



BURCU ÖZTÜRK HİŞMİ DOÇ.DR.

Diger E-posta : burcu.hismi@marmara.edu.tr
Uluslararası Araştırmacı ID'leri
ScholarID: Burcu
ORCID: 0000-0001-7146-0248
Publons / Web Of Science ResearcherID: AAH-6558-2019
ScopusID: 14028278000
Yoksis Araştırmacı ID: 311762



Ögrenim Bilgisi

Tıpta Yandal Uzmanlık 2010 - 2013	Hacettepe Üniversitesi, Tıp Fakültesi (Türkçe), Dahili Tıp Bilimleri Bölümü, Türkiye
Tıpta Uzmanlık 2002 - 2008	Ankara Üniversitesi, Çocuk Sağlığı ve Hastalıkları, Türkiye
Lisans 1996 - 2002	Ankara Üniversitesi, Türkiye

Yaptığı Tezler

Tıpta Yandal Uzmanlık, SİSTATYONİN BETA-SENTAZ EKSİKLİĞİNE BAĞLI HOMOSİTİNÜRİ HASTALARINDA KLINİK, BİYOKİMYASAL, MOLEKÜLER BULGULARIN BELGELENMESİ VE GENOTİP-FENOTİP İLİŞKİSİNİN ARAŞTIRILMASI, Hacettepe Üniversitesi, Tıp Fakültesi (Türkçe), Dahili Tıp Bilimleri Bölümü, 2013
Tıpta Uzmanlık, Çocuk nörolojisi pratiğinde baş ağrılarının sıklık, etyoloji, tedavi yaklaşımları ve izlemleri açısından değerlendirmeleri, Ankara Üniversitesi, Ankara Tıp Fakültesi, Dahili Tıp Bilimleri, 2008

SCI, SSCI ve AHCI İndekslerine Giren Dergilerde Yayınlanan Makaleler

1. **BCKDK deficiency: a treatable neurodevelopmental disease amenable to newborn screening**
Tangeras T., Constante J. R., Backe P. H., Oyarzabal A., Neugebauer J., Weinhold N., Boemer F., Debray F. G., Ozturk-Hism B., Evren G., et al.
BRAIN, cilt.146, sa.7, ss.3003-3013, 2023 (SCI-Expanded)
 2. **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.
Molecular Genetics and Metabolism, cilt.139, sa.3, 2023 (SCI-Expanded)

3. **Management of acute metabolic crisis in TANGO2 deficiency: A case report**
Yilmaz-Gümüş E., ELÇİOĞLU H. N., Genç E., Arıcı Ş., Öztürk G., Yapıcı Ö., Akallı F., ÖZTÜRK HİŞMİ B.
Journal of Pediatric Endocrinology and Metabolism, 2023 (SCI-Expanded)
4. **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, cilt.12, sa.11, 2022 (SCI-Expanded)
5. **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, cilt.37, sa.11, ss.2950, 2022 (SCI-Expanded)
6. **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, cilt.32, 2022 (SCI-Expanded)
7. **A surprising cause of proteinuria: Answers**
DEMİR B. K., Kanik A., Kose M., ÖZTÜRK HİŞMİ B., Baran M.
PEDIATRIC NEPHROLOGY, cilt.37, ss.1033-1039, 2022 (SCI-Expanded)
8. **A surprising cause of proteinuria: Questions**
DEMİR B. K., Kanik A., Kose M., ÖZTÜRK HİŞMİ B., Baran M.
PEDIATRIC NEPHROLOGY, cilt.37, ss.1031-1032, 2022 (SCI-Expanded)
9. **CHARACTERISTICS OF PATIENTS WITH DISTAL RENAL TUBULAR ACIDOSIS AND ITS ASSOCIATION WITH HYPERAMMONEMIA: A SINGLE-CENTER EXPERIENCE**
Bodur E. D., Guven S., ÖZTÜRK HİŞMİ B., Cicek N., Sak M., Turkkan O. N., Pul S., YILDIZ N., GÖKCE İ., ALPAY H.
PEDIATRIC NEPHROLOGY, cilt.36, sa.10, ss.3428, 2021 (SCI-Expanded)
10. **Patients with cerebrotendinous xanthomatosis diagnosed with diverse multisystem involvement.**
Kısa P. T., Yıldırım G. K., Hisimi B., Dorum S., Kusbeci O. Y., Topak A., Baydan F., Celik F. N. D., Gorukmez O., Gulten Z. A., et al.
Metabolic brain disease, cilt.36, ss.1201-1211, 2021 (SCI-Expanded)
11. **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, cilt.64, sa.5, 2021 (SCI-Expanded)
12. **Inflammatory rheumatic diseases in patients with ochronotic arthropathy**
Inel T. Y., Kisa P. T., Balci A., Uslu S., Arslan Z., Hisimi B., Ucar U., Arslan N., Onen F., Sari I.
MODERN RHEUMATOLOGY, cilt.31, sa.5, ss.1031-1037, 2021 (SCI-Expanded)
13. **Neuropsychological attributes of urea cycle disorders: A systematic review of the literature**
Waisbren S. E., Stefanatos A. K., Kok T. M. Y., Ozturk-Hisimi B.
Journal of Inherited Metabolic Disease, cilt.42, sa.6, ss.1176-1191, 2019 (SCI-Expanded)
14. **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., Hisimi B., Bulbul F. S., Kose E., Gunduz M., Canda E., Kucukcongar A., et al.
Journal of Pediatric Endocrinology and Metabolism, cilt.32, sa.7, ss.675-681, 2019 (SCI-Expanded)
15. **Studying the effect of large neutral amino acid supplements on oxidative stress in phenylketonuric patients**
Kumru B., Ozturk Hisimi B., Kaplan D. S., Celik H.
Journal of Pediatric Endocrinology and Metabolism, cilt.32, sa.3, ss.269-274, 2019 (SCI-Expanded)
16. **Effect of Blood Phenylalanine Levels on Oxidative Stress in Classical Phenylketonuric Patients**
Kumru B., Kaplan D. S., Ozturk Hisimi B., Celik H.
Cellular and Molecular Neurobiology, cilt.38, sa.5, ss.1033-1038, 2018 (SCI-Expanded)

