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P-314 Unusual evolution in glycogen storage disease type VI due to growth hormone deficiency
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P-317 The effect of riboflavin is limited in Japanese patients with multiple acyl-CoA dehydrogenase deficiency (MADD)
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P-318 Evaluation of natural or synthetic AMPK activators for correction of inborn fatty acid oxidation defects in patient cells
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P-319 Acute cardiac failure in LCHADD patients caused by parvovirus B19 infection
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P-334 Clinical, biochemical and molecular characterization of very-long-chain acyl-CoA dehydrogenase deficiency (VLCADD) in Saudi Arabia
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P-335 Identification of a novel mutation in Turkish infant with early-onset monocarboxylate transporter 1 (MCT1) deficiency as a cause of recurrent ketoacidosis
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P-350 Clinical, molecular and genetic characteristics of mitochondrial hepatopathy in Japan
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P-356 A patient with mitochondrial disorder and mutation in *MRPS22* gene
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P-357 The new autosomal recessive phenotype of mitochondrial disease caused by *DNA2* gene mutations
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P-367 Two sibling cases of aspartate-glutamate carrier 2 (citrin) deficiency: does diet affect prognosis?
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P-368 Severe infantile epileptic encephalopathy with cerebellar cysts associated with *COQ4* mutations and primary coenzyme Q10 deficiency
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P-371 *COX5* related complex IV deficiency: primary pulmonary hypertension, failure to thrive, and lactic acidosis
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P-372 Two cases with *ELAC2* mutations presenting with isolated infancy-onset cardiomyopathy
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P-376 Acyl-carnitine profile mimicking multiple acyl-CoA dehydrogenase deficiency in a patient with mitochondrial myopathy and a mutation in the *MT-CO2* gene
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P-377 Unusual intragenic *MT-ND1* inversion detected in the eye lens sample from patient with Leigh-like presentation
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P-378 Mitochondrial network properties in cultured myoblasts from patients with mitochondrial disorders
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P-379 3-Methylglutaconic aciduria in a patient with fatal infantile cardiomyopathy due to a m.3303C>T *MT-TL1* mutation
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P-380 A review of anaesthetic outcomes in a cohort of patients with genetically confirmed mitochondrial disorders
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P-381 MELAS-like presentation of mitochondrially-encoded tRNA(Trp) deficiency
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P-383 Mitochondrial optic neuropathies—optimization of diagnosis and management strategy
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P-386 Development of a LC-MS/MS method for the quantitation of purine and pyrimidine metabolites in human urine
A Cremonesi, N Perkins, J Haeberle, M Hersberger

P-387 Altered pre-mRNA splicing due to a novel intronic mutation c.1443+5G>A in the dihydropyrimidinase (*DPYS)* gene
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P-392 Long term outcome of early liver transplantation for a peroxisome biogenesis defect: 15 years follow up
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P-398 Identification of a novel mutation in *PEX10* in a patient with attenuated Zellweger spectrum disorder
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P-401 Homozygous missense mutations in *GPD1* presenting with ichthyosis, hepatosteatosis, and hyperlipidemia
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P-402 D-bifunctional protein deficiency: expanding the phenotypic spectrum
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P-407 Design and rationale of the clinical study programs for BMN 250, a novel investigational enzyme replacement therapy for Sanfilippo B syndrome
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P-410 The reduction of heparan sulfate level in the cerebrospinal fluid and of heparan sulfate content in the brain tissue of *IDS* knockout mice by an intracerebroventricular injection of idursulfase-beta are correlated
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P-412 Nitrosative stress and protein damage in mucopolysaccharidosis type II patients is correlated with dermatan sulphate
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P-413 Multiplex MS/MS method to measure MPS II, MPS IIIB, MPS IVA, MPS VI and MPS-VII enzyme activities in dried blood spots
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P-416 Mucopolysaccharidosis type 2: information on 19 Hunter syndrome patients in Iran
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P-417 34 Maroteaux-Lamy (mucopolysaccharidosis type 6) patients diagnosed in Iran
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P-419 Mucopolysaccharidosis type I from the perspective of phenotype-genotype-therapeutical response correlation
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P-420 Sialidosis type I with nonspecific biochemical and enzymatic patterns caused by two novel, compound heterozygous variants in *NEU1* gene
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P-421 Iminosugar based pharmacological chaperones: selecting new leads to target Gaucher, Morquio A and Hunter diseases
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A Inci, A Kan, B Topuz, I Okur, F S Ezgu, A Bakirtas, L Tumer

P-424 Bone marrow transplantation from heterozygous donors improve IDS deficiency in mucopolysaccharidosis type II mice
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P-425 Sulfated disaccharides improve iduronate-2-sulfatase function in fibroblasts from patients with mucopolysaccharidosis type II
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P-426 From clinical to biochemical—genetic characterisation of Morquio B disease
A Caciotti, R Tonin, S Catarzi, M Vasarri, G La Marca, G Forni, A Paoli, S Bechini, E Procopio, M A Donati, M Rigoldi, M Di Rocco, A Andaloro, D Antuzzi, A Rampazzo, M Scarpa, G Renzo, A Morrone

P-427 Could propionylcarnitine and free carnitine be used as antioxidative markers in mucopolysaccaridosis?
A Inci, G Biberoglu, B Genc, A Karaoglu, I Okur, F S Ezgu, L Tumer

P-428 High-throughput determination of urinary hexosamines in newborns of 2–3 days of age: application for the early diagnosis of mucopolysaccharidoses
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P-429 Determination of total and single species of all uronic acid-bearing glycosaminoglycans in urine of newborns of 2–3 days of age for a possible early diagnosis of mucopolysaccharidoses
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P-430 Development of a routine LC-MS/MS method for the analysis of underivatized urinary oligosaccharides for the diagnosis of oligosaccharidoses
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P-431 Spinal magnetic resonance imaging findings in mucopolysaccharidoses type IVA
E Canda, C Eraslan, H Yazici, S K Ucar, M Coker

P-432 Multidetector computed tomography for the evaluation of the trachea in patients affected by mucopolysaccharidoses
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P-433 Short-term outcome of surgical correction of genu valgum in four patients with mucopolysaccharidosis type IV-A
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P-434 Mucopolysaccharidosis type I due to maternal uniparental disomy of chromosome 4
M Gurner, S Chin, G Owens, T Pyragius, K Brion, M Trinh, K Friend, S Yu, D Ketteridge, S Stark, J Fletcher

P-435 Do cytokine levels play a role in pathogenesis of mucopolysaccaridosis patients?
A Inci, L Tumer, C Y Demirtas, A Karaoglu, I Okur, A Olgac, F S Ezgu, G Biberoglu

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P-438 Mucopolysaccharidosis type II in 44 Czech, Slovak, Serbian and Croatian patients: clinical manifestation and analysis of mutational spectrum
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P-439 Increase of serum levels of matrix metalloproteinase-2 in four paediatric MPS II patients
M Magner, B Asfaw, J Kulhanek, H Poupetova, J Langer, J Zeman, I Svandova

P-440 Effect of enzyme replacement therapy in a 5-year-old boy with mucopolysaccharidosis IV A
M Magner, I Marik, J Kulhanek, S Svecova, M Tesarova, B Asfaw, H Poupetova, J Bartl, J Zeman

