	Wednesday 4 September 2019
9:00-10:30	**Parallel sessions 1A-1D**
Grote Zaal	**Parallel Sessions 1A: Gene therapy**
09:00-09:15	O-018 | A phase 1/2 clinical trial of AAV8 gene therapy in adults with late-onset OTC deficiency: CAPtivate cohort 1 + 2 results ***T. Geberhiwot*** (Birmingham, United Kingdom)
09:15-09:30	O-021 | Hematopoietic stem cell gene therapy for Mucopolysaccharidosis type I, Hurler variant (MPS-IH) ***M.E. Bernardo*** *(Milan, Italy)*
09:30-09:45	O-016 | Lentiviral (LV) hematopoietic stem cell gene therapy (HSC-GT) for metachromatic leukodystrophy (MLD) provides sustained clinical benefit ***V. Calbi*** *(Milan, Italy)*
09:45-10:00	O-019 | Three year open label phase 2a investigation of venglustat safety and exploratory efficacy in classic Fabry patients ***P. Deegan*** *(Cambridge, United Kingdom)*
10:00-10:15	O-015 | Positive cohort 1 results from the phase 1/2, AAV8-mediated liver-directed gene therapy trial in glycogen storage disease type Ia (GSDIa) ***T.G.D. Derks*** *(Groningen, The Netherlands)*
10:15-10:30	O-020 | AAV Gene Therapy For Glycogen Storage Disease Type 3 ***V.P. Vidal*** *(Evry, France)*
Willem Burger Zaal	**Parallel Sessions 1B: Amino acid disorders (including PKU) and urea cycle disorders**
09:00-09:15	O-030 | Targeting CPS1: From CPS1 deficiency to lung tumor regression ***G. Makris*** *(Zurich, Switzerland)*
09:15-09:30	O-032 | Enhancement of hepatic autophagy for therapy of argininosuccinic aciduria ***L.R. Soria*** *(Pozzuoli, Italy)*
09:30-09:45	O-029 | First crystal structure of DHTKD1 provides insight into catalysis and disorders of lysine metabolism ***G.A. Bezerra*** *(Oxford, United Kingdom)*
09:45-10:00	O-027 | A Phase 1/2a Oral Placebo-controlled Study of SYNB1618 in Healthy Adult Volunteers and Subjects with Phenylketonuria ***J. Vockley*** *(Pittsburgh, USA)*
10:00-10:15	O-031 | Pharmacologic rescue of hyperammonemia-induced neurotoxicity by OAT-inhibition in a zebrafish model of acute hyperammonemic encephalopathy ***M. Zielonka*** *(Heidelberg, Germany)*
10:15-10:30	O-028 | Manipulating folate metabolism achieves near-normal homocysteine without methionine restriction in homocystinuric mice ***K.N. MacLean*** *(Aurora, USA)*
Jurriaanse Zaal	**Parallel Sessions 1C: Glycosylation and carbohydrate disorders**
09:00-09:15	O-065 | D-galactose supplementation in SLC35A2-CDG leads to clinical and biochemical improvement ***P. Witters*** *(Leuven, Belgium)*
09:15-09:30	O-062 | Crystal structure of human PMM2 enzyme as a model to evaluate missense

	variants amenable to be rescued using pharmacological chaperones ***A. Briso-Montiano*** *(Madrid, Spain)*
09:30-09:45	O-064 | Glycoproteomics for improved diagnosis of the Congenital Disorders of Glycosylation (CDG) ***M.A. Post*** *(Nijmegen, The Netherlands)*
09:45-10:00	O-063 | Using tracer metabolomics to investigate metabolic effects of galactose in different CDG ***S. Radenkovic*** *(Leuven, Belgium)*
10:00-10:15	O-039 | Nucleotide sugar profile in the galactosemia zebrafish model reveals new pathogenic mechanisms and potential readouts ***M.E. Rubio-Gozalbo*** *(Maastricht, The Netherlands)*
10:15-10:30	O-038 | Biallelic *GALM* pathogenic variants cause type IV galactosemia ***Y.W. Wada*** *(Sendai, Japan)*
Van Weelde Zaal	**Parallel Sessions 1D: Disorders of haem biosynthesis, vitamins, purines and pyrimidines**
09:00-09:15	O-068 | New insights in pyridoxamine 5'-phosphate oxidase (PNPO) deficiency from zebrafish and cell models ***J. Ciapaite*** *(Utrecht, The Netherlands)*
09:15-09:30	O-070 | Gamma aminobutyric acid and tricarboxylic acid pathways in knock-out *aldh7a1* zebrafish for pyridoxine-dependent epilepsy ***S. Mercimek-Andrews*** *(Toronto, Canada)*
09:30-09:45	O-069 | PLPHP deficiency: Clinical, genetic, biochemical, and mechanistic insights ***C.D. van Karnebeek*** *(Amsterdam, The Netherlands)*
09:45-10:00	O-050 | Clinical effectiveness of afamelanotide in patients with erythropoietic protoporphyria: an observational study ***D. Wensink*** *(Rotterdam, The Netherlands)*
10:00-10:15	O-066 | Induced pluripotent stem cells (iPSCs) and iPSC-derived cerebral organoids as a model of Succinic Semialdehyde Dehydrogenase Deficiency ***H. Brennenstuhl*** *(Heidelberg, Germany)*
10:15-10:30	O-049 | Thymidylate kinase deficiency due to mutations in *TYMK* is the cause of a novel vanishing white matter disease. ***J. Bierau*** *(Maastricht, The Netherlands)*

