T., EZGÜ F. S., Hasanoglu A.
TURKISH JOURNAL OF PEDIATRICS, cilt.54, sa.1, ss.35-37, 2012 (SCI-Expanded)

CXIII. **Analysis of acylcarnitine levels by tandem mass spectrometry in epileptic children receiving valproate and oxcarbazepine**
CANSU A., SERDAROĞLU A., BİBEROĞLU G., TÜMER L., HIRFANOĞLU T., EZGÜ F. S., Hasanoglu A.
EPILEPTIC DISORDERS, cilt.13, sa.4, ss.394-400, 2011 (SCI-Expanded)

CXIV. **A girl with spastic tetraparesis associated with biotinidase deficiency**
KÖMÜR M., OKUYAZ Ç., Ezgu F. S., Atici A.
EUROPEAN JOURNAL OF PAEDIATRIC NEUROLOGY, cilt.15, sa.6, ss.551-553, 2011 (SCI-Expanded)

CXV. **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, cilt.210, 2011 (SCI-Expanded)

CXVI. **N-carbamylglutamate treatment for acute neonatal hyperammonemia in isovaleric acidemia**
Kasapkara C. S., EZGÜ F. S., OKUR İ., TÜMER L., BİBEROĞLU G., Hasanoglu A.
EUROPEAN JOURNAL OF PEDIATRICS, cilt.170, sa.6, ss.799-801, 2011 (SCI-Expanded)

CXVII. **A newborn case with perinatal-lethal Gaucher disease due to R463H homozygosity complicated by C677T homozygosity in the MTHFR gene**
Akdag A., Oguz S. S., Ezgu F. S., Erdeve O., Uras N., Dilmen U.
JOURNAL OF PEDIATRIC ENDOCRINOLOGY & METABOLISM, cilt.24, sa.5-6, ss.381-383, 2011 (SCI-Expanded)

CXVIII. **Harderoporphyria due to homozygosity for coproporphyrinogen oxidase missense mutation H327R**
Hasanoglu A., Balwani M., Kasapkara C. S., EZGÜ F. S., OKUR İ., TÜMER L., Cakmak A., Nazarenko I., Yu C., Clavero S., et al.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.34, sa.1, ss.225-231, 2011 (SCI-Expanded)

CXIX. **MOST COMMON LYSOSOMAL STORAGE DISEASES IN OUR COUNTRY: RESULTS OF LAST FIVE YEARS**
Biberoglu G., Ezgu F. S., Hasanoglu A., Tumer L.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.34, 2011 (SCI-Expanded)

CXX. **SRD5A3-CDG: A PATIENT WITH A NOVEL MUTATION**
Leyla T., Seher K. C., Suheyl E. F., Alev H., Valerie R., Gert M., Jacques J.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.34, 2011 (SCI-Expanded)

CXXI. **AN INFANTILE CASE OF ZELLWEGER SYNDROME PRESENTED WITH KABUKI-LIKE PHENOTYPE**
Ezgu F. S., Eminoglu T., OKUR İ., Gunduz M., Tumer L., Hasanoglu A., Dalgic B.
GENETIC COUNSELING, cilt.22, sa.2, ss.217-220, 2011 (SCI-Expanded)

CXXII. **CANAVAN DISEASE: CASE REPORT**
Hasanoglu A., Kucukcongar A., Ezgu F. S., Tumer L., Kasapkara C. S., Biberoglu G., Salomons G. S.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.34, 2011 (SCI-Expanded)

CXXIII. **HALLERMANN-STREIFF SYNDROME WITH HYPERPHENYLALANINEMIA: CASE REPORT**
Ezgu F. S., Kucukcongar A., Hondur A. M., Tumer L., Hasanoglu A.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.34, 2011 (SCI-Expanded)

CXXIV. **THREE SIBLINGS WITH EXT1-CDG**
Suheyl E. F., Seher K. C., Ilyas O., Aynur K., Leyla T., Arzu O., Avni S., Wim W., Els Van H., Alev H.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.34, 2011 (SCI-Expanded)

CXXV. **AN UNUSUAL PRESENTATION OF MUCOPOLYSACCARIDOSIS VI WITH A NOVEL MUTATION**
Kucukcongar A., Tumer L., Ezgu F. S., Biberoglu G., Kasapkara C. S., Gal A., Hasanoglu A.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.34, 2011 (SCI-Expanded)

CXXVI. **OUTCOMES OF ENZYME REPLACEMENT THERAPY FOR MUCOPOLYSACCHARIDOSIS TYPE VI: GAZI UNIVERSITY EXPERIENCE**
Alev H., Suheyl E. F., Leyla T., Seher K. C., Ilyas O., Aynur K.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.34, 2011 (SCI-Expanded)

CXXVII. **Carnitine Palmitoyl Transferase II Deficiency with Renal Failure**
Gok F., Hacihamdioglu D. O., Kalman S., Ezgu F. S.
PEDIATRIC NEPHROLOGY, cilt.25, sa.9, ss.1838, 2010 (SCI-Expanded)

CXXVIII. **THE SCANNING OF COMMONLY SEEN MUTATIONS OF GLUCOSE-6-PHOSPHATASE AND GLUCOSE-6-PHOSPHATASE TRANSLOCASE GENES IN GLYCOGEN STORAGE TYPE 1A AND TYPE 1B DISEASE PATIENTS BY THE MICROELECTRONIC ARRAY TECHNOLOGY**
Eminoglu F. T., Tumer L., Ezgu F. S., Okur İ., Biberoglu G., Hasanoglu A.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.33, 2010 (SCI-Expanded)

CXXIX. **GENOTYPE-PHENOTYPE CORRELATIONS IN TURKISH PATIENTS WITH ALPHA GALACTOSIDASE A DEFICIENCY**
Koca S., Ezgu F. S., Okur İ., Biberoglu G., Tumer L., Hasanoglu A.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.33, 2010 (SCI-Expanded)

CXXX. **HYPERCALCAEMIA IN GLYCOGEN STORAGE DISEASE TYPE 1 PATIENTS OF TURKISH ORIGIN**
Kasapkara C. S., Tumer L., Okur İ., Eminoglu F. T., Ezgu F. S., Hasanoglu A.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.33, 2010 (SCI-Expanded)

CXXXI. **N-CARBAMYLGLUTAMATE TREATMENT FOR ACUTE NEONATAL HYPERAMMONAEMIA IN ISOVALERIC ACIDAEMIA**
Kasapkara C. S., Ezgu F. S., Tumer L., Biberoglu G., Okur İ., Hasanoglu A.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.33, 2010 (SCI-Expanded)

CXXXII. **FOUR CASES OF NIEMANN-PICK TYPE C DISEASE PRESENTED WITH EARLY ONSET CHOLESTASIS**
Kucukcongar A., Okur İ., Ezgu F. S., Turner L., Dalgic B., Hasanoglu A.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.33, 2010 (SCI-Expanded)

CXXXIII. **HARDEROPORPHYRIA PHENOTYPE DUE TO A HOMOZYGOUS H237R MISSENSE MUTATION**
Kasapkara C. S., Hasanoglu A., Ezgu F. S., Okur İ., Tumer L., Cakmak A., Balwani M., Nazarenko I., Clavero S., Yu C., et al.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.33, 2010 (SCI-Expanded)

CXXXIV. **Mucopolysaccharidosis Type IIID: 12 New Patients and 15 Novel Mutations**
Valstar M. J., Bertoli-Avella A. M., Wessels M. W., Ruijter G. J. G., de Graaf B., Olmer R., Elfferich P., Neijs S., Kariminejad R., EZGÜ F. S., et al.
HUMAN MUTATION, cilt.31, sa.5, 2010 (SCI-Expanded)

CXXXV. **Screening for isolated sulfite oxidase/molibden cofactor deficiencies among the pediatric patients with encephalopathy and mental-motor retardation**
Keles E., Ezgu F. S., Tosun M. S., Okur İ., Eminoglu F. T., Tumer L., Hasanoglu A.
EUROPEAN JOURNAL OF MEDICAL RESEARCH, cilt.14, ss.84, 2009 (SCI-Expanded)

CXXXVI. **Homocysteine, Fibrinogen and Anti-ox-LDL Antibody Levels as Markers of Atherosclerosis in Prepubertal Obese Children**
Ezgu F. S., Tumer L., Ozbay F., Hasanoglu A., Biberoglu G., AYBAY C.
JOURNAL OF PEDIATRIC ENDOCRINOLOGY & METABOLISM, cilt.22, sa.10, ss.915-920, 2009 (SCI-Expanded)

CXXXVII. **The Gene of Bloom's Syndrome: An Autosomal Recessive Disorder with Male Dominance**
ASLAN D., EZGÜ F. S.
GENETIC TESTING AND MOLECULAR BIOMARKERS, cilt.13, sa.4, ss.443-444, 2009 (SCI-Expanded)

CXXXVIII. **The Erlenmeyer Flask Bone Deformity In the Skeletal Dysplasias**
Faden M. A., Krakow D., Ezgu F. S., Rimoin D. L., Lachman R. S.
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, cilt.149A, sa.6, ss.1334-1345, 2009 (SCI-Expanded)

CXXXIX. **The same novel mutation determined in 2 Hurler-Scheie patients who are the children of different families**
Hasanoglu A., Okur İ., Eminoglu F. T., Tumer L., Biberoglu G., Bertola F., Ezgu F. S.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.31, ss.112, 2008 (SCI-Expanded)

CXL. **Very long-chain acyl CoA dehydrogenase deficiency which was accepted as infanticide**
Eminoglu F. T., Tumer L., Okur İ., Goekmen Z., Ezgu F. S., Biberoglu G., Hasanoglu A.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.31, ss.33, 2008 (SCI-Expanded)

CXLI. **Acute renal failure due to rhabdomyolysis in a child with McArdle disease**
DELİBAŞ A., BEK K., EZGÜ F. S., Demircin G., Oksal A., Oner A.
EUROPEAN JOURNAL OF PEDIATRICS, cilt.167, sa.8, ss.939-940, 2008 (SCI-Expanded)

CXLII. **The neonatal case diagnosed with nonketotic hyperglycinemia with a preliminary diagnosis of the chloralhydrate intoxication**
Okur İ., Eminoglu F. T., Tumer L., Ezgu F. S., Biberoglu G., Hasanoglu A.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.31, ss.3, 2008 (SCI-Expanded)

CXLIII. **Severe infantile hypotonia with ethylmalonic aciduria: Case report**
OKUYAZ Ç., EZGÜ F. S., Biberoglu G., Zeviani M., Tiranti V., Yilgor E.
JOURNAL OF CHILD NEUROLOGY, cilt.23, sa.6, ss.703-705, 2008 (SCI-Expanded)

CXLIV. **Rapid screening of 10 common mutations in Turkish Gaucher patients using electronic DNA microarray**
Ezgu F. S., Hasanoglu A., OKUR İ., Biberoglu G., Tumer L., Eminoglu T., Dogan H.
BLOOD CELLS MOLECULES AND DISEASES, cilt.40, sa.2, ss.246-247, 2008 (SCI-Expanded)

CXLV. **Investigation of 10 common mutations in Turkish Gaucher patients by use of the nanochip microelectronic array technology**
Hasanoglu A., Ezgu F. S., Okur İ., Eminoglu F. T., Biberoglu G., Tumer L.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.30, ss.108, 2007 (SCI-Expanded)

