P-253 Short-term follow-up in Polish patients with VLCAD deficiency detected by MS/MS screening
J Sykut-Cegielska, A Kowalik, J Taybert, M Olszewski, T Polawski, A Sobczynska-Tomaszewska, K Czerska, B Radomyska, M Gizewska, H Romanowska, E Krzywinska-Zdeb, C Muller, L Kaluzny, J Walkowiak, M Dus-Zuchowska, D Korycinska-Chaaban, K Hozyasz, J Jaglowska, J Wierzba, M Lulek-Ciebiera

P-254 Medium-chain acyl-CoA dehydrogenase deficiency in a patient with a complex congenital heart defect
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P-255 Pilot experience of an external quality assurance for acylcarnitines in serum/plasma
P Ruiz-Sala, C Acquaviva, A Chabli, M G M De Sain-van der Velden, J Garcia-Villoria, M R Heiner-Fokkema, K Leckstrom, F Leifur, J Olesen, W Onkenhout, A Ribes, G J G Ruijter, C Vianey-Saban, B Merinero

P-256 Patients with medium-chain acyl-CoA dehydrogenase deficiency experience oxidative stress – a metabolomic study
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P-257 Favourable long-term evolution of ACAD9 deficient patient treated with riboflavin
C Galimberti, S Gasperini, A Brambilla, C Barboni, R Parini

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M R Heiner-Fokkema, F M Vaz, R Maatman, L Kluijtmans, F J Van Spronsen, D J Reijngoud

P-259 Mitochondrial dysfunction in isolated long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency
H Rocha, R Ferreira, V Almeida, R Vitorino, L Lopes, E Martins, C Nogueira, E Leão-Teles, F Amado, L Vilarinho

15. Disorders of pyruvate metabolism and the Krebs cycle

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M V Kanuka, A A Yanovska, Y B Grechanina, E P Zdubskaya

P-261 Ketone bodies: a therapeutic option to replace ketogenic diet in PDH deficiency?
F Habarou, N Bahi-Buisson, A Boutron, M T Abi-Warde, K H Le Quan Sang, A Brassier, C Broissand, B Chadefaux-Vekemans, C Ottolenghi, P De Lonlay

P-262 Ketogenic diet application in PDH deficiency during the course of 6 years
B Tumiene, J Grikiniene, R Samaitiene, A Utkus

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H Pavlu-Pereira, C Florindo, C Tomé, S Tack, M J Silva, I Tavares de Almeida, P Leandro, I Rivera, J B Vicente

P-264 The spectrum of pyruvate oxidation defects
W Sperl, L Fleuren, R Feichtinger, F Zimmermann, J Koch, E Maier, P Freisinger, H Prokisch, T B Haack, J Smeitink, J A Mayr

16. Mitochondrial disorders: nuclear encoded

P-265 Neonatal liver failure due to deoxyguanosine kinase deficiency; a report of 4 patients
A Tokatli, M Kilic, O Unal, B Hismi, H S Sivri-Kalkanoglu, A Dursun, T Coskun

P-266 Dysphagia, malnutrition and gastrointestinal problems in carriers of the m.3243A>G mutation
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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 Nafisinia, J Li, J Zhang, F Xu, H Jiang, X Xu, W A Gold, L Riley, D R Thorburn, B Keating, Y Guo, H Hakonarson, J Christodoulou

P-269 Whole exome sequencing identifies novel compound heterozygous mutations in PNPT1 in affected siblings with a mitochondrial phenotype
A Alodaib, N Sobreira, W A Gold, L Riley, M Wilson, B Bennetts, 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 Grechanina, E P Zdubskaya

P-271 A new case with resistant hypoglycemia, hypertrophic cardiomyopathy, and encephalopathy due to mitochondrial TSFM gene defect
A Dursun, Ö F Gerdan, M Kızılkan-Pehlivantürk, D Yılmaz-Yücel, B Yuceturk, M Topcu, D Yalnızoglu, S Yigit, D Orhan, M Sagiroglu, R K Ozgul

P-272 Identification of a novel mitochondrial-tRNA modifier (MTO1) gene mutation by exome sequencing analysis
R K Ozgul, D Yucel-Yilmaz, Ö F Gerdan, E Serdaroglu, B Yuceturk, M Sagiroglu, M Topcu, D Yalnizoglu, A Dursun

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M Mohamed, T Gardeitchik, D Dalloyaux, D Karall, E Morava, R A Wevers

P-274 3-Methylglutaconic aciduria as a marker in mitochondrial syndromes
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H I Aydin, V Tiranti, H Kose, E Lamantea, F Aydin, F M Sonmez

P-276 Normal amount of complex III subunits in three patients with TTC19 deficiency
R G Feichtinger, P Freisinger, F A Zimmermann, J Koch, C Rauscher, H Prokisch, T B Haack, U Ahting, H P Wagentristl, V Konstantopoulou, W Sperl, J A Mayr, E Maier