

## Prof. SERAP SİVRİ

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### Education Information

Post Doctorate of Medicine, Hacettepe University, Tıp, Çocuk Sağlığı Ve Hastalıkları, Turkey 1996 - 2012

Expertise In Medicine, Hacettepe University, Çocuk Sağlığı Ve Hastalıkları Anabilim Dalı, Çocuk Metabolizma Hastalıkları, Turkey 1991 - 1996

### Research Areas

Medicine, Health Sciences, Internal Medicine Sciences, Child Health and Diseases

### Academic Titles / Tasks

Professor, Hacettepe University, Tıp Fakültesi, Dahili Tıp Bilimleri Bölümü, 1991 - Continues

### Courses

Rikest, Undergraduate, 2017 - 2018

Fenilketonüri, Undergraduate, 2017 - 2018, 2016 - 2017

Metabolik hastalıklarda hipoglisemi, Undergraduate, 2017 - 2018, 2016 - 2017

Hypoglycemia in IEM, Undergraduate, 2017 - 2018, 2016 - 2017

Protein energy malnutrition, Undergraduate, 2017 - 2018, 2016 - 2017

Büyümenin değerlendirilmesi, Undergraduate, 2017 - 2018, 2016 - 2017

Phenylketonuria, Undergraduate, 2017 - 2018, 2016 - 2017

Problem based learning, Undergraduate, 2017 - 2018, 2016 - 2017

Probleme dayalı öğrenim, Undergraduate, 2017 - 2018, 2016 - 2017

Mukopolisakkaridoz, Undergraduate, 2017 - 2018

Mucopolysaccharidosis, Undergraduate, 2017 - 2018

Obesity and its prevention, Undergraduate, 2017 - 2018, 2016 - 2017

Obezite ve korunma yolları, Undergraduate, 2017 - 2018

Growth and its assessment, Undergraduate, 2017 - 2018

Rickets, Undergraduate, 2016 - 2017

Lizozomal depo hastalıkları : MPS örneği, Undergraduate, 2016 - 2017

Makronütrient eksiklikleri, Undergraduate, 2016 - 2017

Protein enerji malnütrisyonu, Undergraduate, 2016 - 2017

Obesite ve korunma yolları, Undergraduate, 2016 - 2017

Lysosomal storage disease: MPSs , Undergraduate, 2016 - 2017

## Published journal articles indexed by SCI, SSCI, and AHCI

- I. **COVID-19 in inherited metabolic disorders: Clinical features and risk factors for disease severity**  
KAHRAMAN A. B., YILDIZ Y., ÇIKI K., Erdal I., AKAR H. T., DURSUN A., TOKATLI A., SİVRİ S.  
Molecular Genetics and Metabolism, vol.139, no.2, 2023 (SCI-Expanded)
- II. **Successful management of rhabdomyolysis with triheptanoin in a child with severe long-chain 3-hydroxyacyl-coenzyme A dehydrogenase (LCHAD) deficiency**  
KAHRAMAN A. B., YILDIZ Y., GÖKMEN ÖZEL H., KADAYIFÇILAR S., SİVRİ S.  
Neuromuscular Disorders, vol.33, no.4, pp.315-318, 2023 (SCI-Expanded)
- III. **Splenic Gaucheroma Leading to Incidental Diagnosis of Gaucher Disease in a 46-Year-Old Man with a Rare GBA Mutation: A Case Report**  