P-441 Four-years study in 137 Russian patients with mucopolysaccharidosis
A A Pushkov, K V Savostyanov, A K Gevorkyan, L M Kuzenkova, T V Podkletnova, A V Pakhomov, L S Namazova-Baranova, A A Baranov

P-442 Mutation analysis for mucopolysaccharidosis gene's cluster in a southwestern Colombian affected population
A Sanchez, L M Moreno, J M Satizabal

P-443 Outcome of haematopoietic stem cell transplantation in two mucopolysaccharidosis type II severe patients
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P-444 Hematopoietic stem cell transplantation in a patient with mucopolysaccaridosis II
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P-445 Dermatan sulfate and heparan sulfate quantification in CSF, plasma and dried urine spots by UPLC-MS/MS
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P-446 Characteristics of patients aged 5 years and older at first signs and symptoms of mucopolysaccharidosis type II: data from the Hunter Outcome Survey (HOS)
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P-447 Bone crisis in atypical localization in GD patients under long-term enzyme replacement therapy
M Andrade Campos, I Sancho Val, I Garcia, M Roca Espiau, P Giraldo

P-448 Assessment of microvascular endothelial function in children and adolescents with mucopolysaccharidosis type VI
B Ozturk-Hismi, B Kumru, T Kilic, S Sezer, M Keskin, C Bagci

P-449 Health-related quality of life of transplanted MPS I Hurler children
L Scalone, R Ciampichini, L G Mantovani, G Cesana, F Scordo, S Gasperini, C Galimberti, A Biondi, A Rovelli, R Parini

P-450 Clinical and biochemical spectrum of mucopolysaccharidosis type III (Sanfilippo syndrome) in Morocco
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P-451 Targeted population screening for mucopolysaccharidoses—an efficient tool for the diagnosis of patients
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P-452 Using a mathematical–structural model in prediction of pubertal spurt in patients with MPS I and MPS II
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P-453 Four novel mutations in the N-acetylgalactosamine-6-sulfate sulfatase gene among Egyptian patients with Morquio A disease
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P-454 Enzyme replacement therapy for mucopolysaccharidosis type VI: experience from a Brazilian reference center
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P-455 Epileptic seizures profile in patients with mucopolysaccharidosis (MPS) types I, II and VI
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P-456 Nitrative and inflammatory status in long-term idursulfase-treated mucopolysaccharidosis type II patients
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P-458 Natural course of classical and non-classical Fabry disease: a large multicenter cohort study
M Arends, C Wanner, D Hughes, A Mehta, M Biegstraaten, C E M Hollak

P-459 Acid sphingomyelinase deficiency: diverse clinical manifestations and heterogeneous natural history
M McGovern, R Avetisyan, B J Sanson, O Lidove

P-460 A novel mutation in *NPC1* associated with neonatal cholestasis and neurological deterioration despite normal oxysterol and cholesterol esterification findings
S Santra, S Vijay, S Sreekantam, L M Simmons, T Hutchin, J Blundell, E Wright, S Kearney, J Raiman

P-461 High frequency of p.His281Tyr mutation in *GLB1* gene in patients with GM1-gangliosidosis in Ukraine
N I Mytsyk, N V Olkhovych, N S Trofimova, N A Pichkur, N G Gorovenko

P-462 Functional analysis of common splicing mutations detected in *HEXB* gene causing Sandhoff disease
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P-463 Multiplex ligation-dependent probe amplification assay: screening for deletions/duplications in the *GBA1* gene in Gaucher disease patients
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P-464 Molecular mechanism of autophagic pathway in Gaucher cells
S Dokmeci (Emre), O Oral, A Yuce, D Gozuacik

P-465 Consensus recommendation on a diagnostic guideline for acid sphingomyelinase deficiency
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P-466 Technical difficulties in the diagnosis of Krabbe leucodystrophy by enzyme analysis
T Hutchin, F Terry, S Santra, C Egerton, H Church, M A Preece

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P-468 Diagnostic utility of chitotriosidase activity, CCL18/PARC and 7-ketocholesterol concentrations in Gaucher, Niemann-Pick A/B/C and lysosomal acid lipase deficiency
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P-469 Evaluation of bone quality in patients with type 1 Gaucher disease with microindentation. Preliminary results
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P-470 Assessment of diagnostic parameters and disease biomarkers for detecting early-stage Fabry disease and monitoring its progression—results from the SOPHIA study
F Weidemann, M Beer, M Kralewski, C Kampmann

P-471 Study of comorbidities in a Spanish cohort of Gaucher disease type 1 patients
J Perez-Lopez, V Giner, M A Torralba-Cabeza, S J Perez, M Molto-Abad, I Roig, L Vicente, A Luana-Galan, E Cabezudo, X Solanich, B Roig-Espert, E Patera, M L Lozano-Almela, R Hurtado-Garcia, M Reyes, N Revilla, S Novelli, J Esteve

P-472 Characteristics of 27 patients with type 3 Gaucher disease: a descriptive analysis from the Gaucher Outcome Survey
I V D Schwartz, O Goker-Alpan, P Kishnani, A Zimran, L Renault, Z Panahloo, P Deegan

P-473 Clinical use of plasma oxysterols for rapid diagnosis of Niemann-Pick type C
I Lay, D Ardicli, A Samadi, F Akbiyik, E Serdaroglu, G Haliloglu, A Yuce, T Coskun, M Topcu

P-474 Cardiac dysfunction and troponin I levels in patients with Fabry disease followed at Hospital de Clinicas de Porto Alegre, Brazil
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P-475 Metabolic progression to clinical phenotype in classic Fabry disease
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P-476 The ENCORE trial: outcomes in adult patients with Gaucher disease type 1 previously stabilized on enzyme therapy after 4 years of treatment with eliglustat
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P-477 Combination therapy using miglustat and ketogenic diet for treatment of gangliosidoses: overall survival in 10 infants
J R Jarnes Utz, C B Whitley

P-478 Development of a suspicion index tool to help diagnosis of Gaucher disease
D F Bulut, B S Yilmaz, D Kor, G Seydaoglu, M N Ozbek, H I Sasmaz, S Soyupak, S Ceylaner, H N O Mungan

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P-480 Identification of 14 novel mutations in 45 Iranian Niemann-Pick type C patients
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P-481 Expert opinion on the management of CLN2 disease
N Specchio, R Williams, H Adams, M Blohm, J Cohen-Pfeffer, E De los Reyes, J Denecke, K Drago, C Fairhurst, M Frazier, N Guelbert, S Kiss, A Kofler, J Lawson, L Lehwald, M Leung, S Mikhailova, J Mink, M Nickel, R Shediac, K Sims, M Topcu, I Von Lobbecke, A West, A Schulz

P-482 Cardiac manifestations and cardiovascular autonomic functions of 64 Fabry patients
H Goto, K Tsuboi, H Yamamoto

P-483 Severe dilated cardiomyopathy as an unusual clinical presentation in an infant with mucolipidosis type 1
F J M Eyskens, F Marchau, M De Sain, S Ferdinandusse, A B P Van Kuilenburg

P-484 Monitoring oculomotor abnormalities in children with Niemann-Pick type C
J Blundell, S Frisson, A Chakrapani, P Gissen, S Vijay, S Santra, A Olson

