

## Prof. AYŞEGÜL TOKATLI

### Personal Information

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### Education Information

Expertise In Medicine, Hacettepe University, Tıp Fakültesi (Türkçe), Dahili Tıp Bilimleri Bölümü, Turkey 1982 - 1986

### Research Areas

Health Sciences

### Academic Titles / Tasks

Professor, Hacettepe University, Tıp Fakültesi (Türkçe), Dahili Tıp Bilimleri Bölümü, 1989 - Continues

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

- I. **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.  
TURKISH JOURNAL OF PEDIATRICS, vol.63, no.4, pp.691-696, 2021 (Journal Indexed in SCI)
- II. **Sensory, voluntary, and motor postural control in children and adolescents with mucopolysaccharidosis**  
YİÇİT Ö., AKSOY S., Akyol U., TOKATLI A., SİVRİ H. S.  
JOURNAL OF PEDIATRIC ENDOCRINOLOGY & METABOLISM, vol.34, no.5, pp.583-589, 2021 (Journal Indexed in SCI)
- III. **Glutaric aciduria type 1: Genetic and phenotypic spectrum in 53 patients**  
BİLGİNER GÜRBÜZ B., YÜCEL YILMAZ D., COŞKUN T., TOKATLI A., DURSUN A., SİVRİ H. S.  
EUROPEAN JOURNAL OF MEDICAL GENETICS, vol.63, no.11, 2020 (Journal Indexed in SCI)
- IV. **Predictors of acute metabolic decompensation in children with maple syrup urine disease at the emergency department**  
YILDIZ Y., Akcan Yildiz L., DURSUN A., TOKATLI A., COŞKUN T., TEKŞAM Ö., SİVRİ H. S.  
EUROPEAN JOURNAL OF PEDIATRICS, vol.179, no.7, pp.1107-1114, 2020 (Journal Indexed in SCI)
- V. **Retrospective evaluation of 85 patients with urea cycle disorders: one center experience, three new mutations**  
Nakip O. S. , YILDIZ Y., TOKATLI A.  
JOURNAL OF PEDIATRIC ENDOCRINOLOGY & METABOLISM, vol.33, no.6, pp.721-728, 2020 (Journal Indexed in SCI)
- VI. **Comment on: "Multiple acyl-CoA dehydrogenase deficiency in elderly carriers"**  
YILDIZ Y., TOKATLI A.  
JOURNAL OF NEUROLOGY, vol.267, no.4, pp.1209-1210, 2020 (Journal Indexed in SCI)
- VII. **Genotypes and estimated prevalence of phosphomannomutase 2 deficiency in Turkey differ**

**significantly from those in Europe**

YILDIZ Y., Arslan M., Celik G., Kasapkara C. S. , Ceylaner S., DURSUN A., SİVRİ H. S. , COŞKUN T., TOKATLI A.  
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, vol.182, no.4, pp.705-712, 2020 (Journal Indexed in SCI)