	Wednesday 4 September 2019
14:30-16:00	**Parallel sessions 2A-2C**
Grote Zaal	**Parallel Session 2A: Lysosomal storage disorders**
14:30-14:45	O-059 | Benefits of higher and more frequent dosing and immunomodulation on long-term outcome in classic infantile Pompe disease ***E. Poelman*** *(Rotterdam, The Netherlands)*
14:45-15:00	O-057 | A natural disease history study and a meta-analysis of published cases improve clinical knowledge on Multiple Sulfatase Deficiency

	L. Schlotawa *(Goettingen, Germany)*
15:00-15:15	O-055 | Acid ceramidase deficiency presenting as Farber disease: prospective and retrospective clinical data from an ongoing natural history study ***P. Harmatz*** *(Oakland, USA)*
15:15-15:30	O-056 | Persistent treatment effect of cerliponase alfa in children with CLN2 disease: A 3 year update from an ongoing multicenter extension study ***A. Schulz*** *(Hamburg, Germany)*
15:30-15:45	O-054 | Losartan or Propanolol treatment restores Erk1/2 activity during the cardiac remodelling of mucopolysaccharidosis type-I mice ***E. A. Gonzalez*** *(Porto Alegre, Brazil)*
15:45-16:00	O-061 | Intrathecal VTS-270 For The Treatment Of Niemann-Pick Disease, Type C1 ***P. Gissen*** *(London, United Kingdom)*
Willem Burger Zaal	**Parallel Session 2B: Advances in diagnostics (including omics)**
14:30-14:45	O-014 | The 'Cryo-Electron Microscopy Revolution' for structure determination of metabolic enzyme complexes ***H. J. Bailey*** *(Oxford, United Kingdom)*
14:45-15:00	O-012 | Discovery of novel antiquitin deficiency biomarkers through the combination of untargeted metabolomics and infrared ion spectroscopy. ***K.L.M. Coene*** *(Nijmegen, The Netherlands)*
15:00-15:15	O-013 | The Impact of Mitochondrial Bioenergetics on Metabolic Pathways by Real-time NMR of 3D Cell Cultures ***D.H. Hertig*** *(Bern, Switzerland)*
15:15-15:30	O-009 | Multiplex testing for the diagnosis of disorders involving sulfatide degradation ***K.M. Raymond*** *(Rochester, USA)*
15:30-15:45	O-010 | Algorithms for interpretation of (multi)-omics in metabolic diagnostics ***J.J.M. Jans*** *(Utrecht, The Netherlands)*
15:45-16:00	O-011 | Mitochondrial DNA mutation analysis from exome sequencing – a holistic approach in diagnostics of mitochondrial disease ***M. Wagner*** *(Munich, Germany)*
Jurriaanse Zaal	**Parallel Session 2C: Nutrition and Dietetics**
14:30-15:00	Update Lecture: Working mechanism of ketogenic diet ***J. Rho*** *(Calgary, Canada)*
15:00-15:15	O-024 | Outcome after liver transplantation in paediatric IMD- survival, growth and hospital admissions experience from one centre ***A. Daly*** *(Birmingham, United Kingdom)*
15:15-15:30	O-023 | The dietetic and biochemical basis of trimethylaminuria in one Adult Metabolic Centre. ***J. Banks*** *(Salford, United Kingdom)*
15:30-15:45	O-026 | Arginine supplementation's impact on lysine metabolism in humans: A proof-of-concept for lysine-related inborn errors of metabolism ***Z Schmidt*** *(Vancouver, Canada)*
15:45-16:00	O-025 | MyRareDiet®: A diet tracking and monitoring mHEALTH tool for research and clinical care for patients with inborn errors of metabolism. ***D. G. Hook*** *(Davis, USA)*