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P-486 Expert recommendations for the laboratory diagnosis of neuronal ceroid lipofuscinosis type 2 (CLN2 disease): diagnostic algorithm and best practice guidelines for a timely diagnosis
E Izzo, M AlSayed, D G Burke, J Cohen-Pfeffer, J D Cooper, L Dvorakova, M Fietz, R Giugliani, H Jahnova, Z Lukacs, S Mole, I Noher de Halac, D Pearce, A Schulz, N Specchio, W Xin, N J Miller

P-487 A rare lysosomal storage disease: neuronal ceroid lipofuscinosis type 14
T Zubarioglu, G Yesil, E Kiykim, M S Cansever, A C Aktuglu-Zeybek, C Yalcinkaya

P-488 Mesenteric lymphadenopathy and hepatic Gaucheroma in Algerian GD children
A Hadji, N Benali Khoudja, S Sokhal, R Belbouab, F Hassoun, R Boukari

P-489 *GAA* de novo mutation in infantile Pompe disease
A Fiumara, A C Arena, F Raudino, M C Balistreri, G Del Campo, S Catarzi, A Morrone

P-490 A cost-effective case finding study in Fabry disease
O Dursun, S F Bulbul

P-491 Clinical presentation and molecular characterization of children with neuronal ceroid lipofuscinosis (NCL I & II) from India
R Bhavsar, M Mistri, M Kamate, R Shah, S Mehta, H Shah, F Sheth, J Sheth

P-492 Pathophysiology of Niemann-Pick type C revisited: altered protein trafficking is mutation-specific
H Shammas, E M Kuech, A M Das, H Y Naim

P-493 Intracranial hypertension in pediatric patients with cystinosis
M Del Toro, N Martin-Begue, A Felipe, E Lara, J A Arranz, G Ariceta

P-494 New method for molecular genetic diagnosis of glycogen storage disease in Russian patients using next-generation sequencing (NGS)
K V Savostyanov, A A Pushkov, A N Surkov, S I Polyakova, A G Nikitin, L S Namazova-Baranova, A A Baranov

P-495 The activity of chitotriosidase in blood of patients with lysosomal diseases: 10 years of laboratory experience
G Tozzi, A D'Amico, C Dionisi-Vici, E Bertini, E Piermarini, S Petrillo, F Piemonte

P-496 Plasma metabolomic profile in Spanish patients with lysosomal acid lipase deficiency
R Yahyaoui, E Rodriguez-Garcia, M C Garcia-Jimenez, A Dayaldasani, C Munoz-Hernandez, P Ruiz-Sala, M L Garcia-Martin

P-497 Wolman disease—a misdiagnosed case with hemophagocytic lymphohistiocytosis
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P-498 Interpreting cognitive function in children with rapid loss of vision—lessons from the early phase of CLN3 disease
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P-499 Selective screening for Pompe disease in high-risk Russian patients
K V Savostyanov, A A Pushkov, E N Basargina, N V Zhurkova, N D Vashakhmadze, L S Namazova-Baranova, A A Baranov

P-500 Global consensus on barriers to early diagnosis of Gaucher disease*A Mehta, S Salek, D Kuter*

P-501 Early diagnosis in Gaucher disease: findings from a global consensus initiative
A Mehta, S Salek, D Kuter

P-502 Alpha glucosidase on dried blood spot: simple assay in Pompe disease
M Sacchini, E Procopio, E Pasquini, F Pochiero, M Daniotti, D Ombrone, G La Marca, S Catarzi, A Morrone, M A Donati

P-503 Evaluation of different approaches to lysosomal acid lipase deficiency screening
J J Cebolla, P Irun, M Pocovi, P Giraldo

P-504 Dried blood spot screening of LALD and confirmatory studies in Spanish LALD suspected patients
J J Cebolla, P Irun, L Gonzalez-Dieguez, P Del Valle Loarte, M A Barba-Romero, I Garcia-Jimenez, I Ros Arnal, D Ortega Gil, R Tomasini, P Giraldo

P-505 Familial analysis of clinical consequences of type I Gaucher disease
U Mahmood, H A Cheema, M Suleman, S Bukhari, S Mahmood

P-506 Urine dicarboxylic acids and other biomarkers for diagnosis and follow-up of Wolman disease
M R Heiner-Fokkema, K E Niezen-Koning, F J Van der Sluijs, A B Bontekoe, J Janssens-Puister, T J De Koning, T G J Derks, H P J Van der Doef, F J Van Spronsen

P-507 Limb-girdle muscular dystrophy mimicking Pompe disease
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P-508 Spectrum of mutations and biochemical characteristics of 21 Russian patients with lysosomal lipase deficiency
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P-509 A rare presentation of Gaucher type 2 disease in a neonate
N Zdraveska, A Kostovski

P-510 Renal artery involvement in Pompe disease: a case report
E Pappa, C Papadopoulos, P Grimbert, G Bassez, P Laforet

P-511 CLN8 protein is involved in spatial distribution of lysosomes
F Pesaola, G Quassollo, M Remedi, I Noher de Halac, M Bisbal

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P-512 Expert opinion on the management of intracerebroventricular (ICV) drug delivery
I Slavc, J Cohen-Pfeffer, S Gururangan, E Jurecki, J Krauser, T Lester, D Lim, M Maldaun, C Schwering, A Shaywitz, M Westphal

P-513 Long-term galsulfase enzyme replacement therapy in Taiwanese mucopolysaccharidosis VI patients: a case series
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P-514 A national pilot study to investigate the effects of sub-maximal aerobic exercise in adults with late-onset Pompe disease: Salford Royal NHS Foundation Trust (SRFT) cohort results
E J S Silk, M E Roberts, M Meehan, C J Hendriksz

P-515 Efficacy of lentivirus-mediated gene delivery to treat Fabry disease
J R A Lambert, D G Burke, S J Howe, A A Rahim, S J R Heales

P-516 Canadian Fabry Disease Initiative Study (CFDI): 8 year outcomes of a randomized controlled trial of enzyme replacement therapy (ERT)
S Sandra, D Bichet, R Casey, J T R Clarke, R M Iwanochko, A Khan, C Morel, C Auray-Blais, S Doucette, K Lemoine, M L West

P-517 Outcomes of 453 pregnancies in patients with Gaucher disease: an analysis from the Gaucher Outcome Survey

H Lau, N Belmatoug, P Deegan, O Goker-Alpan, I V D Schwartz, S P Shankar, Z Panahloo, A Zimran

P-518 Very early treatment for infantile-onset Pompe disease contributes to better outcomes
C F Yang, C C Yang, H C Liao, L Y Huang, C C Chiang, H C Ho, C J Lai, T H Chu, T F Yang, T R Hsu, W J Soong, D M Niu

P-519 Long-term efficacy and safety results of taliglucerase alfa through 5 years in adult treatment-naïve patients with Gaucher disease
A Zimran, G Duran, P Giraldo, H Rosenbaum, F Giona, M Petakov, S E Solorio-Meza, P A Cooper, S Alon, R Chertkoff

P-520 Sub-analysis of long-term elosulfase alfa treatment outcomes in adults with Morquio A syndrome
D Hughes, R Giugliani, N Guffon, A S Jones, K E Mengel, R Parini, R Matousek, E Jurecki, A Quartel

