BİLİMSEL PROGRAM - POSTER SUNUM PROGRAMI

Tarih: 12 Nisan 2019 **Saat:** 18:30-19:30	
Konu Başlığı: Aminoasidopatiler	
Başkanlar: Nur Arslan, Ertuğrul Kıykım	
Sıra No Bildiri No	**Bildiri Başlığı - Sunan Kişi**
P-1 b-39	**Argininemia: Four Siblings Case Report Sunan Kişi:** Meltem Gümüş
P-2 b-46	**An accidentally discovered non-classical Homocystinuria(MTHFR) Family Sunan Kişi:** Maryem İsmail
P-3 b-51	**Nags Deficiency; A Novel Mutation and Nine Months Follow-Up Sunan Kişi:** Banu Kadıoğlu Yılmaz
P-4 b-54	**MSUD; Three Different Clinic and the Use of Sodium Benzoate/Sodium Phenylacetate Sunan Kişi:** Banu Kadıoğlu Yılmaz
P-5 b-56	**A Case Report: Skeletal Dysplasia in a Patient With Maternal Phenylketonuria Sunan Kişi:** Dilara Kıyak
P-6 b-60	**Nitisinon Experience in Alkaptonuria Patients Sunan Kişi:** Ümmü Alakuş Sarı
P-7 b-73	**Presentation and Management of Urea Cycle Disorders Sunan Kişi:** Çiğdem Seher Kasapkara
P-8 b-95	**Carnitine Deficiency in Cystinosis: Report of 17 Patients Sunan Kişi:** Deniz Kör
P-9 b-114	**Molybdenum Cofactor Deficiency: Report of a Patient with a New Homozygous Mutation in MOCS2 Gene Sunan Kişi:** Mehmet Çalkan
P-10 b-142	**Classic Phenylketonuria (PKU) Responsive To Tetrahydrobiopterin (BH4) Sunan Kişi:** Albandari Alsaban
P-11 b-150	**A Very Rare Disease: Hyperornithinemia-Hyperammonemia-Homocitrullinuria (hhh) Syndrom Sunan Kişi:** Ekin Özsaydı
P-12 b-151	**Two Siblings Two Different Mutation in One Gene: Ornitin Transcarbamylase Deficiency Sunan Kişi:** Ece Öge Enver
P-13 b-155	**A Late-Diagnosed Phenylketonuria Case Presenting with the Autism Spectrum Disorder Sunan Kişi:** Günel Yusifova
P-14 b-156	**Essential Fatty Acids in Phenylketonuria and Their Relations with Clinical Findings Sunan Kişi:** Özlem Ünal Uzun
Konu Başlığı: Aminoasidopatiler	
Başkanlar: Halil Sağlam, Hasan Önal	
P-15 b-166	**The Assessment of Patients with Gyrate Atrophy; Results of a Single Center Sunan Kişi:** Beyza Belde Doğan
P-16 b-167	**Ornithine Transcarbamylase Deficiency Mimicking Glycogen Storage Disease, Case Report Sunan Kişi:** Hanım Babazade
P-17 b-175	**Lysinuric Protein Intolerance in Differential Diagnosis of Hemophagocytic Lymphohistiocytosis Sunan Kişi:** Şule Bektaş
P-18 b-180	**The Luckiest of the Urea Cycle Disorders: N-acetylglutamate Synthase Deficiency Sunan Kişi:** Beste Akdeniz
P-19 b-182	**Clinical and Molecular Features of Seven Patients with Classical Homocystinuria Sunan Kişi:** Havva Yazıcı
P-20 b-184	**Tyrosinemia Type III: Outcome in Two Cases Sunan Kişi:** Havva Yazıcı
P-21 b-185	**Ornithine Aminotransferase Deficiency: Report of Three Cases Sunan Kişi:** Havva Yazıcı
P-22 b-192	**Late Diagnosed Patient With Lysinuric Protein Intolerance Sunan Kişi:** Duygu Ülger Işık
P-23 b-204	**Lysinuric Protein Intolerance: Follow-Up In Pregnancy Sunan Kişi:** Medis Çöllü
P-24 b-212	**Hyperornithinemia-Hyperammonemia-Homocitrullinuria Syndrome In a Turkish Patient Sunan Kişi:** Mustafa Kılıç
Konu Başlığı: Organik Asidemiler	
P-25 b-68	**Determination of Methylmalonic Acid in Urine by Capillary Electrophoresis with Capacitively Coupled Contactless Conductivity Detection Sunan Kişi:** Sirun Özçelik
