BİLİMSEL PROGRAM - SÖZEL SUNUM PROGRAMI

Tarih: 11 Nisan 2019 **Saat:**13:30-15:00 **Salon:** B Salonu	
Oturum Başkanları: Yaşar Cesur, Erdoğan Soyuçen	
Sıra No Bildiri No	**Bildiri Başlığı - Sunan Kişi**
SB-1 b-96	**First Case Report of Methylmalonic Acidemia with Combined Liver/Kidney Transplantation** **Sunan Kişi:** Neslihan Önenli Mungan
SB-2 b-177	**Growth Hormone Treatment: Reverses Catabolic Process in Inborn Errors of Metabolism** **Sunan Kişi:** Aslı İnci
SB-3 b-170	**Mitochondrial Neurogastrointestinal Encephalomyopathy (MNGIE): Our Clinical Experience and a Novel Mutation** **Sunan Kişi:** Esra Er
SB-4 b-106	**Genotypic and Phenotypic Features of 673 Phenylketonuria Patients in Çukurova University** **Sunan Kişi:** Deniz Kör
SB-5 b-186	**Galactosemia Among Turkish Children: Presentation and Outcome from a Pediatric Metabolism Center** **Sunan Kişi:** Beyza Belde Doğan
SB-6 b-193	**Effect of Whole Exome and Mitochondrial DNA Sequencing in Diagnosis of 70 Patients Referred to Çukurova University with a Suspicion of an Inherited Metabolic Disease** **Sunan Kişi:** Sebile Kılavuz
SB-42 b-90	**A Rare Neurodegenerative Metabolic Disorder: Evaluation of Patients With L-2-hydroxyglutaric Aciduria** **Sunan Kişi:** Senanur Şanlı
SB-43 b-36	**Immunological Evaluation of Patients with Organic Acidemia** **Sunan Kişi:** İlayda Uslu
SB-9 b-199	**Glycogen Storage Disease Type Ib And Amyloidosis: a Cause Of Proteinuria** **Sunan Kişi:** Dilek Güneş

Tarih: 11 Nisan 2019 **Saat:**16:30-17:30 **Salon:** B Salonu	
Oturum Başkanları: Gürsel Biberoğlu, Halil İbrahim Aydın	
Sıra No Bildiri No	**Bildiri Başlığı - Sunan Kişi**
SB-10 b-94	**Current Dietary Status of 61 Adult Phenylketonuria Patients** **Sunan Kişi:** Tuğçe Kartal
SB-11 b-153	**With or Without A Diet Ten Years Experience of A Tertiary Center on Bh4 Responsive Phenylketonuria Patients** **Sunan Kişi:** Ece Öge Enver
SB-12 b-183	**Liver Transplantation in Inherited Metabolic Diseases: To Whom and When?** **Sunan Kişi:** Ece Öge Enver
SB-46 b-195	**Clinical and Biochemical Characterization of Patients With 3-Methylcrotonyl-Coa-Carboxylase Deficiency** **Sunan Kişi:** Dilek Güneş
SB-14 b-202	**Nonketotic Hyperglycinemia: Outcome of Patients from a Single Center** **Sunan Kişi:** Seda Güneş
SB-15 b-181	**Clinical Presentation and Follow Up of Patients with Lysinuric Protein Intolerance** **Sunan Kişi:** Zerengiz Bayramlı