P-521 Impact of bortezomib treatment on GAA function in missense murine model of Pompe disease
Y Shimada, T Fukuda, N Ishii, H Hoshina, H Kobayashi, T Higuchi, H Ida, T Ohashi

P-522 Literature review of the prevalence of Fabry disease in dialysis, kidney transplant and chronic kidney disease populations
G Sunder-Plassmann, M Cybulla, S Feriozzi, P Neumann, K Nicholls, M West, J Torras, A Gurevich

P-523 Enzyme replacement therapy for lysosomal acid lipase deficiency: a report of two Japanese patients
J Murakami, N Kuranobu, K Fukushima, R Nishimura, K Amano, Y Eto, S Kanzaki

P-524 Olipudase alfa for the treatment of acid sphingomyelinase deficiency (ASMD): 18-month safety and efficacy data
R Lachmann, M Wasserstein, M H Jouvin, I Nandy, A J Ji, H Inguilizian, A C Puga

P-525 Long-term efficacy and safety of reveglucosidase alfa in subjects with late-onset Pompe disease: 144-week follow-up of the POM-001/002 studies
T Geberhiwot, B Byrne, B A Barshop, R Barohn, D Hughes, D Bratkovic, C Desnuelle, P Laforet, E Mengel, M Roberts, P Haroldsen, L Smith, K Yang, L Walsh

P-526 Morquio A Registry Study (MARS): design and baseline characteristics of enrolled patients
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P-527 Switch of enzyme replacement therapy (ERT) in the Canadian Fabry Disease Initiative Study (CFDI): intermediate follow-up at 3.5 years
C F Morel, D Bichet, R Casey, J T Clarke, M Iwanochko, A Khan, S Sirrs, C Auray-Blais, S Doucette, K LeMoine, M L West

P-528 Prompt agalsidase alfa therapy initiation is associated with improved renal and cardiovascular outcomes in the Fabry Outcome Survey
D Hughes, A Linhart, A Gurevich, A Joseph, M Thakur, S Feriozzi

P-529 30 Infantile Pompe patients with 40 mg/kg/biweekly enzyme replacement treatment and 23 out of 30 survivors: a single center experience from Turkey
N Onenli Mungan, D Kor, B Seker Yilmaz, F D Bulut, S Erdem, F Demir, H Eren, N Ozbarlas

P-530 Autophagy induction as a potential treatment for lysosomal diseases
L Matalonga, J Farrera-Sinfreu, R Pascual, A Arias, F Tort, J Garcia-Villoria, A Ferrer Montiel, B Ponsati, L Gort, A Ribes

P-531 Estimating the value of treatment for Fabry disease: a discrete choice experiment
A Lloyd, K Gallop, A MacCulloch, D Hughes

P-532 Survival in idursulfase-treated and untreated patients with mucopolysaccharidosis type II: data from the Hunter Outcome Survey (HOS)
B K Burton, V Jego, J Mikl, S A Jones

P-533 Real-world treatment patterns from 647 patients with Gaucher disease: an analysis from the Gaucher Outcome Survey
P Deegan, D Fernandez-Sasso, P Giraldo, H Lau, Z Panahloo, A Zimran

P-534 Non-neuronopathic Gaucher disease: a retrospective review comparing clinical outcomes of 2 weekly and 4 weekly enzyme replacement therapy
H Heales, U Ramaswami, D Hughes, M Mckie, A Mehta

P-535 Early treatment with Sebelipase-alfa of two young LAL-D siblings: first outcome data
A Tummolo, F Ortolani, M Masciopinto, L Melpignano, A M Di Mauro, E Piccinno, F Papadia

P-536 Long-term data from patients with Gaucher disease: a descriptive analysis from a single center in the Gaucher Outcome Survey
D Elstein, A Zimran

P-537 Galactosialidosis: moving a step closer towards the development of enzyme replacement therapy with recombinant human protective protein/cathepsin a
V Koppaka, J Cadaoas, S Cullen, E Gomero, C Guzman, C Haller, H Hu, K Jayashankar, M Machado, G Morris, R Mosca, A Natesan, A Schatz, M Vellard, A D'Azzo

P-538 Abnormalities of cellular membranes can be reversed by substrate reduction in Fabry disease
G Brogden, H Shammas, K Maalouf, S L Naim, G Wetzel, M Amiri, M Von Koeckritz Blickwede, A M Das, H Y Naim

P-539 Successful desensitization to enzyme replacement therapy using omalizumab in a patient with late-onset Pompe disease
A Sechi, M De Carli, D Macor, K Bianchi, A Dardis, S Zampieri, G Ciana, S Tripodi, S Galosi, B Bembi

P-540 Efficacy and safety of migalastat, an oral pharmacological chaperone for Fabry disease: results from two randomized phase 3 studies
U Feldt-Rasmussen, R Giugliani, D P Germain, D Hughes, W R Wilcox, R Schiffmann, D G Bichet, A Jovanovic, D Bratkovic, J Castelli, N Skuban, J Barth

P-541 Migalastat improves gastrointestinal symptoms in patients with Fabry disease: results from a double-blind, placebo-controlled phase 3 trial (FACETS)

R Schiffmann, D G Bichet, D Hughes, R Giugliani, W Wilcox, S P Shankar, D P Germain, C Viereck, J Castelli, M Yao, N Skuban, J Barth

P-542 A novel delivery platform for intracellular and extralysosomal targeting of enzyme-replacement therapeutics
D D Armstrong

P-543 Comparison of α-galactosidase A activity in white blood cells of patients with Fabry disease after 2 weeks of exposure to migalastat, agalsidase beta, or agalsidase alfa
F K Johnson, K J Valenzano, J Castelli

P-544 Adaptive functioning and parental stress in patients with lysosomal storage diseases treated with enzymatic replacement therapy
S Caviglia, A Bottari, I Tondo, F Deodato, R Taurisano, C Dionisi-Vici

P-545 Lysosomal acid lipase deficiency (LAL-D; Wolman disease): diagnosed at 2 months needing 3 months to really improve on sebelipase alpha
F J Van Spronsen, T G J Derks, T J De Koning, M Van Rijn, K E Niezen-Koning, M R Heiner-Fokkema, S A Jones, R Scheenstra

P-546 Efficacy of migalastat in a cohort of male patients with the classical form of Fabry disease in a phase 3 study
D P Germain, R Giugliani, D G Bichet, W Wilcox, D Hughes, H M Amartino, R Schiffmann, C Viereck, M Yao, N Skuban, J Castelli, J Barth

P-547 The validation of pharmacogenetics in the identification of patients with Fabry disease for treatment with migalastat
E R Benjamin, C Della Valle, X Wu, E Katz, K J Valenzano, D G Bichet, D P Germain, R Giugliani, D Hughes, R Schiffmann, W R Wilcox, J Yu, J Kirk, J Barth, J Castelli

P-548 Clinical history of a cohort of Gaucher type 1 patients treated with ERT from childhood to adulthood
A Moro, S Cecchinel, D Macor, G Ciana, M R Da Riol, A Dardis, B Bembi