17. **An unusual cause of cavitating leukoencephalopathy: ethylmalonic encephalopathy**
Cavusoglu D., Hismi B., Dundar N. O., Oztekin O., Koc A., CANDA E., Arican P., Gencpinar P.
Acta Neurologica Belgica, cilt.118, sa.2, ss.309-312, 2018 (SCI-Expanded)
18. **Deoxyguanosine kinase deficiency: A report of four patients**
Ünal Ö., Hismi B., KILIÇ M., Gulsen H. H., COŞKUN T., Sivri S. H., DURSUN A., YÜCE A., TOKATLI A.
Journal of Pediatric Endocrinology and Metabolism, cilt.30, sa.6, ss.697-702, 2017 (SCI-Expanded)
19. **A late diagnosis of alkaptonuria in an elderly patient**
Taşkıran E., Yaşar Taş M., Hismi B., Demet İnce F., Ekmekçi S., Solakoğlu Kahraman D., Akar H.
European Geriatric Medicine, cilt.8, sa.2, ss.178-180, 2017 (SCI-Expanded)
20. **Detection of biotinidase gene mutations in Turkish patients ascertained by newborn and family screening**
Karaca M., ÖZGÜL R. K., Ünal Ö., Yücel-Yilmaz D., KILIÇ M., Hismi B., TOKATLI A., COŞKUN T., DURSUN A., SİVRİ H. S.
European Journal of Pediatrics, cilt.174, sa.8, ss.1077-1084, 2015 (SCI-Expanded)
21. **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., Hismi B., COŞKUN T., DURSUN A.
Turkish Journal of Pediatrics, cilt.57, sa.4, ss.388-393, 2015 (SCI-Expanded)
22. **Sapropterin dihydrochloride treatment in Turkish hyperphenylalaninemic patients under age four**
Ünal Ö., Gökmən-Özel H., COŞKUN T., ÖZGÜL R. K., Yücel D., Hismi B., TOKATLI A., DURSUN A., SİVRİ H. S.
Turkish Journal of Pediatrics, cilt.57, sa.3, ss.213-218, 2015 (SCI-Expanded)
23. **Fundus autofluorescence and optical coherence tomography findings in glutathione synthetase deficiency**
Şekeretoğlu H. T., Hismi B., KADAYIFÇILAR S., COŞKUN T.
Journal of AAPOS, cilt.19, sa.1, ss.80-82, 2015 (SCI-Expanded)
24. **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., Yilmaz D. Y., COŞKUN T., SİVRİ H. S., TOKATLI A., DURSUN A.
Gene, cilt.534, sa.2, ss.197-203, 2014 (SCI-Expanded)
25. **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.
Molecular Genetics and Metabolism, cilt.113, sa.4, ss.267-273, 2014 (SCI-Expanded)
26. **Phenotypic and genotypic spectrum of turkish patients with isovaleric acidemia**
Ozgul R. K., Karaca M., Kilic M., Kucuk O., Yucel-Yilmaz D., Unal O., Hismi B., Aliefendioglu D., Sivri S., Tokatli A., et al.
European Journal of Medical Genetics, cilt.57, sa.10, ss.596-601, 2014 (SCI-Expanded)
27. **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, cilt.13, ss.33-36, 2014 (SCI-Expanded)
28. **A patient with pyruvate carboxylase deficiency and nemaline rods on muscle biopsy**
Unal O., ORHAN D., Ostergaard E., TOKATLI A., DURSUN A., Ozturk-Hismi B., COŞKUN T., Wibrand F., Kalkanoglu-Sivri H. S.
Journal of Child Neurology, cilt.28, sa.11, ss.1505-1508, 2013 (SCI-Expanded)
29. **Vanishing white matter with hepatomegaly and hypertriglyceridemia attacks**
Unal O., Ozgen B., ORHAN D., TOKATLI A., Hismi B., DURSUN A., COŞKUN T., Kalkanoglu-Sivri H. S.
Journal of Child Neurology, cilt.28, sa.11, ss.1509-1512, 2013 (SCI-Expanded)
30. **Molecular and clinical evaluation of Turkish patients with lysinuric protein intolerance**
Güzel-Ozantürk A., ÖZGÜL R. K., Ünal Ö., Hismi B., Aydin H. i., Sivri S., TOKATLI A., COŞKUN T., Aksöz E., DURSUN A.
Gene, cilt.521, sa.2, ss.293-295, 2013 (SCI-Expanded)
31. **ENZYME REPLACEMENT THERAPY (ERT)RESULTS OF PATIENTS WITH MUCOPOLYSACCHARIDOSIS TYPE II (HUNTER SYNDROME)**
Unal O., Dogru-Ersöz D., Alehan D., SAĞLAM M. Y., Hismi B., Dursun A., Tokatli A., Coskun T., Sivri H. S.

