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Kişisel Bilgiler

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Eğitim Bilgileri

Doktora, Erciyes Üniversitesi, Sağlık Bilimleri Enstitüsü, Biyokimya (Dr), Türkiye 1988 - 1991

Tıpta Uzmanlık, Erciyes Üniversitesi, Tıp Fakültesi, Temel Tıp Bilimleri Bölümü, Türkiye 1984 - 1987

Lisans, Ankara Üniversitesi, Eczacılık Fakültesi, Eczacılık Pr., Türkiye 1978 - 1982

Yabancı Diller

İngilizce, B2 Orta Üstü

Yaptığı Tezler

Doktora, Behçet hastalarında antioksidan mekanizma, Erciyes Üniversitesi, Siyasi Tarih, Biyokimya (Dr), 1991

Araştırma Alanları

Sağlık Bilimleri

Akademik Unvanlar / Görevler

Prof. Dr., Gazi Üniversitesi, Tıp Fakültesi, Dahili Tıp Bilimleri, 1991 - Devam Ediyor

Yönetilen Tezler

BİBEROĞLU G., Pediatrik renal transplant ve kronik böbrek yetmezliği hastalarında aterosklerotik risk faktörleri, Tıpta Uzmanlık, O.BALCI(Öğrenci), 2008

SCI, SSCI ve AHCI İndekslerine Giren Dergilerde Yayınlanan Makaleler

- I. **Evaluation of Lysosphingolipid Analysis for the Diagnosis of Lysosomal Storage Disease**
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- II. **Clinical and Radiological Profile of Nine Patients with Metachromatic Leukodystrophy**
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- III. **Endocrinological and metabolic profile of Gaucher disease patients treated with enzyme replacement therapy**
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- IV. **Is lysosomal acid lipase activity associated with the presence and severity of coronary artery disease? Steht die Aktivität der lysosomalen sauren Lipase in Zusammenhang mit dem Vorliegen und dem Schweregrad einer koronaren Herzkrankheit?**
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- V. **Pterin Profiling in Serum, Dried Blood Spot, and Urine Samples Using LC-MS/MS in Patients with Inherited Hyperphenylalaninemia**
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Molecular Syndromology, cilt.15, sa.3, ss.185-193, 2024 (SCI-Expanded)
- VI. **Diagnostic value of plasma lysosphingolipids levels in a Niemann-Pick disease type C patient with transient neonatal cholestasis**
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- VII. **Combination of the histone deacetylase inhibitor valproic acid and stopcodon readthrough therapy produces improved alpha-galactosidase activity in Fabry patient-derived R227X fibroblasts**
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- VIII. **An ultra-rare cause of severe hypotonia mimicking Pompe disease in an infant: RRM2B related mitochondrial DNA depletion syndrome with a novel mutation**
İNCİ A., OKUR İ., DEMİR E., BİBEROĞLU G., TÜMER L., SERDAROĞLU A., EZGÜ F. S.
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- IX. **Synergistic action of the chemical chaperone 4-phenylbutyrate and the pharmacological chaperone migalastat on restoration of alpha-galactosidase activity of Fabry G258R mutation**
DÜNDAR H., BİBEROĞLU G., İNCİ A., OKUR İ., TÜMER L., EZGÜ F. S.
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- X. **The first case with FBXL4 mutation successfully treated with a parenteral ketogenic diet for lactic acidosis**
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- XI. **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., Kucukcongari A., TÜMER L., Ezgu F. S.
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- XII. **Ultra-Rare Disorder in a Young Girl with Lipodystrophy: Analbuminemia**
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INDIAN JOURNAL OF PEDIATRICS, cilt.88, ss.723-0, 2021 (SCI-Expanded)
- XIII. **A CASE OF GLYCOGEN STORAGE DISEASE TYPE 1a MIMICKING FAMILIAL CHYLOMICRONEMIA**