Tarih: 11 Nisan 2019 **Saat:** 17:30-18:50 **Salon:** B Salonu	
Oturum Başkanları: Mehmet Nuri Özbek, İlyas Okur	
Sıra No Bildiri No	**Bildiri Başlığı - Sunan Kişi**
SB-16 b-32	**Studying the Effect of Large Neutral Amino Acid Supplements on Oxidative Stress in Phenylketonuric Patients** **Sunan Kişi:** Burcu Kumru
SB-17 b-161	**Reduced Bone Mineral Density in Children With Maple Syrup Urine Disease** **Sunan Kişi:** Burcu Kumru
SB-18 b-144	**Cognitive Assessment of 342 Hyperphenylalaninemia Patients Followed-up in Çukurova University Pediatric Metabolism Department** **Sunan Kişi:** Sibel Öz
SB-19 b-130	**Determining the Knowledge Levels of Parents of Phenyketonurea Patients and Affecting Factors** **Sunan Kişi:** Fatma Selda Hızel Bülbül
SB-20 b-197	**Evaluation of The Phenylalanine Tolerance for Genotype-Phenotype Correlation in P.I48s Mutation for Phenylketonuria** **Sunan Kişi:** Gizem Akın Uslu
SB-21 b-62	**Investigation of Oxidative / Nitrosative Stress in Phenylketonuria and Maple Syrup Urine Diseases** **Sunan Kişi:** Mehmet Keskin
SB-22 b-203	**Citrin Deficiency: The Efficacy of Dietary Treatment** **Sunan Kişi:** Aslı Dudaklı
SB-23 b-103	**Rainbow of Inherited Metabolic Diseases: A Report of Seven Complex CDG Cases** **Sunan Kişi:** Sebile Kılavuz

BİLİMSEL PROGRAM - SÖZEL SUNUM PROGRAMI

Tarih: 13 Nisan 2019 **Saat:** 08:00-09:00 **Salon:** B Salonu

Oturum Başkanları: Işıl Özer, Hasan Önal

Sıra No Bildiri No	Bildiri Başlığı - Sunan Kişi
SB-24 b-38	**Bone Mineral Density in Patients with Cerebrotendinous Xanthomatosis at the Time of Diagnosis** **Sunan Kişi:** Pelin Teke
SB-25 b-43	**Clinical Findings of Cases with Intrahepatic Bile Duct Disease: A One-Year Single Center Experience (Case Series)** **Sunan Kişi:** Hasret Ayyıldız Civan
SB-26 b-71	**Three Cases of Cerebrotendinous Xanthomatosis** **Sunan Kişi:** Mehmet İlker Yön
SB-27 b-72	**Evaluation of Primary Hypertriglyceridemia Patients: Ethiology, Phenotype, Treatment** **Sunan Kişi:** Emel Aytaç
SB-28 b-138	**Outcomes of Patients Referred by Family Physicians Due to Dyslipidemia: Single Center Experience** **Sunan Kişi:** Engin Köse
SB-29 b-198	**Familial Hypercholesterolemia: Factors Associated with Diagnosis and Age at Diagnosis in Children** **Sunan Kişi:** Gülşah Kavrul Kayaalp

Tarih: 13 Nisan 2019 **Saat:** 09:00-10:00 **Salon:** B Salonu

Oturum Başkanları: Ali Dursun, Nesrin Karabul

Sıra No Bildiri No	Bildiri Başlığı - Sunan Kişi
SB-30 b-45	**Fabry Disease: From Gene To Genetic Counseling** **Sunan Kişi:** Özlem Sezer
SB-31 b-50	**Clinical and Molecular Characterization of 10 Patients with Fabry Disease** **Sunan Kişi:** Asburce Olgaç
SB-32 b- 154	**Seventy-Six Fabry Patients Clinical and Genetical Analysis: One Center Experience** **Sunan Kişi:** Ece Öge Enver
SB-33 b-162	**Could Targeted Next Generation Sequencing Be A First Line Diagnostic Method for Lysosomal storage Diseases?** **Sunan Kişi:** Filiz Başak Cengiz
SB-34 b-168	**The Evaluation of Patients with Diagnosis of Infantile, Juvenile and Late-Onset Pompe Disease: A Retrospective Study** **Sunan Kişi:** Beyza Belde Doğan
SB-35 b-188	**Identification of Key Genes by Bioinformatics Analysis in Infantile-Onset Pompe Patients** **Sunan Kişi:** Selçuk Güler

