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Learning Knowledge

Post Doctorate of Medicine
2010 - 2013

Hacettepe University, Tıp Fakültesi (Türkçe), Dahili Tıp Bilimleri Bölümü, Turkey

Expertise In Medicine
2002 - 2008

Ankara University, Pediatrics, Turkey

Undergraduate
1996 - 2002

Ankara University, Turkey

Dissertations

Post Doctorate of Medicine, SİSTATYONİN BETA-SENTAZ EKSİKLİĞİNE BAĞLI HOMOSİSTİNÜRİ HASTALARINDA KLİNİK, BİYOKİMYASAL, MOLEKÜLER BULGULARIN BELGELENMESİ VE GENOTİP-FENOTİP İLİŞKİSİNİN ARAŞTIRILMASI, Hacettepe University, Tıp Fakültesi (Türkçe), Dahili Tıp Bilimleri Bölümü, 2013

Expertise In Medicine, Çocuk nörolojisi pratiğinde baş ağrılarının sıklık, etyoloji, tedavi yaklaşımları ve izlemleri açısından değerlendirilmeleri, Ankara University, Ankara Faculty Of Medicine, Dahili Tıp Bilimleri, 2008

Published journal articles indexed by SCI, SSCI, and AHCI

- Hematopoietic stem cell transplantation in children with mucopolysaccharidosis IVA: single center experience.**
Yalcin K., Uygun V., Ozturk Hismi B., Celen S., Ozturkmen S., Zhumatayev S., Daloglu H., Karasu G., Yesilipek A.
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- Experience with cascade screening: A comprehensive family pedigree analysis of two index patients with Fabry disease**
Kisa P. T., ÖZTÜRK HİŞMİ B., KOCABEY M., Gulten Z. A., Huddam B., Ekinci S., Bozkaya E., Akar H., Pekuz O. K. K., Aydogan A., et al.
American Journal of Medical Genetics, Part A, vol.194, no.7, 2024 (SCI-Expanded)

3. **Clinical, biochemical, and molecular insights into Cerebrotendinous Xanthomatosis: A nationwide study of 100 Turkish individuals**
Zubarioglu T., Kiykim E., Köse E., Eminoğlu F. T., Teke Kısa P., Balcı M. C., Özer I., İnci A., Çilesiz K., Canda E., et al.
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4. **BCKDK deficiency: a treatable neurodevelopmental disease amenable to newborn screening**
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5. **Prevalence of DDC genotypes in patients with aromatic L-amino acid decarboxylase (AADC) deficiency and in silico prediction of structural protein changes**
Himmelreich N., Bertoldi M., Alfadhel M., Alghamdi M. A., Anikster Y., Bao X., Bashiri F. A., Zeev B. B., Bisello G., Ceylan A. C., et al.
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6. **Management of acute metabolic crisis in TANGO2 deficiency: A case report**
Yllmaz-Gümüş E., ELÇİOĞLU H. N., Genç E., Arıclı Ş., Öztürk G., Yapıclı Ö., Akallı F., ÖZTÜRK HİŞMİ B.
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7. **URINARY SYSTEM INVOLVEMENT IN MUCOPOLYSACCHARIDOSIS DISEASE: TWO CASES WITH CHRONIC KIDNEY DISEASE**
Pul S., Cicek N., GÜNAL Ö., Gumus E. Y., ŞEKERCİ Ç. A., DEMİRCİ BODUR E., Guven S., Turkkan O. N., YÜCEL S., ÖZTÜRK HİŞMİ B., et al.
PEDIATRIC NEPHROLOGY, vol.37, no.11, pp.2950, 2022 (SCI-Expanded)
8. **Three-Country Snapshot of Ornithine Transcarbamylase Deficiency**
Seker Yilmaz B., Baruteau J., ARSLAN N., AYDIN H. İ., Barth M., Bozaci A. E., Brassier A., CANDA E., Cano A., Chronopoulou E., et al.
LIFE-BASEL, vol.12, no.11, 2022 (SCI-Expanded)
9. **Improvement in hypertrophic cardiomyopathy after using a high-fat, high-protein and low-carbohydrate diet in a non-adherent child with glycogen storage disease type IIIa**
Akin B. K., ÖZTÜRK HİŞMİ B., Daly A.
MOLECULAR GENETICS AND METABOLISM REPORTS, vol.32, 2022 (SCI-Expanded)
10. **A surprising cause of proteinuria: Questions**
DEMİR B. K., Kanik A., Kose M., ÖZTÜRK HİŞMİ B., Baran M.
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11. **A surprising cause of proteinuria: Answers**
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12. **CHARACTERISTICS OF PATIENTS WITH DISTAL RENAL TUBULAR ACIDOSIS AND ITS ASSOCIATION WITH HYPERAMMONEMIA: A SINGLE-CENTER EXPERIENCE**
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PEDIATRIC NEPHROLOGY, vol.36, no.10, pp.3428, 2021 (SCI-Expanded)
13. **Patients with cerebrotendinous xanthomatosis diagnosed with diverse multisystem involvement.**
Kısa P. T., Yildirim G. K., Hismi B., Dorum S., Kusbeci O. Y., Topak A., Baydan F., Celik F. N. D., Gorukmez O., Gulten Z. A., et al.
Metabolic brain disease, vol.36, pp.1201-1211, 2021 (SCI-Expanded)
14. **Alkaptonuria in Turkey: Clinical and molecular characteristics of 66 patients**
KISA P. T., Gunduz M., Dorum S., Uzun O. U., Cakar N. E., KILIÇ YILDIRIM G., ERDÖL Ş., ÖZTÜRK HİŞMİ B., Tugsal H. Y., Ucar U., et al.
EUROPEAN JOURNAL OF MEDICAL GENETICS, vol.64, no.5, 2021 (SCI-Expanded)
15. **Inflammatory rheumatic diseases in patients with ochronotic arthropathy**
Inel T. Y., Kısa P. T., Balci A., Uslu S., Arslan Z., Hismi B., Ucar U., Arslan N., Onen F., Sari I.
MODERN RHEUMATOLOGY, vol.31, no.5, pp.1031-1037, 2021 (SCI-Expanded)
16. **Neuropsychological attributes of urea cycle disorders: A systematic review of the literature**