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- XIV. **Autism: Screening of inborn errors of metabolism and unexpected results**
İnci A., Özaslan A., Okur İ., Biberoglu G., Güney E., Ezgü F. S., Tümer L., İşeri E.
AUTISM RESEARCH, cilt.14, sa.5, ss.887-896, 2021 (SCI-Expanded)
- XV. **The chemical chaperone 4-phenylbutyrate enhances alpha-galactosidase activity subsequent to stop-codon read-through therapy with triamterene in Fabry R227X fibroblasts**
Dündar H., Biberoglu G., İnci A., Işık Gönül İ., Okur İ., Tümer L., Ezgü F. S.
MOLECULAR GENETICS AND METABOLISM, cilt.132, sa.2, 2021 (SCI-Expanded)
- XVI. **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.
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- XVII. **A rare urea cycle disorder in a neonate: N-acetylglutamate synthetase deficiency**
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- XVIII. **Beneficial Effects of Modified Atkins Diet in Glycogen Storage Disease Type IIIa**
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- XIX. **Vitamin D Levels and Bone Mineral Density in Inborn Errors of Metabolism Requiring Specialised Diets**
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- XX. **A possible biomarker of neurocytolysis in infantile gangliosidoses: aspartate transaminase**
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- XXI. **High incidence of co-existing factors significantly modifying the phenotype in patients with Fabry disease.**
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- XXII. **Neonatal multiple sulfatase deficiency with a novel mutation and review of the literature**
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- XXIII. **Vitamin A status and factors associated in healthy school-age children**
VURALLI KARAOĞLAN D., TÜMER L., Hasanoglu A., BİBEROĞLU G., PAŞAOĞLU H.
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- XXIV. **Could GSD type I expand the spectrum of disorders with elevated plasma chitotriosidase activity?**
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- XXV. **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.
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- XXVI. **Asymmetric dimethylarginine (ADMA) and L-arginine levels in children with glycogen storage disease type I**
Kasapkara C. S., TÜMER L., BİBEROĞLU G., Kasapkara A., Hasanoglu A.
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- XXVII. **Assessment of atherosclerosis risk due to the homocysteine-asymmetric dimethylarginine-nitric**