- JOURNAL OF INHERITED METABOLIC DISEASE, cilt.35, 2012 (SCI-Expanded)
32. 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.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.35, 2012 (SCI-Expanded)
33. ACUTE INTERMITTENT PORPHYRIA: STILL UNCALLED BY PHYSICIANS
Hismi B., Tanriover M. D., Unal O., Sener E., TEMUCİN Ç. M., SİVRİ H. S., DURSUN A., COŞKUN T.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.35, 2012 (SCI-Expanded)
34. 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, cilt.35, 2012 (SCI-Expanded)
35. 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.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.35, 2012 (SCI-Expanded)
36. THE 48-HOUR TETRAHYDOBIOPTERIN (BH4) LOADING TEST AND LONG-TERM OUTCOME OF PATIENTS WITH BH4 TREATMENT IN TURKISH PHENYLKETONURIA (PKU) PATIENTS
Unal O., Gokmen-Ozel H., Coskun T., Hismi B., Tokatli A., Dursun A., Ozgul R. K., Sivri H. S.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.35, 2012 (SCI-Expanded)
37. MOLYBDENUM COFACTOR (MOCO) DEFICIENCY TYPE B; CLINICAL, BIOCHEMICAL AND NEUROIMAGING FEATURES OF FIVE PATIENTS WITH TWO NOVEL MUTATIONS
Hismi B., Unal O., Sass J. O., Beermann F., Ichida K., DURSUN A., SİVRİ H. S., TOKATLI A., COŞKUN T.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.35, 2012 (SCI-Expanded)
38. 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.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.35, 2012 (SCI-Expanded)
39. 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, cilt.35, 2012 (SCI-Expanded)
40. 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, cilt.35, 2012 (SCI-Expanded)
41. Detection of other inborn errors of metabolism in hyperphenylalaninemic babies picked up on narrow-spectrum screening programs
Unal O., Ozturk-Hismi B., COŞKUN T., TOKATLI A., DURSUN A., SİVRİ H. S.
TURKISH JOURNAL OF PEDIATRICS, cilt.54, sa.4, ss.409-412, 2012 (SCI-Expanded)
42. Secondary hemophagocytosis in 3 patients with organic acidemia involving propionate metabolism.
Gokce M., Unal O., Hismi B., Gumruk F., Coskun T., Balta G., Unal S., Cetin M., Kalkanoglu-Sivri H., Dursun A., et al.
Pediatric hematology and oncology, cilt.29, sa.1, ss.92-98, 2012 (SCI-Expanded)
43. A Rare Galactosemia Complication: Vitreous Hemorrhage
Takci S., KADAYIFÇILAR S., COŞKUN T., YİĞİT Ş., Hismi B.
JMD REPORTS - CASE AND RESEARCH REPORTS, 2012/2, cilt.5, ss.89-93, 2012 (SCI-Expanded)
44. CHALLENGES IN THE TREATMENT OF A PATIENT AFFECTED BY BOTH ARGININOSUCCINIC ACIDURIA AND METHYLMALONIC ACIDURIA
Unal O., Hismi B., COŞKUN T., TOKATLI A., DURSUN A., Sivri K. H. S.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.34, 2011 (SCI-Expanded)
45. HEMOPHAGOCYTOSIS IN THREE PATIENTS WITH ORGANIC ACIDEMIA INVOLVING PROPIONATE METABOLISM
Gokce M., Unal O., Hismi B., GÜMRÜK F., COŞKUN T., DURSUN A., TOKATLI A., Sivri K. H. S.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.34, 2011 (SCI-Expanded)