Waisbren S. E., Stefanatos A. K., Kok T. M. Y., Ozturk-Hismi B.

Journal of Inherited Metabolic Disease, vol.42, no.6, pp.1176-1191, 2019 (SCI-Expanded)

17. **Clinical and molecular characteristics and time of diagnosis of patients with classical galactosemia in an unscreened population in Turkey**
Teke Kisa P., Kose M., Unal O., Er E., Hismi B., Bulbul F. S., Kose E., Gunduz M., Canda E., Kucukcongar A., et al.
Journal of Pediatric Endocrinology and Metabolism, vol.32, no.7, pp.675-681, 2019 (SCI-Expanded)
18. **Studying the effect of large neutral amino acid supplements on oxidative stress in phenylketonuric patients**
Kumru B., Ozturk Hismi B., Kaplan D. S., Celik H.
Journal of Pediatric Endocrinology and Metabolism, vol.32, no.3, pp.269-274, 2019 (SCI-Expanded)
19. **Effect of Blood Phenylalanine Levels on Oxidative Stress in Classical Phenylketonuric Patients**
Kumru B., Kaplan D. S., Oztürk Hismi B., Celik H.
Cellular and Molecular Neurobiology, vol.38, no.5, pp.1033-1038, 2018 (SCI-Expanded)
20. **An unusual cause of cavitating leukoencephalopathy: ethylmalonic encephalopathy**
Cavusoglu D., Hismi B., Dundar N. O., Oztekin O., Koc A., CAND A E., Arican P., Gencpinar P.
Acta Neurologica Belgica, vol.118, no.2, pp.309-312, 2018 (SCI-Expanded)
21. **Deoxyguanosine kinase deficiency: A report of four patients**
Ünal Ö., Hişmi B., KILIÇ M., Gulsen H. H., COŞKUN T., Sivri S. H., DURSUN A., YÜCE A., TOKATLI A.
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22. **A late diagnosis of alkaptonuria in an elderly patient**
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23. **Detection of biotinidase gene mutations in Turkish patients ascertained by newborn and family screening**
Karaca M., ÖZGÜL R. K., Ünal Ö., Yücel-Yılmaz D., KILIÇ M., Hişmi B., TOKATLI A., COŞKUN T., DURSUN A., SİVRİ H. S.
European Journal of Pediatrics, vol.174, no.8, pp.1077-1084, 2015 (SCI-Expanded)
24. **Two Turkish siblings with MEGDEL syndrome due to novel SERAC1 gene mutation**
Ünal Ö., Köksal Özgül R., Yücel D., YALNIZOĞLU D., TOKATLI A., Serap Sivri H., Hişmi B., COŞKUN T., DURSUN A.
Turkish Journal of Pediatrics, vol.57, no.4, pp.388-393, 2015 (SCI-Expanded)
25. **Sapropterin dihydrochloride treatment in Turkish hyperphenylalaninemic patients under age four**
Ünal Ö., Gökmen-Özel H., COŞKUN T., ÖZGÜL R. K., Yücel D., Hişmi B., TOKATLI A., DURSUN A., SİVRİ H. S.
Turkish Journal of Pediatrics, vol.57, no.3, pp.213-218, 2015 (SCI-Expanded)
26. **Fundus autofluorescence and optical coherence tomography findings in glutathione synthetase deficiency**
Şekeroğlu H. T., Hismi B., KADAYIFÇILAR S., COŞKUN T.
Journal of AAPOS, vol.19, no.1, pp.80-82, 2015 (SCI-Expanded)
27. **High prevalence of cerebral venous sinus thrombosis (CVST) as presentation of cystathionine beta-synthase deficiency in childhood: Molecular and clinical findings of Turkish probands**
Karaca M., Hismi B., ÖZGÜL R. K., Karaca S., Yılmaz D. Y., COŞKUN T., SİVRİ H. S., TOKATLI A., DURSUN A.
Gene, vol.534, no.2, pp.197-203, 2014 (SCI-Expanded)
28. **Pregnancy and Lactation Outcomes in a Turkish Patient with Lysinuric Protein Intolerance**
Unal O., COŞKUN T., ORHAN D., Tokatl A., DURSUN A., Hismi B., ÖZYÜNCÜ Ö., Sivri S. H. K.
JIMD REPORTS, VOL 13, vol.13, pp.33-36, 2014 (SCI-Expanded)
29. **Phenotypic and genotypic spectrum of turkish patients with isovaleric acidemia**
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30. **Recurrence of carbamoyl phosphate synthetase 1 (CPS1) deficiency in Turkish patients: Characterization of a founder mutation by use of recombinant CPS1 from insect cells expression**
