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P-196 Secondary brain creatine deficiency may lead to diagnosis of ornithine aminotransferase (OAT) deficiency
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P-198 CSF/serum glycine ratios in non-ketotic hyperglycinemia patients
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P-199 Maple syrup urine disease (MSUD) in Ireland: phenotypes, genotypes, and determinants of outcome
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P-200 A novel glycine decarboxylase gene mutation in a neonate with nonketotic hyperglycinemia
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P-203 An autosomal dominant oculocutaneous albinism with a novel *OCA2* mutation in a Korean family
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P-204 Case Report: The Hartnup-disease found by NMR spectroscopic investigations of urine
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P-205 L-carnitine supplementation decreases DNA damage in treated MSUD patients
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P-206 Marple Syrup Urine Disease - New insights from a French cohort
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P-207 Hereditary tyrosinemia type 1 in Algeria: National survey
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P-223 Mild citrullinemia in Turkish patients: Report of three patients
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A Daly, M Assoun, A Belanger-Quintana, S M Bernabei, S Bollhalder, D Cassiman, H Champion, H Chan, J Dalmau, F De Boer, N M De Jong-Ter Horst, A De Meyer, A Desloovere, A Dianin, M Dixon, K Dokoupil, S Dubois, S Evans, F Eyskens, A Faria, I Fasan, E Favre, F Feillet, A Fekete, M Ferreira Almeida, C Fitzachary, G Gallo, C Gingell, J Gribben, C Jankowski, R Janssen-Regelink, I Jones, C Jouault, K Kaalund Hansen, G E Kahrs, I L Kok, A Kowalik, C Laguerre, S Le Verge, R Lilje, C Maddalon, D Mayr, U Meyer, A Micciche, U Och, M Robert, J C Rocha, H Rogozinski, C Rohde, K Ross, I Sarrugia, A Schlune, K Singleton, E Sjoqvist, L H Stolen, A Terry, C Timmer, L Tomlinson, A Tooke, E Van Dam, K Vande Kerckhove, T A M Van Den Hurk, E M C Van Der Ploeg, M Van Driessche, M Van Rijn, A Van Teeffelen-Heithoff, A Van Wegberg, C Vasconcelos, H Vestergaard, I Vitoria, D Webster, F J White, L White, H Zweers, A MacDonald

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P-240 Branched-chain aminoacidopathy: Our experience in Ege University Faculty of Medicine
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P-242 Secondary haemophagocytic lymphohistiocytosis (HLH) and posterior reversible encephalopathy syndrome (PRES) in a patient with B12-responsive late-onset methylmalonic acidemia
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P-243 Two patients with cobalamin A deficiency and novel mutations
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P-245 High prevalence of movement disorders in four inborn errors of metabolism: considerable impact of involuntary movements on quality of life
A Kuiper, M A Coenen, H Eggink, M A J Tijssen, T J De Koning

P-246 Evaluation of the L-carnitine role on the urinary parameters of oxidative stress in maple syrup urine disease patients
G B G Guerreiro, C P Mescka, D M Coelho, A Monalissa, C S Dutra-Filho, M Wajner, C R Vargas

P-247 Impaired mitochondrial oxidative phosphorylation and oxidative damage are detected in brain, heart and muscle of a mouse model of propionic acidemia
L Gallego-Villar, A Rivera-Barahona, C Cuevas, M Sánchez-Aragó, A Guenzel, B Perez, M Barry, M A Martin-Casanueva, J M Cuezva, E Richard, L R Desviat

P-248 Treatment of extrapyramidal disorders in propionic aciduria (PA) and methylmalonic aciduria (MMA)
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P-249 Generation and characterization of iPS cells as a cellular model for propionic acidemia
E Richard, E Alonso-Barroso, M Ugarte, C Pérez-Cerdá, B Perez, L R Desviat

P-250 Chemokine production induced by 2-methylcitrate in 3D organotypic rat brain cell aggregates
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P-251 Excellent outcome of patient with congenital lactic acidosis with riboflavin supplementation
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P-252 Molecular mechanisms and interactions in methylmalonic aciduria due to MMAA deficiency
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P-253 Functional analysis of missense variants in D-2-hydroxyglutarate dehydrogenase gene
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P-254 Genetic diagnosis of cobalamin defects by next generation sequencing
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P-255 Biochemical and molecular analysis of Russian patients with methylmalonic aciduria
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P-256 Late-onset cardiac failure after liver transplantation in one patient with propionic acidemia
M C Nassogne, M F Vincent, R Reding, E M Sokal, C Barrea

P-257 Expression of disease causing isovaleryl-CoA dehydrogenase in *Escherichia coli* for characterization and stability studies
A W Mohsen, V Sharma, M V Amieva, A Karunanidhi, E Naylor, J Vockley

P-258 Molecular genetic characterization of 149 *mut* patients: the Basel/Zurich cohort
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P-259 Molecular genetics of maple syrup urine disease in Brazilian patients
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P-260 The first reported case of pregnancy in 3-methylglutaconic aciduria type I (3-MGA type 1)
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P K Hamilton, K Ryan, C Loughrey, N McStravick, D Cardy, G M Connolly