	Thursday 5 September 2019
11:00-12:30	**Parallel sessions 3A-3D**
Grote Zaal	**Parallel Sessions 3A: Disorders of fatty acid oxidation and metabolic myopathies**
11:00-11:30	Update Lecture: Latest insights into metabolic myopathies ***P. Laforêt*** *(Paris, France)*
11:30-11:45	O-042 | A mutation creating an upstream translation initiation codon in *SLC22A5* 5'UTR is a frequent cause of primary carnitine deficiency ***S. Ferdinandusse*** *(Amsterdam, The Netherlands)*
11:45-12:00	O-041 | Implication of post-translational modifications on ETF, a Multiple Acyl-CoA Dehydrogenase Deficiency related enzyme ***B. J. Henriques*** *(Lisbon, Portugal)*
12:00-12:15	O-008 | Novel genetic discoveries in 14 patients with multiple acyl-CoA dehydrogenation deficiency using whole exome sequencing and RNA-seq. ***S. Mosegaard*** *(Aarhus, Denmark)*
12:15-12:30	O-022 | Modified ketogenic diet in patients with Glycogen Storage Disease type V – a pilot study ***N. Loekken*** *(Copenhagen, Denmark)*
Willem Burger Zaal	**Parallel Session 3B: Organic Acidurias**
11:00-11:30	Update Lecture: Organic acidurias – it's about time to bridge the gaps ***S. Kölker*** *(Heidelberg, Germany)*
11:30-11:45	O-036 | Genetic, structural, and functional analysis of pathogenic variations causing methylmalonyl-CoA epimerase deficiency. ***D.S. Froese*** *(Zurich, Switzerland)*
11:45-12:00	O-034 | Long-term outcome of methylmalonic aciduria after kidney, liver or combined liver-kidney transplantation: the French experience ***A. B. Brassier*** *(Paris, France)*
12:00-12:15	O-033 | DHTKD1 and OGDH form a hybrid ketoacid dehydrogenase complex with functional redundancy toward α-ketoadipic acid ***J. Leandro*** *(New York, USA)*
12:15-12:30	O-037 | Metabolic liver disease in a tailored mouse model of ***mut***-type methylmalonic aciduria ***M. Lucienne*** *(Zurich, Switzerland)*
Jurriaanse Zaal	**Parallel Sessions 3C: Disease modeling**
11:00-11:15	O-060 | Zebrafish as a model to study cystinosis: from pathophysiology to drug screening ***S. P. B. Berlingerio*** *(Leuven, Belgium)*
11:15-11:30	O-035 | Increased ammonium production and reduced appetite in a knock-in rat model for

	glutaric aciduria type I challenged with high lysine diet ***M.G.M. Gonzalez-Melo*** *(Lausanne, Switzerland)*
11:30-11:45	O-040 | CRISPR/Cas9-mediated editing of hepatic *G6pc* in mice allows to investigate the clinical spectrum of glycogen storage disease type Ia ***M.G. S. Rutten*** *(Groningen, The Netherlands)*
11:45-12:00	O-073 | Molecular and metabolic consequences of renal carnosinase deficiency ***V. Peters*** *(Heidelberg, Germany)*
12:00-12:15	O-045 | Brain on a chip; disease modeling and drug screening in a neurophysiological model of MELAS disease ***T. Kozicz*** *(Rochester, USA)*
12:15-12:30	O-017 | Engineered human iPSC-derived skeletal muscles to model Pompe disease ***P. Herrero-Hernandez*** *(Rotterdam, The Netherlands)*
Van Weelde & Mees Zaal	**Parallel Sessions 3D: Patient cohorts and follow-up**
11:00-11:15	O-076 | Patterns of abnormal movements in neonatal inborn errors of metabolism: study following videofilmation analysis and volumetric brain MRI ***A. Darling*** *(Barcelona, Spain)*
11:15-11:30	O-077 | Genotype-phenotype correlation and natural history in NBAS deficiency ***C. S. Staufner*** *(Heidelberg, Germany)*
11:30-11:45	O-058 | Follow-up of patients identified by MPS I newborn screening ***D. Gueraldi*** *(Padova, Italy)*
11:45-12:00	O-067 | Very long-term follow-up of a series of patients with CblA deficiency ***C. Marelli*** *(Montepellier, France)*
12:00-12:15	O-052 | *PEX12* gene mutation causing a peroxisomal biogenesis disorder among Egyptian patients with founder mutations ***M. S. Zaki*** *(Cairo, Egypt)*
12:15-12:30	O-051 | Clinical, biochemical and genetic characteristics of a cohort of 101 French and Italian patients with HPRT deficiency ***A. Madeo*** *(Genoa, Italy)*