P-549 Phenotype of Fabry disease in patients with mutations amenable to migalastat
D Hughes, D G Bichet, D P Germain, R Giugliani, R Schiffmann, W Wilcox, J Castelli, E R Benjamin, N Skuban, J Barth

22. Glycosylation disorders/CDG, protein modification disorders

P-550 Evaluation of the global coagulation balance among PMM2-congenital disorder of glycosylation patients, using the thrombin generation assay
T Pascreau, F Cathala, D Lasne, N Seta, A S Guemann, P De Lonlay, D Borgel

P-551 Liver involvement in congenital disorders of glycosylation: literature review
D Marques-da-Silva, M Monticelli, V Dos Reis Ferreira, T Ferro, P Janeiro, P A Videira, J Jaeken, D Cassiman

P-552 *CCDC115* deficiency causes a disorder of Golgi homeostasis with abnormal protein glycosylation
J C Jansen, S Cirak, M Van Scherpenzeel, F Foulquier, T Marquardt, D J Lefeber

P-553 Exome sequencing of patients with positive screening of congenital disorders of glycosylation (CDG) type I revealed mitochondrial diseases due to *POLG* and *Twinkle* mutations
S Vuillaumier-Barrot, T Dupre, A Bruneel, P De Lonlay, L Servais, S Moore, N Seta

P-554 Facile CDG diagnostics via mass spectrometry and clinical exome sequencing
M Van Scherpenzeel, A AbuBakar, K Huijben, F Zijlstra, A Ashikov, D J Lefeber

P-555 NGS: a new strategy for the molecular diagnosis of α-dystroglycanopathies
C Bouchet-Seraphin, M Chelbi, M Reocreux, S Gazal, S Vuillaumier-Barrot, C Boileau, N Seta

P-556 Transferrin and total serum glycoprofiling for diagnosis and therapy monitoring in phosphoglucomutase 1 deficiency
A AbuBakar, N C Voermans, E Morava, M Van Scherpenzeel, D J Lefeber

P-557 A population based study on congenital defects of protein N-glycosylation experience in clinical and genetic diagnosis
C Perez-Cerda, M L Giros, M Serrano, M J Ecay, L Gort, B Perez Duenas, C Medrano, A Garcia-Alix, R Artuch, P Briones, B Perez

P-558 Galactose supplementation in SLC35A2-CDG: results after 24 weeks of treatment in an Italian patient
R Barone, P Striano, L Sturiale, D Garozzo, A Messina, J Jaeken, E Morava, A Fiumara

P-559 Metabolic insights into the pathomechanism of orofacial malformation: prevalence and clinical variability of cleft palate and other congenital malformations among PGM1-CDG patients
S Y Wong, D Rymen, L Beamer, S J Perez, T Kozicz, E Morava

P-560 Clinical description and long-term outcome in PMM2–congenital disorder of glycosylation: a series of 96 French patients
P De Lonlay, C Roda, M L Monin, T Dupre, S Vuillaumier-Barrot, C De Barace, C Francannet, D Heron, N Seta, N French Metabolic Disease Health Network

P-561 Diminished convergent extension and disturbed TGF-ß/BMP and FGF signaling in *Xenopus* Pmm2-CDG morphants
N Himmelreich, B Dimitrov, L T Kaufmann, C R Bartram, G F Hoffmann, C Thiel

P-562 A new case of SLC35A2-CDG with relatively mild phenotype and our experience with D-galactose treatment
K Ounap, M A Vals, S Pajusalu, D J Lefeber, E Morava, T Reimand

P-563 Screening of FDA approved drugs in fibroblasts derived from PMM2-CDG patients
G Andreotti, C Cimmaruta, V Citro, L Liguori, N Minopoli, M V Cubellis

P-564 Assessment of a CDT kit for screening of congenital disorders of glycosylation and its comparison with transferrin isoform analysis by HPLC
M B Dave, A J Dherai, V P Udani, A U Hegde, N Desai, T F Ashavaid

P-565 Beware of abnormal capillary electrophoretic patterns of serum transferrin: congenital disorder of glycosylation (CDG) type I can be associated with a protein variant
A Bruneel, T Dupre, T Chaabouni, A Dupont, H Mansour, N Seta

P-566 New insights into glycosylation and Na^{+}/Ca^{2+} exchangers in human cells
C A Amorosi, M B Bistue Millon, M Papazoglu, M Siravegna, G Elso-Berberian, C G Asteggiano

P-567 Evaluating galactose treatment in vivo and in vitro in TMEM165-CDG
P Witters, S Y Wong, D Cassiman, G Matthijs, F Foulquier, E Morava

P-568 Expanding phenotype variability in RFT1-CDG: absence of deafness in two patients with novel mutations
A Bandeira, A M Fortuna, D Quelhas, J Jaeken, G Matthijs, E Martins

P-569 Exuberant myopathic phenotype in a DPAGT1-CDG patient
D Quelhas, M Santos, J Jaeken, G Matthijs, L Lacerda, E Martins

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C N Hafifah, D R Sjarif

P-571 Dopamine and serotonin turnover in neuronal cell models of mitochondrial complex I deficiency and Gaucher disease
C De La Fuente Barrigon, M Garcia Gomez, D G Burke, S Eaton, S J R Heales

P-572 Brain development in qdpr (dihydrobiopterin reductase)-deficient zebrafish
M Breuer, T Opladen, M Carl, S W Sauer

P-573 Secondary brain creatine deficiency and neurological impairment in BDL rats, an *in vivo* model of chronic cholestatic liver disease
V Rackayova, M Loup, H Henry, V A McLin, C Cudalbu, O Braissant

P-574 IPSCs from a patient suffering from tyrosine hydroxylase deficiency
S Jung-Klawitter, A Sebe, N Shen, G F Hoffmann, N Blau, T Opladen

P-575 Two siblings with a new genotype of GAMT deficiency and response to sodium benzoate therapy
L Fiori, V Leuzzi, C L Carducci, C A Carducci, C Uggetti, A F Podesta'

P-576 Safety and efficacy of rotigotine in 7 patients with monoaminergic neurotransmitter deficiency
M Mastrangelo, M T Giannini, C L Carducci, C A Carducci, V Leuzzi

P-577 Analyses of cerebrospinal fluid neopterin: inborn errors of metabolism versus neuroinflammatory diseases
M Molero-Luis, A Ormazabal, C Sierra, D Cuadras, A Garcia-Cazorla, Y Jordan, B Perez-Duenas, R Artuch Iriberri

P-578 Two new cases with hereditary dopamine transporter deficiency syndrome
A Tokatli, Y Yildiz, E Pektas, G Haliloglu

P-579 Substantial psychiatric symptoms and reduced quality of life in well-treated patients with GTP-cyhydrolase deficient dopa-responsive dystonia
A Kuiper, M Smit, E R Timmers, A L Bartels, M A J Tijssen, T J De Koning

P-580 Expanded phenotype in creatine transporter deficiency: identification of two novel mutations correlated with mild clinical presentation
M M Mancardi, R Battini, M C Schiaffino, M G Alessandri, M Gherzi, V Viglione, F M Battaglia, C L Carducci, F Moro, C Carducci, G Morana, M Tosetti, G Cioni, V Leuzzi

P-581 Glomerular filtration rate (GFR) strongly influences guanidinoacetic acid in plasma and urine
C L Salvador, A D Rowe, A Bjerre, C Tondel, A Brun, D Brackman, L Morkrid

