
2nd ANNOUNCEMENT



ANNUAL SYMPOSIUM 2019

3-6 SEPTEMBER | ROTTERDAM, THE NETHERLANDS

Building Bridges



www.ssiem2019.org



Welcome Address by Symposium President

Dear Colleagues,

On behalf of the Local Organizing Committee, I have great pleasure in announcing that the SSIEM 2019 Symposium will be held in Rotterdam, the Netherlands, from September 3 to September 6.

We are truly excited to be welcoming you to Rotterdam. Voted number 5 in Lonely Planet's Best in Travel in 2016, Rotterdam is a dynamic city known for its maritime history, multiethnic community, futuristic architecture and spirit of innovation.

As we know from life in a city that's continuously and rapidly renewing itself, innovation is indeed embedded in our culture. Just take our architecture: every year, new buildings and structures are added to our landscape, some of the finest being the bridges connecting different parts of the city.

You might say it's in Rotterdam's genes to build bridges and seek connections. With the biggest port in Europe, we seek connection with the rest of the world. Within the city itself, we connect at various levels, including business, culture and science.

That's why our congress theme is Building Bridges. During SSIEM 2019, we hope to connect new-generation scientists with those who are well established. We hope to build bridges between laboratory and clinic, and to connect up the knowledge which has been generated on a range of diseases. All with the common goal of achieving a better future for our patients.

The congress will be held in De Doelen, a city-center, state-of-the-art venue for conferences and concerts. It's just a short walk from good hotels and Rotterdam Central Station, which is reached by direct bus from Rotterdam The Hague Airport, and is only 20 minutes by train from Schiphol Airport Amsterdam.

We look forward to welcoming you in Rotterdam next year, the city that never sleeps and where innovation never stops.

Kind regards,

A blue ink handwritten signature, appearing to be 'Ans van der Ploeg'.

Ans van der Ploeg
Symposium President



SYMPOSIUM SECRETARIAT
Congress by design
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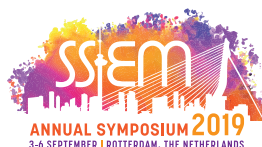
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LATE BREAKING NEWS

Late Breaking News deadline - 22 July 2018

Abstracts of exceptional originality and importance, completed after 23 April 2019 may be considered for oral presentation in the Late Breaking News session on Friday if submitted before 22 July 2019 directly by email to info@ssiem2019.org. These abstracts will not be included in the digital issue of Abstracts. In past years most abstracts submitted late could not be accepted and were rejected.



SCIENTIFIC PRELIMINARY PROGRAM

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: t.b.c.
14:30-15:00	Hans Clevers (Utrecht, The Netherlands) <i>Title: Organoids as models of human disease</i>
15:00-15:30	Jan Hoeijmakers (Rotterdam, The Netherlands) <i>Title: The role of DNA damage repair and transcription stress in aging and the impact of nutrition</i>
15:30-16:00	Andrea Ballabio (Naples, Italy) <i>Title: The lysosome as a control center of cell metabolism</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: t.b.c.
16:30-17:00	Alberto Auricchio (Naples, Italy) <i>Title: AAV gene therapy for inherited diseases</i>
17:00-17:30	Alessandra Biffi (Cambridge, USA) <i>Title: HSC gene therapy for neurometabolic and neurodegenerative disorders</i>
17:30-18:00	Annemieke Aartsma Rus (Leiden, The Netherlands) <i>Title: Opportunities and challenges for RNA therapy development</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
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 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)

<i>Jurjaanse Zaal</i>	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)
<i>Van Weelde & Mees zaal</i>	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 (Vancouver, Canada)
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)
10:30-11:00	COFFEE BREAK

11:00-12:30 <i>Grote Zaal</i>	Plenary Session 3: Regenerative medicine Chairs: t.b.c.
11:00-11:30	Graziella Pellegrini (Modena, Italy) <i>Title: Hurdles in successful examples of stem cell-based regenerative medicine</i>
11:30-12:00	Stephen Strom (Stockholm, Sweden) <i>Title: Ex vivo gene editing studies with human hepatocytes from patients with urea cycle defects</i>
12:00-12:30	Christine Mummery (Leiden, The Netherlands) <i>Title: Human induced pluripotent stem cells come of age in modeling cardiovascular disease</i>
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. Harnatz (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 antitoxin 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 Cross-omics: a diagnostic multi-omics approach 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 (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)
16:00-16:30	COFFEE BREAK
16:30-18:00 <i>Grote Zaal</i>	Plenary Session 4: Mitochondrial Chairs: t.b.c.
16:30-17:00	Shamima Rahman (London, United Kingdom) <i>On The state of art of clinical trials</i>
17:00-17:30	Marni J. Falk (Philadelphia, USA) <i>On Advances in clinical diagnosis, treatment and prevention of mitochondrial disease</i>
17:30-18:00	Eric A. Schon (New York, USA) <i>On Mitochondrial associated membrane and mito-ER links</i>
18:00 – 20:00	POSTER WALK

