

Prof.Dr. TURGAY COŞKUN

Kişisel Bilgiler

E-posta: tcoskun@hacettepe.edu.tr

Web: <https://avesis.hacettepe.edu.tr/tcoskun>

Eğitim Bilgileri

Tıpta Yandal Uzmanlık, Hacettepe Üniversitesi, Tıp Fakültesi, Türkiye 1977 - Devam Ediyor

Tıpta Uzmanlık, Hacettepe Üniversitesi, Tıp Fakültesi, Çocuk Sağlığı Ve Hastalıkları Anabilim Dalı, Türkiye 1977 - 1981

Araştırma Alanları

Tıp, Sağlık Bilimleri, Dahili Tıp Bilimleri, Çocuk Sağlığı ve Hastalıkları, Pediatrik Endokrinoloji ve Metabolizma

Akademik Unvanlar / Görevler

Prof.Dr., Hacettepe Üniversitesi, Hacettepe Tıp Fakültesi, 1977 - Devam Ediyor

Prof.Dr., Hacettepe Üniversitesi, Tıp Fakültesi (İngilizce), Dahili Tıp Bilimleri Bölümü, 1977 - 1981

SCI, SSCI ve AHCI İndekslerine Giren Dergilerde Yayınlanan Makaleler

- I. **Electro-clinical features and long-term outcomes in guanidinoacetate methyltransferase (GAMT) deficiency**
YILDIZ Y., Ardıçlı D., GÖÇMEN R., YALNIZOĞLU D., Topçu M., Coşkun T., TOKATLI A., Haliloğlu G.
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- II. **Predictors of eventual requirement of phenylalanine-restricted diet in young infants with phenylalanine hydroxylase deficiency initially managed with sapropterin monotherapy**
ÇIKI K., YILDIZ Y., KAHRAMAN A. B., ÖZGÜL R. K., COŞKUN T., DURSUN A., TOKATLI A., SİVRİ S.
Molecular Genetics and Metabolism, cilt.140, sa.3, 2023 (SCI-Expanded)
- III. **Organic acidemias in the neonatal period: 30 years of experience in a referral center for inborn errors of metabolism**
ÜNSAL Y., YURDAKÖK M., YİĞİT Ş., ÇELİK H. T., DURSUN A., SİVRİ H. S., TOKATLI A., COŞKUN T.
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- IV. **Recommendations on phenylketonuria in Turkey**
COŞKUN T., ÇOKER M., Mungan N. O., GÖKMEN ÖZEL H., SİVRİ H. S.
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- V. **A new UHPLC-MS/MS method for the screening of urinary oligosaccharides expands the detection of storage disorders**
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- VI. **Autism spectrum disorder in patients with inherited metabolic disorders-a large sample from a tertiary center**
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- VII. **Clinical and molecular characteristics of carnitine-acylcarnitine translocase deficiency with c.270delC and a novel c.408C>A variant**

BİLGİNER GÜRBÜZ B., YÜCEL YILMAZ D., ÖZGÜL R. K., KOŞUKCU C., DURSUN A., SİVRİ H. S., COŞKUN T., TOKATLI A.
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- VIII. **Molecular etiology of isolated congenital cataract using next-generation sequencing: Single center exome sequencing data from Turkey**

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- IX. **Glutaric aciduria type 1: Genetic and phenotypic spectrum in 53 patients**

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- X. **Expanding the clinical spectrum of mitochondrial 3-hydroxy-3-methylglutaryl-CoA synthase deficiency with Turkish cases harboring novel HMGCS2 gene mutations and literature review**

Kilic M., Dorum S., Topak A., Yazici M. U., EZGÜ F. S., COŞKUN T.

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- XI. **Predictors of acute metabolic decompensation in children with maple syrup urine disease at the emergency department**

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- XII. **Two cases of Vici syndrome presenting with corpus callosum agenesis, albinism, and severe developmental delay**

Hızal M., Yeke B., YILDIZ Y., Öztürk A., Gürbüz B. B., COŞKUN T.

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- XIII. **Genotypes and estimated prevalence of phosphomannomutase 2 deficiency in Turkey differ significantly from those in Europe**

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- XIV. **Caring for a Child with Phenylketonuria: Parental Experiences from a Eurasian Country**

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- XV. **Oral health status of children with phenylketonuria**

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- XVI. **Determinants of Riboflavin Responsiveness in Multiple Acyl-CoA Dehydrogenase Deficiency**

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- XVII. **The effectiveness of enzyme replacement therapy on cardiac findings in patients with mucopolysaccharidosis**

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- XVIII. **Imaging liver nodules in tyrosinemia type-1: A retrospective review of 16 cases in a tertiary pediatric hospital**

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- XIX. **International best practice for the evaluation of responsiveness to sapropterin dihydrochloride in patients with phenylketonuria**

