14. Disorders of fatty acid oxidation and ketone body metabolism

A-012 When a common symptom of a neonate becomes an unusual diagnosis: A case report of HMG-CoA lyase deficiency
C S Kasapkara, M Akar, G Biberoglu, M Celik, M N Özbek, H Tüzün

A-013 Neonatal onset VLCADD with a novel mutation
C S Kasapkara, G Baysoy, B Aldudak, M N Özbek, J M Nuoffer, C R Largiadèr

18. Other disorders of energy metabolism, creatine disorders

A-014 Red ragged fibres - A rare finding in muscle biopsies in children
S Sequeira, A C Ferreira, J P Vieira, I Castro, L Lopes, M Grazina, R Roque, T Evangelista

21. Peroxisomal, sterol and bile acid disorders

A-015 Treatment of X-linked childhood cerebral adrenoleukodystrophy by the use of bone marrow transplantation
C S Kasapkara, A Aksoy, M Acar, H G Tanyildiz, M Kilic, V Uygun, M A Yesilipek, S Senel

22. Lysosomal disorders: mucopolysaccharidoses, oligosaccharidoses

A-016 One-year enzyme replacement therapy results in Bulgarian patients with a severe form of Hunter disease
D Avdjieva-Tzavella, H Kathom, I Sinigerska, P Yaneva, R Marinov, I Litvinenko, D Zahariev, E Panteleeva, G Sivrieva, L Spasov, R Tincheva

A-017 Hurler syndrome in a two years old Indonesian girl
M Mexitalia, H P Kahayana, M Ratnasari

A-018 Early laboratory diagnosis and bone marrow transplantation in MPS type I: A case report
K El Moustafa, I Sinici Lay, O Unal, S Sivri, T Coskun

23. Lysosomal disorders: sphingolipidoses

A-019 Stability of miglustat in InOrpha® flavoured suspending excipient for compounding of oral solutions and suspensions
S Riahi, M Ambühl, J Stichler, D Bandilla

A-020 Novel mutation defined in a patient with attenuated form of Niemann-Pick disease
E Kiykim, C Aktuglu Zeybek, S Gunes, T Zubarioglu, A Aydin

A-021 Adult case of metachromatic leukodystrophy
B Bilginer Gurbuz, F Hasiyev, H K Karli Oguz, R Karabudak, H S Sivri

A-022 Important diagnostic clues for Sandhoff disease: Hyperacusis and cherry red macular spots
M Kilic, U Kaya, E Kilic

A-023 A Gaucher patient with portal hypertension: homozygosity for the double D409H+H255Q allele
M Kagnici, M Kose, E Canda, M Karakoyun, A Aykut, E Karaca, S Kalkan Ucar, H Onay, F Ozgenc, E Sozmen, F Ozkinay, M Coker

24. Lysosomal disorders: others

A-024 A novel mutation described in a Turkish patient with infantile neuronal ceroid lipofuscinosis
E Kiykim, C Aktuglu Zeybek, S Gunes, T Zubarioglu

29. Miscellaneous

A-025 Functional independence of Taiwanese children with Down syndrome
H Y Lin, S P Lin, C K Chuang, Y J Chen, R Y Tu, M R Chen, D M Niu

A-026 A child with combined chromosomal abnormality, mitochondrial dysfunction and disordered cobalamin metabolism
N N Kvitchataya, Y B Grechanina, A A Yanovska

A-027 Diagnosis of familial mediterranean fever masked by symptoms of chronic pancreatitis
O V Vasylieva, D V Katsapov

A-028 A case of a possible ciliopathy
S Sequeira, A C Ferreira, A P Serrão, J P Vieira