P-26 b-113	**Acrodermatitis Dysmetabolica in an Infant with Propionic Acidemia Sunan Kişi:** Gonca Kılıç Yıldırım
P-27 b-117	**A Rare Metabolic Disease That May be Considered as Cyclic Vomiting Syndrome: Isovaleric Acidemia Sunan Kişi:** Songül Gökay
P-28 b-122	**A Confusing Case Report of a Methylmalonic Acidemia Sunan Kişi:** Rana İsmayilova

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Başkanlar: Sema Kalkan Uçar, Fatih Kardaş	
Sıra No Bildiri No	**Bildiri Başlığı - Sunan Kişi**
P-29 b-133	**3-Methylcrotonyl-Coa Carboxylase Deficiency Presented with Chest Pain** **Sunan Kişi:** Saffa Ahmadzada
P-30 b-157	**Essential Fatty Acids in Organic Acidemia and Their Relations with Clinical Findings** **Sunan Kişi:** Özlem Ünal Uzun
P-31 b-158	**Late Onset Glutaric Aciduria Type 1: Case Presentations** **Sunan Kişi:** Gülzar Alishbaylı
P-32 b-165	**The Report of The Case Followed with Propionic Acidemia and Applied Liver Transplantation Succesfully** **Sunan Kişi:** Ahmet Onur Yiğit
P-33 b-191	**Ethylmalonic Encephalopathy with Nephrotic Syndrome** **Sunan Kişi:** Emine Ülgen
Konu Başlığı: YAO defektleri	
P-34 b-6	**Evaluation of Clinical Characteristics of Patients with Glutaric Aciduria Type IIc** **Sunan Kişi:** Pelin Teke Kısa
P-35 b-18	**A Novel Mutation Leading to Lethal Form of Carnitine Palmitoyltransferase Type-2 Deficiency and Magnetic Resonance Imaging Findings** **Sunan Kişi:** Sevil Dorum
P-36 b-26	**Three New Cases of Carnitine Palmitoyl-transferase II Deficiency with the Homozygous Point Mutation S113L** **Sunan Kişi: Çiğdem Seher Kasapkara**
P-37 b-34	**Clinical Features and ACADVL Gene Mutation Spectrum Analysis of 4 Turkish Patients with Very Long Chain Acyl-CoA Dehydrogenase Deficiency** **Sunan Kişi: Çiğdem Seher Kasapkara**
P-38 b-35	**Eosinophilia and Systemic Primary Carnitine Deficiency** **Sunan Kişi:** Engin Köse
P-39 b-100	**D-Bifunctional Protein Deficiency: A Case Report of a Turkish Child** **Sunan Kişi:** Neslihan Özcan
P-40 b-111	**Three Carnitine-Acylcarnitine Translocase Deficiency with c.270Delc and Novel c.408C>A Variant** **Sunan Kişi:** Berrak Bilginer Gürbüz
P-41 b-147	**A Case Report of Mitochondrial Short-Chain Enoil-CoA Hydratase Deficiency: Diagnosis and Follow-up** **Sunan Kişi:** Ayşe Ergül Bozacı
P-42 b-189	**Carnitine Palmitoyltransferase 1 Deficiency: A Novel Missense Mutation with Corpus Callosum Dysgenesis** **Sunan Kişi:** Senem Ayça

Konu Başlığı: YAO defektleri	
Başkanlar: Yaşar Cesur, Çiğdem Aktuğlu Zeybek	
P-43 b-171	**Hyperammonemia Due to Carnitine-Acylcarnitine Translocase Deficiency** **Sunan Kişi:** Esra Er
P-44 b-200	**Isobutyryl-Coa Dehydrogenase Deficiency: A Rare Disease Detectable by Tandem Mass Spectrometry** **Sunan Kişi:** Zehra Aslı İşeri Küskü
Konu Başlığı: Mitokondriyal	
P-45 b-27	**Pyruvate Carboxylase Ceficiency in a Child With an Early Diagnosis of Ketolysis Defect** **Sunan Kişi: A**sburce Olgac
P-46 b-17	**A Novel Pus1 Mutation in Two Siblings with Mlasa Syndrome and a Review of Literature** **Sunan Kişi: Ü**mmühan Öncül Demircan
P-47 b-129	**A Novel FBXL4 Mutation in a Turkish Child with Mitochondrial Dna Depletion Syndrome Type 13 and Literature Of Review** **Sunan Kişi:** Ümmühan Öncül Demircan