- VIII. **Oral health status of children with phenylketonuria**  
BALLIKAYA E., YILDIZ Y., SİVRİ H. S. , TOKATLI A., DURSUN A., Olmez S., COŞKUN T., Tekcicek M. U.  
JOURNAL OF PEDIATRIC ENDOCRINOLOGY & METABOLISM, vol.33, no.3, pp.361-365, 2020 (Journal Indexed in SCI)
- IX. **The effectiveness of enzyme replacement therapy on cardiac findings in patients with mucopolysaccharidosis**  
BİLGİNER GÜRBÜZ B., AYPAR E., COŞKUN T., ALEHAN D., DURSUN A., TOKATLI A., SİVRİ H. S.  
JOURNAL OF PEDIATRIC ENDOCRINOLOGY & METABOLISM, vol.32, no.10, pp.1049-1053, 2019 (Journal Indexed in SCI)
- X. **Determinants of Riboflavin Responsiveness in Multiple Acyl-CoA Dehydrogenase Deficiency**  
YILDIZ Y., TALİM B., Haliloglu G., Topaloglu H., AKÇÖREN Z., DURSUN A., SİVRİ H. S. , COŞKUN T., TOKATLI A.  
PEDIATRIC NEUROLOGY, vol.99, pp.69-75, 2019 (Journal Indexed in SCI)
- XI. **Imaging liver nodules in tyrosinemia type-1: A retrospective review of 16 cases in a tertiary pediatric hospital**  
ÖZCAN H. N. , KARÇAALTINCABA M., Pektas E., SİVRİ H. S. , OĞUZ B., DURSUN A., TOKATLI A., COŞKUN T., HALİLOĞLU M.  
EUROPEAN JOURNAL OF RADIOLOGY, vol.116, pp.41-46, 2019 (Journal Indexed in SCI)
- XII. **TWO CASES WITH DIVERSE COURSE OF AHUS RELATED TO COBALAMIN C DEFECT**  
Inozu M., GÜLHAN B., TOKATLI A., COŞKUN T., TOPALOĞLU R.  
PEDIATRIC NEPHROLOGY, vol.33, no.10, pp.1848, 2018 (Journal Indexed in SCI)
- XIII. **Post-mortem detection of FLAD1 mutations in 2 Turkish siblings with hypotonia in early infancy**  
YILDIZ Y., Olsen R. K. J. , SİVRİ H. S. , AKÇÖREN Z., Nygaard H. H. , TOKATLI A.  
NEUROMUSCULAR DISORDERS, vol.28, no.9, pp.787-790, 2018 (Journal Indexed in SCI)
- XIV. **Genotypic-phenotypic features and enzyme replacement therapy outcome in patients with mucopolysaccharidosis VI from Turkey**  
Kilic M., DURSUN A., COŞKUN T., TOKATLI A., ÖZGÜL R. K. , YUCEL-YILMAZ D., Karaca M., Dogru D., ALEHAN D., KADAYIFÇILAR S., et al.  
AMERICAN JOURNAL OF MEDICAL GENETICS PART A, vol.173, no.11, pp.2954-2967, 2017 (Journal Indexed in SCI)
- XV. **Deoxyguanosine kinase deficiency: a report of four patients**  
Unal O., ÖZTÜRK HİŞMİ B., KILIÇ M., HIZARCIOĞLU GÜLŞEN H., COŞKUN T., Sivri S. H. , DURSUN A., YÜCE A., TOKATLI A.  
JOURNAL OF PEDIATRIC ENDOCRINOLOGY & METABOLISM, vol.30, no.6, pp.697-702, 2017 (Journal Indexed in SCI)
- XVI. **Hereditary Dopamine Transporter Deficiency Syndrome: Challenges in Diagnosis and Treatment**  
YILDIZ Y., Pektas E., TOKATLI A., Haliloglu G.  
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- XVII. **Partial hydatidiform mole in a phenylketonuria patient treated with sapropterin dihydrochloride**  
YILDIZ Y., DURSUN A., TOKATLI A., COŞKUN T., Sivri S.  
GYNECOLOGICAL ENDOCRINOLOGY, vol.33, no.1, pp.19-20, 2017 (Journal Indexed in SCI)
- XVIII. **Evaluation and identification of IDUA gene mutations in Turkish patients with mucopolysaccharidosis type I**  
Atceken N., ÖZGÜL R. K. , Yılmaz D. Y. , TOKATLI A., COŞKUN T., SİVRİ H. S. , DURSUN A., Karaca M.  
TURKISH JOURNAL OF MEDICAL SCIENCES, vol.46, no.2, pp.404-408, 2016 (Journal Indexed in SCI)
- XIX. **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**  
ALİKAŞIYOĞLU A., Buyukyılmaz G., GÖNÇ E. N. , ÖZÖN Z. A. , KANDEMİR N., DÜNDAR M., POLAT S., PEKTAŞ E., DURSUN A., Sivri S., et al.  
HORMONE RESEARCH IN PAEDIATRICS, vol.86, pp.281, 2016 (Journal Indexed in SCI)