P-262 Identification of a novel mutation in start codon of *MMAB* gene in an Indian family with methylmalonic acidemia using targeted next generation sequencing and prenatal diagnosis
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P-263 Profound biotinidase deficiency: natural course of the disease and impact of treatment in adult patients
M Demirkol, E Çakar, D Güneş, M Karaca, M C Balcı, Ü Türkoğlu, I Ozer, G Gokcay

P-264 Organic acid disorders: Burden of disease in North India
C Kumari, S Ramji, S Kapoor

P-265 Isovaleric acidemia and nephronophthisis: A case report.
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P-266 A novel frameshift mutation of malonyl-CoA decarboxylase deficiency
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P-267 Functional characterization of novel variants found in *ASPA* gene associated with Canavan disease
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P-268 Living with intoxication-type inborn errors of metabolism - a qualitative analysis of focus group interviews with paediatric patients and their caregivers
N A Zeltner, M A Landolt, M R Baumgartner, S Lageder, J Quitmann, R Sommer, C Mühlhausen, A Schlune, S Scholl-Bürgi, D Karall, M Huemer

P-269 Expanding clinical phenotype in CMAMMA and new tools for fast metabolic diagnostics
M G M De Sain-van der Velden, M Van der Ham, J J Jans, G Visser, H C M Prinsen, N M Verhoeven-Duif, K L I Van Gassen, P M Van Hasselt

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P-271 Ethylmalonic encephalopathy: diagnosis of a turkish case after unknown death of two children in the family
N Onenli Mungan, D Kor, B Seker Yilmaz, D Bulut, G Mert, F Incecik, S Ceylaner

P-272 In vivo magnetic resonance spectrocopy in an adult case of L-2-hydroxyglutaric aciduria
M Ete, B Audoin, P Viout, M Guye, J Pelletier, E Kaphan

P-273 Inborn errors of cerebral L-lysine metabolism: different degradation pathways and the role of brain peroxisomes
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P-274 Anaplerotic therapy in propionic acidemia
N Longo, L B Price, E Gappmaier, N L Cantor, S L Ernst, C Bailey, M Pasquali

P-275 A treatable cause for childhood bulbar pulsy, Fazio-Londe disease.
W Eyaid, M Rifai, H Alem, T Hazwani

P-276 Malonic aciduria: marked clinical variability in a consanguineous family
B Sudrié-Arnaud, S Torre, A Goldenberg, F Kundul, C Brasse-Lagnel, O Duhamel, G S Salomons, E Tourancheau, S Marret

P-277 Expanding the phenotype in aminoacylase 1 (ACY1) deficiency: characterization of the molecular defect in a 63-year-old woman with generalized dystonia
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P-278 Oxidative stress in patients with 3-hydroxy-3-methylglutaric aciduria and the role of L-carnitine therapy
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P-280 A long term follow-up study of 8 individuals with asymptomatic propionic acidemia
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P-281 Ethylmalonic encephalopathy in an Indian boy: a report of mutations in *ETHE1* gene
S Bijarnia-Mahay, D Gupta, Y Shigematsu, S Yamaguchi, R Saxena, I C Verma

P-282 Mutation screening study in Turkish patients with L-2-hydroxyglutaric aciduria

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P-283 Mutation identification and prenatal diagnosis of biotinidase deficiency in Indian patients
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P-284 Organic Acidurias Diagnosis. A critical review of our experience
O Y Echeverri, J M Guevara, N F Pulido, Y A Ardila, E Espinosa, L Cabarcas, L N Correa, L A Barrera

P-285 Behavioural and emotional problems, intellectual impairment and health related quality of life in patients with organic acidurias and urea cycle disorders
D Jamiolkowsky, S Kölker, E M Glahn, I Barić, J Zeman, M R Baumgartner, C Mühlhausen, A Garcia Cazorla, F Gleich, G Haege, P Burgard - on behalf of the E-IMD consortium

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P-287 D-galactose alters behavioral and biochemical parameters in hippocampus and cerebellum of rats
A F Rodrigues, H Biasibetti, E F Sanches, P Pierozan, B S Zanotto, M Sebotaio, A T S Wyse

P-288 Alternative night-time nutrition regimens in glycogen storage disease type I: a controlled crossover study
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P-289 Features of functioning of humoral immunity in children with hepatic forms of glycogen storage disease (GSD).
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P-291 Polish patient diagnosed with glycogen synthase deficiency homozygous for newly identified mutation in *GYS2* gene
E Szymanska, U Watrobinska, D Rokicki, E Ciara, P Halat, R Ploski, A Tylki-Szymanska

P-292 Resting energy expenditure in children with glycogen storage diseases.
E Szymanska, E Ehmke vel Emczynska, D Rokicki, A Tylki-Szymanska, J Ksiazyk