Thursday 5 September 2019	
09:00-10:30 Grote Zaal	Plenary session 5: Metabolomics / System biology Chairs: t.b.c.
09:00-09:30	Ines Thiele (Luxembourg, Luxembourg) <i>Title: Modeling human metabolism in health and disease</i>
09:30-10:00	Thomas Hankemeijer (Leiden, The Netherlands) <i>Title: On Innovative analytical tools for metabolomics-driven systems biology in personalized health strategies. Metabolomics & high throughput analysis to investigate monogenetic and complex disorders</i>
10:00-10:30	Carmen Argmann (New York, USA) <i>Title: On Generating network models in inflammatory bowel disease; finding novel human beta cell regeneration strategies for Type 2 Diabetes and uncovering genetic modifiers of screenable inborn errors of metabolism to explain discrepancies between genotype and phenotype</i>
10:30-11:00	COFFEE BREAK
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)

<i>Jurriaanse Zaal</i>	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)
<i>Van Weelde & Mees Zaal</i>	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. Guerardi (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)
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 KGD4 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 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	R. Hoeben (Leiden, The Netherlands) <i>Title: Basics and clinical applications of gene editing: hopes, hypes and hurdles</i>
14:30-15:00	M. Goncalves (Leiden, The Netherlands) <i>Title: Improving gene editing outcomes: separating the good from the bad and ugly</i>
15:00-15:30	S. Smeekeens (Utrecht, The Netherlands) <i>Title: A bright future for plant genome editing: implications for global food production and societies</i>
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 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)

Friday 6 September 2019	
09:00-10:30 Grote Zaal	Plenary Session 6: Lysosomes-Innovative insights Chairs: t.b.c.
09:00-09:30	<i>Speaker to be announced</i>
9:30-10:00	Marjan Huizing (Bethesda, USA) <i>Title: Disorders of lysosome-related organelle biogenesis</i>
10:00-10:30	Paul Saftig (Kiel, Germany) <i>Title: Challenges and concepts to combat diseases caused by deficiencies of lysosomal hydrolases and membrane proteins</i>
10:30-11:00	COFFEE BREAK
11:00-11:45 Grote Zaal	KOMROWER LECTURE Chair: Gajja Salomons
	Christine Vianey-Saban (Lyon, France) <i>Title: From fatty acid oxidation to riboflavin - Make metabolites great again!</i>
12:00-13:00 Grote Zaal	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 Grote Zaal	Awards Presentation Closing remarks

NETWORKING ACTIVITIES

Young Investigators event

On Monday 2 September there will be a special event for Young Investigators. This event will take place from 20:00 at the Doelen Café.

Young Investigators can register for the event during Symposium registration.

Welcome Reception

On Tuesday 3 September there will be a welcome reception from 18:30 to 20:30 at De Doelen.

You will be welcomed with drinks and small bites.

Networking Evening

On Thursday 5 September a Networking Evening will take place at the harbour of Rotterdam. The Networking Evening will start at 20:30.

You will be transferred to the venue by boat. Please note that you have to register for the Networking Evening during Symposium registration.

