

Tuesday 3 September 2019	
14:00-14:30 <i>Grote Zaal</i>	OPENING CEREMONY SSIEM 2019
14:30-16:00 <i>Grote Zaal</i>	Plenary Session 1: Building Bridges Chairs:
14.30-15.00	Organoids as models of human disease Hans Clevers (<i>Utrecht, The Netherlands</i>)
15.00-15.30	The role of DNA damage repair and transcription stress in aging and the impact of nutrition Jan Hoeijmakers (<i>Rotterdam, The Netherlands</i>)
15.30-16.00	The lysosome as a control center of cell metabolism Andrea Ballabio (<i>Naples, Italy</i>)
16:00-16:30	Coffee Break
16.30-18.00 <i>Grote Zaal</i>	Plenary Session 2: RNA based therapy and Gene therapy Chairs:
16.30-17.00	AAV gene therapy for inherited diseases Alberto Auricchio (<i>Naples, Italy</i>)
17.00-17.30	HSC gene therapy for neurometabolic and neurodegenerative disorders Alessandra Biffi (<i>Cambridge, USA</i>)
17.30-18.00	Opportunities and challenges for RNA therapy development Annemieke Aartsma Rus (<i>Leiden, The Netherlands</i>)
18:00-18:30 <i>Grote Zaal</i>	GARROD AWARD LECTURE
18:30-20:30	WELCOME RECEPTION

Wednesday 4 September 2019	
9:00-10:30	Parallel sessions 1A-1D
<i>Grote Zaal</i>	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 (<i>Birmingham, United Kingdom</i>)
09:15-09:30	O-021 Hematopoietic stem cell gene therapy for Mucopolysaccharidosis type I, Hurler variant (MPS-IH) M.E. Bernardo (<i>Milan, Italy</i>)
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 (<i>Milan, Italy</i>)
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 (<i>Cambridge, United Kingdom</i>)
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 (<i>Groningen, The Netherlands</i>)
10:15-10:30	O-020 AAV Gene Therapy For Glycogen Storage Disease Type 3 V.P. Vidal (<i>Evry, France</i>)
<i>Willem Burger Zaal</i>	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 SYN1618 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)
Juriaanse 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 <i>GALM</i> pathogenic variants cause type IV galactosemia Y.W. Wada (Sendai, Japan)
Van Weelde & Mees 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 <i>aldh7a1</i> 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 <i>TYMK</i> is the cause of a novel vanishing white matter disease. J. Bierau (Maastricht, The Netherlands)

10:30-11:00	Coffee Break
11.00-12.30 <i>Grote Zaal</i>	Plenary Session 3: Regenerative medicine Chairs:
11.00-11.30	Hurdles in successful examples of stem cell-based regenerative medicine Graziella Pellegrini (Modena, Italy)
11.30-12.00	Ex vivo gene editing studies with human hepatocytes from patients with urea cycle defects Stephen Strom (Stockholm, Sweden)
12.00-12.30	Human induced pluripotent stem cells come of age in modeling cardiovascular disease Christine Mummery (Leiden, The Netherlands)
12:30-14:30	LUNCH / POSTER VIEWING
14:30-16:00	Parallel sessions 2A-2D
<i>Grote Zaal</i>	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)
<i>Willem Burger Zaal</i>	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 niti (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)
<i>Jurriaanse Zaal</i>	Parallel Session 2C: Nutrition and Dietetics

14:30-15:00	Update Lecture: Working mechanism of ketogenic diet J. Rho (San Diego, USA)
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)
16:00-16:30	Coffee break
16.30-18.00 <i>Grote Zaal</i>	Plenary Session 4: Mitochondrial Chairs:
16.30-17.00	Moving towards clinical trials for mitochondrial disease Shamima Rahman (London, United Kingdom)
17.00-17.30	The Pursuit of Precision Mitochondrial Medicine: Harnessing pre-clinical cellular and animal models of mitochondrial disease Marni J. Falk (Philadelphia, USA)
17.30-18.00	Mitochondria in Alzheimer disease: It's not what you think Eric A Schon (New York, USA)
18:00 – 20:00	POSTER WALK

	Thursday 5 September 2019
09.00-10.30 <i>Grote Zaal</i>	Plenary session 5: Metabolomics / System biology Chairs:
09.00-09.30	Modeling human metabolism in health and disease Ines Thiele (Galway, Ireland)
09.30-10.00	Innovative analytical tools for metabolomics-driven systems biology to investigate monogenetic and complex disorders Thomas Hankemeier (Leiden, The Netherlands)
10.00-10.30	Integrative network biology approaches to understand rare inborn errors of metabolism Carmen Argmann (New York, USA)
10:30-11:00	Coffee break
11:00-12:30	Parallel sessions 3A-3D
<i>Grote Zaal</i>	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 <i>SLC22A5</i> 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 -ketoacidic acid J. Leandro (New York, USA)
12:15-12:30	O-037 Metabolic liver disease in a tailored mouse model of <i>mut</i> -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 <i>G6pc</i> 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 (Montpellier, France)
12:00-12:15	O-052 <i>PEX12</i> 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)
12:30-14:00	LUNCH / POSTER VIEWING
14:00-15:30	Parallel sessions 4A-4D / EDUCATIONAL SESSION
Grote Zaal	Parallel Session 4A: Mitochondrial disorders
14:00-14:15	O-046 <i>KGD4</i> biallelic variants in two siblings with bilateral striatal necrosis: a new gene of Krebs cycle associated with Leigh syndrome S. Galosi (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 <i>SQOR</i> 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: CRISPR/CAS
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)

<i>Van Weelde & Mees Zaal</i>	Parallel Session 4D: Novel disease genes
14:00-14:15	O-075 <i>RINT1</i> 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 <i>PCYT2</i> 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 <i>IDI1</i> 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 PTC3 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)

Friday 6 September 2019	
09.00-10.30 <i>Grote Zaal</i>	Plenary Session 6: Lysosomes-Innovative insights Chairs:
09.00-09.30	Disorders of lysosome-related organelle biogenesis Marjan Huizing (Bethesda, USA)
9:30-10:00	Challenges and concepts to combat diseases caused by deficiencies of lysosomal hydrolases and membrane proteins Paul Saftig (Kiel, Germany)
10:00-10:30	The future of therapies for Lysosomal disorders, lessons from the past Simon Jones (Manchester, United Kingdom)
10:30-11:00	Coffee break
11:00-11:45 <i>Grote Zaal</i>	KOMROWER LECTURE Chair: Gajja Salomons
	From fatty acid oxidation to riboflavin - Make metabolites great again! Christine Vianey-Saban (Lyon, France)
12:00-13:00 <i>Grote Zaal</i>	Late Breaking News Session
12:00-12:15	Oral presentation TBA
12:15-12:30	Oral presentation TBA
12:30-12:45	Oral presentation TBA
12:45-13:00	Oral presentation TBA
13:00-13:30 <i>Grote Zaal</i>	Awards Presentation Closing remarks