- XX. **Late-diagnosed phenylketonuria in an eight-year-old boy with dyslexia and attention-deficit hyperactivity disorder**  
YILDIZ Y., DURSUN A., TOKATLI A., COŞKUN T., SİVRİ H. S.  
TURKISH JOURNAL OF PEDIATRICS, vol.58, no.1, pp.94-96, 2016 (Journal Indexed in SCI)
- XXI. **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.  
Turkish Journal Of Pediatrics, pp.213-218, 2015 (Journal Indexed in SCI Expanded)
- XXII. **Key features and clinical variability of COG6-CDG**  
Rymen D., Winter J., Van Hasselt P. M. , Jaeken J., Kasapkara C., GÖKÇAY G. F. , Haijes H., Goyens P., TOKATLI A., Thiel C., et al.  
MOLECULAR GENETICS AND METABOLISM, vol.116, no.3, pp.163-170, 2015 (Journal Indexed in SCI)
- XXIII. **Detection of biotinidase gene mutations in Turkish patients ascertained by newborn and family screening**  
KARACA M., ÖZGÜL R. K. , ÜNAL O., Yucel-Yilmaz D., KILIÇ M., Hismi B., TOKATLI A., COŞKUN T., DURSUN A., SİVRİ H. S.  
EUROPEAN JOURNAL OF PEDIATRICS, vol.174, no.8, pp.1077-1084, 2015 (Journal Indexed in SCI)
- XXIV. **Lack of prolidase causes a bone phenotype both in human and in mouse**  
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BONE, vol.72, pp.53-64, 2015 (Journal Indexed in SCI)
- XXV. **Two Turkish siblings with MEGDEL syndrome due to novel SERAC1 genemutation.**  
ÜNAL Ö., ÖZGÜL R. K. , YÜCEL YILMAZ D., YALNIZOĞLU D., TOKATLI A., SİVRİ H. S. , HİŞMİ B., COŞKUN T., DURSUN A.  
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- XXVI. **Phenotypic and genotypic spectrum of Turkish patients with isovaleric acidemia**  
ÖZGÜL 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, vol.57, no.10, pp.596-601, 2014 (Journal Indexed in SCI)
- XXVII. **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.  
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- XXVIII. **A Patient With Pyruvate Carboxylase Deficiency and Nemaline Rods on Muscle Biopsy**  
Unal O., ORHAN D., Ostergaard E., TOKATLI A., DURSUN A., ÖZTÜRK HİŞMİ B., COŞKUN T., Wibrand F., Kalkanoglu-Sivri H. S.  
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- XXIX. **Cobalamin C defect: a patient of late-onset type with homozygous p. R132\*mutation**  
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TURKISH JOURNAL OF PEDIATRICS, vol.55, no.6, pp.633-636, 2013 (Journal Indexed in SCI)
- XXX. **Vanishing White Matter With Hepatomegaly and Hypertriglyceridemia Attacks**  
Unal O., ÖZGEN MOCAN B., ORHAN D., TOKATLI A., Hismi B. O. , DURSUN A., COŞKUN T., Kalkanoglu-Sivri H. S.  
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- XXXI. **Molecular and clinical evaluation of Turkish patients with lysinuric protein intolerance**  
Guzel-Ozanturk A., ÖZGÜL R. K. , Unal O., Hismi B., Aydin H. I. , Sivri S., TOKATLI A., COŞKUN T., Aksoz E., DURSUN A.  
GENE, vol.521, no.2, pp.293-295, 2013 (Journal Indexed in SCI)
- XXXII. **Multisystem fatal infantile disease caused by a novel homozygous EARS2 mutation**  
TALİM B., Pyle A., Griffin H., Topaloglu H., TOKATLI A., Keogh M. J. , Santibanez-Koref M., Chinnery P. F. , Horvath R.  
BRAIN, vol.136, 2013 (Journal Indexed in SCI)
- XXXIII. **MOLECULAR CHARACTERISATION OF BIOTINIDASE GENE MUTATIONS IN TURKISH PATIENTS; AN UPDATE OF THE RESULTS**