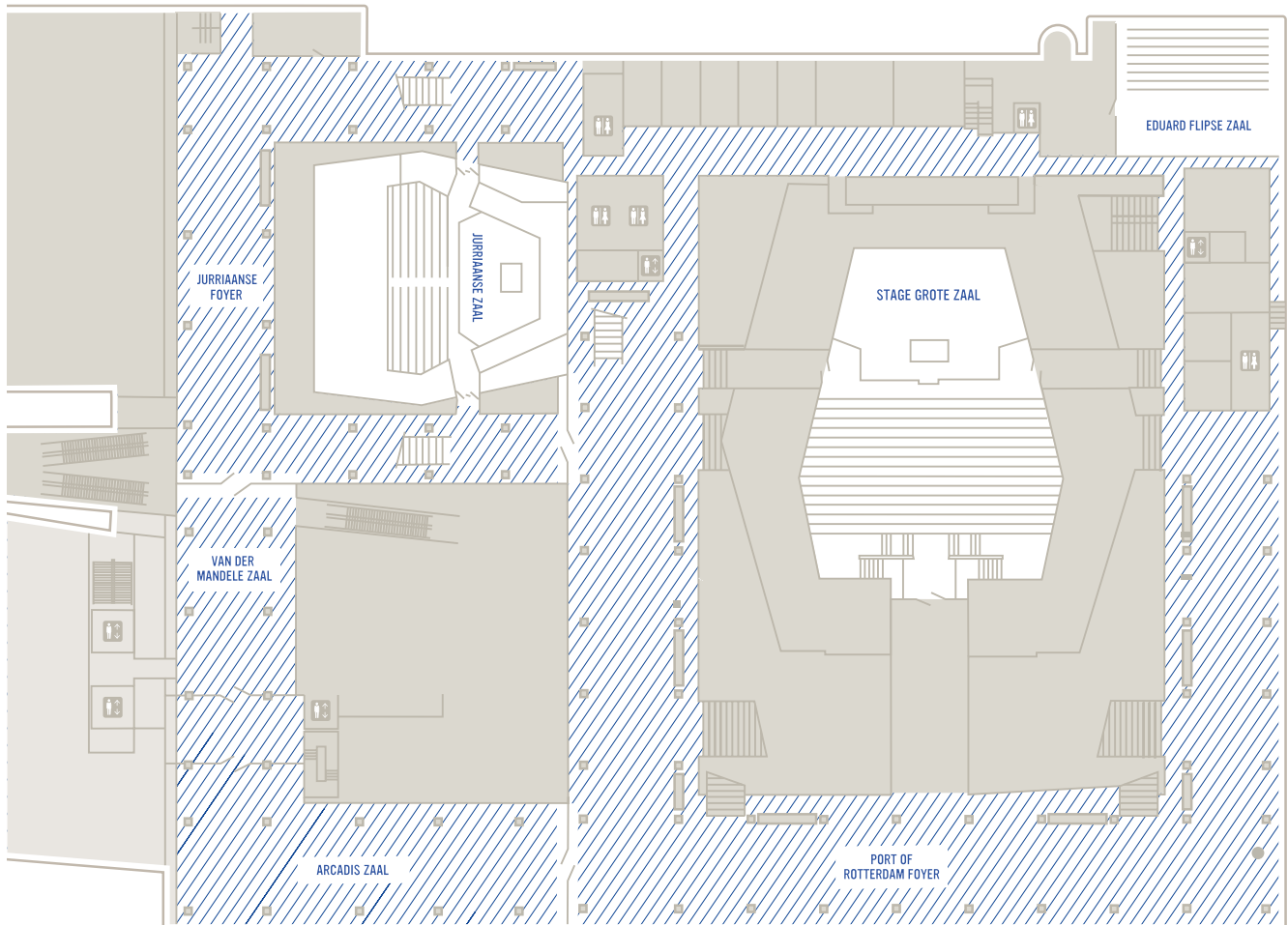
Networking activities

Several **networking activities** will be organized on Thursday 5 September. We hope that you will take advantage of the many opportunities for learning, networking, and socializing at SSIEM 2019. You can sign up for the social events during the online registration.