CXLVI. **The first results of 18 months experience with lysosomal storage disease**
Biberoglu G., Hasanoglu A., Ezgu F. S., Tumer L., Okur İ., Eminoglu F. T., Yalcinkaya D.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.30, ss.95, 2007 (SCI-Expanded)

CXLVII. **Incidence of osteoporosis in a metabolic unit**
Hasanoglu A., Turner L., Ezgu F. S., Eminoglu F. T., Okur İ.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.29, ss.159, 2006 (SCI-Expanded)

CXLVIII. **Ethylmalonic encephalopathy due to ETHE1 mutation**
Turner L., Ezgu F. S., Gunduz M., Hasanoglu A., Tiranti V., Zeviani M.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.29, ss.159, 2006 (SCI-Expanded)

CXLIX. **The effects of laronidase treatment in a patient with Hurler syndrome: Results of one year therapy**
Hasanoglu A., Tumer L., Ezgu F. S., Gunduz M., Okur İ., Eminoglu T.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.29, ss.149, 2006 (SCI-Expanded)

CL. **Severe renal tubulopathy and cholestasis due to BCSIL gene mutation of complex III: Experience with high dose sodium bicarbonate therapy**
Ezgu F. S., Tumer L., Hasanoglu A., Gunduz M., Tyras U., Unsal R., Seneca S.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.29, ss.116, 2006 (SCI-Expanded)

CLI. **Effects of oral glutamine supplementation on children with solid tumors receiving chemotherapy**
Okur A., Ezgu F. S., Tumer L., Cinasal G., Oguz A., Hasanoglu A., Karadeniz C.
PEDIATRIC HEMATOLOGY AND ONCOLOGY, cilt.23, sa.4, ss.277-285, 2006 (SCI-Expanded)

CLII. **Cholesteryl ester storage disease in a young child presenting as isolated hepatomegaly treated with**

simvastatin.
Dalgic B., Sari S., Gunduz M., Ezgu F. S., Tumer L., Hasanoglu A., Akyol G.
The Turkish journal of pediatrics, cilt.48, sa.2, ss.148-51, 2006 (SCI-Expanded)

CLIII. **Cirrhosis in an infant heterozygous for classical citrullinaemia**
Ezgu F. S., Tumer L., Dalgic B., Hasanoglu A., Kobayashi K., Saheki T.
ACTA PAEDIATRICA, cilt.94, sa.12, ss.1849-1851, 2005 (SCI-Expanded)

CLIV. **Endothelial activation and inflammation in prepubertal obese Turkish children**
Ezgu F. S., Hasanoglu A., Tumer L., Ozbay F., Aybay C., Gunduz M.
METABOLISM-CLINICAL AND EXPERIMENTAL, cilt.54, sa.10, ss.1384-1389, 2005 (SCI-Expanded)

CLV. **Postoperative secondary glaucoma and anterior staphyloma in a patient with homocystinuria**
Ozdek Ş., Bahceci U., Onol M., Ezgu F. S., Hasanreisoglu B.
JOURNAL OF PEDIATRIC OPHTHALMOLOGY & STRABISMUS, cilt.42, sa.4, ss.243-246, 2005 (SCI-Expanded)

CLVI. **Effect of enteral. and parenteral nutrition on growth parameters of hospitalized Turkish children**
Ezgu F. S., Tumer L., Cinasal G., Hasanoglu A., Gunduz M.
INDIAN PEDIATRICS, cilt.42, sa.5, ss.469-472, 2005 (SCI-Expanded)

CLVII. **THE RESULTS OF HIGH DOSE IMIGLUCERASE TREATMENT IN A TYPE THREE GAUCHER PATIENT ADMITTED WITH OCULOMOTOR APRAXIA**
Tumer L., Ezgu F. S., Aksu T., Gunduz M., Hasanoglu A.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.28, ss.174, 2005 (SCI-Expanded)

CLVIII. **THE CO-EXISTENCE OF SATOYOSHI SYNDROME AND MYOADENYLATE DEAMINASE DEFICIENCY**
Ezgu F. S., Tumer L., Serdaroglu A., Hasanoglu A., Cansu A., Hirfanoglu T., Dalgic B.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.28, ss.253, 2005 (SCI-Expanded)

CLIX. **TWO CASES OF GM1 GANGLIOSIDOSIS WITH NOVEL MUTATIONS**
Ezgu F. S., Gunduz M., Tuemer L., Fatih A., Paschke E., Hasanodlu A.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.28, ss.157, 2005 (SCI-Expanded)

CLX. **Serum carnitine levels in newborns with perinatal asphyxia and relation to neurologic prognosis**
Ezgu F. S., Atalay Y., Hasanoglu A., Gucuyener K., Biberoglu G., Koc E., Ergenekon E., Tumer L.
NUTRITIONAL NEUROSCIENCE, cilt.7, sa.5-6, ss.351-356, 2004 (SCI-Expanded)

CLXI. **Vertebra and femur neck bone mineral density values in healthy Turkish children**
Hasanoglu A., Tumer L., Ezgu F. S.
TURKISH JOURNAL OF PEDIATRICS, cilt.46, sa.4, ss.298-302, 2004 (SCI-Expanded)

CLXII. **Investigating for inborn errors of metabolism with tandem mass spectrometry in Ankara Region, Turkey: A 2 years evaluation**
Tumer L., Biberoglu G., Hasanoglu A., Ezgu F. S., Atalay Y.
JOURNAL OF TROPICAL PEDIATRICS, cilt.50, sa.3, ss.189-190, 2004 (SCI-Expanded)

CLXIII. **The co-existence of Fabry and celiac diseases: a case report**
Tumer L., Ezgu F. S., Hasanoglu A., Dalgic B., Bakkaloglu S. A., Memis L., Dursun A.
PEDIATRIC NEPHROLOGY, cilt.19, sa.6, ss.679-681, 2004 (SCI-Expanded)

CLXIV. **Vitamin D intoxication and hypercalcaemia in an infant treated with pamidronate infusions**
Ezgu F. S., Buyan N., Gunduz M., Tumer L., OKUR İ., Hasanoglu A.
EUROPEAN JOURNAL OF PEDIATRICS, cilt.163, sa.3, ss.163-165, 2004 (SCI-Expanded)

CLXV. **Intracranial venous thrombosis after hypoxic-ischemic brain insult in two newborns: Could low serum carnitine levels have contributed?**
Ezgu F. S., Atalay Y., Hasanoglu A., Gucuyener K., Koc E., Ergenekon E.
NUTRITIONAL NEUROSCIENCE, cilt.7, sa.1, ss.63-65, 2004 (SCI-Expanded)

CLXVI. **Neuron-specific enolase levels and neuroimaging in asphyxiated term newborns**
Ezgu F. S., Atalay Y., Gucuyener K., Tunc S., Koc E., Ergenekon E., Tiras U.
JOURNAL OF CHILD NEUROLOGY, cilt.17, sa.11, ss.824-829, 2002 (SCI-Expanded)

CLXVII. **Leptin levels in children with insulin dependent diabetes mellitus**
Bideci A., Cinaz P., Ezgu F. S.
TURKISH JOURNAL OF PEDIATRICS, cilt.44, sa.3, ss.211-214, 2002 (SCI-Expanded)

CLXVIII. **Plasma leptin levels of infants after feeding**
Cinaz P., Bideci A., Ezgu F. S.
ACTA PAEDIATRICA, cilt.89, sa.6, ss.746, 2000 (SCI-Expanded)

CLXIX. **Cerebrospinal fluid and serum nitric oxide levels in asphyxiated newborns**
Ergenekon E., Gucuyener K., Erbas D., Ezgu F. S., Atalay Y.
BIOLOGY OF THE NEONATE, cilt.76, sa.4, ss.200-206, 1999 (SCI-Expanded)

CLXX. **Plasma leptin levels of large for gestational age and small for gestational age infants**
Cinaz P., Sen E., Bideci A., Ezgu F. S., Atalay Y., Koca E.
ACTA PAEDIATRICA, cilt.88, sa.7, ss.753-756, 1999 (SCI-Expanded)

CLXXI. **Suspected allergy to lamotrigine**
Gucuyene K., Turktas I., Serdaroglu A., Ezgu F. S.
ALLERGY, cilt.54, sa.7, ss.767-768, 1999 (SCI-Expanded)

CLXXII. **Autoimmune polyglandular syndrome type I - A case report**
Cinaz P., Bideci A., Haznedaroglu A., Ezgu F. S., Agaoglu O., Kursaklioglu S.
TURKISH JOURNAL OF PEDIATRICS, cilt.39, sa.2, ss.271-275, 1997 (SCI-Expanded)

Diğer Dergilerde Yayınlanan Makaleler

I. **Simultaneous succinylacetone-nitisinone measurement in tyrosinemia type I patients and evaluation of the nitisinone therapeutic range**
Öktem R. M., İnci A., Biberoğlu G., Okur İ., Ezgü F. S., Tümer L.
Biochimica Clinica, cilt.47, sa.3, ss.340-345, 2023 (Scopus)

II. **Dietary Fiber Supplementation in Type I Glycogen Storage Disease; Could it Contribute to a Better Metabolic Control?**
Emecen Şanlı M., Aktaş E., İnci A., Okur İ., Ezgü F. S., Tümer L.
GUNCEL PEDIATRI, cilt.21, 2023 (ESCI)

III. **Variants of Fumarate Hydratase Gene in Uterine Disorders: A Clinical Trial**
KAZANCI F., YÜKSEL D., EZGÜ F. S., ERDEM O., ONAN M. A.
Journal of Clinical Obstetrics & Gynecology, cilt.33, sa.2, ss.81-87, 2023 (ESCI)

IV. **MİTOKONDRİYAL HASTALIK NEDENİYLE TETKİK EDİLEN HASTALARDA M.16189T>C DEĞİŞİKLİĞİNİN METABOLİK SENDROM AÇISINDAN İNCELENMESİ**
İNCİ A., Hasanoğlu A., OKUR İ., BİBEROĞLU G., TÜMER L., EZGÜ F. S.
Kocatepe Tıp Dergisi, cilt.23, sa.3, ss.322-325, 2022 (Hakemli Dergi)

V. **PROPIONYLCARNITINE AND FREE CARNITINE ARE NEW BIOMARKERS IN THE FOLLOW-UP PERIOD OF MUCOPOLYSACCARIDOSIS TO SCREEN OXIDATIVE STRESS**
İNCİ A., OLGAÇ A., GENÇ DERİN B., BİBEROĞLU G., OKUR İ., EZGÜ F. S., TÜMER L.
Süleyman Demirel Üniversitesi Tıp Fakültesi Dergisi, cilt.28, sa.4, ss.565-571, 2021 (Hakemli Dergi)

VI. **The effect of a geometric-shaped tool with blue led light on the activation of human dermal fibroblasts and cancer cells**
Kılıçarslan Törüner E., Kayhan H., Ezgu F. S.
Journal of Photochemistry and Photobiology, cilt.8, 2021 (Scopus)

VII. **Do cytokines play role in the pathogenesis of mucopolysaccharidosis**
İNCİ A., OLGAÇ KILIÇKAYA M. A. B., YILMAZ DEMİRTAŞ C., OKUR İ., BİBEROĞLU G., EZGÜ F. S., TÜMER L.
Medicine Science, cilt.10, sa.4, ss.1492-1497, 2021 (Hakemli Dergi)

VIII. **The Evaluation of Skeletal Manifestations in Patients with Gaucher Disease**
Kasapkara C. S., Olgac A., OKUR İ., EZGÜ F. S., TÜMER L.
JOURNAL OF PEDIATRIC RESEARCH, cilt.8, sa.3, ss.257-261, 2021 (ESCI)