P-582 5-Oxoprolinase deficiency: report of three siblings and a case with two new compound heterozygous mutations and clinical diversity in the same family
N Onenli Mungan, B Seker Yilmaz, F D Bulut, M Oktem, D Kor, G Ceylaner, O Herguner

P-583 Abnormal CSF phenylalanine level in patients presenting with disorders of tetrahydrobiopterin metabolism with hyperphenylalaninemia
A Celato, M Mastrangelo, A P Burlina, G Polo, C Carducci, C Carducci, V Leuzzi, A B Burlina

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T Hoshina, S Nozaki, S Kudo, T Hamazaki, Y Nakatani, E Hayashinaka, Y Wada, H Kodama, Y Watanabe, H Shintaku

P-585 Secondary coenzyme Q_{10} deficiencies in mitochondrial and non-mitochondrial diseases
R Artuch, D Yubero, R Montero, M A Martin, M Grazina, I Hargreaves, L Salviati, A Ribes, P Navas

P-586 Rare inborn error of cobalamin metabolism (cobalamin J deficiency) presenting as deficiency of vitamin B_{12}
P Jesina, P Chrastina, J Krijt, J Bartl, L Dvorakova, H Vlaskova, M R Baumgartner, B Fowler, P Burda, M Hrebicek, J Stary, V Kozich

P-587 Molecular characterization of the cblC disease reveals new pathways in pathogenesis
L Hannibal, S Behringer, M Klenzendorf, D W Jacobsen, U Spiekerkoetter, H J Blom

P-588 Disruption of the metabolome in a zebrafish model of PNPO deficiency
M Albersen, S M C Savelberg, M Bosma, J Gerrits, M Willemsen, M G M De Sain-van der Velden, H C M Prinsen, M Van der Ham, M L Pras-Raves, J Bakkers, F G Tessadori, J J M Jans, G Van Haaften, N M Verhoeven-Duif

P-589 Vitamin B6 is essential for serine de novo biosynthesis
R J Ruben, M L Pras-Raves, J Gerrits, M Bosma, H C M Prinsen, J J M Jans, N M Verhoeven-Duif

P-590 Long-term visual and electrophysiological follow-up in early onset cblC patients
N Garcia Segarra, F Rigaudiere, J F Benoist, E Delouvrier, S Pichard, H Ogier De Baulny, M Schiff

P-591 Role of intramuscular levofolinate in treatment of hereditary folate malabsorption
E Manea, P Gissen, S Pope, S J R Heales, S Batzios

P-592 Early onset of cobalamin C deficiency in 3 neonates: clinical and biochemical features following parenteral hydroxycobalamin dose intensification strategy
E Scalais, E Osterheld, D Amrom, C Geron, C Pierron, L Bindl, V Schlesser, L Regal, G Martens, L De Meirleir
A Olgac, L Tumer L, A Inci, B Karaoglu, I Okur, F S Ezgu

P-594 Evaluation of genetic and biochemical profiles of patients with biotinidase deficiency
E Kose, H Onay, O Unal, M Gunduz, S Bulbul, N Arslan

P-595 Abnormal folate metabolism is associated with metabolic syndrome components in spontaneous hypertensive rats
V Kozich, M Pravenec, J Krijt, L Kazdova, J Sokolova

P-596 Postnatal hepatocyte transplantation in a child with molybdenum cofactor deficiency type B
B C Schwahn, S Bansal, E Fitzpatrick, H Lemonde, M Champion, C Turner, L Fairbanks, R Mitry, S Lehec, C Filippe, K Chong, F White, E V E Okokon, A Dhawan

P-597 Preponderance of c.394C>T mutation in *MMACHC* gene in Indian patients with combined methylmalonic aciduria and homocystinuria due to cobalamin C deficiency
S Bijarnia-Mahay, D Gupta, R D Puri, S Kohli, R Saxena, Y Shigematsu, S Yamaguchi, O Sakamoto, R Deb, V Udani, I C Verma

P-598 The challenge of treatments in three patients affected by pyridoxine dependent epilepsy
A Celato, I Fasan, E Zanonato, C Cazzorla, I Toldo, S Sartori, G Polo, A B Burlina

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P-599 Recessive pathogenic variants in the *MICU1* gene: expanding the phenotypic and genotypic spectrum
T Ben-Omran

P-600 *MBTPS2* mutations in X-linked osteogenesis imperfecta
U Lindert, W A Cabral, S Ausavarat, S Tongkobpetch, K Ludin, A M Barnes, P Yeetong, M Weis, B Krabichler, C Srichomthong, E Makareva, A R Janecke, S Leikin, B Roethlisberger, M Rohrbach, I Kennerknecht, D R Eyre, K Suphapeetiporn, C Giunta, J C Marini, V Shotelersuk

P-601 Clinical and biochemical spectrum of metabolic cardiomyopathies in Egyptian children
N M Al Menabawy, R I Ismail, M A Mohamed, D Mehany, S ElSaeedy, I Abdel Sattar, R Amar, L A Selim, Z S Seliem

P-602 Mutations in *PYCR2* define a lethal syndrome of failure to thrive, microcephaly, and brain atrophy in 10 Egyptian families
M S Zaki, G Bhat, M Y Issa, E Dikoglu, L A Selim, I Gamal, M S Abdel Hamid, I Marin-Valencia, J G Gleeson

P-603 Asfotase alfa treatment in a prematurely born baby with hypophosphatasia
H Niinikoski, K Nanto-Salonen, K Korhonen

P-604 Gene therapy for Canavan disease in the knockout mouse using rAAVs at a 20-fold lower dose
R Matalon, S Ahmed, Q Su, J Trapasso, G Gao

P-605 Increase in the diagnostic rate by exome sequencing in patients with neurometabolic disorders
E Kilic, M Kilic, R K Ozgul, D Yucel-Yilmaz, P Kavak, B Yuceturk, H Demirci, M S Sagiroglu

P-606 A floppy infant with slit ventricles on brain imaging—absence of SLC4A10 may cause the first human cerebrospinal fluid synthesis disorder
S B Wortmann, J A Mayr, J Spenger, D Mayr, W Sperl, J Koch, B Alhaddad, T Meitinger, H Prokisch, C Huebner, N Wolf, T B Haack

P-607 Rotavirus: the implications in children with inherited metabolic disorders
M A Mannion, A Smith, J Hughes, I Knerr, A Monavari

P-608 High incidence of hereditary liver diseases as an indication for pediatric liver transplant in Saudi Arabia: comparison to the Hungarian data
M A Barr, J Burkholder, M A Shagrani, T Algoufi, A Dezsofi, L Szonyi

P-609 Glycolysis and the formation of building blocks in proliferating epithelial cells
S A Fuchs, I Schene, P M Hasselt, R H L Houtkooper, N M Verhoeven-Duif, E E S Nieuwenhuis

P-610 Diagnostic biochemical abnormalities masked by early testing or prospective treatment in newborns at risk of multiple acyl-coA dehydrogenase deficiency and maple syrup urine disease
H Aitkenhead, A A J Lam, H Prunty, M A Cleary, J Davison