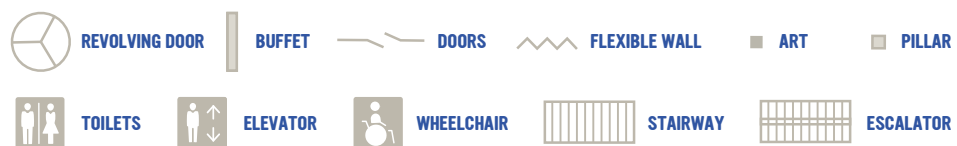


VENUE FLOOR PLANS

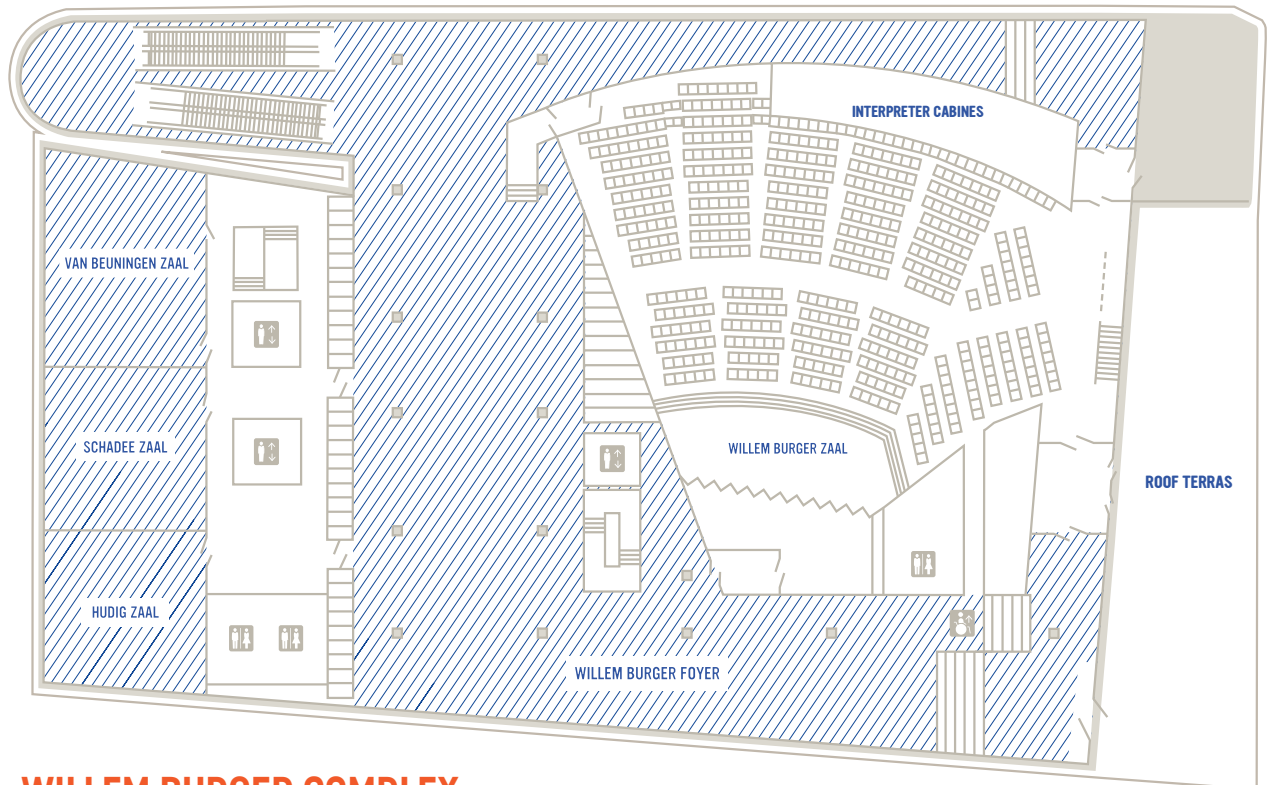
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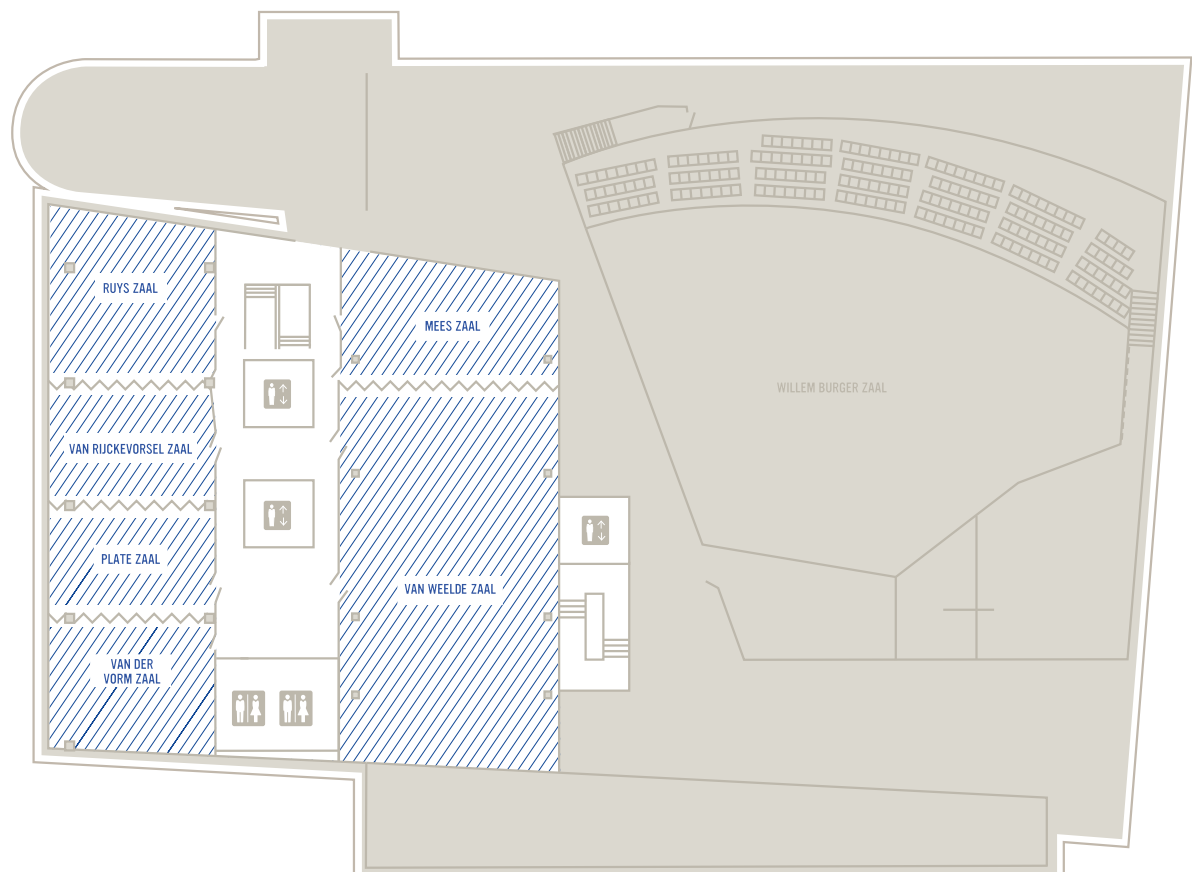
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WILLEM BURGER COMPLEX THIRD FLOOR