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P-297 A novel mutation in the *ABCC8* gene causing a variable phenotype of impaired glucose metabolism in the same family
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P-299 Glucose tetrasaccharide biomarker in 24 hour urine collections from patients with glycogen storage disease IIIa
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P-300 A patient with a novel mutation in the *GALT* gene, and initially misdiagnosed with a Congenital Glycolysation Defect
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P-301 A patient with Fanconi Bickel syndrome and a novel mutation
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P-302 The diagnosis and monitoring of Galactosemia using NMR spectroscopy
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P-682 The diagnostic value of the vitamin B6 profile in epileptic encephalopathies
D Mathis, L Abela, M Albersen, C Bürer, H Hartmann, M Hersberger, N M Verhoeven-Duif, B Plecko

P-683 Wilson Disease. Further evidence of phenotypic heterogeneity in two sisters
A Fiumara, A SAPUPPO, V Brafa Musicoro, M B Lepori, G Loudianos, F Raudino, G Bertino

P-684 Identification of new protein members of the methionine synthase interactome
C Bassila, R Ghemrawi, J Flayac, J L Guéant, D Coelho

P-685 Cobalamin X (*HCFC1* deficiency) mimicking nonketotic hyperglycinemia with increased CSF glycine and methylmalonic acid-a case report
E Scalais, C Weitzel, L De Meirleir, G Maertens, T Shaikh, C Coughlin, M Swanson, M Friederich, S Gunter, D Helbling, J Wendt-Andrea, C Acquaviva-Bourdain, J Van Hove

P-686 6 novel mutations in Menkes disease in russian patients
E A Kamenets, N A Demina, N V Milovanova, E Y Zakharova

P-687 S-Sulfocysteine excitotoxicity provides the molecular basis of neurodegeneration in molybdenum cofactor deficiency
G Schwarz, A Kumar, B Dejanovic, M Semtner, D Fusca, S Arjune, P Kloppenburg, J C Meier, A A Abdel

P-688 MEDNIK syndrome: clinical and biochemical delineation of the response to long-term zinc therapy - A case report
D Martinelli, C Dionisi-Vici

P-689 New insights into the frequency of molybdenum cofactor deficiency from the Exome Aggegation Consortium
G Schwarz, D Lal, S Arjune, S Mayr, N Havarushka, P Nuernberg

P-690 Extensive characterization of 14 CblC (Cobalamin C)-defective patients: clinical signs and neurocognitive outcome, biological, molecular and ophthalmologic findings
M Paviolo, N Garcia, A Imbard, E Delouvrier, F Rigaudière, O Rigal, H Ogier de Baulny, J F Benoist, M Schiff

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D L Green, C Bleakly, C Hendriksz, L Thompson, S Kempshall, I Anand

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R Matalon, D Gessler, J Li, Q Su, G Gao

P-692 Energy expenditure in chilean children with maple syrup urine disease (MSUD)
K Campo, Pérez, V Cornejo, Espinoza, M G Castro, Chaves, V Hamilton, Viollier, J F Cabello, Andrade, C Arias, Pefaur

P-693 Application of whole exome sequencing to a rare inherited metabolic disease with neurological and gastrointestinal manifestations: a congenital disorder of glycosylation mimicking glycogen storage disease
H D Park, R Choi, H I Woo, B H Choe, S Park, Y Yoon, C S Ki, S Y Lee, J Song, J W Kim

P-694 Autism and inherited metabolic disease
M Ersoy

P-695 Qualitative urinary organic acid analysis: 10 years of quality control
V Peters, J Bonham, G F Hoffmann, C Scott, C D Langhans

P-696 Is long-term ketogenic diet treatment hepatotoxic for children with intractable epilepsy?
N Arslan, O Guzel, U Yılmaz, T Calik, Z Akisin

P-697 Genotype and phenotype analysis in Taiwanese patients with osteogenesis imperfecta
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P-698 Effect of purified α1-antitrypsin (AAT) on expression of AAT in normal (PiMM) and AAT deficient (PiZZ) primary human hepatocytes
A Karadagi, H Johansson, H Zemack, C Jorns, R Gramignoli, K Stokkeland, S Strom, B G Ericzon, G Nowak, E Ellis

P-699 Dihydrolipoamide dehydrogenase deficiency diagnosed by using new generation sequencing technology
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P-700 A metabolic disease as an underlying cause of a child psychiatric disorder: literature review
F Eyskens, A Simons

P-701 Juvenile hemochromatosis type 2b: a case report
Y Aydemir, A Yuce, B Talim, A Pietrangelo, G Hizal, B Berberoglu-Ates, H Demir, I N Saltik-Temizel, T Bayhan, H Ozen

P-702 Wasted time devasted children: Effect of war on inborn errors of metabolism
A C Aktuglu Zeybek, E Kiykim, T Zubarioglu, M S Cansever, H Cam, A Aydin

P-703 Molecular diagnosis of lactic acidosis by using a clinical exome-sequencing panel
R Navarrete, A Oyarzabal, A Vega, I Bravo-Alonso, B Merinero, C Pérez-Cerdá, M Ugarte, B Perez, P Rodríguez-Pombo

P-704 Molecular genetic study of congenital adrenal hyperplasia in Serbia: novel p.Leu129Pro and p.Ser165Pro *CYP21A2* gene mutations
M Stojiljkovic, I Milacic, M Barac, T Milenkovic, M Ugrin, K Klaassen, A Skakic, M Jesic, I Joksic, K Mitrovic, S Todorovic, S Vujovic, S Pavlovic