P-48 b-55	**A Novel Rars2 Mutation in Two Siblings With Microcephaly, Seizures and Liver Involvement** **Sunan Kişi:** Selin Sevinç
P-49 b-63	**A Patient Presented with Coarse Face Diagnosed as Combined Oxidative Phosphorylation Deficiency-3** **Sunan Kişi:** Huseyin Bilgin
P-50 b-76	**A Rare Case Presentation: A Novel Mutation in GTPBP3 Gene** **Sunan Kişi: D**ilek Çavuşoğlu
P-51 b-92	**MPV17-related Hepatocerebral Mitochondrial DNA Depletion Syndrome: Report of Three Cases** **Sunan Kişi: B**erna Şeker Yılmaz
P-52 b-102	**A Patient with Phenotypic Characteristics of Mitochondrial Disorders: 3-Hydroxyisobutyryl-CoA Hydrolase Deficiency** **Sunan Kişi:** Hüseyin Bilgin
P-53 b-104	**Rare Combination of Oculo Cutameous Albinism and Megdel Syndrome in One Patient** **Sunan Kişi:** Ayşe Aysima Özçelik
P-54 b-126	**A Rare Cause of Vision Loss in Childhood: Two Case Reports with Leber's Hereditary Optic Neuropathy** **Sunan Kişi:** Caner Hacıoğlu
P-55 b-128	**A Case of Genetically Diagnosed Leigh Syndrome, Resembling Rohhad Syndrome** **Sunan Kişi:** Begüm Murt

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Konu Başlığı: Mitokondriyal	
Başkanlar: Halil İ. Aydın, Mehmet Gündüz	
Sıra No Bildiri No	**Bildiri Başlığı - Sunan Kişi**
P-56 b-52	**Mitochondrial DNA Depletion Syndrome-12B and Hypomagnesemia: Two Cases** **Sunan Kişi:** Banu Kadıoğlu Yılmaz
P-57 b-196	**Coenzym Q-10 Deficiency Due To Coq4 Gene Defect** **Sunan Kişi:** Dilek Güneş
Konu Başlığı: Enerji Metab Boz.	
P-58 b-20	**Clinical and Molecular Characterization of Patients with Fructose 1,6- Bisphosphatase Deficiency** **Sunan Kişi:** Çiğdem Seher Kasapkara
P-59 b-44	**Succinyl-CoA:3-oxoacid CoA Transferase Deficiency: An Atypical Case with a Novel Mutation** **Sunan Kişi:** Yılmaz Yıldız
P-60 b-53	**Presentation of Two Cases with Ketolysis Defects** **Sunan Kişi:** Ümmü Alakuş Sarı
P-61 b-164	**A Rare Cause of Life-Threatening Ketoacidosis: Succinyl-CoA:3-Ketoacid CoA Transferase Deficiency** **Sunan Kişi:** Berrak Bilginer Gürbüz
P-62 b-174	**Novel Mutation in FBP1 Gene Presenting with Recurrent Episodes of Vomiting in a Child** **Sunan Kişi:** Merve Emecen Şanlı
P-63 b-206	**β-ketothiolase deficiency in Three Turkish Patients** **Sunan Kişi:** Mustafa Kılıç
P-64 b-208	**Hyperammonemia Secondary to Mitochondrial HMG-Coa Synthase Deficiency** **Sunan Kişi:** Mustafa Kılıç
Konu Başlığı: GSD ve KH Metab. Boz.	
P-65 b-207	**Exon 1 Deletion Represents a Founder Mutation in Turkish Patients with Fructose 1-6 Bisphosphatase Deficiency** **Sunan Kişi:** Mustafa Kılıç
P-66 b-210	**PHGK2 Gene Mutation in a Turkish Siblings with Glycogen storage disorder IXc** **Sunan Kişi:** Mustafa Kılıç
P-67 b-211	**PHKB gene mutation in a Turkish Patient with Glycogen Storage Disorder IXb** **Sunan Kişi:** Mustafa Kılıç
P-68 b-47	**Fanconi-Bickel Syndrome: A Patient with Novel Mutation in SLC2A2 Gene** **Sunan Kişi:** Zumrut Arslan Gülten
P-69 b-7	**A Rare Cause of Foamy Cells in the Bone Marrow Aspiration: Glycogen Storage Disease Type IV** **Sunan Kişi:** Zümrüt Gülten Arslan

Konu Başlığı: GSD ve KH Metab. Boz.	