WILLEM BURGER COMPLEX FOURTH FLOOR



GENERAL INFORMATION

Congress Venue

SSIEM 2019 will be held at De Doelen, a city-centre, state-of-the-art venue for conferences and concerts, which is just a short walk from good hotels and Rotterdam Central Station. The latter is reached by direct bus from Rotterdam The Hague Airport, and is only 25 minutes by train from Schiphol Airport Amsterdam.

Address

De Doelen, Schouwburgplein 50
3012 CL Rotterdam - The Netherlands
www.dedoeleniccrotterdam.nl/en/

Public Transport

Public transport is the best option for all those travelling to Rotterdam. De Doelen is just a few minutes' walk from Rotterdam Central Station where trains, metros, buses and trams all stop.

Walking from Rotterdam Central Station

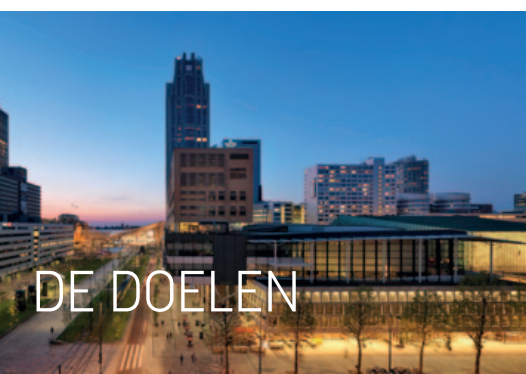
Click [here](#) for the walking route from Rotterdam Central Station to De Doelen.
This route will take you to the entrance of the Grote Zaal Complex.

Parking

There are several secure car parks in the immediate vicinity. Tickets for Parking Schouw-burgplein 1 and 2 are available at reduced rates from De Doelen.

Cloakrooms

At the conference centre various cloakrooms are available.



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Endless innovation brings powerful solutions.

* The residual amount of Phe is due to the presence of minor amounts of other proteins/peptides.



How to reach De Doelen

The symposium will be held in De Doelen Rotterdam, Schouwburgplein 50 in Rotterdam, The Netherlands. For your flight, you can fly to Amsterdam Schiphol Airport. Although most of the flights go to Amsterdam, for some flights it is also possible to fly to Rotterdam The Hague Airport. From Amsterdam Schiphol Airport you can take the Intercity Direct train that takes you to downtown Rotterdam under 30 minutes!

If your flight goes to Rotterdam The Hague Airport you can take a taxi or bus nr. 33 to Rotterdam Central Station from where it is just a few minutes walk to De Doelen.

Check [this video](#) for travel instructions.

For more information about the time tables please check www.ns.nl/en.

Language

The official language of the Symposium is English.

Time

Rotterdam is two hours ahead of Greenwich Mean Time, six hours ahead of Eastern Standard Time and has the Central European Time.