- oxide cascade in children taking antiepileptic drugs**
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SEIZURE-EUROPEAN JOURNAL OF EPILEPSY, cilt.22, sa.2, ss.124-127, 2013 (SCI-Expanded)
- XXVIII. **The effect of zinc on ethanol-induced oxidative stress in rat liver**
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- XXIX. **Analysis of acylcarnitine levels by tandem mass spectrometry in epileptic children receiving valproate and oxcarbazepine**
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- XXX. **The levels of asymmetric dimethylarginine, homocysteine and carotid intima-media thickness in hypercholesterolemic children**
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- XXXI. **Very long-chain acyl CoA dehydrogenase deficiency which was accepted as infanticide**
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- XXXII. **N-carbamylglutamate treatment for acute neonatal hyperammonemia in isovaleric acidemia**
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- XXXIII. **Carnosine supplementation protects rat brain tissue against ethanol-induced oxidative stress**
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- XXXIV. **3-Methylcrotonyl-CoA Carboxylase Deficiency: Phenotypic Variability in a Family**
Eminoglu F. T., Ozcelik A. A., OKUR İ., TÜMER L., BİBEROĞLU G., DEMİR E., Hasanoglu A., Baumgartner M. R.
JOURNAL OF CHILD NEUROLOGY, cilt.24, sa.4, ss.478-481, 2009 (SCI-Expanded)
- XXXV. **Asymmetric dimethylarginine and coronary collateral vessel development**
Kocaman S. A., ŞAHİNARSLAN A., BİBEROĞLU G., Hasanoglu A., Akyel A., Timurkaynak T., Cengel A.
CORONARY ARTERY DISEASE, cilt.19, sa.7, ss.469-474, 2008 (SCI-Expanded)
- XXXVI. **Asymmetrical dimethylarginine level in atrial fibrillation**
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ACTA CARDIOLOGICA, cilt.63, sa.1, ss.33-37, 2008 (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., Biberoglu G., Okur İ., Ezgü F. S., Tümer L.
Biochimica Clinica, cilt.47, sa.3, ss.340-345, 2023 (Scopus)
- II. **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)
- III. **m.3010G>A Değişikliğinin Türk Populasyonunda Siklik Kusma Sendromuna Etkisi**
ERGİN F. B., İNCİ A., OKUR İ., BİBEROĞLU G., TÜMER L., EZGÜ F. S.
Celal Bayar Üniversitesi Sağlık Bilimleri Enstitüsü Dergisi, 2022 (Hakemli Dergi)
- IV. **PROPIONYL CARNITINE AND FREE CARNITINE ARE NEW BIOMARKERS IN THE FOLLOW-UP PERIOD OF MUCOPOLYSACCHARIDOSIS TO SCREEN OXIDATIVE STRESS**
İNCİ A., OLGAC A., GENÇ DERİN B., BİBEROĞLU G., OKUR İ., EZGÜ F. S., TÜMER L.
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- V. **Do cytokines play role in the pathogenesis of mucopolysaccharidosis**
İNCİ A., OLGAC KILIÇKAYA M. A. B., YILMAZ DEMİRTAŞ C., OKUR İ., BİBEROĞLU G., EZGÜ F. S., TÜMER L.
Medicine Science, cilt.10, sa.4, ss.1492-1497, 2021 (Hakemli Dergi)
- VI. **The Relationship between Inflammation and Serum Estrogen, Testosterone, and DHEA-S Levels in Obstructive Coronary Artery Disease**
Karaaslan O. C., ÜNLÜ S., TOPAL S., BİBEROĞLU G., Biberoglu K., ÇENGEL A.
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- VII. **"Double Hit" Homozygous Mutations for Two Different Rare Inborn Errors of Metabolism: A Burden for Countries with High Prevalences of Consanguineous Marriages**
Olgac A., TÜMER L., Ceylaner S., BİBEROĞLU G., Hasanoglu A.
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- VIII. **L carnitine L propionyl carnitine and malondialdehyde levels of pediatric patients with solid tumor**
OKUR A., HASANOĞLU A., OĞUZ A., BİBEROĞLU G., ERTEM U., TÜMER L.
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- IX. **Analysis Of Acylcarnitine Profiles In Children With Idiopathic Epilepsy Using Valproic Acid**
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Hakemli Kongre / Sempozyum Bildiri Kitaplarında Yer Alan Yayınlar