Başkanlar: Fatma Tuba Eminoğlu, Şahin Erdöl	
P-70 b-209	**Galaktokinase Deficiency: Missed Diagnosis in a Case with Early Presentation** **Sunan Kişi:** İrem Kalay
P-71 b-82	**Two Different Subtypes of Glycogen Storage Diseases in the Same Family** **Sunan Kişi:** Nurbanu Bilgin
P-72 b-98	**Phenotypic and Genotypic Features of Classic Galactosemia Patients in Osmangazi University** **Sunan Kişi:** Gonca Kılıç Yıldırım
P-73 b-121	**A GSD Type 1b Case with Atipic Clinic Presentation** **Sunan Kişi:** Aynur Küçükçongar Yavaş
P-74 b-139	**Phenotypic Heterogenity of Four Cases with Glycogen Storage Disease Type 1b** **Sunan Kişi:** Sultan Ceren Yıldırım
P-75 b-143	**Hereditary Fructose Intolerance: An Interesting Case Report with Liver Failure** **Sunan Kişi:** Ali Karakaş
P-76 b-149	**Cornelia de Lange Syndrome and Glycogen Storage Disease Together in a Patient** **Sunan Kişi:** Ayşe Kılıç
P-77 b-169	**Effects of High-Fat and Low Carbohydrate Diet in Patients with Glycogen Storage Disease Type III and Type IX: A Single Center Experience** **Sunan Kişi:** Beyza Belde Doğan
P-78 b-172	**Glycogen Storage Disease Type VI and IX: Ege University Experience** **Sunan Kişi:** Esra Er
P-79 b-21	**Glycogen Storage Disease Type IX: High Variability in Clinical Phenotype** **Sunan Kişi:** Çiğdem Seher Kasapkara
Konu Başlığı: Koles-safra asit-peroksiz	
P-80 b-24	**Zellweger Spectrum Disease with a Mutation in PEX6** **Sunan Kişi:** Çiğdem Seher Kasapkara
P-81 b-12	**Two Sibling Diagnosed with Cerebrotendinous Xanthomatosis: Case Report** **Sunan Kişi:** Abdurrahman Akgün
P-82 b-16	**Clinical Characteristics of Childhood Cerebral X-linked Adrenoleukodystrophy Patients and Early Outcomes of Hematopoietic Stem Cell Transplantation** **Sunan Kişi:** Asburçe Olgaç
P-83 b-48	**A Rare Case of Rhizomelic Chondrodysplasia Punctata Type 1 due to PEX7 Mutation** **Sunan Kişi:** Asburçe Olgaç

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Konu Başlığı: Koles-Safra Asit-Peroksiz	
Başkanlar: Mehmet Nuri Özbek, Mehmet Cihan Balcı	
Sıra No Bildiri No	**Bildiri Başlığı - Sunan Kişi**
P-84 b-64	**The Presence of Bilateral Cataract and Chronic Diarrhea: Cerebrotendinous Xanthomatosis Sunan Kişi:** Hüseyin Bilgin
P-85 b-86	**Delta (4)-3-Oxosteroid 5 Beta-Reductase Deficiency: A Rare Cause of Neonatal Cholestasis Sunan Kişi:** Merve Usta
P-86 b-97	**Cerebrotendinous Xanthomatosis: Diagnosis After Juvenile Cataract Sunan Kişi:** Gonca Kılıç Yıldırım
P-87 b-101	**Familial Hypobetalipoproteinemia: Clinical Presentation of a Family Sunan Kişi:** Işıl Çulha
P-88 b-107	**Alpha-methylacyl-CoA Racemase Deficiency: Report of an Adult Patient Sunan Kişi:** Deniz Kör
P-89 b-108	**A Rare Metabolic Disorder: Chanarin Dorfman Syndrome Sunan Kişi:** Burcu Tabakcı
P-90 b-112	**Patients with Hypertriglyceridemia Due to Lipoprotein Lipase and Apoc2 Deficiency Sunan Kişi:** Ebru Canda
P-91 b-118	**Clinical Profile of Two Siblings with Cerebrotendinous Xanthomatosis: A Novel Mutation in The CYP27A1 Gene Sunan Kişi:** Esra Er
P-92 b-120	**Evaluation of Elevated Serum Lipoprotein a in Children: Which Options Are Available in Treatment? Sunan Kişi:** Burcu Akbaba