Climate

Rotterdam enjoys an average temperature of 15°C to 19°C/59° F to 66° F during the month of September.

Electricity

The electrical power supply "voltage" in The Netherlands is: 220-240 Volts. .
(U.S./Canada: 110-120 Volts)

Emergency Telephone Numbers

The emergency phone number for the police, fire department and ambulance is 112.

Travel and Health Insurance

It is strongly recommended that participants take out insurance to cover loss (including registration fees) incurred in case of cancellation, medical expenses and damage to or loss of personal effects. The organizers of the SSIEM Annual Symposium will not be responsible for any medical costs incurred by participants. The Local Organizing Committee will not accept liability for personal injuries or for loss or damage to property belonging to Congress participants, either during or as a result of the Congress or during any tours.





- Sobi is an international rare disease company dedicated to making a significant difference to patients with **Genetic and Metabolic diseases** by providing access to innovative treatments



Currency

The Netherlands is a full member of the European Union, and its currency is the Euro (€). All major credit cards are widely accepted in The Netherlands.

Visa Requirements/Letter of Invitation

Visa regulations depend on your nationality and country of origin. Please contact your local Embassy / Consulate for full and official instructions on the specific visa regulations and application procedures that apply to you. It is the responsibility of the participant to obtain a visa, if required. Individuals requiring an official Letter of Invitation can contact the Symposium Secretariat. The Letter of Invitation does not financially obligate the Congress organizers in any way. All expenses incurred in relation to the Congress are the sole responsibility of the attendee. If you wish to receive a Visa invitation letter, please contact the Symposium Secretariat.

Lost and Found

Lost and found items can be recovered at the Registration Desk.





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Amicus Therapeutics is a global, patient-dedicated biotech company focused on discovering, developing, and delivering novel high-quality medicines for people living with rare metabolic diseases.

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**To learn more, visit
www.amicusrx.com**

Amicus is proud to be a sponsor of SSIEM 2019.
We look forward to seeing you in Rotterdam.

NP-NN-ALL-00050419
Date of Preparation: May 2019

LOCAL INFORMATION

Host City

Welcome to Rotterdam: a young, dynamic metropolis that continuously and rapidly renews itself at a rapid rate. A city with sparkling skyscrapers, a world-class port, famous museums, awesome festivals, trendy restaurants and fabulous food markets – each the product of Rotterdammers and their celebrated “can-do” mentality.

Experience the city’s many extraordinary attractions and tours – by bike, bus, boat or on foot – and you’ll see why Rotterdam was voted one of the world’s top ten cities in Lonely Planet’s Best in Travel 2016.

Innovation

After the bombing of the Second World War, Rotterdam reinvented itself with characteristic energy and innovation. Working under a then ground-breaking motto – light, air and space – it broke with the past and welcomed the new age. And in a city that still embraces experimentation, anything is possible. From a rooftop city farm to a music festival in the harbour, from an exclusive rooftop bar to a brewery in a former warehouse, you’ll be enthralled by its vigour and inventiveness.

City of architecture

If you love architecture, Rotterdam won’t disappoint you. Its patchwork of styles combines icons of the Nieuwe Bouwen (c. 1920-1960) with buildings of the post-war reconstruction period (c. 1960-1980). These clash cheerfully with the hypermodern landmarks of recent decades Erasmusbrug, Rotterdam Central Station, De Rotterdam building, and the Timmerhuis complex. Then, of course, there’s the Markthal. In the first indoor market hall in the Netherlands. Here, alongside, nearly 100 stalls selling fresh produce you’ll find food shops and a range of restaurants that includes Jamie Oliver’s Jamie’s Italian. When you’re there, don’t forget to look up to the ceiling where you will find amazing artwork.

Port City

As a port city, Rotterdam has indissoluble ties to the water. The Maas River flows through the city, supplying its many harbours, such as historic Delfshaven. Downstream, these merge seamlessly with the industrial port, which stretches 40 kilometres to Hoek van Holland and the new harbour complex at Maasvlakte 2.