Tarih: 13 Nisan 2019 **Saat:** 10:30-11:30 **Salon:** B Salonu

Oturum Başkanları: Halil Sağlam, İkbal Süheyla Altay

Sıra No Bildiri No	Bildiri Başlığı - Sunan Kişi
SB-36 b-148	**Determining Nutritional Risks of Hospitilized Patients and Their Risk Perceptions** **Sunan Kişi:** Fatma Selda Hızel Bülbül
SB-37 b-85	**The Relationship Between Breastfeeding Times and the Success of Children in the 7-10 Age Range** **Sunan Kişi:** Büşra Yılmaz
SB-38 b-58	**Epileptic and Non Epileptic Abnormal Movements Due to Nutritional Cobalamin Deficiency in Infancy** **Sunan Kişi:** Ahmet Yaramış
SB-39 b-84	**Melanocortin-4 Receptor Gene Mutations in Obese Children and Adolescents** **Sunan Kişi:** Gülcan Seymen Karabulut
SB-40 b-5	**Assessment of Malnutrition and Nutritional Status of Hospitalized and Treated Children Aged between 12 and 60 Months** **Sunan Kişi:** Burcu Kumru
SB-41 b-33	**Assessment of Nutrition Status in Maple Syrup Urine Disease Patients** **Sunan Kişi:** Burcu Kumru

Tarih: 13 Nisan 2019 **Saat:** 14:30-15:30 **Salon:** B Salonu

Oturum Başkanları: Mustafa Kendirci, Fatma Tuba Eminoğlu

Sıra No Bildiri No	Bildiri Başlığı - Sunan Kişi
SB-7 b-28	**Clinical and Molecular Characterization of 11 Turkish Patients with PMM2-CDG** **Sunan Kişi:** Yılmaz Yıldız
SB-8 b-29	**Spectrum of Multiple Acyl-Coa Dehydrogenase Deficiency from Neonatal Period to Adulthood** **Sunan Kişi:** Yılmaz Yıldız
SB-44 b-15	**Adaptive Functions of Children with Organic Acidemias** **Sunan Kişi:** Ezgi Özalp Akın
SB-45 b-152	**The Evaluation Of Glutaric Aciduria Type 1 Patients Which Were Diagnosed And Followed At Harran Univercity Medical Faculty Hospital Between January 2017 And December 2018** **Sunan Kişi:** Meryem Karaca
SB-13 b-30	**Clinical and Laboratory Predictors of Acute Metabolic Decompensation in Children with Maple Syrup Urine Disease** **Sunan Kişi:** Yılmaz Yıldız
SB-47 b-201	**L2-Hydroxyglutaric Aciduria: Clinical and Biochemical Evaluation of 33 Patients from a Single Center** **Sunan Kişi:** Nebahat Ceyda Bayraktar Eltutan

BİLİMSEL PROGRAM - SÖZEL SUNUM PROGRAMI

Tarih: 13 Nisan 2019 **Saat:** 16:00-17:00 **Salon:** B Salonu	
Oturum Başkanları: Çiğdem Seher Kasapkara, Fatih Kardaş	
Sıra No **Bildiri No**	**Bildiri Başlığı - Sunan Kişi**
SB-48 **b-115**	**An Alternative Method for Diagnosing Inborn Metabolic Diseases; Capillary Electrophoresis** **Sunan Kişi:** Mehmet Şerif Cansever
SB-49 **b-8**	**Evaluation of Respiratory Manifestations in Inherited Metabolic Diseases: Six-Year Single-Center Experience** **Sunan Kişi:** Nisa Eda Cullas Ilarslan
SB-50 **b-131**	**Investigation of Lysosomal Acid Lipase Activity in 3D Cell Culture Model** **Sunan Kişi:** Kübranur Kaplan
SB-51 **b-134**	**Rare Metabolic Diseases in Etiology of Early Infantile Epileptic Encephalopathies: Yield of Array CGH and Whole Exome Sequencing Analysis** **Sunan Kişi:** Güneş Sağer
SB-52 **b-14**	**Whole Mitochondrial Genome Screening in Children with Suspected Mitochondrial Disease** **Sunan Kişi:** Emine Begüm Gencer Öncül
SB-53 **b-137**	**Metabolik Hastaliklara Bağli Akut Karaciğer Yetmezlikli Olgularimiz; 10 Yillik Tek Merkez Deneyimi** **Sunan Kişi:** Burcu Güven