P-93 b-127	**A Rare Cause of Fat Malabsorption in Children: Chylomicron Retention Disease Sunan Kişi:** Gökhan Tümgör
P-94 b-163	**Efficacy and Complications of Plasmapheresis in Two Patients with Familial Homozygous Hyperlipidemia Sunan Kişi:** Esin Eyican
Konu Başlığı: Beslenme	
P-95 b-109	**Sjögren-Larrson Syndrome: One Year Follow-Up with Fat-Restricted Diet Sunan Kişi:** Hatice Mutlu Albayrak
P-96 b-176	**A New Dietary Approach to a Case with Dgat 1 Enzyme Deficiency Sunan Kişi:** Hanim Şeyma Özmen

Konu Başlığı: Beslenme	
Başkanlar: İlyas Okur, Mehmet Şerf Cansever	
P-98 b-11	**The Effects of Nutritional Knowledge and Applications of Mothers Whose Child Aged Between 3 to 5 on Child's Nutritional Habits and Growth Curves Sunan Kişi:** Esma Uygur
P-99 b-31	**Initiation of Classic Ketogenic Diet in a 16-Year-Old Boy Newly Diagnosed with Glucose Transporter 1 Deficiency Syndrome Sunan Kişi:** Yılmaz Yıldız
P-100 b-40	**Does Body Mass Index Affect Lung Function in Patients with Cystic Fibrosis? Sunan Kişi:** Burcu Kumru
P-101 b-41	**Energy and Macronutrıent Intakes in Children with Cystic Fibrosis Sunan Kişi:** Burcu Kumru
P-102 b-42	**Malnutrıtıon in Children with Cystic Fibrosis Sunan Kişi:** Burcu Kumru
P-103 b-93	**Ketogenic Diet and Cognitive Development Sunan Kişi:** Tuğçe Kartal
Konu Başlığı: Lizozomal Depo Hastalık.	
P-104 b-13	**Two Cousins with Joint Stiffness: Mucolipidosis Type III Gamma Sunan Kişi:** Berrak Bilginer Gürbüz
P-105 b-123	**Early Clinical Findings of Infantile-Onset Pompe Disease: A Case Report Sunan Kişi:** Berrak Bilginer Gürbüz
P-106 b-19	**A Rare Cause of Protein Losing Enteropathy: Gaucher Disease Sunan Kişi:** Mehmet Akif Göktaş
P-107 b-61	**Double Curse: Co-existence of Morquio type a Disease (Mucopolysaccharidosis type IVA) and Phenylketonuria Sunan Kişi:** Kısmet Çıkı
P-108 b-70	**Neuronal Ceroid Lipofuscinosis Type 5 Caused by a Novel Mutation: A Case Report Sunan Kişi:** Gonca Bektaş
P-109 b-88	**Two Different Clinic Entities Related To The Same Enzyme Deficiency: Mucolipidosis Type II and Type III Sunan Kişi:** Nesrin Kaya
P-110 b-116	**Acute Noronopatic Type Gaucher : Case Report Sunan Kişi:** Abdurrahman Akgün
P-111 b-140	**Clinical Utility of Total Level of Globotriaosylsphingosine (Lyso-Gb3) and its Analogues in Diagnosing Fabry Disease Sunan Kişi: Fahad J Alharbi**

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Konu Başlığı: Lizozomal Depo Hastalıkları	
Başkanlar: Erdoğan Soyucen, Gürsel Biberoğlu	
Sıra No Bildiri No	**Bildiri Başlığı - Sunucu**
P-97 b-178	**Screening of Twelve Lysosomal Storage Diseases with LC-MS/MS in Gazi University Hospital: The First Results of Validation** **Sunan Kişi:** Aslı İnci
P-112 b-145	**Clinical Presentation and Follow Up of Patients with Infantile Krabbe: Single Center Experience** **Sunan Kişi:** Ayşe Ergül Bozacı
P-113 b-194	**Essential Aminoacid Deficiency is Life-threatining Situation: 3 Cases Under Dietary Treatment** **Sunan Kişi:** Meryem Karaca
P-114 b-187	**Early Infantile Period Niemann Pick Type C Cases** **Sunan Kişi:** Nafiye Emel Çakar
Konu Başlığı: Diğer	