46. **ACUTE RESPIRATORY DISTRESS SYNDROME IN TWO PATIENTS WITH ORGANIC ACIDEMIA INVOLVING PROPIONATE METABOLISM**
Unal O., Hismi B., Sivri K. H. S., Dursan A., TOKATLI A., COŞKUN T.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.34, 2011 (SCI-Expanded)
47. **A NOVEL MUTATION IN BETA KETOTHIOLASE DEFICIENCY**
Unal O., Hismi B., KILIÇ M., DURSUN A., Kalkanoglu-Sivri H. S., TOKATLI A., COŞKUN T., Sass O.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.33, 2010 (SCI-Expanded)
48. **GALACTOSEMIA IN A TURKISH POPULATION WITH A HIGH PREVALENCE OF Q188R MUTATION**
Guzel A., ÖZGÜL R. K., DÜNDAR H., COŞKUN T., SİVRİ H. S., TOKATLI A., Goksun E., Hismi B., DURSUN A.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.33, 2010 (SCI-Expanded)
49. **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, cilt.33, 2010 (SCI-Expanded)
50. **POLYNEUROPATHY AS THE MAIN PRESENTING SYMPTOM IN PDH DEFICIENCY**
Unal O., Hismi B., Kilie M., DURSUN A., Kalkanoglu-Sivri H. S., TOKATLI A., COŞKUN T., Zeviani M.
JOURNAL OF INHERITED METABOLIC DISEASE, cilt.33, 2010 (SCI-Expanded)
51. **A Truncating Mutation in SERPINB6 Is Associated with Autosomal-Recessive Nonsyndromic Sensorineural Hearing Loss**
Sirmaci A., Erbek S., Price J., Huang M., Duman D., Cengiz F. B., Bademci G., Tokgöz-Yilmaz S., Hismi B., Özdağ H., et al.
American Journal of Human Genetics, cilt.86, sa.5, ss.797-804, 2010 (SCI-Expanded)
52. **Mutations in TMC1 contribute significantly to nonsyndromic autosomal recessive sensorineural hearing loss: A report of five novel mutations**
Sirmaci A., Duman D., Öztürkmen-Akay H., Erbek S., Incesulu A., Öztürk-Hismi B., Arici Z. S., Yüksel-Konuk E. B., Taşır-Yilmaz S., Tokgöz-Yilmaz S., et al.
International Journal of Pediatric Otorhinolaryngology, cilt.73, sa.5, ss.699-705, 2009 (SCI-Expanded)
53. **Renal replacement therapies in pediatric intensive care patients: Experiences of one center in Turkey**
Kendirli T., Ekim M., Özçakar Z. B., Yüksel S., Acar B., Öztürk-Hismi B., Derelli E., Kavaz A., Yalaki Z., Yalçinkaya F.
Pediatrics International, cilt.49, sa.3, ss.345-348, 2007 (SCI-Expanded)
54. **Homozygous mutations in fibroblast growth factor 3 are associated with a new form of syndromic deafness characterized by inner ear agenesis, microtia, and microdontia**
Tekin M., Hismi B., Fitoz S., Özdağ H., Cengiz F. B., Sirmaci A., Aslan I., Inceoğlu B., Yüksel-Konuk E. B., Yilmaz S. T., et al.
American Journal of Human Genetics, cilt.80, sa.2, ss.338-344, 2007 (SCI-Expanded)
55. **Mechanical ventilation in children**
Kendirli T., Kavaz A., Yalaki Z., Öztürk Hismi B., Derelli E., Ince E.
Turkish Journal of Pediatrics, cilt.48, sa.4, ss.323-327, 2006 (SCI-Expanded)
56. **Antibiotic resistance of urinary tract pathogens and evaluation of empirical treatment in Turkish children with urinary tract infections**
Yuksel S., Ozturk B., Kavaz A., Ozcakar Z. B., Acar B., Guriz H., Aysev D., Ekim M., Yalcinkaya F.
INTERNATIONAL JOURNAL OF ANTIMICROBIAL AGENTS, cilt.28, sa.5, ss.413-416, 2006 (SCI-Expanded)
57. **Clinical improvement with infliximab in a child with amyloidosis secondary to familial Mediterranean fever**
Yuksel S., Yalcinkaya F., Acar B., Ozcakar Z. B., Ozturk B., Ekim M.
RHEUMATOLOGY, cilt.45, sa.10, ss.1307-1308, 2006 (SCI-Expanded)
58. **Effects of GJB2 genotypes on the audiological phenotype: Variability is present for all genotypes**
Hismi B., Yilmaz S. T., Incesulu A., Tekin M.
International Journal of Pediatric Otorhinolaryngology, cilt.70, sa.10, ss.1687-1694, 2006 (SCI-Expanded)
59. **A germline PTEN mutation with manifestations of prenatal onset and verrucous epidermal nevus**
Tekin M., Hismi B., Fitoz S., Yalçinkaya F., Ekim M., Kansu A., Ertem M., Deda G., Tutar E., Arsan S., et al.