P-705 Clinical utility of a new metabolic NGS panel in diagnostic service
L Heptinstall, A Ghosh, H Schlecht, S Bhaskar, J Urquhart, A Broomfield, A Morris, E Jameson, B Schwahn, J Walter, S Ramsden, S Jones, S Banka

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P-707 Two years experience of selective screening for organic acidurias (OA) and amino acidopathies (AA) in Pakistani pediatric population
N Sherazi, A H Khan, A Jamil, R Mehar, F Jehan, N Yousufzai, L Jafri, I Siddiqui, F Ghani, B Afroze

P-708 Exome sequencing results in unknown genetic, metabolic/ neurometabolic disorders
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P-709 Therapeutic management in pediatric intensive care unit of inherited metabolic diseases of intoxication by proteins: retrospective study of 53 cases
L Abily-Donval, C Joffre, F Lesage, M Oualha, L De Saint Blanquat, S Renolleau, V Valayannopoulos, P De Lonlay, L Dupic

P-710 Prenatal diagnosis on 143 cases with organic and fatty acid diseases by biochemical and gene analyses with amniocentesis
S Yamaguchi, Y Hasegawa, R Bo, K Yamada, H Kobayashi, T Taketani, S Fukuda

P-711 *RARS*: a novel disease-causing gene for Pelizaeus–Merzbacher disease
M Nafisinia, N Sobreira, W A Gold, L G Riley, R Ouvrier, C Boehm, B Uhlenberg, J Christodoulou

P-712 Paeonol can stimulate SIRT1 via activation of the NAD+ salvage pathway
A B Potthast, A M Das

P-713 Neurometabolic disorders associated with early childhood epilepsy: A single center experience in Saudi Arabia
S Mohamed, E El Melegy, I Talaat, H Hosny, K K Abu-Amero

P-714 Do the depression and anxiety levels of parents differ in different Inherited Metabolic Disorders (IMD)?
S Bülbül, N A Arslan,Nur, M G Gündüz,Mehmet, Ö Ü Ünal, Özlem

P-715 Liver transplantation in paediatric patients with metabolic disease: 20 years' experience in Melbourne, Australia
J Yaplito-Lee, R Jones, H Peters, L Herd, A Boneh, W Hardikar

P-716 Parent coping and the behavioural outcomes of children diagnosed with inherited metabolic disorders
A Brown, L Crowe, V Anderson, A Boneh

P-717 A longitudinal, prospective, long-term registry of patients with hypophosphatasia
P Kishnani, C Langman, A Linglart, E Mornet, K Ozono, C Rockman-Greenberg, L Seefried, C Bedrosian, K P Fujita, A Cole, W Högler

P-718 Does Aicardi-Goutière Syndrome present with High Neopterin and Biopterin?
D H Shinde, A B Jalan, M M Joshi, M A Borugale, J M Mahakal, N D Sonalkar, R A Jalan, K V Kudalkar, S V Damle, R H Merchant

P-719 Dunnigan syndrome - an unusual cause of severe and premature cardiovascular events
S Sequeira, O Moldovan, A C Alves, A M Medeiros, A C Ferreira, M Bourbon

P-720 Next generation sequencing role in diagnosis of mitochondrial disease in Serbian patient
M Djordjevic, M Stojiljkovic, A Sarajlija, B Kecman, A Skakic, S Pavlovic

P-721 Inborn errors of metabolism in the molecular era: will biochemical analyses become a second-tier test?
F B Piazzon, F P Monteiro, L S A Costa, C Bueno, J P Kitajima, R L Borges, M Dellamano, E L Freitas, B A C Santos, I Corrêa, F Kok

P-722 Protective role of *CNDP1* genotype on renal survival in children with glomerulopathies
V Peters, M Kebbewar, B Janssen, G F Hoffmann, K Möller, S Wygoda, M Charbit, A Fernandes-Teixeira, T Urasinski, N Jeck, J Zschocke, C P Schmitt, F Schaefer, E Wühl

P-723 Can the same platform of TMS for newborn screening of amino acid and acylcarnitine be used for investigation of symptomatic patients? A 4-year-experience.
F B Piazzon, L R G Garcia, D Y Arita, M A M Iskandar, H H Martins, E N Silva, C Bueno, F Kok, S M Hadachi

P-724 A case of metaphyseal chondromatosis with high levels of D-2 hydroxyglutaric acid
J J O'Byrne, P E Fitzsimons, S Unger, J M Croft, P D Mayne, E Moylett, C Mc Donnell, S A Lynch

P-725 Low plasma aromatic L-amino acid decarboxylase (AADC) activity in children and adolescents with high functioning autism.
E J Footitt, P B Mills, D Skuse, I P Hargreaves, G Allen, S J R Heales, P T C Clayton

P-726 Designing tools for multiorgan-directed neonatal gene therapy in neuro-hepatotropic inherited metabolic diseases: interest of an ubiquitous promoter "elongation factor 1α"
J Baruteau, J Hanley, S M Buckley, P Gissen, S J Howe, R Karda, P B Mills, J Ng, D Perocheau, A A Rahim, N Suff, S N Waddington