IX. **Pierson Syndrome Characterized by Mild Renal Variant: A Case Report**
BÜYÜKKARAGÖZ B., BAKKALOĞLU EZGÜ S. A., ÖZMEN M. C., EZGÜ F. S.
GAZI MEDICAL JOURNAL, cilt.32, sa.3, ss.461-463, 2021 (ESCI)

X. **Acute Stroke in a Patient with Mucopolysaccharidosis Type I with Increased Carotid Intima-Media Thickness**
Olgac A., TÜMER L., Damar C., Hasanoglu A., Ezgu F. S.
EURASIAN JOURNAL OF MEDICINE AND ONCOLOGY, cilt.2, sa.2, ss.114-116, 2018 (ESCI)

XI. **Clinical course and outcome of glycogen-storage disease type 1a and type 1b**
Eminoglu F. T., TÜMER L., OKUR İ., EZGÜ F. S., Hasanoglu A.
TURK PEDIATRI ARSIVI-TURKISH ARCHIVES OF PEDIATRICS, cilt.48, sa.2, ss.117-122, 2013 (ESCI)

XII. **Isolated sulfite oxidase deficiency**
Senbil N., Tosun M. S., EZGÜ F. S., Gurer Y. K. Y.
TURK PEDIATRI ARSIVI-TURKISH ARCHIVES OF PEDIATRICS, cilt.40, sa.2, ss.105-108, 2005 (ESCI)

XIII. **Türk Toplumunda Tip I Mukopolisakkaridoz un Moleküler Analizi**
YILMAZ A., ERGÜN M. A., ezgü F. S., menevşe a., hasanoğlu a.
Turkiye Klinikleri J Pediatr Sci, cilt.1, ss.127-128, 2005 (Hakemli Dergi)

Hakemli Kongre / Sempozyum Bildiri Kitaplarında Yer Alan Yayınlar

I. **Ultra-Nadir Bir Hastalık ve Yeni Bir Mutasyon; Smg9 Eksikliği, Bir Aile 4Etkilenmiş Birey**
Ergin F. B., İnci A., Ezgü F. S.
6. Ulusal Çocuk Genetik Kongresini 09-12 Kasım 2023, Aydın, Türkiye, 9 - 11 Kasım 2023

II. **Fenilketonüri Tanısı İle İzlenen Hastalarda Visseral Adipositenin Değerlendirilmesi**
Gökalp S., Bostancı F., Aktaş E., İnci A., Okur İ., Ezgü F. S., Tümer L.
I. Ulusal Çocuk Beslenme Kongresi, Gaziantep, Türkiye, 25 Ekim 2023

III. **Glikojen Depo Tip 1 Hastalığında Lif Takviyesinin Metabolik Kontrole Etkisi**
Emecen Şanlı M., Aktaş E., İnci A., Okur İ., Ezgü F. S., Tümer L.
I. Ulusal Çocuk Beslenme Kongresi, Gaziantep, Türkiye, 25 Ekim 2023

IV. **Bone Turnover in Patients with Lysosomal Storage Disorders**
Gökalp S., İnci A., Okur İ., Ezgü F. S., Tümer L.
Annual Symposium 2023, Jerusalem, Yerushalayim, İsrail, 29 Ağustos - 01 Eylül 2023

V. **EVENT BASED TREATMENT OUTCOMES OF PATIENTS WITH GAUCHER DISEASE: ADIFFERENT PERSPECTIVE**
Kilic A., İnci A., Okur İ., Tümer L., Ezgü F. S.
Annual Symposium 2023, Jerusalem, Yerushalayim, İsrail, 29 Ağustos 2023

VI. **Intestinal Microbiota Composition of Children with Glycogen Storage Disease Type 1**
Gökalp S., Dinleyici E. Ç., Muluk C., İnci A., Aktaş E., Okur İ., Ezgü F. S., Tümer L.
SSIEM 2023, Yerushalayim, İsrail, 29 Ağustos 2023

VII. **An Alternative for Early Detection of Cardiac Involvement in Gaucher Type 1 Disease: Speckle Tracking Echocardiograpy**
GÖKALP S., ÜNLÜ S., İNCİ A., OKUR İ., EZGÜ F. S., TÜMER L.
Annual Symposium SSIEM 2023, İsrail, 29 Ağustos - 01 Eylül 2023

VIII. **Pterin Profiling in Serum, Dry Blood Spot and Urine using LC-MS/MS in Patients withHyperphenylalaninemia**
Öktem R. M., İnci A., Bayrak H., Demir F., Biberoğlu G., Mavis M. E., Okur İ., Ezgü F. S., Tümer L.
Annual Symposium 2023, Jerusalem, Yerushalayim, İsrail, 29 Ağustos 2023

IX. **How about new treatment in rare disease?**
EZGÜ F. S.
The National Conference On Rare diseases, Libya, 27 - 28 Aralık 2022

X. **Retargeting phenylbutyrate, ursodeoxycholic acid, pyrimethamine and betaine for beta-glucocerebrosidaserecovery in gaucher disease fibroblasts resulting from homozygous p.L483P mutation**
Kiliç A., BİBEROĞLU G., ÖKTEM R. M., İNCİ A., Aydogdu S., Udgu Isik B., IŞIK GÖNÜL İ., OKUR İ., TÜMER L., EZGÜ F. S.

SSIEM Annual Symposium, Almanya, 30 Ağustos - 02 Eylül 2022, sa.1418955

XI. **İNFANTİL TİP POMPE HASTALIĞI ULUSAL KONSENSUS ÇALIŞMASI**
Aktaşoğlu E., İNCİ A., OKUR İ., BİBEROĞLU G., ÖKTEM R. M., EZGÜ F. S., TÜMER L., Kılıç M., Güneş S., KAĞNICI M., et al.
VII. Uluslararası Katılımlı Lizozomal Hastalıklar Kongresi, Türkiye, 25 - 27 Kasım 2021

XII. **Gaucher Tip I Hastalığında Kardiyak Tutulumun Erken Saptanması için Bir Alternatif: Speckle Tracking Ekokardiyografi**
GÖKALP S., ÜNLÜ S., İNCİ A., OKUR İ., EZGÜ F. S., TAÇOY G., EMİNOĞLU F. T., KASAPKARA Ç. S., TÜMER L.
VII. Uluslararası Katılımlı Lizozomal Hastalıklar Kongresi 25-27 Kasım 2021 Çevrimiçi Kongre http://lizozomal2021.org/, Türkiye, 25 - 27 Kasım 2021

XIII. **MPS 6 Hastalarında Klinik Bulgular, ERT önce ve Sonrası Olay Bazlı Değerlendirme**
İNCİ A., OKUR İ., TÜMER L., BİBEROĞLU G., ÖKTEM R. M., EZGÜ F. S.
VII. Uluslararası Katılımlı Lizozomal Hastalıklar Kongresi, Türkiye, 25 - 27 Kasım 2021

XIV. **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.
14th International Congress Of Inborn Errors Of Metabolism, Sidney, Avustralya, 21 - 23 Kasım 2021

XV. **A PATIENT WITH ADENOSINE KINASE DEFICIENCY DUE TO A NOVEL MUTATION PRESENTING WITH NOVEL DYSMORPHIC AND CARDIAC FINDINGS**
Aktaşoğlu E., Kılıç A., Emecan Şanlı M., GÖKALP S., İNCİ A., OKUR İ., EZGÜ F. S., SARI S., DALGIÇ B., CEYLANER S., et al.
14th International Congress of Inborn Errors of Metabolism 2021, Avustralya, 21 - 24 Kasım 2021

XVI. **Pompe Hastalarında Enzim Replasman Tedavisine Bağlı Anafilaksi ve Yönetimi:Tek Merkez Deneyim**
ERTOY KARAGÖL H. İ., İNCİ A., Polat Tecere S., KILIÇ A., Demir F., YAPAR D., OKUR İ., EZGÜ F. S., TÜMER L., BAKIRTAŞ A.
XXVIII. Ulusal Alerji ve Klinik İmmünoloji kongresi, Türkiye, 13 - 17 Ekim 2021

XVII. **Triamterene-induced suppression of R227X premature termination codon in Fabry disease**
Dündar H., Udgu B., Biberoğlu G., Inci A., Ezgu F. S., Işık Gönül İ., Okur İ., Tümer L.
16th Annual Research Meeting of the WORLDSymposium(TM), Florida, Amerika Birleşik Devletleri, 10 - 14 Şubat 2020, cilt.129

XVIII. **Anatomy of MPS Patients**
EZGÜ F. S.
MPS Anesthesia Course - Istanbul, Turkey - 13-14th December 2019, 13 - 14 Aralık 2019

XIX. **Diagnostic workshop: 'Solve the mystery case'**
EZGÜ F. S.
MPS Masterclass 2019, 03–05th November 2019, Budapest-Hungary, 3 - 05 Kasım 2019

XX. **Laboratory diagnosis of Mucopolysaccharidosis disorders**
EZGÜ F. S.
MPS Masterclass 2019, 03–05th November 2019, Budapest-Hungary, 3 - 05 Kasım 2019

XXI. **Analysis of hereditary nephrotic diseases through next generation DNA sequencing**
YAZICIOĞLU B., BAKKALOĞLU EZGÜ S. A., BÜYÜKKARAGÖZ B., EZGÜ F. S.
18th Congress of the International Pediatric Nephrology Association, Venice, ITALY, 17 - 21 Ekim 2019

XXII. **Case Study: Switch from agalsidase alfa to beta**
EZGÜ F. S.
Fabry Academy South Asia, 18-19th October, 2019, Taipei, Taiwan, 18 - 19 Ekim 2019

XXIII. **Fabry Hastalığı'nda Doğal Seyir**
EZGÜ F. S.
Enine Boyuna Fabry Hastalığı, 10-12 Ekim 2019, Ankara, Türkiye, 10 - 12 Ekim 2019

XXIV. **Fabry mi? Değil mi?**
EZGÜ F. S.
Enine Boyuna Fabry Hastalığı, 10-12 Ekim 2019, Ankara, Türkiye, 10 - 12 Ekim 2019

XXV. **Fabry Hastalığı'nda Kalıtım**

EZGÜ F. S.
Enine Boyuna Fabry Hastalığı, 10-12 Ekim 2019, Ankara, Türkiye, 10 - 12 Ekim 2019

XXVI. **Fabry Hastalığı'nda Tedavi Seçenekleri, Endikasyonlar ve Sorunlar**
EZGÜ F. S.
Enine Boyuna Fabry Hastalığı, 10-12 Ekim 2019, Ankara, Türkiye, 10 - 12 Ekim 2019

XXVII. **Akut Porfiria Benzeri Periferik Nöropati Gelişen Tirozinemi Tip 1 Olgusu**
AKKUZU E., ÖZEN DEMİRCİOĞLU P., İNCİ A., OKUR İ., EZGÜ F. S.
16. Çocuk Acil Tıp ve Yoğun Bakım Kongresi, Antalya, Türkiye, 2 - 05 Ekim 2019, ss.327-328

XXVIII. **Genetik Hastalıklarda Yeni Tedaviler**
EZGÜ F. S.
4.Ulusal Çocuk Genetik Kongresi, 25-27 Eylül 2019, Ankara, Türkiye, 25 - 27 Eylül 2019