- American Journal of Medical Genetics, Part A, cilt.140, sa.13, ss.1472-1475, 2006 (SCI-Expanded)
60. Seronegative spondyloarthropathy associated with Takaya-su's arteritis in a child
Acar B., Yalcinkaya F., Ozturk B., Yuksel S., Ozcakar Z., Fitoz S., Buyukcelik M., Noyan A., Sanlidilek U., Ekim M.
CLINICAL AND EXPERIMENTAL RHEUMATOLOGY, cilt.23, sa.2, ss.278-279, 2005 (SCI-Expanded)

Diger Dergilerde Yayınlanan Makaleler

1. Molecular Aspects of Distal Kidney Tubular Acidosis in Children, Its Long-Term Outcome, and Relationship with Hyperammonemia
Güven S., Gökçe İ., Alavanda C., Öztürk Hışmî B., Çiçek N., Bodur Demirci E., Sak M., Yıldız N., Ata P., Alpay H.
TURKISH ARCHIVES OF PEDIATRICS, cilt.57, sa.4, ss.432-440, 2022 (ESCI)
2. Diagnostic Yield of Neuroimaging and Electroencephalography in Children with Recurrent Headaches
ÖZTÜRK HİŞMÎ B., Teber S., Ozkan M., Unal O., Deda G.
Journal of Pediatric Neurology, cilt.19, ss.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.
IZMIR DR BEHCET UZ COCUK HASTANESİ DERGİSİ, cilt.11, sa.2, ss.198-201, 2021 (ESCI)
4. Erişkin başlangıçlı kalıtsal metabolik hastalıklar: Tek merkez deneyimi
ÖZTÜRK HİŞMÎ B.
Pamukkale Medical Journal, cilt.14, sa.3, ss.692-705, 2021 (Hakemli Dergi)
5. Investigation of L-Carnitine Concentrations in Treated Patients with Maple Syrup Urine Disease
Kumru B., Hismi B.
JOURNAL OF PEDIATRIC GENETICS, cilt.8, sa.3, ss.133-136, 2019 (ESCI)
6. Renal–Hepatic–Pancreatic Dysplasia: An Ultra-Rare Ciliopathy with a Novel NPHP3 Genotype
Cagan A. Y., Baran M., ÖZTÜRK HİŞMÎ B., Ozyilmaz B., Vardi K., Ozer K. O., Aksoy B., Kasap D. B.
Journal of Pediatric Genetics, 2019 (ESCI)
7. Association of esophageal atresia without fistula and methylmalonic acidemia: The first case in the literature
Sayan A., Mert M., Oztan M. O., Hismi B., Akbay S., Koyluoglu G.
IZMIR DR BEHCET UZ COCUK HASTANESİ DERGİSİ, cilt.8, sa.3, ss.239-242, 2018 (ESCI)
8. Severe vitamin B12 deficiency with pancytopenia, hepatosplenomegaly and leukoerythroblastosis in two Syrian refugee infants: A challenge to differentiate from acute leukaemia
Belen B., Hismi B., KOÇAK Ü.
BMJ Case Reports, 2014 (Scopus)
9. Evde Parenteral Beslenme
ÖZTÜRK HİŞMÎ B.
Katkı Pediatri Dergisi, cilt.34, sa.2, ss.249-264, 2012 (Hakemsiz Dergi)
10. A life-saving line in resuscitation and shock management of the critically ill child: intraosseous infusion
KENDİRLİ T., YALAKİ Z., ÖZTÜRK HİŞMÎ B., KAVAZ TUFAN A., OKULU E., İNCE E.
Ankara Üniversitesi Tıp Fakültesi Mecmuası, cilt.58, sa.4, ss.176-179, 2005 (Hakemli Dergi)
11. Severe myocardial injury in an infant with hemorrhagic shock and encephalopathy syndrome
KENDİRLİ T., GÜMÜŞ DOĞAN D., ÖZTÜRK HİŞMÎ B., İNCE E., DEDA G., TUTAR H. E.
Journal of Pediatric Neurology, cilt.3, sa.2, ss.113-116, 2005 (Scopus)
12. Tanınız nedir?
KENDİRLİ T., ÇİFTÇİ E., DOĞAN D., ÖZTÜRK HİŞMÎ B., DEDA G., İNCE E.
Klinik Pediatri, cilt.3, sa.1, ss.42-44, 2004 (Hakemli Dergi)