- Karaca M., Yucel D., Unal O., Guzel A., TOKATLI A., COŞKUN T., DURSUN A., SİVRİ H. S. , ÖZGÜL R. K.  
JOURNAL OF INHERITED METABOLIC DISEASE, vol.35, 2012 (Journal Indexed in SCI)
- XXXIV. **PROPIONIC ACIDEMIA PRESENTING WITH PERSISTENT PULMONARY HYPERTENSION IN TWO NEONATES**  
Hismi B., TEKŞAM Ö., Unal O., Takci S., Ertugrul İ., SİVRİ H. S. , DURSUN A., TOKATLI A., COŞKUN T.  
JOURNAL OF INHERITED METABOLIC DISEASE, vol.35, 2012 (Journal Indexed in SCI)
- XXXV. **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.  
TURKISH JOURNAL OF PEDIATRICS, vol.54, no.4, pp.409-412, 2012 (Journal Indexed in SCI)
- XXXVI. **Identification of Mutations and Evaluation of Cardiomyopathy in Turkish Patients with Primary Carnitine Deficiency**  
KILIÇ M., ÖZGÜL R. K. , COŞKUN T., Yucel D., KARACA M. A. , SİVRİ H. S. , TOKATLI A., ŞAHİN M., KARAGÖZ T., DURSUN A.  
JIMD REPORTS - CASE AND RESEARCH REPORTS, 2011/3, vol.3, pp.17-23, 2012 (Journal Indexed in SCI)
- XXXVII. **MUTATION ANALYSIS IN ARSB GENE IN TURKISH PATIENTS WITH MPS TYPE VI: HIGH PREVALENCE OF L321P MUTATION**  
ÖZGÜL R. K. , Karaca M., SİVRİ H. S. , TOKATLI A., COŞKUN T., DURSUN A.  
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- XXXVIII. **A novel mutation in the DGUOK gene in a Turkish newborn with mitochondrial depletion syndrome**  
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- XXXIX. **Mutation Spectrum of Fumarylacetoacetase Gene and Clinical Aspects of Tyrosinemia Type I Disease**  
DURSUN A., ÖZGÜL R. K. , Sivri S., TOKATLI A., Guzel A., Mesci L., KILIÇ M., Aliefendioglu D., Ozcay F., Gunduz M., et al.  
JIMD REPORTS: CASE AND RESEARCH REPORTS, 2011/1, vol.1, pp.17-21, 2011 (Journal Indexed in SCI)
- XL. **IDENTIFICATION OF NOVEL MUTATIONS IN PROMOTER AND CODING REGIONS IN ALDOB GENE CAUSING HEREDITARY FRUCTOSE INTOLERANCE**  
Yucel D., ÖZGÜL R. K. , COŞKUN T., SİVRİ H. S. , TOKATLI A., DURSUN A.  
JOURNAL OF INHERITED METABOLIC DISEASE, vol.34, 2011 (Journal Indexed in SCI)
- XLI. **VITAMIN B6 AND B12 STATUS IN TURKISH CHILDREN WITH PHENYLKETONURIA**  
Buyuktuncer Z., GOKMEN-OZEL H., KUCUKKASAP T., KOKSAL G., KILIÇ M., DURSUN A., KALKANOGLU-SIVRI H. S. , TOKATLI A., COŞKUN T., BESLER H. T.  
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- XLII. **BLOOD PHENYLALANINE CONTROL IN TURKISH PHENYLKETONURIC CHILDREN**  
GOKMEN-OZEL H., Buyuktuncer Z., KOKSAL G., KILIC M., DURSUN A., KALKANOGLU-SIVRI H. S. , TOKATLI A., COSKUN T.  
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- XLIII. **GROWTH AND PROTEIN INTAKE IN PHENYLKETONURIA: RESULTS OF 398 TURKISH CHILDREN**  
GOKMEN-OZEL H., Buyuktuncer Z., KOKSAL G., KILIÇ M., DURSUN A., KALKANOGLU-SIVRI S., TOKATLI A., COŞKUN T.  
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- XLIV. **Mucopolysaccharidosis Type IIID: 12 New Patients and 15 Novel Mutations**  
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- XLV. **The myopathic form of coenzyme Q10 deficiency is caused by mutations in the electron-transferring-flavoprotein dehydrogenase (ETFDH) gene**  
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- XLVI. **Acetaminophen-induced hepatotoxicity in a glutathione synthetase-deficient patient**  
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- XLVII. **Assessment of tetrahydrobiopterin-responsiveness in Turkish hyperphenylalaninemic patients**  
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- XLVIII. **Phenylketonuria in pediatric neurology practice: A series of 146 cases**  
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- XLIX. **Haematological findings in children with inborn errors of metabolism**  
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- L. **Molecular Analysis of Turkish Mucopolysaccharidosis IVA (Morquio A) Patients: Identification of Novel Mutations in the N-acetylgalactosamine-6-sulfate sulfatase (GALNS) Gene**  
Terzioglu M., TOKATLI A., COŞKUN T., Emre S.  
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## Articles Published in Other Journals

- I. **Evaluation of Cardiac Findings in Mucopolysaccharidosis Type III Patients**  
BİLGİNER GÜRBÜZ B., AYPAR E., ALEHAN D., TOKATLI A., COŞKUN T., DURSUN A., SİVRİ H. S.  
JOURNAL OF PEDIATRIC RESEARCH, vol.8, no.2, pp.195-201, 2021 (Journal Indexed in ESCI)

## Books & Book Chapters

- I. **Identification of mutations and evaluation of cardiomyopathy in Turkish patients with primary carnitine deficiency.**  
KILIÇ M., ÖZGÜL R. K. , COŞKUN T., YÜCEL YILMAZ D., KARACA M., SİVRİ H. S. , TOKATLI A., ŞAHİN M., KARAGÖZ T., DURSUN A.  
in: JIMD Reports Case and Research Reports 2011 3, , Editor, SPRINGER, 2011