XXIX. **Screening of twelve lysosomal storage diseases with LC-MS/MS in Gazi university hospital in Turkey: The firstresults of validation**
BİBEROĞLU G., İNCİ A., DERİN B., OKUR İ., EZGÜ F. S., TÜMER L.
SSIEM, 3 - 06 Eylül 2019

XXX. **Benefficial effects of Modified Atkins Diet in Glygocen Storage Disorder Type IIIa**
OLGAÇ KILIÇKAYA M. A. B., İNCİ A., OKUR İ., KASAPKARA Ç. S., BİBEROĞLU G., OĞUZ A. D., AKTAŞ E., EZGÜ F. S., TÜMER L.
SSIEM Annual Symposium 2019, Rotterdam, Hollanda, 3 - 06 Eylül 2019

XXXI. **Mild hyperammonemia due to Antiquitin deficiency**
kılıç m., Kasapkara Ç. S., OLGAÇ M. A. B., aksoy a., EZGÜ F. S.
SSIEM 2019, 3-6th September, 2019, Rotterdam-The Netherlands, 3 - 06 Eylül 2019

XXXII. **A Rare Case of Primary Coenzyme Q10 deficiency due to COQ9 gene mutation**
OLGAÇ M. A. B., Öztoprak Ü., Kasapkara Ç. S., Yüksel D., DERİNKUYU B. E., EZGÜ F. S.
SSIEM 2019, 3-6th September, 2019, Rotterdam-The Netherlands, 3 - 06 Eylül 2019

XXXIII. **Next generation DNA sequencing as an initial diagnostic method for congenital defects of glycosylation**
EZGÜ F. S., İNCİ A., Çiftçi B., TÜMER L., OKUR İ., Topçu B., Hasanoğlu A.
SSIEM 2019, 3-6th September, 2019, Rotterdam-The Netherlands, 3 - 06 Eylül 2019

XXXIV. **Beneficial Effects of Modified Atkins Diet in Glycogen Storage Disease Type IIIa**
OLGAÇ M. A. B., İNCİ A., OKUR İ., Kasapkara Ç. S., BİBEROĞLU G., OĞUZ A. D., Aktaş E., EZGÜ F. S., TÜMER L.
SSIEM 2019, 3-6th September, 2019, Rotterdam-The Netherlands, 3 - 06 Eylül 2019

XXXV. **Natural History in Fabry Disease: Classic vs Late-onset phenotype-Paediatrics**
EZGÜ F. S.
6th European Fabry Summer School27th – 29th June 2019, Paris-France, 27 - 29 Haziran 2019

XXXVI. **Metabolic and rare diseases: new effective treatments**
EZGÜ F. S.
Europaediatrics 2019, 13-15th June, 2019, Dublin-Ireland, 13 - 15 Haziran 2019

XXXVII. **Population Genetics and Inborn errors of Metabolism**
EZGÜ F. S.
Zone 4 Meeting, 10-12th June 2019, İstanbul-Turkey, 10 - 12 Haziran 2019

XXXVIII. **Hafif Renal Varyant ile Karakterize Pİerson Sendromu: Olgu Sunumu**
BÜYÜKKARAGÖZ B., BAKKALOĞLU EZGÜ S. A., ÖZMEN M. C., EZGÜ F. S.
10. Uluslararası Katılımlı Çocuk Nefroloji Kongresi, Bodrum, Türkiye, 1 - 04 Mayıs 2019

XXXIX. **Fabry Hastalığı**
EZGÜ F. S.
2. Eskişehir Romatoloji Günleri, 3-5 Mayıs 2019, Eskişehir, Türkiye, 3 - 05 Mayıs 2019

XL. **Screening of Twelve Lysosomal Storage Diseases with LC-MS/MS in Gazi University Hospital: The First Results of Validation.**
BİBEROĞLU G., İNCİ A., DERİN B., OKUR İ., EZGÜ F. S., TÜMER L.
INTERNATIONAL INBORN ERRORS OFMETABOLISM AND NUTRITION CONGRESS10 - 14 April 2019 Istanbul-

Turkey, 10 - 14 Nisan 2019

XLI. **Cornelia de Lange Syndrome and Glycogen Storage Disease Together in a Patient**
KILIÇ A., EMECAN ŞANLI M., ÖZSAYDI E., İNCİ A., OKUR İ., TÜMER L., EZGÜ F. S.
International Inborn Errors Of Metabolism And Nutrition Congress, İstanbul, Türkiye, 10 - 14 Nisan 2019

XLII. **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.
International Inborn Errors of Metabolism and Nutrition Congress, 10 - 14 Nisan 2019

XLIII. **Physiopathology and Diagnostic Methods in CDG**
EZGÜ F. S.
International Inborn Errors Of Metabolism And Nutrition Congress 10 - 14 April 2019, Istanbul-Turkey, 10 - 14 Nisan 2019

XLIV. **Familial Hyperphosphatemic Tumoral Calcinosis in an Unusual Site**
Emecan Şanlı M., Özsaydı E., kılıç m., İNCİ A., OKUR İ., EZGÜ F. S., TÜMER L.
International Inborn Errors Of Metabolism And Nutrition Congress 10 - 14 April 2019, Istanbul-Turkey, 10 - 14 Nisan 2019

XLV. **Hyperinsulinemic Hypoglycemia: Think of GLUD1 Gene Mutation Leading To Hyperinsulinism/Hyperammonemia (HI/HA) Syndrome**
Emecan Şanlı M., kılıç m., Özsaydı E., İNCİ A., OKUR İ., TÜMER L., EZGÜ F. S.
International Inborn Errors Of Metabolism And Nutrition Congress 10 - 14 April 2019, Istanbul-Turkey, 10 - 14 Nisan 2019

XLVI. **Marinesco-Sjögren Syndrome: Case Report**
Kasapkara Ç. S., OLGAÇ M. A. B., GENÇ SEL Ç., DERİNKUYU B. E., kılıç m., EZGÜ F. S.
International Inborn Errors Of Metabolism And Nutrition Congress 10 - 14 April 2019, Istanbul-Turkey, 10 - 14 Nisan 2019

XLVII. **Adenylosuccinate Lyase Deficiency in A Turkish Siblings**
kılıç m., YILDIZ Y., OLGAÇ M. A. B., Kasapkara Ç. S., EZGÜ F. S.
International Inborn Errors Of Metabolism And Nutrition Congress 10 - 14 April 2019, Istanbul-Turkey, 10 - 14 Nisan 2019

XLVIII. **Novel Mutation in FBP1 Gene Presenting with Recurrent Episodes of Vomiting in A Child**
Emecan Şanlı M., kılıç m., Özsaydı E., İNCİ A., OKUR İ., EZGÜ F. S., TÜMER L.
International Inborn Errors Of Metabolism And Nutrition Congress 10 - 14 April 2019, Istanbul-Turkey, 10 - 14 Nisan 2019

XLIX. **Novel Mutation in Two Siblings with Normouricemic Lesch Nyhan Syndrome**
Emecan Şanlı M., Özsaydı E., kılıç m., İNCİ A., OKUR İ., EZGÜ F. S., TÜMER L.
International Inborn Errors Of Metabolism And Nutrition Congress 10 - 14 April 2019, Istanbul-Turkey, 10 - 14 Nisan 2019

L. **A Very Rare Disease: Hyperornithinemia-Hyperammonemia-Homocitrullinuria (Hhh) Syndrome**
Özsaydı E., Emecan Şanlı M., kılıç m., İNCİ A., OKUR İ., TÜMER L., EZGÜ F. S.
International Inborn Errors Of Metabolism And Nutrition Congress 10 - 14 April 2019, Istanbul-Turkey, 10 - 14 Nisan 2019

LI. **Hyperammonemia Secondary to Mitochondrial HMG-Coa Synthase Deficiency**
kılıç m., YILDIZ Y., OLGAÇ M. A. B., Kasapkara Ç. S., EZGÜ F. S.
International Inborn Errors Of Metabolism And Nutrition Congress 10 - 14 April 2019, Istanbul-Turkey, 10 - 14 Nisan 2019

LII. **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.
International Inborn Errors Of Metabolism And Nutrition Congress 10 - 14 April 2019, Istanbul-Turkey, 10 - 14 Nisan 2019

LIII. **Pyruvate Carboxylase Ceficiency in A Child with an Early Diagnosis of KetolysisDefect**
OLGAÇ M. A. B., Kasapkara Ç. S., Öztoprak Ü., Yüksel D., Akçaboy M., EZGÜ F. S.

International Inborn Errors Of Metabolism And Nutrıtıon Congress 10 - 14 April 2019, Istanbul-Turkey, 10 - 14 Nisan 2019

LIV. **Hyperinsulinemic Hypoglycemia: Think of GLUD1 dgene mutation leading to Hyperinsulinemic hyperammonemia (HI/HA syndrome)**
EMECAN ŞANLI M., KILIÇ A., AKTAŞOĞLU E., İNCİ A., OKUR İ., TÜMER L., EZGÜ F. S.
INTERNATIONAL INBORN ERRORS OFMETABOLISM AND NUTRITION CONGRESS 10 - 14 April 2019 Istanbul-Turkey, Türkiye, 10 - 14 Nisan 2019

LV. **Growth Hormone Treatment: Reverses Catabolic Process in Inborn Errors of Metabolism**
İNCİ A., OKUR İ., AKKUZU E., DÖĞER E., BİBEROĞLU G., KALKAN G., TÜMER L., EZGÜ F. S.
International Inborn Errors Of Metabolism And Nutrıtıon Congress 10 - 14 April 2019, Istanbul-Turkey, 10 - 14 Nisan 2019

LVI. **Could Targeted Next Generation Sequencing Be A First Line Diagnostic Method for Lysosomal storage Diseases**
İNCİ A., OKUR İ., AKKUZU E., DÖĞER E., BİBEROĞLU G., KALKAN G., TÜMER L., EZGÜ F. S.
INTERNATIONAL INBORN ERRORS OFMETABOLISM AND NUTRITION CONGRESS10 - 14 April 2019 Istanbul-Turkey, 10 - 14 Nisan 2019

LVII. **Screening Approaches and Laboratory Diagnosis in Gaucher Disease**
EZGÜ F. S.
Gaucher Disease Workshop, 22-23rd March 2019, Riyadh, Saudi Arabia, 22 - 23 Mart 2019

LVIII. **Case Presentations and Discussion**
EZGÜ F. S.
Gaucher Disease Workshop, 22-23rd March 2019, Riyadh, Saudi Arabia, 22 - 23 Mart 2019

LIX. **Insights into Lysosomal Storage Diseases**
EZGÜ F. S.
Gaucher Disease Workshop, 22-23rd March 2019, Riyadh, Saudi Arabia, 22 - 23 Mart 2019

LX. **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 Mart 2019

LXI. **The Practical Aspects of Diagnosis Protocols Implementation**
EZGÜ F. S.
Rare Diseases Annual Forum, 15-16th March, 2019, İstanbul-Turkey, 15 Mart 2019

LXII. **Safety and tolerability of SOBI003 in pediatric MPS IIIA patients key study design features of the ongoing first-in-human study**
Broijersen A., Dalen P., Ezgu F. S., Huledal G., Lindqvist D., Wiken M., Harmatz P.
15th Annual Research Meeting of the WORLDSymposium(TM), Florida, Amerika Birleşik Devletleri, 4 - 07 Şubat 2019, cilt.126

LXIII. **ICV-administered tralesinidase 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, Amerika Birleşik Devletleri, 4 - 07 Şubat 2019, cilt.126

