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P-002 Updates in lysinuric protein intolerance, a multi-faceted disease
W Mauhin, F Habarou, S Gobin, A Servais, A Brassier, C Grisel, C Roda, D Moshous, C Ottolenghi, J De Blic, J B Arnoux, P De Lonlay

P-003 Ambulatory performance in adolescents and adults with hypophosphatasia treated with asfotase alfa: data from a phase II, randomized, dose-ranging, open-label, multi-center study
P S Kishnani, C Rockman-Greenberg, K L Madson, M Gayron, U Iloeje, M P Whyte

P-004 Hypogonadotropic hypogonadism in males with glycogen storage disease type 1
S M Sirrs, E M Wong, A Lehman, P Acott, J Gillis, D L Metzger

P-005 *ELOVL4* mutation in a family with dominant cerebellar and brainstem atrophy (SCA34): clinical, radiological, and metabolic findings
C Marelli, D Cheillan, C Guissart, M Koenig, P Labauge

P-006 Transition in patients with inborn errors of metabolism: a continuous challenge
D Piel, F K Trefz, D Haas, S Koelker, E Vogel, H Fluhr, P Nawroth, G F Hoffmann

P-007 French cohort of maple syrup urine disease: assessment of neuropsychiatric outcome
M T Abi-Warde, C Roda, J B Arnoux, F Habarou, C Grisel, A S Guemann, A Brassier, V Barbier, S Dubois, M Assoun, S Leverge, V Leboeuf, M Bayart, J M Alili, R Barouki, F Lesage, L Dupic, V Valayannopoulos, A Servais, C Ottolenghi, P De Lonlay

P-008 Cerebrospinal fluid neurotransmitter depletion in adult PKU patients
A Pilotto, N Blau, E Charyasz, P Freisinger, G Gramer, G F Hoffmann, K Scheffler, D Berg, F K Trefz

P-009 A coordinated transition model for patients with cystinosis in Spain: from pediatric to adult care
G Ariceta, J A Camacho, A Fernandez-Polo, J Gamez, J Garcia-Villoria, E Lara, P Leyes, N Martin-Begue, M Perello, G Pintos-Morell, R Torra, J V Torregrosa, S Torres-Sierra, A Vila-Santandreu, A Guell

P-010 Nitisinone in alkaptonuria—quantifying the pigmentary pathway
L R Ranganath

P-011 Transition from pediatric to adult care in patients with inborn errors of metabolism in Spanish referral centers
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P-012 Intrafamilial phenotypic variations in adult onset classical homocystinuria
C Tran, C Buerer, M R Baumgartner, D Ballhausen

P-013 Clinical characteristics of adult patients with inborn errors of metabolism from Spanish referral centers

J Perez-Lopez, L Ceberio, J S Garcia-Morillo, J M Grau-Junyent, A Hermida Ameijeiras, M Lopez-Rodriguez, J C Milisenda, M Molto-Abad, M Morales-Conejo, J J Nava Mateos

P-014 Cystinosis in adult and adolescent patients: recommendations for the comprehensive care of the disease in Spain
G Ariceta, J A Camacho, M Fernandez-Obispo, A Fernandez-Polo, J Gamez, J Garcia-Villoria, E Lara, P Leyes, N Martin-Begue, F Oppenheimer, M Perello, G Pintos-Morell, R Torra, A Vila-Santandreu, A Guell

P-015 Hyperammonaemia secondary to chemotherapy: unmasking and exacerbation of underlying urea cycle defects in two patients
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P-016 Severe missense *ASL* mutation causing relatively mild argininosuccinic aciduria in a Cypriot adult patient with learning difficulties, epilepsy and generalized whole-body tremor
G A Tanteles, T Georgiou, G Mavrikiou, V Christophidou-Anastasiadou, Y Christou, S S Papacostas, A Drousiotou

P-017 Phenylketonuria: an analysis of 67 adult patients after a transition process from a pediatric hospital
N Bulto, G Garrabou, J M Grau, F Cardellach

P-018 Altered osteoclast activity in a group of young patients affected by alkaptonuria
A Tummolo, G Brunetti, S Colucci, M Grano, L Piacente, A M Ventura, C Rizzo, M F Faienza, F Papadia

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P-020 Psychiatric and neurological symptoms in patients with Niemann-Pick disease type C: findings from the International NPC Registry
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P-021 Food neophobia, olfactory and gustatory functions in Brazilian patients with hepatic glycogen storage disease type I
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P-022 New consensus recommendations for the detection and diagnosis of Niemann-Pick disease type C
T Marquardt, P Clayton, P Gissen, M C Patterson, N P C Diagnostics Working Group

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P-024 A ceroid lipofuscinosis 11 patient in a frontotemporal lobar degeneration family due to progranulin gene mutation
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P-025 Woodhouse-Sakati syndrome: a rare cause of leukodystrophy
P Louro, J Duraes, S Paiva, P Tavares, M C Macario

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P-028 Diagnosing inborn errors of metabolism using next generation sequencing
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P-032 A new multiplex method for the diagnosis of peroxisomal disorders allowing simultaneous determination of plasma very-long-chain fatty acids, phytanic, pristanic, docosahexaenoic and bile acids by LC-MS/MS with atmospheric pressure chemical ionization
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P-033 Analysis of bile acid profiles by liquid chromatography–tandem mass spectrometry (LC-MS/MS)
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P-034 A new optimization approach for liquid chromatography ion mobility–mass spectrometry untargeted metabolomics method using experimental design
A Tebani, I Schmitz-Afonso, D N Rutledge, B J Gonzalez, S Bekri, C Afonso