P-611 Developing next-generation pharmacological chaperones by fragment screening and crystallography
S R Mackinnon, J Kopec, F Von Delft, P E Brennan, W W Yue

P-612 Follow-up of a patient with cobalamin F deficiency
M El Habbas, C Sechter, M Joncquel, M Fontaine, G Morin, D Dobbelaere, J F Benoist, K Mention

P-613 Minimal prevalence and incidence of inherited metabolic disorders in Austria
D Karall, S Herbst, S Scholl-Buergi, M Brunner-Krainz, J Emhofer, M Huemer, S G Kircher, J Koch, V Konstantopoulou, F Lagler, W N Loescher, E M Maier, J A Mayr, D Moeslinger, G Sunder-Plassmann, W Sperl, T M Stulnig, S B Wortmann, J Zschocke

P-614 Artefactual increase in urine thymine concentration affecting GC-MS but not HPLC analysis
E Mozley, L Fairbanks, R Carling

P-615 GMP isolation and biodistribution of human amnion epithelial cell for clinical therapy
C Raghuraman Srinivasan, K Kannisto, C Hammarstedt, M Zabulica, C S Strom, R Gramignoli, U Askelof

P-616 Congenital myopathy associated with splice mutation in *PAX7* gene
U Ahting, C Makowski, H Juenger, B Schoser, J Mayr, B Alhaddad, H Prokisch, B Rolinski, T Haack, P Freisinger, T Meitinger

P-617 *HACE1* deficiency mimicking mitochondrial disorder
D Petkovic Ramadza, J A Mayr, T Haack, H Prokisch, K Zarkovic, K Fumic, I Baric

P-618 Bi-allelic variants in *PRUNE* cause early manifestation of severe epileptic encephalopathy with muscular hypotonia
B Alhaddad, A Schossig, T Haack, R Kovacs-Nagy, W Sperl, H Prokisch, J Mayr, J Senderek, J Zschocke, F Distelmaier, J Koch

P-619 Mutations in *RC3H1* in a boy with ethylmalonic aciduria and hemophagocytic lymphohistiocytosis
P Verloo, D Bogaert, F Haerynck, A Vanlander, B Menten, R Van Coster, M Dullaers

P-620 Diagnosis and discovery of treatable neurometabolic diseases via an integrated -omics approach
T Dalkeith, K Bhattacharya, A Biggin, J Christodoulou, Y Zurynski

P-623 The burden endured by caregivers of metabolic patients: mucopolysaccharidoses compared to intoxication disorders
F Nichelli, P Meregalli, S Gasperini, C Galimberti, E Schivalocchi, R Parini

P-624 A homozygous splice mutation in *SLC25A42*, encoding a mitochondrial transporter of coenzyme A and adenosine 3′,5′-diphosphate, causes metabolic crises and epileptic encephalopathy
A Iuso, A Bader, T Meitinger, H Prokisch, T M Strom, C Weigel, T B Haack

P-625 Modulation of sirtuins under ketogenic diet
A B Potthast, A M Das

P-626 Influence of altered ROS levels on expression and activity of sirtuins
A B Potthast, A Fitter, A M Das

P-627 Sirtuin 4 is up-regulated in the HELLP-syndrome
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P-628 Basal ganglia in neurometabolic disorders
J Paprocka, M Machnikowska-Sokolowska, B Rzepka

P-629 Synthetic cyclic pyranopterin monophosphate (scPMP) rescues the lethal phenotype of molybdenum cofactor (MoCo) deficient mice: relationship of scPMP doses, liver sulfite oxidase activity, body weight, and the detoxification of sulfite in vivo
S Liu-Chen, E Watsky, D Devore, N Kuklin, A Marozsan, Y Wang

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A-001 Screening for Fabry disease using dried blood spots: an Australasian experience
S L Stark, J R Dobbins, B Fong, S Chin, M Fuller, J M Fletcher

A-002 The importance of *MTHFR* testing in woman with recurrent spontaneous abortions *D Serapinas, A Bartkeviciute, D Bartkeviciene*

A-003 Valproic acid-induced severe hyperammoniemia unmasked by a protein loading test
C Tran, B Royer Bertrand, A O Rossetti, L Bonafe

A-004 An unexpected differential diagnosis for a severe deterioration in an adult with glycogen storage disease type 1
L Bosanska, T Schroeder, U Hoff, R Koerner, J M Kruse, R Senf, P Enghard, M Vietzke, N Tiling, U Ploeckinger

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A-005 GC/MS analysis of urinary mono- and disaccharides following mixed-bed resin purification and acetylation in methyl sulfoxide/1-methylimidazole reagent
M Z Habbal

A-006 Metabolome analysis by tandem mass spectrometry as a diagnostic tool for inborn errors of metabolism—preliminary studies
T Polawski, K Kusmierska, J Sykut-Cegielska, M Oltarzewski

A-007 Spectrum of metabolic diseases identified by clinical whole exome sequencing *H Lee, Y Ko, C Lee, J S Lee, H D Kim, J S Lee*

A-008 UV-light microscopy and its application to visualize detailed cataractogenic structures in the zebrafish lens
L Rumping, A Graaff, de, P A W Schellekens, F Tessasori, G W Haaften, van, R H J Houwen, J J M Jans, P M Hasselt, van, N M Verhoeven-Duif

A-009 Complementary test for the diagnosis of Pompe disease
A B Schenone, J Frabasil, C Durand, C Bambara, S B Sokn

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A-010 Withdrawn

A-011 Classical PKU with unusual neonatal presentation
R Fingerhut, N Lehnherr, V Pfeifle, S Holland-Cunz, S Sluka, G Szinnai, M Huemer, M Rohrbach

A-012 Free ethylmalonic acid (EMA) measurement for newborn screening of short chain CoA dehydrogenase deficiency: preliminary results
F Baronio, F Righetti, I Bettocchi, R Ortolano, G Monti, A Pession, A Cassio

A-013 Ratio C8/C10 as a discriminative predictor for MCADD in NBS
E Glab-Jablonska, T Polawski, J Sykut-Cegielska, M Oltarzewski

A-014 Newborn screening may detect inappropriately high protein intake in newborn infants
E Maines, G Gugelmo, M Vincenzi, N Campostrini, F Teofoli, M Camilot, A Bordugo

A-015 Biotinidase deficiency: evaluation of patients diagnosed with newborn screening
M Demirkol, E Cakar, K Bas, M Karaca, C Balci, G Gokcay

A-016 Profile of patients with hyperphenylalaninemia followed by the neonatal screening program of Minas Gerais, Brazil
V C Kanufre, M R A Alves, R D L Soares, G C Camatta, L M Almeida, C A A Souza, M J B Aguiar, R C Norton, A L P Starling

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A-017 Phenylketonuria and obesity in pregnancy: a case study
A Guerra, P A Nunes, A Oliveira, D C Gomes

A-018 Impact of specific diet therapy on controlling late onset glutaric acidemia type II (a case report)
M Ziadlou, M Shakiba

05. Phenylketonuria: general

A-019 Does metabolic control influence bone mineral density in adult PKU patients?
A Barta, E Kiss, E Simon, A Patocs, P Reismann

A-020 Spectrum of *PAH* variants in phenylketonuria patients from the Republic of Moldova and identification of a novel p.M1T missense allele
C Boiciuc, C Gemperle-Britschgi, D Sato, N Rimann, D Badicean, D Blanita, V Hlistun, V Sacara, B Thony, N Usurelu

