16. Mitochondrial disorders: nuclear encoded

P-265 Neonatal liver failure due to deoxyguanosine kinase deficiency: a report of 4 patients
A Tokadı, M Kilic, O Unal, B Hızlı, H S Sovt-Kalkanoglu, A Dursun, F Cosan

P-266 Dysphagia, malnutrition and gastrointestinal problems in carriers of the m.3243A>G mutation
H E E Zweers, P Last de, S Kneijt, J A M Smeitink, G J A Wante, M C H Janssen

P-267 Withdrawn
Panel-based Next Generation Sequencing: a powerful tool for genetic diagnostic testing of patients with mitochondrial disorders

P-268 Whole exome sequencing confirms Leigh syndrome in a patient showing little biochemical evidence of a mitochondrial disorder
M Nagstasa, J Li, J Zhang, F Xu, H Jiang, X Xu, W A Gold, L Riley, D R Thorburn, K Keating, Y Guo, H Hakenberg, J Christodoulou

P-269 Whole exome sequencing identifies novel compound heterozygous mutations in PNP1 in affected siblings with a mitochondrial phenotype
A Adladı, N Sobirea, W A Gold, L Riley, M Wilson, B Benoı, C Boehm, J Christodoulou

P-270 An early manifestation of LBSL (leukoencephalopathy with brainstem and spinal cord involvement and lactate elevation) syndrome, case description
E Y Grecianast, E P Zabdałlak

P-271 A new case with resistant hypoglycemia, hypertrophic cardiomyopathy, and encephalopathy due to mitochondrial TSFM gene defect
A Dursun, O F Gerdan, M Kızılkaya-Pehlivan, D Yılmaz-Yıldız, B Yıcenar, M Topçu, D Yılanızoğlu, S Yigit, D Orkan, M Sığıroğlu, R K Özgül

P-272 Identification of a novel mitochondrial RNA modifier (M101) gene mutation by exome sequencing analysis
R K Özgül, D Yıcenar, O F Gerdan, E Sendaroğlu, B Yıcenar, M Sığıroğlu, M Topçu, D Yılanızoğlu, A Dursun

P-273 A novel mitochondrial translation defect leading to deafness and cutis laxa is caused by mutations in a mitochondrial ribosomal protein subunit
M Mohamed, T Gardestich, D Dalayra, D Karrel, E Morava, R A Wevers

P-274 3-Methylglutaconic aciduria as a marker in mitochondrial encephalomyopathies
S Sequeira, A C Ferreira, A M Moreira, S B Wörtmann

P-275 Ethynalactone encephalopathy: a novel deletion mutation in the ETHIE1 gene
H I Aydin, V Tirant, H Kose, E Lamantia, F Aydin, F M Sonmez

P-276 Normal amount of complex III subunits in three patients with TTC19 deficiency