Erdal İ., YILDIZ Y., ÖNAL G., AKTEPE O. H., Düzgün S. A., Sağlam A., DÖKMECİ S., SİVRİ H. S.  
Endocrine, Metabolic and Immune Disorders - Drug Targets, vol.23, no.2, pp.230-234, 2023 (SCI-Expanded)
- IV. **Expert-opinion-based guidance for the care of children with lysosomal storage diseases during the COVID-19 pandemic: An experience-based Turkey perspective**  
Akgun A., Gokcay G., Mungan N. O., Sivri H. S., TEZER H., Zeybek C. A., EZGÜ F. S.  
Frontiers in Public Health, vol.11, 2023 (SCI-Expanded)
- V. **Efficacy of Tele-CO-OP in Children With Organic Acidemia: A Pilot Randomized Controlled Trial**  
Dursun E. L., BUMİN G., ÇIKI K., SİVRİ S.  
OTJR Occupation, Participation and Health, 2023 (SSCI)
- VI. **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.  
JOURNAL OF PEDIATRIC ENDOCRINOLOGY & METABOLISM, vol.35, no.11, pp.1345-1356, 2022 (SCI-Expanded)
- VII. **Management of early treated adolescents and young adults with phenylketonuria: Development of international consensus recommendations using a modified Delphi approach**  
Burton B. K., Hermida A., Belanger-Quintana A., Bell H., Bjoraker K. J., Christ S. E., Grant M. L., Harding C. O., Huijbregts S. C. J., Longo N., et al.  
MOLECULAR GENETICS AND METABOLISM, vol.137, no.1-2, pp.114-126, 2022 (SCI-Expanded)
- VIII. **Does glutaric aciduria type 1 affect hearing function?**  
ÖZGEDİK D., TOKGÖZ YILMAZ S., BİLGİNER GÜRBÜZ B., SİVRİ H. S., SENNAROĞLU G.  
METABOLIC BRAIN DISEASE, vol.37, no.6, pp.2121-2132, 2022 (SCI-Expanded)
- IX. **Recommendations on phenylketonuria in Turkey**  
COŞKUN T., ÇOKER M., Mungan N. O., GÖKMEN ÖZEL H., SİVRİ H. S.  
TURKISH JOURNAL OF PEDIATRICS, vol.64, no.3, pp.413-434, 2022 (SCI-Expanded)
- X. **A non-interventional observational study to identify and validate clinical outcome assessments for adults with phenylketonuria for use in clinical trials**  
Burton B. K., Skalicky A., Baerwald C., Bilder D. A., Harding C. O., Ilan A. B., Jurecki E., Longo N., Madden D. T., SİVRİ H. S., et al.  
MOLECULAR GENETICS AND METABOLISM REPORTS, vol.29, 2021 (SCI-Expanded)
- XI. **Long-term efficacy and safety of sapropterin in patients who initiated sapropterin at <4 years of age with phenylketonuria: results of the 3-year extension of the SPARK open-label, multicentre, randomised phase IIIb trial**  
Muntau A. C., Burlina A., Eyskens F., Freisinger P., Leuzzi V., SİVRİ H. S., Gramer G., Pazdirkova R., Cleary M., Lotz-Havla A. S., et al.  
ORPHANET JOURNAL OF RARE DISEASES, vol.16, no.1, 2021 (SCI-Expanded)
- XII. **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 (SCI-Expanded)