A-021 Spectrum and outcome of phenylketonuria (PKU) in India
R A Jalan, A B Jalan, K V Kudalkar, D H Shinde, M M Joshi, M A Borugale, S M Shirke, A P Mahamunkar, R J Tawde

A-022 Bone mineral density in children, adolescents and young adults with phenylketonuria and hyperphenylalaninemia
C A Leiva, V Cornejo, P Bravo

A-023 PKU is still late diagnosed in Morocco: urgency to establish a systematic neonatal screening
S Dahri, H Talbaoui, Y Kriouile, A Agadr, S Chaouki, L Chabraoui

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D N Salimbayeva, G S Svyatova, A Ormankyzy, G Musabalaeva

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A-025 Thromboembolic manifestations in classical homocystinuria
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A-026 Sulfite intrastriatal administration induces mitochondrial dysfunction and alters antioxidant defenses in rat striatum: protective effects of bezafibrate
M Grings, A P Moura, B Parmeggiani, L M Alvorcem, R Boldrini, M M Motta, J T Pletsch, G M Cardoso, P M August, C Matte, A T S Wyse, M Wajner, G Leipnitz

08. Other amino acid disorders

A-027 The effects of methylphenidate on processing speed and executive functioning in a 12-year-old girl with tyrosinemia type 1: a case report
E Raets, F J M Eyskens, A Simons

A-028 The multiple faces of lysinuric protein intolerance with a novel mutation
E Ozaydin, M Gunduz, D Tural, H Altan, O Unal

A-029 Pyroglutamic aciduria: a splice site mutation (IVS9 G>A) in *GSS* in a Tunisian child
E Kerkeni, N Ghedira, R Sakka, H Ben Hmida, K Ben Ameur, M Bizid, N Kaabachi, F Z Chioukh, K Monastiri

A-030 Behavioral responses in an animal model of maple syrup urine disease treated with tianeptine
G Scaini, F A Morais, H M Abelaira, G Z Reus, J Quevedo, P F Schuck, G C Ferreira, E L Streck

A-031 Nonketotic hyperglycinemia: different and atypical manifestations of the disease
C Galimberti, A Brambilla, M Pasetti, S Tursi, C Corbetta, V Ravazzani, R Pretese, S Gasperini

A-032 Nutritional monitoring of patients with maple syrup urine disease
J C C Lopes, V C Kanufre, A F Cruz, T U Maioli, R R Arantes, E R Valadares

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A-033 Clinical, laboratory data, molecular features and outcome of 14 citrullinemia type 1 patients
T Zaman, S H Moarefian, B Behnam, A Rahmanifar, M Nagel

A-034 Autism spectrum disorder as a clinical presentation of undiagnosed urea cycle defects
W G Wilson, E D Black

A-035 Argininosuccinic aciduria: from identification to management of paucisymptomatic late onset forms
S Paquay, B Desnous, M Paviolo, A Imbard, S Pichard, J F Benoist, M Schiff

A-036 Identification of three novel mutations in patients with citrullinemia type 1
N Arslan, E Kose, O Unal, M Gunduz, J Haberle, S Bulbul

10. Organic acidurias: branched-chain

A-037 Different clinical course of two Korean maple syrup urine disease patients with novel mutations of *BCKDHB* gene
Y M Kim, Y S Park, S W Nam, G H Kim, H W Yoo, C K Cheon

A-038 Influenza A (H1N1) virus-associated acute pancreatitis and posterior reversible encephalopathy syndrome (PRES) in a patient with mut0 methylmalonic acidemia (MMA)
O Unal, M Gunduz, N Koc, U Bayrakci, H Genc, A Yaman, D Ademhan

A-039 Disorders of isoleucine degradation: diagnostic problems in two patients
M Sredzinska, D Rokicki, M Pajdowska, D Kozlowski, A Bogdanska, D Piekutowska-Abramczuk

A-040 Insulin resistant hyperglycemia in methylmalonic acidemia decompensation in a 1-year-old child
E Chavez Lopez, S Redecillas, R Lorite, J Ortega Lopez, I Valenzuela, J A Arranz Amo, M Del Toro

A-041 Propionic acidemia with severe decompensation despite normal newborn screening results
D Olsson, R H Zetterstrom, R Nergardh, C Heidenborg, S Lajic, A Nordenstrom

A-139 Two patients with neonatal-onset MSUD in Slovakia with different clinical courses
Brennerova K, Bzduch V, Behulova D, Skodova J, Krajciova A, Hikkelova M

11. Organic acidurias: others

A-042 Internal quality control (IQC) of qualitative organic acid analysis using numerics allows improved monitoring
G Urbano Blanco, P E Fitzsimons, P D Mayne

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A-043 Detection through newborn screening versus clinical symptoms of galactosemia: phenotype and genotype of two cases
A Lopez-Uriarte, M R Torres-Sepulveda, A B Hinojosa-Amaya, M C Ruiz-Herrera, E De la Rosa-Marban, A Sanchez-Pena, L E Martinez-Garza

A-044 Pompe disease and Hirschsprung disease: a case report of a rare association
C Kasapkara, F Ozbay-Hosnut, G Evirgen-Sahin, B Ardicli, M Kilic, E Aksoy

A-045 Exercise induced fatigue: McArdle disease in an adult patient with myopathy
S Aydogdu, D Uskudar Cansu, G Kilic Yildirim, B Erdogan

A-046 Coexistence of glycogen storage disease type III and haemophilia B in two brothers
D F Bulut, B S Yilmaz, D Kor, Y S Shin, G Leblebisatan, G Tumgor, S Gonkek, B Yuksel, H N O Mungan

A-047 Sinus bradycardia revealing hypertrophic cardiomyopathy in glycogen storage disease type III
A Ben Chehida, H Mansouri, R Ben Abdelaziz, H Hajji, H Boudabous, M S Abdelmoula, H Ben Turkia, H Azzouz, N Tebib

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A-048 A single paediatric centre experience of L-carnitine supplementation in medium-chain acyl-CoA dehydrogenase deficiency (MCADD)
W A Batten, E Chronopoulou, G Pierre

A-049 Medium-chain acyl-CoA dehydrogenase activity in correlation with acylcarnitines in dry blood spot of patients with MCAD deficiency detected by newborn screening
J Sykut-Cegielska, T Polawski, E Jablonska, J Taybert, A Kowalik, M Oltarzewski

A-050 10-year follow-up of 3 patients with long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency treated with heptanoate
M Zlamy, K Pichler, M Michel, S Scholl-Buergi, D Karall

A-051 Severe rhabdomyolysis and dilated cardiomyopathy: unusual clinical presentation of primary carnitine deficiency
A Madeo, A S Guemann, F Iserin, J B Arnoux, A Brassier, P De Lonlay

A-052 Prenatal findings and autopsy examination in a newborn with multiple acyl-CoA dehydrogenase deficiency
Y Yildiz, R Sirma Dokuzboy, B Talim, S Yigit, S Ceylaner, H S Sivri, A Tokatli, T Coskun, A Dursun

A-053 First Slovak patients affected by two primary defects in mitochondrial energy metabolism
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