P-727 Validation of an NGS-panel for routine diagnosis of lysosomal and peroxisomal disorders
M Blomqvist, J Lindgren, L Olsson, J E Månsson, J Asin Cayuela

P-728 Awareness amongst medical doctors of inherited metabolic disorders
S Bülbül, N A Arslan,Nur, O D Dursun, Osman, M G Gündüz,Mehmet, E D Dursun, Esra

P-729 Vacuolar storage material in a family with juvenile parkinsonism and mutations in *FBXO7*
E Serdaroglu, R K Ozgul, D Yalnizoglu, M Madeo, A Malandrini, E Klee, Y Li, T N Jepperson, K K Oguz, D Yucel-Yilmaz, M Patterson, M C Kruer, A Dursun

P-730 Value of Whole Exome Sequencing in the diagnosis of neurometabolic disorders with unusual clinical presentation
L A Selim, I G Mahmoud, J Gleeson, A Rolfs

P-731 Elevated pipecolic acid in cerebro-facial-ocular skeletal syndrome
D Trujillano, R Abou Jamra, O Brandau, J Berger, C Wiesinger, C Lampe, A Rolfs, M Scarpa

P-732 Rett-like clinical picture as an entry to detect inborn errors of neurotransmission
J Armstrong, S Vidal, N Brandi, E Gerotina, M O'Callaghan, M S Perez-Poyato

P-733 Neuroimaging findings in patients with GLUT1 deficiency syndrome
E A Larshina, S V Mikhailova, L P Andreeva, E Y Zakharova

P-734 A completely new approach to the diagnosis of inborn errors: development of a 450-gene (all metabolic disorders) next-generation sequencing panel
F Ezgu, B Ciftci, B Topcu, I Okur, A Inci, A Olgac, A Karaoglu, G Biberoglu, L Tumer, A Hasanoglu

P-735 *DNM1* mutations: from exercise-induced collapse syndrome to a metabolic cause of severe epileptic encephalopathy
C Lourenco, C Leprevost, M Almeida, L Nobre, W Marques Jr

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A-001 Withdrawn

02. Novel diagnostic/laboratory methods

A-002 Comparison of non-derivatization and derivatization tandem mass spectrometry research methods for analysis of amino acids, acylcarnitines, and succinylacetone in dried blood spots
V Thibert, X Xie, M Kozak

A-003 Direct and fully automated extraction/analysis of Dried Blood Spots (DBS)
S Gaugler

03. Newborn screening

A-004 Moved to P-736

A-005 The first index case with coinsidence of phenylketonuria, polysplenia syndrome and methylene tetrahydrofolate reductase mutation from newborn screening
I Ozer, S Çam, Ç Durakbaşa, A Kıral

A-006 Current state of newborn screening in Slovenia
A Smon, U Groselj, M Zerjav Tansek, A Bicek, A Oblak, M Zupancic, C Krzisnik, B Repic Lampret, S Murko, S Hojker, T Battelino

A-007 Newborn screening using the DBS-MS 500 from CAMAG
S Gaugler

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A-008 Intravenous and enteral ketogenic diet in a patient with cow milk allergy
A Desloovere, P Verloo

06. Phenylketonuria: general

A-009 Nutritional evaluation and metabolic profile in children affected by phenylketonuria (PKU): case-control study
F Moretti, E Verduci, S Paci, E Salvatici, J Zuvadelli, S Bellini, E Riva

A-010 Vitamin D levels in an adult PKU population
D L Green, M McLoughlin, S McDarby, S Ripley

A-011 Marmara metabolism group consensus report for large neutral amino acids
I Ozer, T Zübarioğlu, H Önal, G Gökçay, H Sağlam, Y Cesur, I Taş, A Aydın, M C Balcı, E Kıykım, Ç Aktuğlu Zeybek, M Demirkol

A-012 First day phenylalanine levels in the offspring may reflect maternal phenylketonuria
O Unal, G Celen

A-013 Moved to P-739 06. Phenylketonuria: General

A-014 Supporting patients with a congenital metabolic disease and their social environment: the development of an educational book for children and adolescents
A Boury

A-015 Metabolic group study for consensus development on the management of phenylketonuria
M Demirkol, M C Balcı, I Ozer, A C Aktuglu Zeybek, H Saglam, C Yasar, H Onal, E Kıykım, S Erdol, A Yesil, M S Cansever, T Zubarioglu, Ü Türkoğlu, G Gokcay

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A-016 Prevalence, frequency and phenotype of BH4 deficiencies identified in a Neonatal Screening Program for hyperphenylalaninemia
C A A Souza, R D L Soares, M R A Alves, V C Kanufre, R C Norton, A L P Starling, M J B Aguiar

08. Sulphur amino acid disorders

A-017 Trimethylaminuria (Fish odor syndrome) in a adolescent boy
M Kilic, P Celik-Babalioglu, B Tokgoz-Cuni

A-018 Withdrawn

09. Other amino acid disorders

A-019 Hereditary tyrosinemia type 1, effectiveness of early treatment
B Sadaoui, Z Arrada