P-035 Mass spectrometry based metabolomics: a promising tool for the diagnosis of inborn errors of metabolism
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P-039 The effectiveness of whole exome sequencing in unsolved patients with the clinical suspicion of mitochondrial disease
K Reinson, S Pajusalu, S Puusepp, U Murumets, R J Rodenburg, K Ounap

P-040 Quantification of plasma lysosphingolipids using LC-MS/MS: a new tool for diagnoses of sphingolipidoses
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P-041 Diagnosis of cerebrotendinous xanthomatosis using untargeted mass spectroscopy-based metabolomics—next generation metabolic screening (NGMS)
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P-044 Monitoring bile acid replacement treatment for 3β-hydroxy-Δ5-C27-steroid dehydrogenase deficiency
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P-045 Knowledgebase of inborn errors of metabolism (IEMBASE): validation and performance
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P-046 Redox profiles in patients with isolated complex I mutations
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P-047 Targeted NGS for 3-methylglutaconic aciduria
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P-056 Fibroblast metabolic flux analysis for the work-up of inherited metabolic disorders
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P-059 Quantitation of plasma organic acids (OAs) in patients with mitochondrial disorders and other inborn errors of metabolism (IEMs) by gas chromatography-mass spectrometry (GC-MS)
W J Laxen, P R Loken, M A Danielson, D Gavrilov, K Raymond, P Rinaldo, S Tortorelli, D Matern, D Oglesbee

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R Fingerhut, P Joset, S Sluka, T Herget, S M Azzarrello-Burri, A Rauch, M Baumgartner

P-067 Japan-wide gene panel study for target metabolic diseases in newborn mass screening using tandem mass spectrometry
H Sasai, H Otsuka, R Fujiki, O Ohara, Y Nakajima, T Ito, M Kobayashi, G Tajima, O Sakamoto, S Matsumoto, K Nakamura, T Hamazaki, H Kobayashi, Y Hasegawa, T Fukao

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N Ishige, K Watanabe, S Hasegawa, K Konishi, M Mashita, Y Sera, M Ishige, M Owada

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P-070 Mimicking medium chain acyl CoA dehydrogenase deficiency (MCADD) on newborn screening (NBS): an unusual iatrogenic cause of deranged acylcarnitines
S L Hulley, J Dalley, L Shakespeare, M Downing, J R Bonham, M Sharrard

P-071 A two-tier biochemical genetic approach to newborn screening for mucopolysaccharidosis type I
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P-073 Massive parallel sequencing-based genetic confirmation of metabolic disorders detected by the neonatal screeening program
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A B Burlina, G Polo, E Viggiano, M Zampieri, E Zanonato, C Cazzorla

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P-493 Intracranial hypertension in pediatric patients with cystinosis
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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: ten years of laboratory experience
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P-496 Plasma metabolomic profile in Spanish patients with lysosomal acid lipase deficiency
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P-497 Wolman disease—a misdiagnosed case with hemophagocytic lymphohistiocytosis
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W F E Kuper, M M Van Genderen, P M Van Hasselt

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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
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J J Cebolla, P Irun, M Pocovi, P Giraldo

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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

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E Pappa, C Papadopoulos, P Grimbert, G Bassez, P Laforet

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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
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P-516 Canadian Fabry Disease Initiative Study (CFDI): 8 year outcomes of a randomized controlled trial of enzyme replacement therapy (ERT)
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H Lau, N Belmatoug, P Deegan, O Goker-Alpan, I V D Schwartz, S P Shankar, Z

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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

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A Zimran, G Duran, P Giraldo, H Rosenbaum, F Giona, M Petakov, S E Solorio-Meza, P A Cooper, S Alon, R Chertkoff

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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
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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
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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
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N Guffon, G Baujat, M B Bober, B K Burton, L Clarke, P Garcia, R Giugliani, C J Hendriksz, C Lavery, J Raiman, E Jurecki, Z Sisic, A Waite

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 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

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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)
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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
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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
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P-537 Galactosialidosis: moving a step closer towards the development of enzyme replacement therapy with recombinant human protective protein/cathepsin a
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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
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P-541 Migalastat improves gastrointestinal symptoms in patients with Fabry disease: results from a double-blind, placebo-controlled phase 3 trial (FACETS)
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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
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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
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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 (CDG): 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
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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
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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
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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

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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
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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
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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 (GAA) in plasma and urine
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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
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P-583 Abnormal CSF phenylalanine level in patients presenting with disorders of tetrahydrobiopterin (BH_4) 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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P-584 Biodistribution of copper with disulfiram administered perorally in Menkes disease model mouse: new potential treatment to avoid subcutaneous injection
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
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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

P-593 Bone mineral density and vitamin D status in inborn errors of metabolism
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 (SHR)
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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 (PDE)
A Celato, I Fasan, E Zanonato, C Cazzorla, I Toldo, S Sartori, G Polo, A B Burlina

25. Miscellaneous/new disease group

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T Ben-Omran

P-600 *MBTPS2* mutations in X-linked osteogenesis imperfecta
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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
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P-617 *HACE1* deficiency mimicking mitochondrial disorder
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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
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