Kitap & Kitap Bölümleri

1. **FENİLKETONÜRİ ve BESLENME TEDAVİSİ**
ÖZTÜRK HİŞMİ B., KUMRU B.
Kalıtsal Metabolik Hastalıklarda Beslenme Tedavisi, Fatma Tuba Eminoğlu, Editör, Orient Ulusal Yayınevi, Ankara, ss.159-201, 2022
2. **Beslenme-Vitamin E**
ÖZTÜRK HİŞMİ B.
Yurdakök Pediatri, H.Serap Sivri, Editör, Güneş Kitabevi, Ankara, ss.1547-1551, 2017
3. **Kalıtsal Metabolik Hastalıklar - Diğer Aminoasit Metabolizması Bozuklukları**
Kılıç M., ÖZTÜRK HİŞMİ B., Ünal Ö., AYDIN H. İ.
Yurdakök Pediatri, Turgay Coşkun, Editör, Güneş Kitabevi, Ankara, ss.1617-1656, 2017
4. **Kalıtsal Metabolizma Hastalıkları- Pürin ve Pirimidin Metabolizması Bozuklukları**
ÖZTÜRK HİŞMİ B.
Yurdakök Pediatri, Turgay Coşkun, Editör, Güneş Kitabevi, Ankara, ss.1859-1869, 2017
5. **Metabolik Otopsi**
ÖZTÜRK HİŞMİ B.
Yenidoğanda Kalıtsal Metabolik Hastalıklar, Coşkun Turgay, Yurdakök Murat, Editör, Güneş Tıp Kitabevi, Ankara, ss.383-387, 2014
6. **Bebeklik ve Erken Çocukluk Döneminde Yaygın Gelişimsel Bozukluk ve Otizm**
ÖZTÜRK HİŞMİ B., Öztürk Ertem İ.
Gelişimsel Pediatri, İlgi Ertem, Editör, Çocuk Sağlığı ve Hastalıkları Araştırma Vakfı, Ankara, ss.388-413, 2005

Metrikler

Yayın: 130
Atıf (WoS): 483
Atıf (Scopus): 607
H-İndeks (WoS): 11
H-İndeks (Scopus): 12

Araştırma Alanları

Sağlık Bilimleri

Akademi Dışı Deneyim

Harvard Medical School, Boston Children's Hospital, Department of Genetics and Genomics
İZMİR TEPECİK EĞİTİM VE ARAŞTIRMA HASTANESİ
Ankara Dr. Hulusi Alataş Elmadağ Devlet Hastanesi
GAZİANTEP ÇOCUK HASTALIKLARI HASTANESİ
MARDİN KADIN DOĞUM VE ÇOCUK HASTALIKLARI HASTANESİ