A-020 Drug development for paediatric patients with inborn errors of metabolism (IEM) – A development program for an oral suspension of nitisinone for hereditary tyrosinemia type 1 (HT-1)
M Rudebeck, L Svensson, M Sahlberg, B Olsson

A-021 Clinical case report: maple syrup urine disease treatment
V C Kanufre, A F Cruz, J C C Lopes, M C Alves, E R Valadares

A-022 MSUD presenting as an episodic ataxia
P Verloo, H Verhelst

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A-023 Continuous renal replacement therapy for nenatal hyperammonemia due to ornithine transcarbamylase deficiency
J Y Han

A-024 Clinical and biochemical features, molecular diagnosis and management of a Moroccan female with *OTC* gene mutation
S Dahri, H Talbaoui, Y Kriouile, C Acquaviva, C Vianey-Saban, L Chabraoui

A-025 Recurrent vomiting and somnolence in a 18-month old girl: ornithine transcarbamylase deficiency due to de novo heterozygous mutation
S Grkovic, M Djordjevic, A Sarajlija, B Kecman

11. Organic acidurias: branched-chain

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M Z Habbal

A-027 Patient with MSUD presenting with diabetic ketoacidosis
A Olgac, I Okur, E D Akbas, E Doger, F S Ezgu, A Bideci, L Tumer

A-028 Propionic acidemia and humoral immune deficiency
C F M Souza, F O Poswar, L F Refosco, A Sitta, M Wajner, M S L Jobim, A N R Taniguchi, F P Vairo, I V D Schwartz

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M Ersoy, S Yılmaz

A-030 A novel mutation for L-2 hydroxyglutaric aciduria in a 7 year old patient
A Olgac, L Tumer, F Ezgu, G Biberoglu, A Hasanoglu

A-031 Evaluating laboratory performance in external quality assessment schemes for urine organic acids by GCMS
L Jafri, N Shirazi, A Jamil, F Jehan, R Mehar, N Yousufzai, B Afroze, A H Khan

A-032 Ethylmalonic encephalopathy with a homozygous mutation in C.554T>G in the exon 5 of *ETHE1* gene
S Lougani, L Moussa, K Neggazi, A SAADI, M Chaoch, V Tiranti, W Ameur Elkhedoud, M Bendib

A-033 The Acute Management in Intercurrent illness of Isovaleric Acidemia Associated with Typhoid Fever
N A Widjaja, E Puspitaningtyas, R Irawan

A-034 Associated Glutaric aciduria type 1 and celiac disease
P Kuyum, S Cakar, Y Ozturk

13. Carbohydrate disorders

A-035 Clinical, laboratory data, molecular features, and follow up of 3 Iranian GSD type IIIa patients
T Zaman, S Moarefian, F Saeed Tehrani, R Moradian

A-036 Galactosemia case with a novel mutation
M Kilic, A Zenciroglu, E Goksun, D Y Yilmaz, R K Ozgul

A-037 Genotype-phenotype characteristics of Turkish patients with glycogen storage disease type I at a single centre
M Kilic, P Ozbudak, E Demir, Ç S Kasapkara, H G Tanyildiz, C Aytekin, F Özbay-Hosnut, G Sahin, K Karaer, S Ceylaner

A-038 Hepatic glycogen storage diseases in Macedonian patients: A single centre experience
A Kostovski, N Zdraveska

A-039 Successful dietary management of severe hyperlipidemia in patients with glycogen storage disease type 1
B Kecman, M Djordjevic, A Skakic, M Stojiljkovic, A Sarajlija, S Grkovic

14. Disorders of fatty acid oxidation and ketone body metabolism

A-040 Primary systemic carnitine deficiency: two turkish cases with two novel *SLC22A5* mutations
B Seker Yilmaz, D Kor, O Kucukosmanoglu, D Bulut, G Ceylaner, M Oktem, N Onenli Mungan

A-041 Screening for inherited metabolic disorders in patients with familial Mediterranean fever
E Kiykim, A C Aktuglu Zeybek, K Barut, M S Cansever, T Zubarioglu, O Kasapcopur, A Aydin

A-042 Is there any effect of acylcarnitines on proinflammatory process in obese children
G Biberoglu, B Derin Genc, A Inci, E Doger, I Okur, F S Ezgu, L Tumer

A-043 Screening of free carnitine and acylcarnitine status in patients with familial Mediterranean fever
E Kiykim, A C Aktuglu Zeybek, K Barut, T Zubarioglu, M S Cansever, S Alsancak, O Kasapcopur, A Aydin

A-044 Determination of free carnitine and acyl-carnitine status in patients with juvenile idiopathic arthritis
E Kiykim, A C Aktuglu Zeybek, K Barut, T Zubarioglu, M S Cansever, S Alsancak, O Kasapcopur, A Aydin

A-045 Challenge to detect the suspected of carnitine palmitoyl transferase-1 (CPT-1) deficiency with limited resources
L C Gultom, D R Sjarif

A-046 Two novel mutations in clinically detected MCADD
A Smon, U Groselj, B Repic Lampret, M Zerjav Tansek, T Battelino