## Refereed Congress / Symposium Publications in Proceedings

- I. **Hyperphenylalaninemia due to novel JCDNA12 mutation**  
SİVRİ H. S. , ÇIKI K., YÜCEL YILMAZ D., GÜRSES CİLA H. E. , ÖZGÜL R. K. , TOKATLI A., COŞKUN T., DURSUN A.  
SSIEM 2019: Annual Symposium of the Society for the Study of Inborn Errors of Metabolism, Rotterdam, Netherlands, 3 - 06 September 2019, vol.42, pp.324
- II. **Acute Metabolic Decompensations of Branched-Chain Organic Acidemias in the Pediatric Emergency Department: Clinical Presentation and Outcomes**  
SİVRİ H. S. , YILDIZ Y., AKCAN L., PEKTAŞ E., BİLGİNER GÜRBÜZ B., DURSUN A., TOKATLI A., COŞKUN T., TEKŞAM Ö.  
13th International Congress of Inborn Errors of Metabolism, 5 - 08 September 2017
- III. **Three-year experience of pediatric physicians with adult inpatient consultations**  
YILDIZ Y., PEKTAŞ E., BİLGİNER GÜRBÜZ B., DURSUN A., TOKATLI A., COŞKUN T., SİVRİ H. S.  
13th International Congress of Inborn Errors of Metabolism (ICIEM), Rio de Janeiro, Brazil, 5 September - 08 May 2017
- IV. **The clinical, biochemical features, and mutational analyses in glutaric acid type 1 patients**  
BİLGİNER GÜRBÜZ B., YILDIZ Y., GOKSOY E., YÜCEL YILMAZ D., DURSUN A., TOKATLI A., COŞKUN T., SİVRİ H. S.

- V. **Etilmelonik ensefalopati: vaka sunumu**  
PEKTAŞ E, Yoldaş T. Ç. , BİLGİNER GÜRBÜZ B., YILDIZ Y., DURSUN A., SİVRİ H. S. , TOKATLI A., COŞKUN T.  
XIV. Ulusal Metabolik Hastalıklar ve Beslenme Kongresi, Turkey, 26 - 30 April 2017
- VI. **Erişkinlerde kalıtsal metabolik hastalıklar: yatan hasta konsültasyonları ile üç yıllık deneyim**  
YILDIZ Y., PEKTAŞ E., BİLGİNER GÜRBÜZ B., DURSUN A., TOKATLI A., COŞKUN T., SİVRİ H. S.  
XIV. Ulusal Metabolik Hastalıklar ve Beslenme Kongresi, Turkey, 26 - 30 April 2017
- VII. **Tedavi Almayan Hiperfenilalaninemili Çocuklarda Nörokognitif Fonksiyonların Değerlendirilmesi: İlk Sonuçlar**  
PEKTAŞ E., Evinc G., FOTO ÖZDEMİR D., Karaboncuk Y., BİLGİNER GÜRBÜZ B., YILDIZ Y., TOKATLI A., COŞKUN T., Öktem F., SİVRİ H. S.  
14. Ulusal Metabolik Hastalıklar ve Beslenme Kongresi, Turkey, 26 - 30 April 2017
- VIII. **Fenilketonürlü bireylerde besin gruplarının enerji, protein ve fenilalanin alımına katkısı**  
YILMAZ Ö., BİLGİNER GÜRBÜZ B., YILDIZ Y., GÖKSOY E., DURSUN A., SİVRİ H. S. , TOKATLI A., COŞKUN T., GÖKMEN ÖZEL H.  
XIV Ulusal Metabolik Hastalıklar ve Beslenme Kongresi, Muğla, Turkey, 26 - 30 April 2017, pp.150
- IX. **Fenilketonürlü bireylerde diyet enerji ve protein alımlarının antropometrik ölçümlere etkisi var mıdır?**  
YILMAZ Ö., BİLGİNER GÜRBÜZ B., YILDIZ Y., GÖKSOY E., DURSUN A., SİVRİ H. S. , TOKATLI A., COŞKUN T., GÖKMEN ÖZEL H.  
XIV Ulusal Metabolik Hastalıklar ve Beslenme Kongresi, Muğla, Turkey, 26 - 30 April 2017, pp.149
- X. **Fenilketonürlü bireylerde beslenme ve diyet hasta destek programının değerlendirilmesi**  
GÖKMEN ÖZEL H., YILMAZ Ö., YILDIZ Y., GÖKSOY E., BİLGİNER GÜRBÜZ B., DURSUN A., SİVRİ H. S. , TOKATLI A., COŞKUN T.  
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- XI. **Fenilketonürlü bireylerde Türkiye'ye Özgü Beslenme Rehberi'ne göre enerji ve bazı besin öğeleri alımının değerlendirilmesi**  
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