- XIII. **Complicated peripartum course in a patient with very long-chain acyl-coenzyme A dehydrogenase (VLCAD) deficiency**  
AKAR H. T., Çağan M., YILDIZ Y., SİVRİ H. S.  
NEUROMUSCULAR DISORDERS, vol.31, no.6, pp.566-569, 2021 (SCI-Expanded)
- XIV. **Clinical characteristics and journey to diagnosis in patients with mucopolysaccharidosis type VII**  
SİVRİ H. S., ERDÖL Ş.  
MOLECULAR GENETICS AND METABOLISM, vol.132, no.2, 2021 (SCI-Expanded)
- XV. **Sensory, voluntary, and motor postural control in children and adolescents with mucopolysaccharidosis**  
YİĞİT Ö., AKSOY S., Akyol U., TOKATLI A., SİVRİ H. S.  
JOURNAL OF PEDIATRIC ENDOCRINOLOGY & METABOLISM, vol.34, no.5, pp.583-589, 2021 (SCI-Expanded)
- XVI. **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 (SCI-Expanded)
- XVII. **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 (SCI-Expanded)
- XVIII. **Consensus guideline for the diagnosis and treatment of tetrahydrobiopterin (BH4) deficiencies**  
Opladen T., Lopez-Laso E., Cortes-Saladelafont E., Pearson T. S., SİVRİ H. S., YILDIZ Y., Assmann B., Kurian M. A., Leuzzi V., Heales S., et al.  
ORPHANET JOURNAL OF RARE DISEASES, vol.15, no.1, 2020 (SCI-Expanded)
- XIX. **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 (SCI-Expanded)
- XX. **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 (SCI-Expanded)
- XXI. **Inborn errors of metabolism in the differential diagnosis of fatty liver disease**  
YILDIZ Y., SİVRİ H. S.  
TURKISH JOURNAL OF GASTROENTEROLOGY, vol.31, no.1, pp.3-16, 2020 (SCI-Expanded)
- XXII. **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 (SCI-Expanded)
- XXIII. **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 (SCI-Expanded)
- XXIV. **Maternal phenylketonuria in Turkey: outcomes of 71 pregnancies and issues in management**  
YILDIZ Y., SİVRİ H. S.  
EUROPEAN JOURNAL OF PEDIATRICS, vol.178, no.7, pp.1005-1011, 2019 (SCI-Expanded)
- XXV. **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 (SCI-Expanded)
- XXVI. **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 (SCI-Expanded)
- XXVII. **Oral health status in patients with mucopolysaccharidoses**  
BALLIKAYA E., Eymirli P. S., YILDIZ Y., AVCU N., SİVRİ H. S., Uzamis-Tekcicek M.  
TURKISH JOURNAL OF PEDIATRICS, vol.60, no.4, pp.400-406, 2018 (SCI-Expanded)
- XXVIII. **Evaluation of spinal involvement in children with mucopolysaccharidosis VI: the role of MRI**  
Bulut E., Pektas E., SİVRİ H. S., BİLGİNER B., UMAROĞLU M. M., Ozgen B.  
BRITISH JOURNAL OF RADIOLOGY, vol.91, no.1085, 2018 (SCI-Expanded)
- XXIX. **Hyperinsulinemic Hypoglycemia in Congenital Disorder of Glycosylation Type-1a (CDG-1a)**  
Vuralli D., YILDIZ Y., SİVRİ H. S., ALİKAŞİFOĞLU A.  
HORMONE RESEARCH IN PAEDIATRICS, vol.90, pp.362-363, 2018 (SCI-Expanded)
- XXX. **Efficacy, safety and population pharmacokinetics of sapropterin in PKU patients < 4 years: results from the SPARK open-label, multicentre, randomized phase IIIb trial**  
Muntau A. C., Burlina A., Eyskens F., Freisinger P., De Laet C., Leuzzi V., Rutsch F., SİVRİ H. S., Vijay S., Bal M. O., et al.  
ORPHANET JOURNAL OF RARE DISEASES, vol.12, 2017 (SCI-Expanded)
- XXXI. **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 (SCI-Expanded)
- XXXII. **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 (SCI-Expanded)
- XXXIII. **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 (SCI-Expanded)
- XXXIV. **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.  
EUROPEAN JOURNAL OF PEDIATRICS, vol.174, no.8, pp.1077-1084, 2015 (SCI-Expanded)
- XXXV. **Prevalence of Anti-Adeno-Associated Virus Serotype 8 Neutralizing Antibodies and Arylsulfatase B Cross-Reactive Immunologic Material in Mucopolysaccharidosis VI Patient Candidates for a Gene Therapy Trial**  
FERLA R., CLAUDIANI P., SAVARESE M., KOZARSKY K., PARINI R., SCARPA M., DONATI M. A., SORGE G., HOPWOOD J. J., PARENTI G., et al.  
HUMAN GENE THERAPY, vol.26, no.3, pp.145-152, 2015 (SCI-Expanded)
- XXXVI. **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.  
Turkish Journal Of Pediatrics, vol.57, pp.388-393, 2015 (SCI-Expanded)
- XXXVII. **Mucopolysaccharidosis: Otolaryngologic findings, obstructive sleep apnea and accumulation of glucosaminoglycans in lymphatic tissue of the upper airway**  
GONULDAS B., YILMAZ T., SİVRİ H. S., GÜÇER K. Ş., KILINC K., GENÇ G. A., KILIÇ M., COŞKUN T.  
INTERNATIONAL JOURNAL OF PEDIATRIC OTORHINOLARYNGOLOGY, vol.78, no.6, pp.944-949, 2014 (SCI-Expanded)
- XXXVIII. **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)
- XXXIX. **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 (SCI-Expanded)

- XL. **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 (SCI-Expanded)
- XLI. **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 (SCI-Expanded)
- XLII. **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 (SCI-Expanded)
- XLIII. **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 (SCI-Expanded)
- XLIV. **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.  
JOURNAL OF INHERITED METABOLIC DISEASE, vol.34, 2011 (SCI-Expanded)
- XLV. **A novel mutation in the DGUOK gene in a Turkish newborn with mitochondrial depletion syndrome**  
KILIÇ M., SİVRİ H. S., DURSUN A., TOKATLI A., De Meirleir L., Seneca S., AKÇÖREN Z., YİĞİT Ş., Topaloglu H., COŞKUN T.  
TURKISH JOURNAL OF PEDIATRICS, vol.53, no.1, pp.79-82, 2011 (SCI-Expanded)