A-047 Medium/short chain 3-hydroxyacyl-coA dehydrogenase deficiency *M/SCHAD:* A case report
M Kendirci, F Kardas, P Ustkoyuncu, S Gokay, B N Akyildiz, N Tekerek

A-048 Systemic primary carnitine deficiency - early treatment with a good outcome in first case from India!
S Bijarnia-Mahay, S Deswal, K Hara, Y Shigematsu, I C Verma

A-049 A case of severe rhabdomyolysis in early childhood: *LPIN1* gene mutation
T Zubarioglu, A C Aktuglu Zeybek, E Kiykim, M S Cansever, D Demirkol, A Aydin

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A-050 PolG deficiency revealed by valproic acid in a 3-year-old child
S Torre, K Cassinari, L Abily-Donval, A Tebani, A M Guerrot, P Gaignard, S Marret, A Slama, S Allouche, S Bekri

A-051 Rare cause of congenital lactic acidosis: A case of TMEM70 - ATP synthase deficiency
M Çoker, E Canda, S Güneş, Ö Altun Köroglu, T Tanyalçın, N Kültürsay

A-052 Mitochondrial DNA depletion syndrome: A case with DGUOK mutation
G Kilic Yildirim, S Durmus Aydogdu, N Tekin

A-053 Deoxyguanosine kinase deficiency in a Turkish infant with a novel mutation
M Gunduz, E Ozaydin, M Bastemur, C T Kirsaclioglu, O Unal

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A-054 A case with Kearns Sayre Syndrome and definition of an atypical mutation
M Köse, E Canda, M Kagnıcı, S Bozkurt Gökçe, L Erturk, S Kalkan Ucar, S Kalkan Ucar, S Ceylaner, R Ozyurek, M Çoker

A-055 Mitochondrial respiratory chain disorders in the Old Order Amish population
J Vockley, L Ghaloul Gonzalez, A Goldstein, S Dobrowolski, C Walsh Vockley, A Irani, B Wayburn, H Morton

A-056 Clinical variability of mitochondrial encephalopathy, lactic acidosis, and stroke-like (MELAS) syndrome
S Lee, J Yu

A-057 Mitochondrial hepatopathy as an unusual presentation of mtDNA mutation
H Kathom, D Avdjieva-Tzavella, S Mihailova, R Tincheva

A-058 Patient with *DGUOK* gene mutation diagnosed by using new generation sequence technology scanning 500 metabolic diseases
I Okur, A Inci, L Tumer, A Olgac, S Sari, B Ciftci, A B Topcu, C Turkyilmaz, F S Ezgu

18. Other disorders of energy metabolism, creatine disorders

A-059 Mitochondrial deficiency in two siblings with Cockayne syndrome
M Mexitalia, J C Susanto, A Utari, A Purwanti

20. Lipid and lipoprotein disorders, porphyrias

A-060 Abetalipoproteinemia: A case report
G Kilic Yildirim, S Durmus Aydogdu, M Eren, T Tekin

21. Peroxisomal, sterol and bile acid disorders

A-061 X-linked dominant condrodysplasia punctata: clinical phenotype in an affected female
V Plaiasu, D Ochiana, G Motei, A Coltoiu

22. Lysosomal disorders: mucopolysaccharidoses, oligosaccharidoses

A-062 p.258delY: A novel mutation of *IDUA* gene with atypical cardiac involvement in an infant with Mucopolysaccaridosis-I
M Ersoy, M B Akyol, S Yaroglu Kazancı

A-063 A carrier detection of Hunter syndrome (MPSII) in Mongolia
Z Doljoo, M Tumurkhuu, S Jav, P Ichinkhorloo, M Puntsag, N Tsedenbal, M Bazarragchaa

A-064 Long-term clinical course of a patient with mucopolysaccharidosis type IIIB
J H Lee, J H Kim

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A-066 Moved to P-738

A-067 Clinical and radiologic findings of Morquio A disease and spondylo-epi-metaphyseal dysplasia of two prepubertal girls
M Mexitalia, F M Wahyuni, J C Susanto, A Purwanti

A-068 A novel aspartylglucosaminuria mutation in a patient with co-existence of Gaucher disease
E Kiykim, A C Aktuglu Zeybek, O Gorukmez, T Zubarioglu, S Gunes, M S Cansever, A Aydin

A-069 Mucopolysaccharidosis type VI Maroteaux-Lamy Syndrome: A rare case report
A Juliaty

A-070 A case of α-mannosidosis
E Canda, M Köse, M Kağnıcı, S Kalkan Ucar, M Çoker

A-071 Growth curves in mexican children with mucopolysaccharidosis I receiving enzyme replacement therapy
E D Ruiz-Cruz, M A Márquez-Gutierrez, M E Vega-Ramírez, S J Franco-Ornelas

A-072 Mucopolysaccharidosis Type VII at an Early Age: A good candidate for investigational enzyme replacement therapy
A Karaoglu, F S Ezgu, G Biberoglu, A Olgac, A Inci, I Okur, L Tumer

A-073 Bilateral tarsal tunnel syndrome in mucopolysaccharidosis type VI
A L Barth, D S Silva, A C Esposito, A R Bellas, J F M Salomão, D D G Horovitz, J C Llerena