Hu L., Diez-Fernandez C., Rüfenacht V., Hismi B., Ünal Ö., Soyucen E., Çoker M., Bayraktar B. T., Gunduz M., Kiykim E., et al.
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31. **A patient with pyruvate carboxylase deficiency and nemaline rods on muscle biopsy**
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32. **Vanishing white matter with hepatomegaly and hypertriglyceridemia attacks**
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Journal of Child Neurology, vol.28, no.11, pp.1509-1512, 2013 (SCI-Expanded)
33. **Molecular and clinical evaluation of Turkish patients with lysinuric protein intolerance**
Güzel-Ozantürk A., ÖZGÜL R. K., Ünal Ö., Hişmi B., Aydın H. i., Sivri S., TOKATLI A., COŞKUN T., Aksöz E., DURSUN A.
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34. **ENZYME REPLACEMENT THERAPY (ERT) RESULTS OF PATIENTS WITH MUCOPOLYSACCHARIDOSIS TYPE II (HUNTER SYNDROME)**
Unal O., Dogru-Ersoz D., Alehan D., SAĞLAM M. Y., Hismi B., Dursun A., Tokatli A., Coskun T., Sivri H. S.
JOURNAL OF INHERITED METABOLIC DISEASE, vol.35, 2012 (SCI-Expanded)
35. **PROPIONIC ACIDEMIA PRESENTING WITH PERSISTENT PULMONARY HYPERTENSION IN TWO NEONATES**
Hismi B., TEKŞAM Ö., Unal O., Takci S., Ertugrul I., SİVRİ H. S., DURSUN A., TOKATLI A., COŞKUN T.
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36. **A PATIENT WITH PYRUVATE CARBOXYLASE DEFICIENCY AND NEMALINE RODS ON MUSCLE BIOPSY**
Unal O., Hismi B., DURSUN A., TOKATLI A., COŞKUN T., Wibrand F., SİVRİ H. S.
JOURNAL OF INHERITED METABOLIC DISEASE, vol.35, 2012 (SCI-Expanded)
37. **SEVERE AZOTEMIA AND HYPERNATREMIC DEHYDRATION IN AN INFANT WITH PHENYLKETONURIA**
Unal O., DÜZOVA A., Hismi B., DURSUN A., TOKATLI A., COŞKUN T., SİVRİ H. S.
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38. **ACUTE INTERMITTENT PORPHYRIA: STILL UNCALLED BY PHYSICIANS**
Hismi B., Tanriover M. D., Unal O., Sener E., TEMUÇİN Ç. M., SİVRİ H. S., DURSUN A., COŞKUN T.
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39. **THE 48-HOUR TETRAHYDOBIOPTERIN (BH4) LOADING TEST AND LONG-TERM OUTCOME OF PATIENTS WITH BH4 TREATMENT IN TURKISH PHENYLKETONURIA (PKU) PATIENTS**
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40. **FEVER OF UNKNOWN ORIGIN IN A YOUNG ADULT WITH END-STAGE RENAL DISEASE, PREMATURE CORONARY ARTERY DISEASE AND POLYNEUROPATHY**
Hismi B., Yasar Y., Unal O., KILIÇ L., Turkmen E., ERDEM Y., DURSUN A., SİVRİ H. S., COŞKUN T.
JOURNAL OF INHERITED METABOLIC DISEASE, vol.35, 2012 (SCI-Expanded)
41. **3-HYDROXY-3-METHYLGLUTARYL-COENZYME A (HMG-COA)-LYASE DEFICIENCY: A DISORDER OF KETOGENESIS AND LEUCINE CATABOLISM**
Sass J. O., Beermann F., Spiekerkoetter U., Hismi B., COŞKUN T., Wegener V. M., Baehr L.
JOURNAL OF INHERITED METABOLIC DISEASE, vol.35, 2012 (SCI-Expanded)
42. **AUDIOLOGICAL OUTCOMES OF MPS II: BEFORE AND AFTER ENZYME REPLACEMENT THERAPY**
Yigit-Duran O., Unal O., GENÇ A., Hismi B., Dursun A., Tokatli A., Coskun T., Sivri H. S.
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43. **MOLYBDENUM COFACTOR (MOCO) DEFICIENCY TYPE B; CLINICAL, BIOCHEMICAL AND NEUROIMAGING FEATURES OF FIVE PATIENTS WITH TWO NOVEL MUTATIONS**
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44. **Detection of other inborn errors of metabolism in hyperphenylalaninemic babies picked up on narrow-spectrum screening programs**
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45. **A Rare Galactosemia Complication: Vitreous Hemorrhage**