	Thursday 5 September 2019
14:00-15:30	**Parallel sessions 4A-4D / EDUCATIONAL SESSION**
Grote Zaal	**Parallel Session 4A: Mitochondrial disorders**
14:00-14:15	O-046 | *KGD4* biallelic variants in two siblings with bilateral striatal necrosis: a new gene of Krebs cycle associated with Leigh syndrome ***S. Garosi*** *(Rome, Italy)*
14:15-14:30	O-048 - Interferon signature: a new biomarker to follow disease progression in Pearson and Kearns-Sayre syndrome ***D. Martinelli*** *(Rome, Italy)*
14:30-14:45	O-078 | Assay for simultaneous evaluation of aminoacyl-tRNA synthetase activities: application to mitochondrial aspartyl tRNA synthetase ***M. I. Mendes*** *(Amsterdam, The Netherlands)*

14:45-15:00	O-047 | No effect of Resveratrol supplementation in patients with mitochondrial myopathy – a randomized, double-blind, cross-over study ***N. Loekken*** *(Copenhagen, Denmark)*
15:00-15:15	O-001 | A six year prospective follow-up study in 151 carriers of the m.3243A>G mutation. ***P. De Laat*** *(Nijmegen, The Netherlands)*
15:15-15:30	O-044 | Mutations in *SQOR* encoding sulfide:quinone oxidoreductase are a potentially treatable cause of Leigh disease ***J. L. K. Van Hove*** *(Aurora, USA)*
Willem Burger Zaal	**Parallel Session 4B: EDUCATIONAL SESSION**
14:00-14:30	Basics and clinical applications of gene editing: hopes, hypes and hurdles ***R. Hoeben*** *(Leiden, The Netherlands)*
14:30-15:00	Improving gene editing outcomes: separating the good from the bad and ugly ***M. Goncalves*** *(Leiden, The Netherlands)*
15:00-15:30	A bright future for plant genome editing: implications for global production and societies ***S. Smeekens*** *(Utrecht, The Netherlands)*
Jurriaanse Zaal	**Parallel Session 4C: Metabolic disorders in adults**
14:00-14:15	O-003 | Eye movement disorders as an aid to early detection of late-onset inborn errors of metabolism ***L. H. Koens*** *(Groningen, The Netherlands)*
14:15-14:30	O-002 | Does nitisinone modify ochronosis in alkaptonuria - experience from the United Kingdom National Alkaptonuria Centre ***L. Ranganath*** *(Liverpool, United Kingdom)*
14:30-14:45	O-006 | Atypical cerebral palsy: yield of genetic diagnostics in the adult metabolic disease clinic ***G. Horvath*** *(Vancouver, Canada)*
14:45-15:00	O-005 | Longitudinal cohort study of cardiac complications in Fabry disease ***M. Langeveld*** *(Amsterdam, The Netherlands)*
15:00-15:15	O-007 | Metabolic control modulates neuropsychiatric involvement, brain damage and neurodegenerative markers in adult Phenylketonuria patients ***A. Pilotto*** *(Brescia, Italy)*
15:15-15:30	O-004 | Are we failing to support the cognitive and mental health of adults with IMDs? Developing neuropsychology services in one Metabolic Centre. ***C. Chen*** *(Salford, United Kingdom)*
Van Weelde & Mees Zaal	**Parallel Session 4D: Novel disease genes**
14:00-14:15	O-075 | *RINT1* biallelic variants cause a novel disorder of infantile onset recurrent acute liver failure and skeletal anomalies ***M. A. Cousin*** *(Rochester, USA)*
14:15-14:30	O-071 | Mutations in *PCYT2* disrupt etherlipid biosynthesis and cause a complex hereditary spastic paraplegia ***F. M. Vaz*** *(Amsterdam, The Netherlands)*
14:30-14:45	O-074 | Possible new peroxisomal disorder associated with *IDI1* mutation and identified on global metabolomics study ***H. Alharbi*** *(Los Angeles, USA)*

14:45-15:00	O-072 | MDH1 deficiency is a metabolic disorder of the malate-aspartate shuttle associated with early onset severe encephalopathy ***M. H. Broeks*** *(Utrecht, The Netherlands)*
15:00-15:15	O-043 | Identification of mutations in mitochondrial ribosomal protein PTCD3 as a novel cause of Leigh syndrome ***Y. Sugiyama*** *(Chiba, Japan)*
15:15-15:30	O-053 | Identification of an autosomal dominant Zellweger Spectrum Disorder ***H. R. Waterham*** *(Amsterdam, The Netherlands)*