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Education Information

Undergraduate Minor, Gazi University, Tıp Fakültesi, Dahili Tıp Bilimleri Bölümü, Turkey 2014 - 2017

Academic Titles / Tasks

Lecturer, Gazi University, Tıp Fakültesi, Dahili Tıp Bilimleri Bölümü, 2018 - Continues

Articles Published in Journals That Entered SCI, SSCI and AHCI Indexes

- I. **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.
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- II. **Ultra-Rare Disorder in a Young Girl with Lipodystrophy: Analbuminemia**
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- III. **The first case with FBXL4 mutation successfully treated with a parenteral ketogenic diet for lactic acidosis**
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- VI. **Two patients from Turkey with a novel variant in the GM2A gene and review of the literature**
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- VII. **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., TÜMER L.
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- VIII. **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.
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- IX. **A new NBIA patient from Turkey with homozygous C19ORF12 mutation.**
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- X. **Vitamin D Levels and Bone Mineral Density in Inborn Errors of Metabolism Requiring Specialised Diets**
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- I. **Citrullinemia with an Atypical Presentation: Paroxysmal Hypoventilation Attacks**
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- I. **Yağ Asidi Oksidasyon Bozuklukları**
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in: TEMEL PEDIATRİ, HASANOĞLU ENVER, DÜŞÜNSEL RUHAN, BİDECİ AYSUN, BODUROĞLU KORAY, Editor, GÜNEŞ TIP KİTAPEVLERİ, Ankara, pp.1126-1130, 2020
- II. **Normal Çocuklukta Beslenme ve Beslenme Bozuklukları**
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- III. **Mitokondriyal Hastalıklar**
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- IV. **Vitamin K**
İNCİ A.
in: Yurdakök Pediatri, Murat Yurdakök, Editor, Güneş Tıp Kitapevi, Ankara, pp.1552-1556, 2017

Refereed Congress / Symposium Publications in Proceedings

- I. **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.
SSIEM, 3 - 06 September 2019
- II. **Beneficial Effects of Modified Atkins Diet in Glycogen Storage Disease Type IIIa**
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- III. **Next generation DNA sequencing as an initial diagnostic method for congenital defects of**

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- V. **Cornelia de Lange Syndrome and Glycogen Storage Disease Together in a Patient**
KILIÇ A., EMECAN ŞANLI M., ÖZSAYDI E., İNCİ A., OKUR İ., TÜMER L., EZGÜ F. S.
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- VI. **A Very Rare Disease: Hyperornithinemia-Hyperammonemia-Homocitrullinuria (Hhh) Syndrome**
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- VII. **Novel Mutation in FBP1 Gene Presenting with Recurrent Episodes of Vomiting in A Child**
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- IX. **Hyperinsulinemic Hypoglycemia: Think of GLUD1 Gene Mutation Leading To Hyperinsulinism/Hyperammonemia (HI/HA) Syndrome**
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- X. **Novel Mutation in Two Siblings with Normouricemic Lesch Nyhan Syndrome**
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- XI. **A Novel Rars2 Mutation in Two Siblings with Microcephaly, Seizures and Liver Involvement**
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- XII. **Growth Hormone Treatment: Reverses Catabolic Process in Inborn Errors of Metabolism**
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- XIII. **Could Targeted Next Generation Sequencing Be A First Line Diagnostic Method for Lysosomal storage Diseases**
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- XIV. **RAR2 mutation in two siblings with microcephaly, seizures and liver involvement**
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- XV. **Respiratory system involvement of 41 Mucopolysaccharidosis patients with the evaluation of KL-6, SPA and SPD levels**
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- 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., Hasanođlu A., EZGÜ F. S.
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- XVII. Gaucher Disease and Pregnancy**
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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. The clinical evaluation of Fabry patientswith Mainz severity score index and DS3 score**
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- XX. Determination of succinylacetone in dried blood spot: preliminary results of our laboratory**
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- XXI. Glycogen storage disease type 9: Insidious onset,mild form**
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- XXII. An early diagnosis cerebretendinous xanthomatosis in a patient at the age of 15 years**
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- XXIII. RENAL INVOLMENT IN FABRY DİSEASE**
İNCİ A., BİBEROĞLU G., PAŞAOĞLU Ö. T. , TÜMER L., PAŞAOĞLU H., EZGÜ F. S.
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- XXIV. Carnitine Acyl Carnitine TranslocaseDeficiency With Severe Hyperammonemiaand Hypoglycemia**
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- XXV. Renal Involvement in Fabry Disease**
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- XXVI. Preliminary Results of Our Laboratoryfor Bile Acid Metabolism Disorders**
BİBEROĞLU G., DERİN B., İNCİ A., OKUR İ., EZGÜ F. S. , TÜMER L.
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- XXVII. Short Chain Fatty Acid OxidationDefect in an Adult Patient With RefractorySeizures**
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- XXVIII. Screening ALPL Gene Differences byNext Generation Sequence Techonology inPatients Having Low ALP Levels**
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- XXIX. DiagnosticCapability ofNextGenerationDNA Sequencing With A 450 Gene Panel forInborn Errors of Metabolism**
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- XXXI. 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.
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- XXXII. **Ailevi Hiperkolesterolemi Olan Türk Hastalarda LDLR Gen Mutasyonlarının Araştırılması**
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2. Ege Endokrin Hastalıklar ve Genetik Sempozyumu, Turkey, 23 - 25 February 2017
- XXXIII. **Evaluation of chitotriosidase and high sensitive c reactive protein levels in mucopolysaccharidosis**
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- XXXVI. **Could propionylcarnitine and free carnitine be used as antioxidative markers in mucopolysaccharidosis**
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- XXXVII. **Do cytokine levels play a role in the pathogenesis of mucopolysaccharidosis patients**
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- XXXVIII. **Early initiation of investigational enzyme replacement therapy in a nine month old infant with mucopolysaccharidosis type VII**
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- XL. **Early initiation of investigational enzyme replacement therapy in a 9 month old infant with mucopolysaccharidosis type VII**
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- XLI. **Bone mineral density and vitamin D status in inborn errors of metabolism**
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- XLIII. **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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- XLIV. **Type 1 hypersensitivity reaction and desensitization with Elosulphase alpha**
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- XLV. **BonemineradensityandvitaminDstatusininbornerrorsofmetabolism**
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- XLVII. **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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- XLVIII. **Is there any effect of acylcarnitines on proinflammatory process in obese children**
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- XLIX. **Patient with Niemann Pick type C presenting with lymphatic involvement with Niemann Pick cells in the left jaw**
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- L. **Mucopolysaccharidosis Type VII at an Early Age A good candidate for investigational enzyme replacement therapy**
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- LI. **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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- LII. **Dihydrolipoamide dehydrogenase deficiency diagnosed by using new generation sequencing technology**
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- LIII. **Sol çenede Lenfatik tutulum ile giden Niemann Pick tip C olgusu**
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Citations

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