A-099 Biodistribution of idursulfase in cynomolgus monkeys after intrathecal-lumbar administration
J K Chung, M A Mascelli, E Brown, R Crooker, K J Palmieri, T G McCauley

23. Lysosomal disorders: sphingolipidoses

A-074 A novel mutation in a patient with early infantile type GM1 gangliosidosis
S Erdöl, S Dorum, H Saglam

24. Lysosomal disorders: others

A-075 Two cases of Pompe's disease
M Kilic, B Tokgoz-Cuni, E Polat, I Ertugrul, M Akcaboy, P Zorlu

A-076 Cases of neuronal ceroid lipofuscinosis
M Çoker, E Canda, M Kağnıcı, M Köse, A Tosun, M Polat, S Kalkan Ucar

A-077 Novel small deletion of *GALC* gene in a young Russian boy with Krabbe disease
N V Jourkova, A A Pushkov, K V Savostyanov, N L Nechaeva, L M Kuzenkova

A-078 Clinical spectrum of Fabry disease In Egyptian children : report of 6 cases
N M Al Menebawy, M Y Girgis, L A Selim, I G Mahmoud, A I El Badawy, M Abdelmoneim

25. Lysosomal disorders: treatment, enzyme replacement therapy

A-079 Histologic examination of the effect of a proprietary recombinant human acid α-glucosidase co-administered with a pharmacological chaperone on glycogen reduction in disease-relevant muscles of Pompe mice
Y Lun, S Xu, R Gotschall, A Schilling, R Soska, M Frascella, A Garcia, J Feng, K Chang, H Do, K J Valenzano, R Khanna

A-080 Hematological manifestations and enzyme replacement therapy outcomes of Gaucher disease: experience of eighteen years
T Zubarioglu, E Kiykim, M S Cansever, A C Aktuglu Zeybek, A Aydin

A-100 Comorbidities and use of concomitant medications in adults with Gaucher disease (GD): a MarketScan™ US claims database analysis
R Pleat, L Underhill, L Ross, L Nalysnyk, J Ibrahim, S Tcherny-Lessenot, L Wang

26. Glycosylation disorders/CDG, protein modification disorders

A-081 Our patients with congenital disorders of glycosylation
M Köse, E Canda, M Kagnici, S Kalkan Ucar, M Çoker

A-082 CDG incidence in unknown genetic-metabolic/neurometabolic disorders
M Kilic, Ç S Kasapkara, E Kilic, J Jaeken

28. Disorders of vitamins, cofactors and trace elements

A-083 Evaluation of vitamin B12, methylmalonic acid and homocysteine levels in obese adolescents
R Turan, M Akis, P Kuyum, S Cakar, M Kant, N Arslan, H Islekel

A-084 5,10-Methylenetetrahydrofolate reductase deficiency with different pictures
M Çoker, M Köse, E Canda, M Kagnici, D E C Smith, M I Mendes, S Kalkan Ucar, G S Salomons

A-085 Reappearance of neurological symptoms in a patient with a remethylation defect due to a cerebral folate deficiency
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A-087 Cobalamin (Cbl) D deficiency: A case reportt
M Kendirci, F Kardas, P Ustkoyuncu, S Gokay, H Per, A Kacar Bayram

A-088 The clinical predictive power in terms of the clinical results and family history of the MTHFR thermolabile variants
I Ozer, F Ozen

29. Miscellaneous

A-089 The inborn errors of metabolism emergency folder: making life easier for health professionals and families
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A-090 Application of sigma metrics for the assessment of quality assurance for plasma amino acid analysis in biochemical genetics laboratory in Pakistan
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A-091 Netherton syndrome: *SPINK5* gene mutation found through whole exome sequencing
B Seker Yilmaz, D Kor, S Ceylaner, D Bulut, M Yilmaz, N Onenli Mungan

A-092 Coexistence of pseudohypoaldosteronism and cholelithiasis, presenting with metabolic acidosis, dystrophy and vomiting in a neonate
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A-093 Pelizaeus-Merzbacher Disease (PMD) and Mitochondrial Dysfunction
P E Fitzsimons, P D Mayne, K Shadani, I Robinson, B Lynch

A-094 Clinical experience in an Inherited Metabolic Diseases Unit of a high complexity hospital in Medellin, Colombia, between 2010 to 2014
B L A Ortiz, C L A Garcia

A-095 Clinical and psychophysiological aspects of l-dopa treatment in ataxia-telangectasia patients: preliminary observations
D D Daniela, d'agnano, D M Daniela, Mannarelli, V L Vincenzo, Leuzzi, C C Caterina, Caputi, F F Francesco, Fattapposta

A-096 The experience of erythrocyte-delivered dexamethasone in ataxia-teleangiectasia
D D'Agnano, R Micheli, P Alessandro, C Luciana, M Mauro, L Vincenzo

A-097 Whole exome sequencing in clinical context identifies treatable intellectual disability
O Brandau, D Trujillano, A Rolfs, R Abou Jamra

A-098 Atypical presentation of Lowe syndrome
K Brennerova, A Šalingova, J Škodova, A Gerinec, V Bzduch