METABOLICS

Tuesday
September 3rd
12.45 – 13.45

JOIN THE NUTRICIA METABOLICS SATELLITE SYMPOSIUM

Setting Solid Foundations in Early Life

The importance of nutrition for the fetus,
infant and growing child with IEM

Session Chair

Clara D.M. van Karnebeek, MD PhD

Pre- and postnatal nutritional interaction between mother and child

François Feillet, MD PhD

Relevance of precision nutrition in early life

Bernd Stahl, PhD

The impact of nutrition in the growing child

Anita MacDonald, PhD

MUST SEES ROTTERDAM

Rotterdam Centraal

Rotterdam Central Station is one of the most iconic architectural sites in Rotterdam. The roof over the tracks is covered in solar panels and the striking hall roof points towards the city centre. A number of historic elements from the former station building (1957) by Sybold van Ravesteyn have been re-used, like the original clock in the front façade and the letters spelling out 'Centraal Station'. In the main hall you can find several shops, information about public transport and the Rotterdam Tourist Information. If you're looking for original souvenirs, don't miss shopping at 'Love Rotterdam. Gifts, Food & More.' Need time to take in the architectural masterpiece? Visit rooftopbar The Suicide Club for a beautiful view of Rotterdam Central Station.

Markthal

You'll find an indoor market hall in various world-class cities, but the combination with luxury housing makes Rotterdam's Markthal the first of its kind. The apartments are arched over the food market in a horseshoe configuration. The main hall houses the market itself, as well as shops and various restaurants, with a supermarket and a four-storey car park situated below. As you wander amongst the market stands, look up to enjoy the massive artwork sprawled across the ceiling: the 'Horn of Plenty' by Arno Coenen and Iris Roskam. Don't forget to visit one of the country's biggest outdoor markets which is held on the large square (Binnenrotte) in front of the Markthal (Tuesday and Saturday).

Cube Houses

The Cube Houses (Piet Blom, 1978-1984) are part of the Blaakse Bos (Blaakse Wood) development which borders on the Laurenskwartier district and the Waterfront area. The architect wanted to design a kind of village within the city and saw the houses as trees and the whole area as a wood. The Cube Houses are tipped to one side as it were, making three sides face the ground and three face the sky. Don't forget to visit the Kijk-Kubus (Show-Cube) to experience what life is like in a Cube House.





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- **Gaucher disease**
- **Fabry disease**

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C-ANPROM/INT//5506 | Date of preparation: May 2019

ss Rotterdam

The ss Rotterdam (1959) is the former flagship of the Holland-America Line and boasts a wealth of history. The steamship was built in Rotterdam, returning to its home port in 2008 after many voyages and nautical miles. Since then, it has been moored at the tip of the Katendrecht peninsula. It's used as a hotel, restaurant and events venue. The terrace on deck, including a small pool, offers a unique view of the city. The general areas on the ship, including the restaurants, are open to the public. The ss Rotterdam also offers various tours of the decks, corridors, staterooms and platforms, some lasting only an hour and others as long as two; the tours include an audiophone and additional explanations by the ship hosts. The tours show the amazing interior of a large luxury cruise liner and let visitors explore what life used to be like on board. The Rotterdam Complete Tour also includes a peek inside the engine room.

Erasmusbrug

The 800-metre long Erasmus Bridge (Ben van Berkel, 1996) spans the Nieuwe Maas River and links the northern and southern parts of Rotterdam. The bridge is part of the redevelopment of an old port area and was specifically designed to be an iconic structure to give the area a strong presence. The suspension bridge has a 139-metre high steel pylon which is secured with 40 cables. The shape of the pylon gave the bridge its nickname: The Swan.

Euromast

The Euromast observation tower was constructed to mark the Floriade gardening extravaganza in 1960. The tower, made from reinforced concrete, has an inner diameter of 9 metres with walls that are 30 cm thick. Its floor hangs 96 metres above the ground. The Euromast lift takes just 30 seconds to whisk visitors to a height of 100 metres, where there is an observation deck and brasserie restaurant. The Euroscope revolving lift takes people up to 185 metres. Euromast is Europe's only attraction from which you can abseil to the ground. Of course, this only takes place under the guidance of a professional.

Rotterdamse Haven

The Rotterdam harbour is one of the largest seaports in the world amid the hustle and bustle of various inland and seagoing vessels. In the harbour there are unique views of wharfs, docks and ultramodern transshipment operations involving thousands of containers.





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Learn More About Our Programmes:

- MPSVII: Mucopolysaccharidosis VII
- LC-FAOD: Long-Chain Fatty Acid Oxidation Disorders
- OTC: Ornithine Transcarbamylase Deficiency
- GSDIa: Glycogen Storage Disease Type Ia
- GSDIII: Glycogen Storage Disease Type III
- CTD: Creatine Transporter Deficiency
- WD: Wilson Disease

Visit us at our SSIEM Booth
ultragenyx.com

De Rotterdam

De Rotterdam was designed by internationally well-known Rotterdam-based architectural firms OMA/Rem Koolhaas and MVRDV. The 31st floor of De Rotterdam – at a height of 105 meters – offers a stunning 360° view of the city.