## Articles Published in Other Journals

- I. **Difficulties Associated with Enzyme Replacement Therapy for Mucopolysaccharidoses**  
YILDIZ Y., SİVRİ H. S.  
TURKISH ARCHIVES OF PEDIATRICS, vol.56, no.6, pp.602-609, 2021 (ESCI)
- II. **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 (ESCI)
- III. **Infant Acute Lymphoblastic Leukemia with Atypical Presentation**  
YAMAN BAJİN H. İ., YILDIZ Y., AKIN Ş., AYTAÇ EYÜPOĞLU Ş. S., ÜNAL CANGÜL Ş., KUŞKONMAZ B. B., Cetin M., SİVRİ H. S., GÜMRÜK F.  
Acta Medica, vol.50, no.4, pp.57-59, 2019 (Peer-Reviewed Journal)
- IV. **Mukopolisakkaridozlarda ortopedik sorunlar**  
YILDIZ Y., SİVRİ H. S.  
TOTBİD Dergisi, vol.15, no.4, pp.303-310, 2016 (Peer-Reviewed Journal)
- V. **Kalıtısal Metabolik Hastalıklarda Dental Bulgular**  
YILDIZ Y., SİVRİ H. S.  
Türkiye Klinikleri Çocuk Diş Hekimliği - Özel Konular, vol.2, no.2, pp.28-33, 2016 (Peer-Reviewed Journal)
- VI. **Rizomelik kondrodizplazi punktata: Bir vaka takdimi**  
ZENGİN AKKUŞ P., TAKCI Ş., UTİNE G. E., SİVRİ H. S., YURDAKÖK M.  
Çocuk Sağlığı ve Hastalıkları Dergisi, vol.56, no.4, pp.188-191, 2013 (Scopus)

## Books & Book Chapters

- I. **Nörotransmitter Bozukluklarında Klinik Yaklaşım**  
Yıldız Y., Sivri H. S.  
in: Kalıtsal Metabolik Hastalıklarda Hareket Bozuklukları, Dursun, Ali, Editor, Türkiye Klinikleri Yayınevi, Ankara, pp.20-29, 2020
- II. **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. **Listening Parents Caring A Child With Phenylketonuria**  
ZENGİN AKKUŞ P., BİLGİNER GÜRBÜZ B., ÇIKI K., İLTER BAHADUR E., KARAHAN S., ÖZMERT E. N., COŞKUN T., SİVRİ H. S.  
3rd International Developmental Pediatrics Association Congress, 9 - 12 December 2019
- II. **Fenilketonüri ile Yaşamak: Anne Ve Babaların Penceresinden**  
ZENGİN AKKUŞ P., BİLGİNER GÜRBÜZ B., ÇIKI K., İLTER BAHADUR E., KARAHAN S., ÖZMERT E. N., COŞKUN T., SİVRİ H. S.  
63. Türkiye Milli Pediatri Kongresi, Turkey, 30 October - 03 November 2019
- III. **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
- IV. **THE BURDEN OF ILLNESS IN ADULTS WITH PHENYLKETONURIA (PKU): INTERIM ANALYSIS OF A CROSS-SECTIONAL STUDY CONDUCTED IN NORTH AMERICA AND EUROPE**  
Burton B., Longo N., Maillot F., Rahman Y., Singh R., SİVRİ H. S., Stuy M., Vockley J., Van Backle J., Jha A., et al.  
41st Annual Meeting of the Society-for-Inherited-Metabolic-Disorders (SIMD), Washington, United States Of America, 6 - 09 April 2019, vol.126, pp.293
- V. **Oxysterol levels as oxidative stress biomarkers in organic acidemia patients**  
Eraslan Y., Lay İ., Samadi A., Gürbüz B., DURSUN A., SİVRİ H. S., COŞKUN T.  
SSIEM 2018: Annual Symposium of the Society for the Study of Inborn Errors of Metabolism, 04 September 2018 - 07 September 2017, vol.41, pp.37-219
- VI. **Mukopolisakkaridozlu (MPS) Çocuklarda Otomatik ve İstemli Motor Kontrol Performanslarının İncelenmesi: Pilot Çalışma**  
YİĞİT Ö., AKSOY S., SİVRİ H. S.  
13. Uluslararası Kulak Burun Boğaz ve Baş Boyun Cerrahisi Kongresi, 5 - 07 April 2018, pp.52
- VII. **Presenting signs and symptoms of MPS: Results of an international physician survey**  
Mitchell J., Clarke L., Ellaway C., Foster H., Giugliani R., Goizet C., Goring S., Hawley S. M., Jurecki E., Khan Z., et al.  
We're Organizing Research for Lysosomal Diseases (WORLD) Symposium, California, United States Of America, 5 - 09 February 2018, vol.123
- VIII. **Presenting signs and symptoms of MPS: Results of a systematic literature analysis**  
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