LXIV. **Natural history data for young subjects with Sanfilippo syndrome type B (MPS IIIB)**
Villarreal M. S., OKUR İ., Cleary M., Lopez M. J. d. C., Harmatz P., Lee J., Lin S., Couce M. L., Muschol N., Peters H., et al.
15th Annual Research Meeting of the WORLDSymposium(TM), Florida, Amerika Birleşik Devletleri, 4 - 07 Şubat 2019, cilt.126

LXV. **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.
15th Annual Research Meeting of the WORLDSymposium(TM), Florida, Amerika Birleşik Devletleri, 4 - 07 Şubat 2019, cilt.126

LXVI. **Pompe hastalığının tedavisinde güncel gelişmeler**
EZGÜ F. S.
Korkut Yaltkaya XIII. Klinik Nörofizyoloji Sempozyumu, Antalya, Türkiye, 21 - 23 Aralık 2018

LXVII. **Fabry hastalığının tedavisi ve seçenekler**
EZGÜ F. S.
Korkut Yaltkaya XIII. Klinik Nörofizyoloji Sempozyumu, Antalya, Türkiye, 21 - 23 Aralık 2018

LXVIII. **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 Aralık 2018

LXIX. **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 MeetingLysosomal Storage Disorders, Beirut, 2018, 14 Aralık 2018

LXX. **Gaucher Disease: from Diagnosis to Treatment**
EZGÜ F. S.
1st Levant Regional Lysosomal Storage Disease Expert Meeting, Beirut, 14 Aralık 2018

LXXI. **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 MeetingLysosomal Storage Disorders, Beirut, 2018, 14 Aralık 2018

LXXII. **Forum conclusion and Take Home Messages**
EZGÜ F. S.
1st Levant Regional Lysosomal Storage Disease Expert Meeting, 14th December 2018, Beirut, 14 Aralık 2018

LXXIII. **LSDs Landscape**
EZGÜ F. S.
1st Levant Regional Lysosomal Storage Disease Expert Meeting, Beirut, 14 Aralık 2018

LXXIV. **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 Aralık 2018

LXXV. **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 Aralık 2018

LXXVI. **RAR2mutation in two siblingswith microcephaly,seizures and liver involvement**
EMİNOĞLU F. T., s s., gök t., EZGÜ F. S., İNCİ A., TÜMER L.
15 th MEMG, Beyrut, Lübnan, 29 Kasım - 02 Aralık 2018

LXXVII. **Respiratory system involvement of 41 Mucopolysaccaridosis patients with the evaluation of KL-6, SPA and SPD levels**
İNCİ A., OKUR İ., Yılmaz Demirtaş C., BİBEROĞLU G., ASLAN A. T., EZGÜ F. S., TÜMER L.
15 th MEMG, Beyrut, 29 Kasım - 02 Aralık 2018

LXXVIII. **Closing Day 1**
EZGÜ F. S.
4th Gaucher Disease Regional Forum, İstanbul, 2 - 03 Kasım 2018

LXXIX. **Interview with Gaucher Experts**
EZGÜ F. S.
4th Gaucher Disease Regional Forum, 2 - 03 Kasım 2018

LXXX. **Wrap Up**
EZGÜ F. S.
4th Gaucher Disease Regional Forum, İstanbul, 2 - 03 Kasım 2018

LXXXI. **Advances in Gaucher Disease Management: Available Choices and Direction of Future Research**

EZGÜ F. S.
4th Gaucher Disease Regional Forum, İstanbul, 2 - 03 Kasım 2018

LXXXII. **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 Kasım 2018

LXXXIII. **UNIQUE CLINICAL AND MOLECULAR FINDINGS IN LARGE COHORT OF PATIENTS WITH GAUCHER DISEASE FROM TURKEY**
Akay Tayfun G., OKUR İ., BİBEROĞLU G., TÜMER L., İNCİ A., Küçükcongar A., Hasanoğlu A., EZGÜ F. S.
Gaucher Symphosium, İstanbul, Türkiye, 21 - 22 Ekim 2018

LXXXIV. **Diagnosis and Testing**
EZGÜ F. S.
Gaucher Disease symposium 2018, İstanbul, 21 - 22 Ekim 2018

LXXXV. **FMF'e genetik yaklaşım**
EZGÜ F. S.
VI. Çocuk genetik hastalıkları sempozyumu, Gaziantep, 2018, Türkiye, 19 Ekim 2018

LXXXVI. **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 Ekim 2018

LXXXVII. **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.
51st ESPN Congress, 2018, ANTALYA, 3 - 06 Ekim 2018

LXXXVIII. **Multidisipliner bir hastalık: Deneyimler ışığında Fabry**
EZGÜ F. S.
35. Ulusal nefroloji, hipertansiyon, diyaliz ve transplantasyon kongresi, 2018, Antalya, Türkiye, 3 - 07 Ekim 2018

LXXXIX. **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, Amerika Birleşik Devletleri, 15 - 18 Ekim 2018, cilt.84

XC. **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, Amerika Birleşik Devletleri, 15 - 18 Ekim 2018, cilt.84

XCI. **Çocuklarda spontan pnömotoraks nedeni olarak marfan sendromu**
RAMASLI GÜRSOY T., ŞİŞMANLAR EYÜBOĞLU T., ASLAN A. T., EZGÜ F. S.
Çocuk Göğüs Hastalıkları 3. Kongresi, Türkiye, 26 - 28 Eylül 2018

XCII. **Glycogen storage disease type 9: Insidious onset,mild form**
TÜMER L., İNCİ A., OKUR İ., BİBEROĞLU G., EZGÜ F. S.
SSIEM, 4 - 07 Eylül 2018

XCIII. **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 Eylül 2018

XCIV. **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 Eylül 2018

XCV. **Respiratory system involvement of mucopolysaccaridosis patients with the evaluation of KL-6, SPA and SPD levels**
İNCİ A., OKUR İ., YILMAZ-DEMİRTAŞ C., BİBEROĞLU G., aslan A. T., EZGÜ F. S., TÜMER L.
SSIEM, 4 - 07 Eylül 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 Eylül 2018

XCVII. **Laboratory Diagnosis of MPS Disorders**
EZGÜ F. S.
MPS Masterclass 2018, Belgrade, 8 - 10 Temmuz 2018

XCVIII. **Next generation sequencing in the diagnosis of metabolic diseases**
EZGÜ F. S.
Organelle disease revisited, 2018, Antwerp, 18 Mayıs 2018

XCIX. **Erken Başlangıçlı Lizozomal Asit Lipaz Eksikliğinde Enzim Replasman Tedavisi Sonuçları**
EZGÜ F. S.
VI. Uluslararası katılımlı Lizozomal Hastalıklar Kongresi, Türkiye, 11 - 15 Nisan 2018

C. **Natural history data for young subjects with Sanfilippo Syndrome Type B (MPS IIIB)**
OKUR İ., Cleary M., de Castro Lopez M. J., Harmatz P., Lees J., Lin S., Luz Couce M., Muschol N., Peters H., Solano Villarrea M., et al.
40th Annual Meeting of the Society-for-Inherited-Metabolic-Disorders (SIMD), California, Amerika Birleşik Devletleri, 11 - 14 Mart 2018, cilt.123, ss.255-256

CI. **RENAL INVOLMENT IN FABRY DİSEASE**
İNCİ A., BİBEROĞLU G., PAŞAOĞLU Ö. T., TÜMER L., PAŞAOĞLU H., EZGÜ F. S.
14 th middle east metabolic group (MEMG) meeting Athens GREECE, Atina, Yunanistan, 9 - 11 Şubat 2018

CII. **Natural history data for young subjects with Sanfilippo syndrome type B (MPS IIIB)**
Okur İ., Cleary M., de Castro Lopez M. J., Harmatz P., Lee J., Lin S., Luz Couce M., Muschol N., Peters H., Solano Villarreal M., et al.
We're Organizing Research for Lysosomal Diseases (WORLD) Symposium, California, Amerika Birleşik Devletleri, 5 - 09 Şubat 2018, cilt.123

CIII. **Düşündüğüm metabolik hastalığa nasıl tanı koyabilirim?**
EZGÜ F. S.
IV.Hassas Dokunuş Toplantısı, Ankara, Türkiye, 5 - 06 Ocak 2018

CIV. **Atipik hemolitik üremik sendromda kompleman faktör H mutasyonu: Vaka sunumu**
ÇELEBİ TAYFUR A., ÇALTIK YILMAZ A., KARAOKUR E., İNAN Y., BÜYÜKKARAGÖZ B., KOÇAK M., EZGÜ F. S.
Çocuk Nefroloji Derneği 4. Olgu Panayırı, İzmir, Türkiye, 3 - 04 Kasım 2017

CV. **Nadir Bir Akut Böbrek Yetmezliği Nedeni: Primer Hiperokzalüri**
SÜRMELİ DÖVEN S., DELİBAŞ A., ARSLANKÖYLÜ A. E., EZGÜ F. S.
4. ÇOCUK NEFROLOJİ OLGU PANAYIRI, İZMİR, Türkiye, 3 - 04 Kasım 2017, ss.41

CVI. **Nefronofitizis ve İnkontinensiya Pigmenti Birlikteliği**
TÜRSEN Ü., SÜRMELİ DÖVEN S., DELİBAŞ A., EZGÜ F. S.
çocuk nefroloji derneği 4. olgu panayırı, İzmir, Türkiye, 3 - 04 Aralık 2017

CVII. **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 Eylül 2017

CVIII. **Renal Involvement in Fabry Disease**
İNCİ A., BİBEROĞLU G., OKUR İ., PAŞAOĞLU Ö. T., TÜMER L., PAŞAOĞLU H., EZGÜ F. S.
ICIEM, 5 - 08 Eylül 2017

CIX. **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 Eylül 2017

CX. **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 Eylül 2017

CXI. **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 Eylül 2017

CXII. **Preliminary Results of Our Laboratoryfor Bile Acid Metabolism Disorders**
BİBEROĞLU G., DERİN B., İNCİ A., OKUR İ., EZGÜ F. S., TÜMER L.
ICIEM, 5 - 08 Eylül 2017

CXIII. **Carnitine Acyl Carnitine TranslocaseDeficiency With Severe Hyperammonemiaand Hypoglycemia**
İNCİ A., OKUR İ., OLGAÇ M. A. B., AKKUZU E., BİBEROĞLU G., EZGÜ F. S., TÜMER L.
ICIEM, 5 - 08 Eylül 2017

CXIV. **Population Medical Genetics and IEM**
EZGÜ F. S.
13th International Congress of Inborn Errors of Metabolism, 5 - 08 Eylül 2017

CXV. **İnvestigation of LDLR Gene Mutations in Turkish Patients With Familial Hypercholesterolemia**
OKUR İ., İNCİ A., OLGAÇ M. A. B., ÇİFTÇİ B., TOPÇU B., TÜMER L., EZGÜ F. S.
13th International Congress of Inborn Errors of Metabolism - ICIEM 2017, 5 - 08 Eylül 2017, cilt.5

CXVI. **Neuronopathic Gaucher Disease**
EZGÜ F. S.
Gaucher Disease Symposium 2017, 14 Haziran - 15 Mayıs 2017

CXVII. **Mutation analysis of cystic fibrosis patients in the middle region of Turkey: Three centers results**
ASLAN A. T., ŞİŞMANLAR EYÜBOĞLU T., PEKCAN S., KÖSE M., EZGÜ F. S.
40th European Cystic Fibrosis Conference, 7 - 10 Haziran 2017