- Takci S., KADAYIFÇILAR S., COŞKUN T., YİĞİT Ş., Hismi B.
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46. **Secondary hemophagocytosis in 3 patients with organic acidemia involving propionate metabolism.**
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47. **ACUTE RESPIRATORY DISTRESS SYNDROME IN TWO PATIENTS WITH ORGANIC ACIDEMIA INVOLVING PROPIONATE METABOLISM**
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48. **HEMOPHAGOCYTOSIS IN THREE PATIENTS WITH ORGANIC ACIDEMIA INVOLVING PROPIONATE METABOLISM**
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50. **GALACTOSEMIA IN A TURKISH POPULATION WITH A HIGH PREVALENCE OF Q188R MUTATION**
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51. **A NOVEL MUTATION IN BETA KETOTHIOLASE DEFICIENCY**
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52. **POLYNEUROPATHY AS THE MAIN PRESENTING SYMPTOM IN PDH DEFICIENCY**
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53. **IDENTIFICATION OF MUTATIONS IN THE PCCA AND PCCB GENES CAUSING PROPIONIC ACIDEMIA IN TURKISH PATIENTS**
ÖZGÜL R. K., Yucel D., Hismi B., Karaca M., SİVRİ H. S., COŞKUN T., TOKATLI A., DURSUN A.
JOURNAL OF INHERITED METABOLIC DISEASE, vol.33, 2010 (SCI-Expanded)
54. **A Truncating Mutation in SERPINB6 Is Associated with Autosomal-Recessive Nonsyndromic Sensorineural Hearing Loss**
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55. **Mutations in TMC1 contribute significantly to nonsyndromic autosomal recessive sensorineural hearing loss: A report of five novel mutations**
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56. **Renal replacement therapies in pediatric intensive care patients: Experiences of one center in Turkey**
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60. **Effects of GJB2 genotypes on the audiological phenotype: Variability is present for all genotypes**
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63. **Seronegative spondyloarthropathy associated with Takaya-su's arteritis in a child**
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1. **Molecular Aspects of Distal Kidney Tubular Acidosis in Children, Its Long-Term Outcome, and Relationship with Hyperammonemia**
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2. **Diagnostic Yield of Neuroimaging and Electroencephalography in Children with Recurrent Headaches**
ÖZTÜRK HİŞMİ B., Teber S., Ozkan M., Unal O., Deda G.
Journal of Pediatric Neurology, vol.19, pp.76-82, 2021 (ESCI)
3. **A Mortal Complication in a Case with Mucopolysaccharidosis Type I Following Hematopoietic Stem Cell Transplantation: Pulmonary Haemorrhage**
YAZICI H., CANDA E., Er E., Malbora B., Hismi B., Onay H., AKSOYLAR S., Ucar S. K., Ozkinay F., ÇOKER M.
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Research Areas

Health Sciences

Non Academic Experience

Harvard Medical School, Boston Children's Hospital, Department of Genetics and Genomics

İZMİR TEPECİK EĞİTİM VE ARAŞTIRMA HASTANESİ

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MARDİN KADIN DOĞUM VE ÇOCUK HASTALIKLARI HASTANESİ