- I. **Pterin Profiling in Serum, Dry Blood Spot and Urine using LC-MS/MS in Patients with Hyperphenylalaninemia**
Öktem R. M., İnci A., Bayrak H., Demir F., Biberoglu G., Mavis M. E., Okur İ., Ezgü F. S., Tümer L.
Annual Symposium 2023, Jerusalem, Yerushalayim, İsrail, 29 Ağustos 2023
- II. **3-O Metil Dopa ölçümü ile AADC eksikliği taraması**
Öktem R. M., Biberoglu G., İnci A., Okur İ., Ezgü F. S., Tümer L.
KBUD Kongre, Lab EXPO 2022, Antalya, Türkiye, 03 Ekim 2022
- III. **Lysosphingolipids in the screening of sphingolipidoses**
Öktem R. M., İnci A., Biberoglu G., Okur İ., Ezgü F. S., Tümer L.
360 LYSOSOME_FEBS Advanced Lecture Course_2022, İzmir, Türkiye, 04 Ekim 2022
- IV. **Retargeting phenylbutyrate, ursodeoxycholic acid, pyrimethamine and betaine for beta-glucocerebrosidase recovery in gaucher disease fibroblasts resulting from homozygous p.L483P mutation**
Kiliç A., BİBEROĞLU G., ÖKTEM R. M., İNCİ A., Aydogdu S., Udgu Isik B., IŞIK GÖNÜL İ., OKUR İ., TÜMER L., EZGÜ F. S.
SSIEM Annual Symposium, Almanya, 30 Ağustos - 02 Eylül 2022, sa.1418955
- V. **İNFAÑTİL TİP POMPE HASTALIĞI ULUSAL KONSENSUS ÇALIŞMASI**
Aktaşoğlu E., İNCİ A., OKUR İ., BİBEROĞLU G., ÖKTEM R. M., EZGÜ F. S., TÜMER L., KILIÇ M., GÜNEŞ S., KAĞNICI M., et al.
VII. Uluslararası Katılımlı Lizozomal Hastalıklar Kongresi, Türkiye, 25 - 27 Kasım 2021
- VI. **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
- VII. **Triamterene-induced suppression of R227X premature termination codon in Fabry disease**
Dündar H., Udgu B., Biberoglu G., İnci 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
- VIII. **Beneficial effects of Modified Atkins Diet in Glycogen Storage Disorder Type IIIa**
OLGAÇ KILIÇKAYA M. A. B., İNCİ A., OKUR İ., KASAPKARA Ç. S., BİBEROĞLU G., OĞUZ A. D., AKTAŞ E., EZGÜ F. S., TÜMER L.
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- IX. Screening of twelve lysosomal storage diseases with LC-MS/MS in Gazi university hospital in Turkey: The first results of validation**
BİBEROĞLU G., İNCİ A., DERİN B., OKUR İ., EZGÜ F. S., TÜMER L.
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- X. Beneficial Effects of Modified Atkins Diet in Glycogen Storage Disease Type IIIa**
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- XI. Screening of Twelve Lysosomal Storage Diseases with LC-MS/MS in Gazi University Hospital: The First Results of Validation.**
BİBEROĞLU G., İNCİ A., DERİN B., OKUR İ., EZGÜ F. S., TÜMER L.
INTERNATIONAL INBORN ERRORS OF METABOLISM AND NUTRITION CONGRESS 10 - 14 April 2019 Istanbul-Turkey, 10 - 14 Nisan 2019
- XII. Growth Hormone Treatment: Reverses Catabolic Process in Inborn Errors of Metabolism**
İNCİ A., OKUR İ., AKKUZU E., DÖĞER E., BİBEROĞLU G., KALKAN G., TÜMER L., EZGÜ F. S.
International Inborn Errors Of Metabolism And Nutrition Congress 10 - 14 April 2019, Istanbul-Turkey, 10 - 14 Nisan 2019
- XIII. Could Targeted Next Generation Sequencing Be A First Line Diagnostic Method for Lysosomal storage Diseases?**
ERGİN F. B., İNCİ A., BİBEROĞLU G., ÇİFTÇİ B., TOPÇU YÜCE A. B., TOKGÖZ D., YAZAR Ö. F., GÖKMENOĞLU H., RAJ Y., OKUR İ., et al.
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- XIV. Could Targeted Next Generation Sequencing Be A First Line Diagnostic Method for Lysosomal storage Diseases**
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INTERNATIONAL INBORN ERRORS OF METABOLISM AND NUTRITION CONGRESS 10 - 14 April 2019 Istanbul-Turkey, 10 - 14 Nisan 2019
- XV. Respiratory system involvement of 41 Mucopolysaccharidosis patients with the evaluation of KL-6, SPA and SPD levels**
İNCİ A., OKUR İ., Yılmaz Demirtaş C., BİBEROĞLU G., ASLAN A. T., EZGÜ F. S., TÜMER L.
15 th MEMG, Beyrut, 29 Kasım - 02 Aralık 2018
- XVI. UNIQUE CLINICAL AND MOLECULAR FINDINGS IN LARGE COHORT OF PATIENTS WITH GAUCHER DISEASE FROM TURKEY**
Akay Tayfun G., OKUR İ., BİBEROĞLU G., TÜMER L., İNCİ A., Küçükcongür A., Hasanoğlu A., EZGÜ F. S.
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- XVII. An early diagnosis cerebretendinous xanthomatosis in a patient at the age of 15 years**
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- XVIII. Respiratory system involvement of mucopolysaccharidosis patients with the evaluation of KL-6, SPA and SPD levels**
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- XXIII. **Determination of succinylacetone in dried blood spot: preliminary results of our laboratory**
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- XXIV. **The Relationship between Inflammation and Serum Estrogen, Testosterone, and Dhea-S Levels in Obstructive Coronary Artery Disease**
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Metrikler

Yayın: 112

Atıf (WoS): 241

Atıf (Scopus): 272

H-İndeks (WoS): 9

H-İndeks (Scopus): 10