CXVIII. **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, İspanya, 22 - 25 Haziran 2017, cilt.102, ss.829-830

CXIX. **Laboratory Diagnosis of Mucopolysaccharidosis (MPS) Disorders**
EZGÜ F. S.
MPS Masterclass 2017, 14 - 16 Mayıs 2017

CXX. **Ciddi hiperammonemi ve hipoglisemi ile giden karnitin-açil translokaz olgusu**
İNCİ A., OLGAÇ KILIÇKAYA M. A. B., OKUR İ., AKKUZU E., BİBEROĞLU G., EZGÜ F. S., TÜMER L.
14. Ulusal Metabolik Hastalıklar ve Beslenme Kongresi, Muğla, Türkiye, 26 - 30 Nisan 2017

CXXI. **MUTATION ANALYSIS OF CYSTIC FIBROSIS PATIENTS: THREECENTERS RESULTS IN THE MIDDLE REGION OF TURKEY**
ŞİŞMANLAR EYÜBOĞLU T., PEKCAN S., ASLAN A. T., KÖSE M., EZGÜ F. S., ERDOĞAN M.
Erciyes Pediatrics Academy Winter Congress, Kayseri, Türkiye, 9 - 11 Mart 2017, cilt.39, ss.4

CXXII. **Kistik fibrozis hastalarının mutasyon analizleri: İç Anadolu bölgesinde üç merkezin sonuçları**
ŞİŞMANLAR EYÜBOĞLU T., PEKCAN S., ASLAN A. T., KÖSE M., EZGÜ F. S., ERDOĞAN M.
3. Erciyes Pediatri Akademisi Kış Kongresi, Kayseri, Türkiye, 9 - 11 Mart 2017

CXXIII. **Ailevi Hiperkolesterolemi Olan Türk Hastalarda LDLR Gen Mutasyonlarinin Araştırılması**
OKUR İ., EZGÜ F. S., İNCİ A., OLGAÇ M. A. B., TÜMER L.
2. Ege Endokrin Hastalıklar ve Genetik Sempozyumu, Türkiye, 23 - 25 Şubat 2017

CXXIV. **In vitro translational readthrough by gentamicin and geneticin improves GLA activity in Fabry disease**
Dündar H., Biberoğlu G., Okur İ., Tümer L., Ezgü F. S.
13th Annual Research Meeting on We're Organizing Research for Lysosomal Diseases (WORLD), California, Amerika Birleşik Devletleri, 13 - 17 Şubat 2017, cilt.120

CXXV. **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 Ekim 2016

CXXVI. **Evaluation of chitotriosidase and high sensitivity c reactive protein levels in mucopolysaccaridosis patients**
İNCİ A., DERİN B., YILMAZ C., udgu b., KARAOĞLU A., OKUR İ., EZGÜ F. S., BİBEROĞLU G., TÜMER L.
MEMG, 28 - 30 Ekim 2016

CXXVII. **Could propionylcarnitine and free carnitinebe used as antioxidative markers in mucopolysaccaridosis**
İNCİ A., BİBEROĞLU G., DERİN B., KARAOĞLU A., OKUR İ., EZGÜ F. S., TÜMER L.
MEMG, 28 - 30 Ekim 2016

CXXVIII. **Do cytokine levels play a role in the pathogenesis of mucopolysaccharidosis patients**
İNCİ A., TÜMER L., YILMAZ-DEMİRTAŞ C., KARAOĞLU A., OKUR İ., OLGAÇ M. A. B., EZGÜ F. S., BİBEROĞLU G.
13th Middle East Metabolic Group Meeting/Amman -Jordan, 28 - 30 Ekim 2016

CXXIX. **Evaluation of chitotriosidase and high sensitive c reactive protein levels in mucopolysaccarıdosıs**
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 Ekim 2016

CXXX. **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 Ekim 2016

CXXXI. **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 Ekim 2016

CXXXII. **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 Ekim 2016

CXXXIII. **Recent Advances in the Diagnosis and the Management of Inherited Metabolic Diseases**
EZGÜ F. S.
The 16th International Conference of Jordan Pediatric Society In collaboration with International Pediatric Association Union of Arab Pediatric Societies, 13 Ekim 2016

CXXXIV. **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 Eylül 2016

CXXXV. **Do cytokine levels play a role in pathogenesis of mucopolysaccaridosis 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 Eylül 2016

CXXXVI. **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 Eylül 2016

CXXXVII. **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 Eylül 2016

CXXXVIII. **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

Eylül 2016

CXXXIX. **Evaluation of chitotriosidase and high sensitivity c reactive protein levels in mucopolysaccaridosis**
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 Eylül 2016

CXL. **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 Eylül 2016

CXLI. **Identification of a novel mutation in Turkish infant with early onset monocarboxylatetransporter1 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 Eylül 2016, cilt.39, ss.35-284

CXLII. **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 Eylül 2016

CXLIII. **Fabry Hastalarında Subklinik Sol Ventrikül Disfonksiyonunun Speckle Tracking Ekokardiyografi ile Değerlendirilmesi**
GÖKALP G., OKUR İ., ÜNLÜ S., İNCİ A., EZGÜ F. S., ŞAHİNARSLAN A., TÜMER L.
V. Uluslararası Katılımlı Lizozomal Hastalıklar Kongresi, Türkiye, 14 - 17 Nisan 2016

CXLIV. **Kistik fibrozisin ilk bulgusu 'agır anemi': Altta yatan ne?**
ŞİŞMANLAR EYÜBOĞLU T., ASLAN A. T., KÖSE M., PEKCAN S., EZGÜ F. S., BUDAKOĞLU I. İ., YENİCESU İ.
Türk Toraks Dernegi 19. Yıllık Kongresi, Türkiye, 6 - 10 Nisan 2016

CXLV. **Fibulanın situs inversusu**
ATALAR H., TURANLI S., ATİK O. Ş., EZGÜ F. S.
1. Ulusal Çocuk Ortopedi Kongresi, Türkiye, 11 - 13 Mart 2016

CXLVI. **At risk screening for Fabry Disease**
EZGÜ F. S.
2015 LSD Masterclass, Abu Dhabi, Birleşik Arap Emirlikleri, 20 - 21 Kasım 2015

CXLVII. **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 Ekim - 01 Kasım 2015

CXLVIII. **Severe anemia in infancy may be the first sign of cystic fibrosis**
ŞİŞMANLAR T., ASLAN A. T., EZGÜ F. S., çiftçi b.
European Cystic Fibrosis Society Congress 2015, 10 - 13 Haziran 2015, cilt.14, ss.142

CXLIX. **Hepatopulmonary syndrome may mask cystic fibrosis**
ASLAN A. T., ŞİŞMANLAR T., SARI S., DALGIÇ B., OĞUZ A. D., EZGÜ F. S.
European Cystic Fibrosis Society Congress 2015, 10 - 13 Haziran 2015, cilt.14, ss.143

CL. **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 Eylül 2015

CLI. **Lysinuric protein intolerance An overlooked diagnosis**
TÜMER L., OLGAÇ M. A. B., ÖZGÜL R. K., YENİCESU İ., EZGÜ F. S., BİBEROĞLU G., hasanoğlu a.
SSIEM Annual Symposium, 1 - 04 Eylül 2015

CLII. **Dihydrolipoamide dehydrogenase deficiency diagnosed byusing new generation sequencing technology**
İNCİ A., TÜMER L., OKUR İ., OLGAÇ M. A. B., SARI S., çiftçi b., topçu b., EZGÜ F. S.
SSIEM Annual Symposium, 1 - 04 Eylül 2015

CLIII. **Impact of sebelipase alfa on survival and liver function ininfants with rapidly progressive lysosomal**

acid lipasedeficiency
simon j., sandra r., anthony q., EZGÜ F. S., zaki o., Gargus J., Hughes J., Dominique P., Vara R., Eckert S., et al.
SSIEM Annual Symposium, 1 - 04 Eylül 2015

CLIV. **Patient with Niemann Pick type C presenting with lymphatic in volvement 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, Fransa, 1 - 04 Eylül 2015

CLV. **Mucopolysaccharidosis Type VII at an Early Age A good candidate for investigational enzyme replacement therapy**
Abdulbaki K., EZGÜ F. S., BİBEROĞLU G., OLGAÇ M. A. B., İNCİ A., TÜMER L.
SSIEM, 1 - 04 Eylül 2015

CLVI. **A novel mutation for L 2 hydroxyglutaric aciduria in a 7 year old patient**
OLGAÇ M. A. B., TÜMER L., EZGÜ F. S., BİBEROĞLU G., alev h.
SSIEM, 1 - 04 Eylül 2015

CLVII. **A completely new approach to the diagnosis of inbornerrors development of a 450 gene all metabolic disorders next generation sequencing panel**
EZGÜ F. S., çiftçi b., topçu b., OKUR İ., İNCİ A., OLGAÇ M. A. B., KARAOĞLU A., BİBEROĞLU G., TÜMER L., hasanoğlu a.
SSIEM Annual Symposium, 1 - 04 Eylül 2015

CLVIII. **EFFICACY AND SAFETY OF SEBELIPASE ALFA IN CHILDREN AND ADULTS WITH LYSOSOMAL ACID LIPASE DEFICIENCY: RESULTS OF A PHASE 3 TRIAL**
Rojas-Caro S., Balwani M., Bialer M., Camarena Grande C., Consuelo Sanchez A., EZGÜ F. S., Kostyleva M., Laukaitis C., Malinova V., Neilan E., et al.
83rd Congress of the European-Atherosclerosis-Society (EAS), Glasgow, Birleşik Krallık, 22 - 25 Mart 2015, cilt.241

CLIX. **Sol çenede Lenfatik tutlum ile giden Niemann Pick tip C olgusu**
İNCİ A., OKUR İ., ESENDAĞLI G., OKUR A., OLGAÇ M. A. B., EZGÜ F. S., TÜMER L.
XIII.Ulusal Metabolik Hastalıklar ve Beslenme Kongresi, Adana, Türkiye, 14 - 18 Nisan 2015

CLX. **EFFICACY AND SAFETY OF SEBELIPASE ALFA IN CHILDREN AND ADULTS WITH LYSOSOMAL ACID LIPASE DEFICIENCY: RESULTS OF A PHASE 3 TRIAL**
Rojas-Caro S., Baric I., Camarena Grande C., Coker M., Deegan P., DiRocco M., Ezgu F. S., Feillet F., Malinova V., Mengel E., et al.
50th International Liver Congress of the European-Association-for-the-Study-of-the-Liver, Vienna, Avusturya, 22 - 26 Nisan 2015, cilt.62

CLXI. **Identification of novel mutations and prevalence for Fabry disease (FD) via screening studies using dried blood samples (DBS) among hemodialysis patients in Turkey**
Okura I., BİBEROĞLU G., EZGÜ F. S., Turner L., ERTEN Y., Bicik Z., Akin Y., Ecder T., Hasanoglu A.
11th Annual WORLD Symposium of the Lysosomal-Disease-Network, Florida, Amerika Birleşik Devletleri, 9 - 13 Şubat 2015, cilt.114

CLXII. **The results of enzyme studies in the diagnosis of lysosomal diseases: 8 years experience of Gazi University, Ankara, Turkey**
Hasanoglu A., BİBEROĞLU G., OKUR İ., Turner L., EZGÜ F. S., Udgu B., Olgac A.
11th Annual WORLD Symposium of the Lysosomal-Disease-Network, Florida, Amerika Birleşik Devletleri, 9 - 13 Şubat 2015, cilt.114