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- XX. **Do not Miss Rare and Treatable Cause of Early-Onset Hemolytic Uremic Syndrome: Cobalamin C Deficiency**
TOPALOĞLU R., Inozu M., GÜLHAN B., BİLGİNER GÜRBÜZ B., TALİM B., COŞKUN T.
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- XXI. **TWO CASES WITH DIVERSE COURSE OF AHUS RELATED TO COBALAMIN C DEFECT**
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- XXII. **The potential role of gut microbiota and its modulators in the management of propionic and methylmalonic acidemia**
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- XXIII. **Genotypic-phenotypic features and enzyme replacement therapy outcome in patients with mucopolysaccharidosis VI from Turkey**
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- XXIV. **Deoxyguanosine kinase deficiency: a report of four patients**
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- XXV. **Screening for mucopolysaccharidoses in the Turkish population: Analytical and clinical performance of an age-range specific, dye-based, urinary glycosaminoglycan assay**
EL MOUSTAFA K., Sivri S., KARAHAN S., COŞKUN T., Akbilyik F., LAY İ.
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- XXVI. **Partial hydatidiform mole in a phenylketonuria patient treated with sapropterin dihydrochloride**
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- XXVII. **Riboflavin-Responsive and -Non-responsive Mutations in FAD Synthase Cause Multiple Acyl-CoA Dehydrogenase and Combined Respiratory-Chain Deficiency**
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- XXVIII. **Clinical phenotype, biochemical profile, and treatment in 19 patients with arginase 1 deficiency**
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- XXIX. **Brown-Vialetto-Van Laere syndrome: two siblings with a new mutation and dramatic therapeutic effect of high-dose riboflavin**
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- XXX. **Diagnostic and management practices for phenylketonuria in 19 countries of the South and Eastern European Region: survey results**
Gizewska M., MacDonald A., Belanger-Quintana A., Burlina A., Cleary M., COŞKUN T., Feillet F., Muntau A. C., Trefz F. K., van Spronsen F. J., et al.
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- XXXI. **A Nonvirilized form of Classic 3 beta-Hydroxysteroid Dehydrogenase Deficiency Due to a Homozygous S218P Mutation in the HSD3B2 Gene in a Girl with Classic Phenylketonuria**
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- XXXII. **Late-diagnosed phenylketonuria in an eight-year-old boy with dyslexia and attention-deficit hyperactivity disorder**
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- XXXIII. **Evaluation and identification of IDUA gene mutations in Turkish patients with mucopolysaccharidosis type I**
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- XXXIV. **Sapropterin dihydrochloride treatment in Turkish hyperphenylalaninemic patients under age four**
ÖZLEM U., GÖKMEN ÖZEL H., COŞKUN T., ÖZGÜL R. K., YÜCEL YILMAZ D., BURCU H., TOKATLI A., DURSUN A., SİVRİ H. S.
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- XXXV. **Detection of biotinidase gene mutations in Turkish patients ascertained by newborn and family screening**
KARACA M., ÖZGÜL R. K., ÜNAL O., Yucel-Yılmaz D., KILIÇ M., Hismi B., TOKATLI A., COŞKUN T., DURSUN A., SİVRİ H. S.
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- XXXVI. **Ailevi Hiperkolesterolemili Hastaların Mutasyon Analiz Sonuçlarının Simone-Broome Kriterleriyle Değerlendirilmesi**
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- XXXVII. **Conventional and advanced MR imaging in infantile Refsum disease**
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- XXXVIII. **Fundus autofluorescence and optical coherence tomography findings in glutathione synthetase deficiency**
TAYLAN ŞEKEROĞLU H., ÖZTÜRK HİŞMİ B., KADAYIFÇILAR S., COŞKUN T.
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- XXXIX. **Two Turkish siblings with MEGDEL syndrome due to novel SERAC1 genemutation.**
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- XL. **Phenotypic and genotypic spectrum of Turkish patients with isovaleric acidemia**
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- XLI. **A case of fucosidosis type II: diagnosed with dysmorphological and radiological findings**
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- XLII. **Mucopolysaccharidosis: Otolaryngologic findings, obstructive sleep apnea and accumulation of glucosaminoglycans in lymphatic tissue of the upper airway**
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- XLIII. **High prevalence of cerebral venous sinus thrombosis (CVST) as presentation of cystathionine beta-synthase deficiency in childhood: Molecular and clinical findings of Turkish probands**
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- XLIV. **Guanidinoacetate methyltransferase (GAMT) deficiency: Outcomes in 48 individuals and recommendations for diagnosis, treatment and monitoring**

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- XLV. **Pregnancy and Lactation Outcomes in a Turkish Patient with Lysinuric Protein Intolerance**
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- XLVI. **A Patient With Pyruvate Carboxylase Deficiency and Nemaline Rods on Muscle Biopsy**
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- XLVII. **Vanishing White Matter With Hepatomegaly and Hypertriglyceridemia Attacks**
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- XLVIII. **Cobalamin C defect: a patient of late-onset type with homozygous p. R132*mutation**
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- XLIX. **Galactosemia in the Turkish population with a high frequency of Q188R mutation and distribution of Duarte-1 and Duarte-2 variations**
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- L. **Molecular and clinical evaluation of Turkish patients with lysinuric protein intolerance**
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- LI. **Genetic basis of hyperlysinemia**
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- LII. **Serum alpha-fetoprotein levels in neonatal cholestasis**
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- LIII. **PROPIONIC ACIDEMIA PRESENTING WITH PERSISTENT PULMONARY HYPERTENSION IN TWO NEONATES**
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- LIV. **MOLECULAR CHARACTERISATION OF BIOTINIDASE GENE MUTATIONS IN TURKISH PATIENTS; AN UPDATE OF THE RESULTS**
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- LV. **Detection of other inborn errors of metabolism in hyperphenylalaninemic babies picked up on narrow-spectrum screening programs**
Unal O., ÖZTÜRK HİŞMİ B., COŞKUN T., TOKATLI A., DURSUN A., SİVRİ H. S.
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- LVI. **When do we need to perform a diagnostic lumbar puncture for neurometabolic diseases? Positive yield and retrospective analysis from a tertiary center**
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- LVII. **Genetic basis of cystinosis in Turkish patients: a single-center experience**
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- LVIII. A Rare Galactosemia Complication: Vitreous Hemorrhage**
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- LIX. Identification of Mutations and Evaluation of Cardiomyopathy in Turkish Patients with Primary Carnitine Deficiency**
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- LX. Effects of probiotic (Primalac 454) on nonalcoholic fatty liver disease in broilers**
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- LXI. Annual symposium of the SSIEM 2010**
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- LXII. Home visits in phenylketonuria: a 12-month longitudinal study**
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