

## Prof. Dr. GÜRSEL BİBEROĞLU

### 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

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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**  
Öktem R. M., İnci A., BAYRAK H., DEMİR F., BİBEROĞLU G., Maviş M. E., Gürsu G. G., Yılmaz H., OKUR İ., EZGÜ F. S., et al.  
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- 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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- IX. **Synergistic action of the chemical chaperone 4-phenylbutyrate and the pharmacological chaperone migalastat on restoration of alpha-galactosidase activity of Fabry G258R mutation**  
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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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- 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.  
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- 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.  
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- 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**  
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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**  
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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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### **XXVIII. The effect of zinc on ethanol-induced oxidative stress in rat liver**

Caglar O. M., Bilgihan A., ÖZEL TÜRKÇÜ Ü., BİBEROĞLU G., TAKE KAPLANOĞLU G.

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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**

ÖZEL TÜRKÇÜ Ü., Bilgihan A., BİBEROĞLU G., Caglar O. M.

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### **XXXIV. 3-Methylcrotonyl-CoA Carboxylase Deficiency: Phenotypic Variability in a Family**

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### **XXXV. Asymmetric dimethylarginine and coronary collateral vessel development**

Kocaman S. A., ŞAHİNARSLAN A., BİBEROĞLU G., Hasanoglu A., Akyel A., Timurkaynak T., Cengel A.

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### **XXXVI. Asymmetrical dimethylarginine level in atrial fibrillation**

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## **Diğer Dergilerde Yayınlanan Makaleler**

- I. **Simultaneous succinylacetone-nitisinone measurement in tyrosinemia type I patients and evaluation of the nitisinone therapeutic range**  
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- 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.  
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- IV. **PROPIONYL CARNITINE AND FREE CARNITINE ARE NEW BIOMARKERS IN THE FOLLOW-UP PERIOD OF MUCOPOLYSACCHARIDOSIS TO SCREEN OXIDATIVE STRESS**  
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- V. **Do cytokines play role in the pathogenesis of mucopolysaccharidosis**  
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- VI. **The Relationship between Inflammation and Serum Estrogen, Testosterone, and DHEA-S Levels in Obstructive Coronary Artery Disease**  
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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**  
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- VIII. **L carnitine L propionyl carnitine and malondialdehyde levels of pediatric patients with solid tumor**  
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JOURNAL OF PEDIATRIC SCIENCES, cilt.4, sa.3, 2012 (Hakemli Dergi)
- 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. **Benefficial 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**  
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- X. 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.  
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- XI. Screening of Twelve Lysosomal Storage Diseases with LC-MS/MS in Gazi University Hospital: The First Results of Validation.**  
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- 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.  
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- 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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- 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**  
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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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- XIX. Sphingolipidosis : phenotypic and genotyping spectrum of patients from a single centre experience**  
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- XX. Glycogen storage disease type 9: Insidious onset, mild form**  
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- XXI. The clinical evaluation of Fabry patients with Mainz severity score index and DS3 score**  
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- XXII. **A new case from Turkey with glutathione synthetase deficiency complicated by necrotizing enterocolitis**  
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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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- XXV. **RENAL INVOLMENT IN FABRY DİSEASE**  
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- XXVI. **In Vitro Stopcodon Readthrough ofAlfa-Galactosidase and Alfa-GlucosidasePremature Termination Codons UsingGentamicin, Geneticin, and Ataluren:Therapeutic Potential for Fabry and PompeDiseases**  
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- XXVII. **Renal Involvement in Fabry Disease**  
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- XXVIII. **Short Chain Fatty Acid OxidationDefect in an Adult Patient With RefractorySeizures**  
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- XXIX. **Screening ALPL Gene Differences byNext Generation Sequence Techonology inPatients Having Low ALP Levels**  
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- XXX. **DiagnosticCapability ofNextGenerationDNA Sequencing With A 450 Gene Panel forInborn Errors of Metabolism**  
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- XXXI. **Preliminary Results of Our Laboratoryfor Bile Acid Metabolism Disorders**  
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- XXXII. **Carnitine Acyl Carnitine TranslocaseDeficiency With Severe Hyperammonemiaand Hypoglycemia**  
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- XXXV. **Evaluation of chitotriosidase and high sensitive c reactive protein levels in mucopolysaccharidosis**  
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- XXXVIII. **Could propionylcarnitine and free carnitine be used as antioxidative markers in mucopolysaccharidosis**  
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- XXXIX. **Do cytokine levels play a role in the pathogenesis of mucopolysaccharidosis patients**  
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- XL. **The specificity and sensitivity of next generation semiconductor DNA sequencing in detecting heteroplasmic mitochondrial**  
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- XLI. **Evaluation of gentamycin for stop codon readthrough therapy in Fabry disease**  
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- XLII. **Early initiation of investigational enzyme replacement therapy in a nine month old infant with mucopolysaccharidosis type VII**  
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- XLV. **Identification of a novel mutation in Turkish infant with early onset monocarboxylate transporter 1 MCT1 deficiency as a cause of recurrent ketoacidosis**  
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- XLVIII. **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.  
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- XLIX. Is there any effect of acylcarnitines on proinflammatory process in obese children**  
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- L. 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.  
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- LI. Mucopolysaccharidosis Type VII at an Early Age A good candidate for investigational enzyme replacement therapy**  
Abdubaki K., EZGÜ F. S., BİBEROĞLU G., OLGAÇ M. A. B., İNCİ A., TÜMER L.  
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- LII. A completely new approach to the diagnosis of inborn errors development of a 450 gene all metabolic disorders next generation sequencing panel**  
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- LIII. Lysinuric protein intolerance An overlooked diagnosis**  
TÜMER L., OLGAÇ M. A. B., ÖZGÜL R. K., YENİCESU İ., EZGÜ F. S., BİBEROĞLU G., hasanoğlu a.  
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- LIV. The results of enzyme studies in the diagnosis of lysosomal diseases: 8 years experience of Gazi University, Ankara, Turkey**  
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- LV. Isovaleric acidemia and Niemann Pick disease type C coexistence and new mutation for Niemann Pick disease type C**  
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- LVI. 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.  
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- LVII. 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
- LVIII. 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.  
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## **Desteklenen Projeler**

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## **Metrikler**

Yayın: 112

Atıf (WoS): 241

Atıf (Scopus): 272

H-İndeks (WoS): 9

H-İndeks (Scopus): 10