CLXIII. **Importance of family screening in Fabry disease: Reaching the bottom of the iceberg**
Ezgu F. S., Koca S., OKUR İ., BİBEROĞLU G., TÜMER L., Bakkaloglu S. A., ERTEN Y., Hasanoglu A.
11th Annual WORLD Symposium of the Lysosomal-Disease-Network, Florida, Amerika Birleşik Devletleri, 9 - 13 Şubat 2015, cilt.114

CLXIV. **PREVALENCE OF FABRY DISEASE AMONG HEMODIALYSIS PATIENTS IN TURKEY**
Okur İ., BİBEROĞLU G., Ezgu F. S., TÜMER L., Hasanoglu A., Bicik Z., Akin Y., Mumcuoglu M., Ecder T.
50th European-Renal-Association - European-Dialysis-and-Transplant-Association Congress, İstanbul, Türkiye, 18 - 21 Mayıs 2013, cilt.28, ss.321

CLXV. **Phase 3 study of migalastat HCl for Fabry disease: Stage 1 results**

Nicholls K., Germain D. P., Feliciani C., Shankar S., Ezgu F. S., Janmohamed S. G., Laing S. M., Schroyer R. O., Bragat A. C., Sitaraman S., et al.
9th Annual World Symposium of the Lysosomal-Disease-Network (LDN), Florida, Amerika Birleşik Devletleri, 12 - 15 Şubat 2013, cilt.108

CLXVI. **Apheresis inducible cytokine pattern change in children with homozygous familial hypercholesterolemia**
KUCUKCONGAR A., YENİCESU İ., TÜMER L., KASAPKARA S., EZGÜ F. S., PAŞAOĞLU Ö. T., YILMAZ-DEMİRTAŞ C., ÇELİK B., DİLSİZ G., HASANOGLU A.
14. International Congress of the world Apheresis society /İstanbul, 13 - 15 Eylül 2012

CLXVII. **Fabry Disease Mutations Addressable with Migalastat HCl, an Investigational Chaperone Therapy. Screening Results from FACETS, a Phase 3 Study in Male and Female Patients**
Bichet D., Boudes P., El Din S., Ezgu F. S., Feliciani C., Zampetti A., Lourenco C. M., Nicholls K., Castelli J., Overton C., et al.
8th Anuual Research Meeting of the WORLD Symposium on Lysosomal Disease Networks, California, Amerika Birleşik Devletleri, 7 - 10 Şubat 2012, cilt.105

CLXVIII. **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, İsviçre, 30 Ağustos - 02 Eylül 2011

CLXIX. **The consistency of tutors' and committee members' scores related to small groups**
COŞKUN Ö., BUDAKOĞLU I. İ., HAZNEDAROĞLU Ş., EZGÜ F. S., YETKİN İ., BİDECİ A.
AMEE2011, 27 - 31 Ağustos 2011

CLXX. **TWO NOVEL MUTATIONS IN TWO PATIENTS WITH MEDIUM-CHAIN ACYL-CoA DEHYDROGENASE DEFICIENCY**
Hasanoglu A., Okur İ., Largiader C., Biberoglu G., Tumer L., Eminoglu F. T., EZGÜ F. S.
11th International Conference of Inborn Errors of Metabolism, California, Amerika Birleşik Devletleri, 29 Ağustos - 02 Eylül 2009, cilt.98, ss.52

CLXXI. **Down syndrome: Disassociation from dementia in a 65 year-old man with partial trisomy 21 including SOD1-Telomere but not APP**
Dai L., Doran E., Ezgu F. S., Korbel J. O., Korenberg J. R., Lott I. T., Shaffer L. G., Snyder M., Tirosh-Wagner T., Urban A. E.
37th Annual Meeting of the Child-Neurology-Society, Santa-Clara, Küba, 5 - 08 Kasım 2008, cilt.64

CLXXII. **The Co-existence of Satoyoshi Syndrome and Myoadenylate Deaminase Deficiency**
EZGÜ F. S., TÜMER L., SERDAROĞLU A., HASANOĞLU A., CANSU A., HIRFANOĞLU T., DALGIÇ B.
SSIEM 42st Annual Symposium, Paris, France, 6-9 September, 2005, Paris, Fransa, 6 - 09 Eylül 2005, cilt.28, ss.253

Desteklenen Projeler

Yazol M., Ezgü F. S., Diğer Uluslararası Fon Programları, Osteogenesis Imperfecta'lı Gönüllülerde Setrusumab'ın Etkililiğini ve Güvenliliğini Değerlendiren, Bir Faz 2 Tek Kör, Doz Değerlendirme Fazı ile Bir Faz 3 Çift Kör, Plasebo Kontrollü Faz'dan Oluşan Operasyonel Olarak Kesintisiz, Randomize Faz 2/3 Çalışma, 2023 - 2026
Okur İ., Tümer L., İnci A., Ezgü F. S., Diğer Ülkelerin Sanayi Kuruluşları Tarafından Desteklenmiş Proje, Fenilketonüride PTC923-MD-PKU Faz 3 Açık Etiketli Uzatma Çalışması, 2022 - 2025
Ezgü F. S., Okur İ., İnci A., Tümer L., Arhan E., Soysal Acar A. Ş., Diğer Ülkelerin Sanayi Kuruluşları Tarafından Desteklenmiş Proje, Venglustatın geç başlangıçlı GM2 gangliosidoz (Tay-Sachs hastalığı ve Sandhoff hastalığı) ile ayrı bir kolda juvenil/adolesan geç başlangıçlı GM2 gangliosidozda ve, aynı ve benzer glukozilseramid bazlı sfingolipid yolağı içindeki ultra-nadir hastalıklardaki etkinlik, farmakodinamik, farmakokinetik, güvenlilik ve tolere edilebilirliğini değerlendirmek için çok merkezli, uluslararası, randomize, çift-kör, plasebo kontrollü bir çalışma, EFC15299, 2021 - 2025
Okur İ., Arhan E., İnci A., Tümer L., Ezgü F. S., Diğer Uluslararası Fon Programları, En az 6 Aydır Tedavi Almamış veya Hiç

Tedavi Edilmemiş Fabry Hastalığı Bulunan Erkek ve Kadın Yetişkinlerde Venglustat'ın Nöropatik Ağrı ve Karın Ağrısı üzerindeki Etkisini Değerlendiren Randomize,Çift Kör, Plasebo Kontrollü, 12 Aylık Faz 3 Çalışma-PERIDOT, 2023 - 2024
Öktem R. M., Ezgü F. S., Tümer L., Biberoğlu G., TÜBİTAK Projesi, Kütle Spektrometresi İçin Biyokimyasal Tanı Kitlerinin Geliştirilmesi Hiperfenilalaninemi Ayırıcı Tanısında Kritik Role Sahip Pterin Biyobelirteçlerinin Ölçülmesine Yönelik LC-MS/MS Temelli Tanı Kitinin Geliştirilmesi, 2022 - 2022
Okur İ., Ezgü F. S., Tümer L., İnci A., Diğer Ülkelerin Sanayi Kuruluşları Tarafından Desteklenmiş Proje, K020-218---Standart Bakımda Yeterince Kontrol Altına Alınamayan Bir Üre Döngüsü Bozukluğu Olan Gönüllülerde KB195'in Etkinliğinin ve Güvenliğinin Değerlendirildiği Faz 2, Açık Etiketli Çalışma, 2021 - 2021
Okur İ., Ezgü F. S., Dündar H., Tümer L., TÜBİTAK Projesi, Metilmalonil Koenzim A Mutaz Enziminin Konformasyonel Bozukluğuna Bağli Olarak Gelişen Metilmalonik Asidemi Hastaliğin'xxda Farmakolojik Şaperon Uygulamasi İle Enzim Aktivitesinin Yeniden Kazandırılması, 2017 - 2021
Okur İ., Ezgü F. S., Tümer L., Özger İlhan S., Şıvgın V., Soysal Acar A. Ş., İnci A., Diğer Ülkelerdeki Özel Organizasyonlar Tarafından Desteklenmiş Proje, Pediyatrik MPSIIIA hastalarında SOBİ003'ün güvenliğini, tolerabilitesini, farmakokinetik ve farmakodinamik özelliklerini değerlendiren açık, kontrollü olmayan, paralel, artan çoklu doz, çok merkezli bir çalışma (Faz 1-2) (SOBI003-001) ClinicalTrials.gov Identifier: NCT03423186), 2018 - 2020
Okur İ., Börcek A. Ö., Ezgü F. S., Tümer L., İnci A., Soysal Acar A. Ş., Arhan E., Tutar H., Gündüz B., Diğer Ülkelerdeki Özel Organizasyonlar Tarafından Desteklenmiş Proje, Mukopolisakaridoz Tip IIIB (MPS IIIB, Sanfilipo Sendromu Tip B) hastalarında intraserebroventriküler BMN 250 nin güvenliliği, tolerabilitesi, farmakokinetiği ve etkililiğinin değerlendirildiği faz 1/2 açık etiketli doz artırma çalışması(BMN250-201) https://clinicaltrials.gov/ct2/show/NCT02754076?term=BMN250draw=2rank=2, 2017 - 2020
Okur İ., Ezgü F. S., Tümer L., Soysal Acar A. Ş., Şıvgın V., Kurtipek Ö., Gündüz B., İnci A., Tutar H., Arhan E., Diğer Uluslararası Fon Programları, BMN250-901-A Study of Mucopolysaccharidosis Type IIIB MPS IIIB (Mukopolisakkaridoz tip IIIB'xxye yönelik uluslararası çok merkezli, prospektif gözlemsel çalışma)-(BMN250-901 nolu proje kodu), 2015 - 2020
Okur İ., Ezgü F. S., Tümer L., İnci A., Diğer Ülkelerdeki Özel Organizasyonlar Tarafından Desteklenmiş Proje, HAUSER-OLE---Heterezigot Ailevi Hiperkolesterolemisi (HeAH) veya Homozigot Ailevi Hiperkolesterolemisi (HoFH) Olan 10 ve 17 yaşları arasındaki pediatrik Gönüllülerde LDK-K ' nin azaltılmasında diyete 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., Diğer Uluslararası Fon Programları, 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., Yükseköğretim Kurumları Destekli Proje, MOLEKÜLER YA DA ENZİMATİK ANALİZ İLE MUKOPOLİSAKKRİDOZ TANISI ALMIŞ OLGULARDA BÖBREK VE ÜRİNER SİSTEM TUTULUMUNUN ARAŞTIRILMASI, 2015 - 2016
EZGÜ F. S., Yükseköğretim Kurumları Destekli Proje, BİRDEN FAZLA BİREYİNDE "KARDİYOFASİYOKUTANÖZ SENDROM" FENOTİPİ GÖZLENEN BİR AİLEDE MOLEKÜLER BAĞLANTI ANALİZİ İLE SENDROMA YOL AÇAN GEN/GENLERİN BELİRLENMESİ, 2010 - 2015
EZGÜ F. S., Yükseköğretim Kurumları Destekli Proje, Hiperfenilalaninemili Hastalarda Kan ve Beyin Fenilalanin Düzeylerinin Değerlendirimesi ve Tedavi Etkinliğinin Saptanması, 2011 - 2014
EZGÜ F. S., Yükseköğretim Kurumları Destekli Proje, ALFA GALAKTOSİDAZ A EKSİKLİĞİNDE GENOTİP-FENOTİP İLİŞKİSİ, 2010 - 2011
EZGÜ F. S., Yükseköğretim Kurumları Destekli Proje, Prebubertal Obez Çocuklarda Karotis İntima-Media Kalınlığı ve Kan Okside LDL Düzeyleri, 2004 - 2006