P-115 b-1	**Acute Intermittent Porphyria and Drugs** **Sunan Kişi:** Gülbüz Sezgin
P-116 b-10	**A Rare Cause of Atypical Autism: Succinic Semialdehyde Dehydrogenase Deficiency** **Sunan Kişi:** Sevil Dorum
P-117 b-22	**MAN1B1-CDG with Increased Serum Transaminases** **Sunan Kişi:** Çiğdem Seher Kasapkara
P-118 b-23	**Marinesco-Sjögren Syndrome: Case Report** **Sunan Kişi:** Çiğdem Seher Kasapkara
P-119 b-25	**SRD5A3-CDG: a Patient with a Novel Variant** **Sunan Kişi:** Çiğdem Seher Kasapkara
P-120 b-57	**The Relation of Gestational Diabetes and Sarcopenia** **Sunan Kişi:** Mustafa Behcet Demirbas
P-121 b-59	**Severe Motor Mental Retardation with Microcepahly and Hypomyelination due to PYCR2 Gene variant in a Large Family** **Sunan Kişi:** Berrak Bilginer Gürbüz
P-122 b-81	**A Rare Case Mimicking Osteogenesis Imperfecta and Storage Disease-Like Clinic: Yunis-Varón Syndrome** **Sunan Kişi:** Berrak Bilginer Gürbüz
P-123 b-80	**The Importance of Screening Inborn Errors of Metabolism in Autism Spectrum Disorders: Case Report** **Sunan Kişi:** İnci Pinar Bilen
P-124 b-83	**Two Cases of Metabolic Myopathies: Glutaric Aciduria Type 2 and Mc Ardle Disease** **Sunan Kişi:** Lütfiye Şahin Keskin
P-125 b-89	**Hallervorden-Spatz Disease: Magnetic Resonance Imaging and Diagnostic Clues** **Sunan Kişi:** Şenanur Şanlı
Konu Başlığı: Diğer	
Başkanlar: Işıl Özer, Tanyel Zübarioğlu	
P-126 b-91	**Neurodegeneration with Brain Iron Accumulation Type-8: Two Siblings with a Novel Mutation** **Sunan Kişi:** Berna Şeker Yılmaz
P-127 b-99	**Cerebral Creatine Deficiency Syndrome: A Case Report** **Sunan Kişi:** Neslihan Özcan
P-128 b-105	**Comparison of Serum Homocysteine Levels in Active and Sedentary Young Adults** **Sunan Kişi:** Yakup Tuncer
P-129 b-124	**A Rare Bone Metabolism Disorder: Osteopetrosis Congenita** **Sunan Kişi:** Döndü Nilay Yıldırım
P-130 b-132	**Visibility of Rare Diseases and Newborn Screening in Turkish Instagram Posts** **Sunan Kişi:** Fatma Selda Hızel Bülbül
P-131 b-136	**Valproic Acid Induced Hyperammonemia and Treatment Management** **Sunan Kişi:** Elif Dede
P-132 b-141	**Tetrahydrobiopterin (BH4) Responsiveness Test Revealed 6 - Pyruvoyl - Tetrahydrobiopterin Synthase (PTPS) Deficiency in a Saudi Infant** **Sunan Kişi:** Albandari Alsaban
P-133 b-146	**DPAGT1-CDG (CDG-Ij)-Mild Form Case Report** **Sunan Kişi:** Ayşe Ergül Bozacı
P-134 b-159	**Novel Mutation in Two Siblings with Normouricemic Lesch Nyhan Syndrome** **Sunan Kişi:** Merve Emecen Şanlı
P-135 b-160	**Familial Hyperphosphatemic Tumoral Calcinosis in an Unusual Site** **Sunan Kişi:** Merve Emecen Şanlı
P-136 b-173	**Hyperinsulinemic Hypoglycemia: Think of Glud1 Gene Mutation Leading to Hyperinsulinism/Hyperammonemia (Hi/Ha) Syndrome** **Sunan Kişi:** Merve Emecen Şanlı
P-137 b-179	**A Rare Inborn Error of Metabolism: Aromatic L-Amino Acid Decarboxylase Deficiency** **Sunan Kişi:** Sabina Sharifova
P-138 b-205	**Adenylosuccinate Lyase Deficiency in a Turkish Siblings** **Sunan Kişi:** Mustafa Kılıç
P-139 b-49	**Porphyrias: What a Pediatric Metabolism Specialist Needs to Know ?** **Sunan Kişi:** İsmail Kurt