Bilimsel Dergilerdeki Faaliyetler

TURKISH JOURNAL OF PEDIATRICS, Editörler Kurulu Üyesi, 2022 - Devam Ediyor
Turkish Journal Of Medical Sciences, Editörler Kurulu Üyesi, 2018 - Devam Ediyor

Türkiye Klinikleri Pediatri Dergisi, Yayın Kurul Üyesi, 2013 - Devam Ediyor

Bilimsel Kuruluşlardaki Üyelikler / Görevler

Çocuk Genetik Hastalıkları Derneği Yönetim Kurulu, Yönetim Kurulu Üyesi, 2018 - 2023, Türkiye

Bilimsel Hakemlikler

TURKISH JOURNAL OF PEDIATRICS, SCI-E Kapsamındaki Dergi, Kasım 2023
ORPHANET JOURNAL OF RARE DISEASES, SCI-E Kapsamındaki Dergi, Kasım 2023
ORPHANET JOURNAL OF RARE DISEASES, SCI-E Kapsamındaki Dergi, Haziran 2023
UFUK AVRUPA Projesi, UFUK AVRUPA Projesi, Avrupa Birliği, Hollanda, Mart 2023
TURKISH JOURNAL OF PEDIATRICS, SCI-E Kapsamındaki Dergi, Aralık 2022
ORPHANET JOURNAL OF RARE DISEASES, SCI-E Kapsamındaki Dergi, Kasım 2022
TURKISH JOURNAL OF PEDIATRICS, SCI-E Kapsamındaki Dergi, Eylül 2022
TURKISH JOURNAL OF PEDIATRICS, SCI-E Kapsamındaki Dergi, Eylül 2022
ORPHANET JOURNAL OF RARE DISEASES, SCI-E Kapsamındaki Dergi, Haziran 2022
TURKISH JOURNAL OF PEDIATRICS, SCI-E Kapsamındaki Dergi, Nisan 2022
ORPHANET JOURNAL OF RARE DISEASES, SCI-E Kapsamındaki Dergi, Nisan 2022

Bilimsel Danışmanlıklar

Ankara Üniversitesi Nadir Hastalıklar Uygulama ve Araştırma Merkezi, Kurum veya Organizasyonlar İçin Yapılan Danışmanlık, Gazi Üniversitesi, Tıp Fakültesi, Dahili Tıp Bilimleri, Türkiye, 2023 - Devam Ediyor
ACURARE, Kurum veya Organizasyonlar İçin Yapılan Danışmanlık, Gazi Üniversitesi, Tıp Fakültesi, Dahili Tıp Bilimleri, Türkiye, 2021 - Devam Ediyor
TİTCK, Kurum veya Organizasyonlar İçin Yapılan Danışmanlık, Gazi Üniversitesi, Tıp Fakültesi, Dahili Tıp Bilimleri, Türkiye, 2020 - Devam Ediyor

Etkinlik Organizasyonlarındaki Görevler

Ezgü F. S., EMPOWER MPS Meeting, Bilimsel Kongre / Sempozyum Organizasyonu, London, İngiltere, Kasım 2020

Metrikler

Yayın: 406
Atıf (WoS): 1494
Atıf (Scopus): 1595
H-İndeks (WoS): 17
H-İndeks (Scopus): 18

Davetli Konuşmalar

Overview of Current & Future Therapies of MPSII, Konferans, Hong Kong, Aralık 2021
Overview: State of the Field in AADCd , Konferans, PTC-Experiental Learning Program, İngiltere, Aralık 2021
Patient Identification and Referral , Konferans, PTC-Experiental Learning program, İngiltere, Aralık 2021

Niemann-Pick Tip C: Tanı, İzlem ve Tedavide Neler Değişti?, Konferans, VII. Lizozomal Hastalıklar Kongresi, Türkiye, Kasım 2021
Approach to MPSII: Diagnosis from organomegaly & short stature, Konferans, takeda-MPS II Tutorial, Hong Kong, Kasım 2021
AADC deficiency: a disorder with multiple signs and symptoms, Konferans, PTC Therapeutics- Treatment Advances in AADC Deficiency: a rare neurotransmitter disorder, Kuveyt, Ekim 2021
Lizozomal depo hastalıklarında yeni tedavi stratejileri, Konferans, 5. ulusal Çocuk Genetik Kongresi, Türkiye, Ekim 2021
What do you do with a floppy infant?„ Konferans, 10th Europaediatrics, Hırvatistan, Ekim 2021
Current & Future Treatment Options in MPS II, Konferans, MPS Academy Digital 2021 "Current & Future Treatment Options in MPS II, Ukrayna, Eylül 2021
Makale Okurken, Seminer, Klinik araştırmalar Derneği - Akademika- İleri Düzey İyi Klinik Uygulamalar (İKU) Eğitimi, Türkiye, Eylül 2021
Nadir Genetik Hastalık Araştırmalarında Özellikler, Seminer, Klinik Araştırmalar Derneği-Akademika-İleri Düzey İyi Klinik Uygulamalar (İKU) Eğitimi, Türkiye, Eylül 2021
Viewpoints: Management/Practices Pattern, Konferans, Takeda-LSD 21, İngiltere, Ağustos 2021
Fabry Gastropathy: prevalence and response to ERT, Konferans, İngiltere, Ağustos 2021
Case examples of differential diagnoses, Konferans, Biomarin-Differential diagnosis of skeletal dysplasias, İngiltere, Temmuz 2021
Diagnosis of skeletal dysplasias, Konferans, Biomarin-Differential diagnosis of skeletal dysplasias, İngiltere, Temmuz 2021
Nadir nörotransmitter hastalıklar; Hareket bozuklukları belirti ve bulguları, Konferans, Türk Pediatri Kurumu-Nörotransmitter Hastalıklar Toplantıları , Türkiye, Haziran 2021
Diagnostic and therapeutic update on Neurotransmitter disorders, Konferans, The 1ST MENA Metabolic & Genetic Conference, Qatar, Haziran 2021
Challenges associated with MPS IVA: transition to adult care , Konferans, İngiltere, Haziran 2021
Challenges in management of patients during Covid-19 pandemic, Konferans, Sanofi-Practical Solutions for Managing Patients with Gaucher Disease During COVID-19 Pandemic, İngiltere, Haziran 2021
Guidance for the management of MPS IVA: key takeaways , Konferans, ISMPS 2021-Improving patient care in MPS IVA through knowledge and empowerment , İngiltere, Haziran 2021
AADC eksikliğinin etyolojisi, genetik ve metabolik temelleri-AADC eksikliğinin prevalans ve temel klinik belirtileri, Konferans, Türk Pediatri Kurumu-Nörotransmitter Hastalıklar Toplantıları , Türkiye, Haziran 2021
Fabry Hastalığı, Konferans, 43.Pediatri Günleri, Türkiye, Mayıs 2021
Bebeklik Dönemi Karaciğer Hastalıklarında Genetik ve Metabolik Testler, Konferans, 13. ulusal Çocuk Gastroenteroloji, Hepatoloji ve Beslenme Kongresi, Türkiye, Mayıs 2021
Neurotransmitter-related disorders – a focus on non-ketotic hyperglycinemia, Konferans, PTC-The road less travelled:Finding a path to diagnosis in rare neurotransmitter disorders, İngiltere, Mayıs 2021
Yeni Normal ile Tecrübeler Güncellemesi, Konferans, Sanofi-Yeni Normal ile Tecrübeler, Türkiye, Mayıs 2021
• Fabry disease in general (briefly with Fabry awareness month perspective) • Challenges in the diagnosis of Fabry disease, Konferans, Fabry Tedavisinde Fark Yaratan Bilgiler |20.Yılında Fabrazyme Global Deneyimi , Türkiye, Nisan 2021
Fabry Hastalığında Spesifik Tedaviler Ve Güncel Yaklaşımlar, Konferans, PARTNERSHIP IN FABRY, Türkiye, Nisan 2021
Kadın Hastaya Tanısal Yaklaşım, Konferans, PARTNERSHIP IN FABRY, Türkiye, Nisan 2021
Overview of Gaucher disease and its current management, Konferans, Takeda-ICMEA Engage VPRIV Speaker Program, İngiltere, Mart 2021
Genetik ve Klinik Bağlantısı, Konferans, Türk Hematoloji Derneği-Gaucher Hastalığı: Sık Belirtilerin Nadir Sebepleri, Türkiye, Mart 2021
Fabry Hastalığında Genetik, Kardiyak Varyantlar ve Temel Pedigri, Konferans, PARTNERSHIP IN FABRY, Türkiye, Mart 2021
Management Options for MPS II: A Practical "Step by Step" Approach, Konferans, Takeda-MPS II Tutorial, Hong Kong, Mart 2021
Gen Tedavisi İle İlgili Son Gelişmeler, Konferans, Prof. dr. Hıfzı Özcan 8. Uluslararası Katılımlı Cerebral Palsy ve Gelişimsel

Bozukluklar Kongresi, Türkiye, Şubat 2021
Long-term management of Gaucher – what we've learned so far, Konferans, 6th Gaucher Disease Regional Forum, İngiltere, Aralık 2020
Pitfalls: MPS II can be missed as it is less common than other MPS diseases, Konferans, The Yin and Yang of Diagnosing MPS II, İngiltere, Aralık 2020
Gaucher disease biomarkers – available options and utility in clinical practice, Konferans, 6th Gaucher Disease Regional Forum, İngiltere, Kasım 2020
Diagnostic challenges: -Laboratory testing, - Classical and non-classical cases, Konferans, EMPOWER MPS Meeting, İngiltere, Kasım 2020
Challenges in Diagnosis of Gaucher Disease, Konferans, Tayland, Eylül 2020
Mukopolisakkaridoz ve Oligosakkaridozların Tedavisinde Yeni Ufuklar, Konferans, ÇUKUROVA, GÜNEYDOĞU ANADOLU ve DOĞU ANADOLU BÖLGELERİ ÇOCUK METABOLİZMA ONLİNE OLGU SUNUMLARI – II, , Türkiye, Eylül 2020
Given Its rarity-Are Gaucher Disease Patients under-Recognized?, Konferans, 56th Annual Meeting of the Thai Society of Hematology, Tayland, Eylül 2020
Çocuklarda Büyüme Geriligi ve Organomegalinin Görünmeyen Nedenleri, Konferans, İstanbul Üniversitesi, Türkiye, Eylül 2020
Overview of treatment options for management of MPS II patients, Konferans, MPS Academy: "Mucopolysaccharidosis type II: past, present and future", Kazakistan, Ağustos 2020
Practical aspects of ERT applications, Konferans, MPS Academy: "Mucopolysaccharidosis type II: past, present and future", , Kazakistan, Ağustos 2020

Akademi Dışı Deneyim

Bakanlık, Tıpta Uzmanlık Kurulu, Çocuk Metabolizma Hastalıkları Tıpta Uzmanlık Kurulu Müfredat Oluşturma Komisyonu
GAZİ ÜNİVERSİTESİ TIP FAKÜLTESİ
Bakanlık, Nadir Hastalıklar Bilimsel Danışma Komisyonu, Nadir Hastalıklar Bilimsel Danışma Komisyonu
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