Fenix Loods & Food Hallen

Good news! Now that the Foodhallen on Wilhelmina Pier are open, visiting the Markthal, Foodhallen and Fenix Food Factory is a must. You have the chance to explore different kinds of food and cuisines at the Food Hallen and Fenix Loods.

Coolhaven Island

Coolhaven Island is a vibrant multicultural neighbourhood. Coolhaven Island is a working-class neighbourhood currently undergoing development, with multicultural diversity and the latest trends and artistic movements. Living at Coolhaven Island is El Aviva/hairdresser Tati – the inventor of the famous fast food dish known as ‘kapsalon’ (hair salon).

Water Plaza

Water Plaza is a world famous project. Intervention, architectural design, climate adaptation, leisure and water management have converged perfectly in this project, setting an example for the world.

St. Lawrence

The Laurenskerk, or Church of St. Lawrence, originally arose on the banks of the River Rotte and its location can truly be called the very birthplace of Rotterdam. It is an imposing church built between 1449 and 1525, and it is Rotterdam’s only surviving late Gothic building. After the bombing of Rotterdam in the Second World War, the image of the heavily damaged church and the reconstruction works that followed became a symbol of all that the city and its people had endured. Outside of the St Lawrence there is the oldest statue in the Netherlands, Desiderius Erasmus.



Spido

As a port city, Rotterdam has unbreakable ties to the Nieuwe Maas River. There are all sorts of ways to explore and experience the river and the port. Go on a tour with Spido, one of Rotterdam's biggest attractions. The Port of Rotterdam tour (75 minutes) sails year-round and gives you a short but powerful impression of Rotterdam's container ports. The route also passes sights like the Hotel New York, the Euromast, and the ss Rotterdam.

Witte de Withstraat

The ultimate arts and culture street in Rotterdam is known for its artistic, multicultural and lively character. Referred to as the city's 'Axis of Art', it runs from the Maritime Museum to the Museumpark and is lined with galleries, avantgarde fashion boutiques, restaurants and (art) cafés. Fan of street art? Discover impressive wall paintings from local artists with the Rotterdam Street Art Route.

Diergaarde Blijdorp

Rotterdam Zoo is situated in the district of Blijdorp, in Rotterdam Noord. Stroll across the African Savannah and stand face-to-face with the giraffes. Visit Bokito the gorilla and his family. Walk on the seabed in the indoor Oceanarium and meet stingrays and sharks. Stroll through the largest butterfly paradise of Europe: Amazonica, filled with fragrant flowers, thousands of South American butterflies, anacondas and piranhas that love to bite. Discover the special Monuments & Botanical route by downloading the English version of the Diergaarde Blijdorp app.

Historisch Delfshaven

This picturesque yacht marina is one of the few parts of the old city that survived the 1940 bombardment of Rotterdam. It has a remarkable history, both as the departure point from which the Pilgrim Fathers sailed for America and as the birthplace of prominent Dutch maritime hero Piet Hein. Delfshaven locals once earned their living by fishing for herring and distilling gin. The historic canal houses now hold cosy shops selling antiques and bric-a-brac, as well as lovely cafés and restaurants like brewery De Pilgrim and café De Oude Sluis (with waterside patio dining).

For more information about how to visit one of the Rotterdam Must Sees, please go the SSIEM 2019 registration page.





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With You



Innovation in Nutrition
A Nestlé Health Science Company

Please join us for a lunchtime
satellite symposium

**Hepatic Glycogen Storage Disease:
The latest research on dietary therapy.**

Dr Helen Mundy

Consultant, Evelina Children's Hospital,
London, UK

Tuesday 3rd September 2019

**Hall TBC,
De Doelen Exhibition Centre, Rotterdam, NL**

Vitaflo is proud to be a platinum sponsor of SSIEM 2019

REGISTRATION

Registration Category:	Early Registration (until 7 July 2019)	Late Registration (from 8 July 2019 – onsite)
SSIEM Member	€ 400	€ 550
Non-member	€ 650	€ 750
Students/ Residents -in- Training (<35 yrs)	€ 250	€ 400
SSIEM Member Single Day Ticket	€ 120	€ 200
Non-Member Single Day Ticket	€ 200	€ 300
Full Group Registration	€ 550	€ 650
Accompanying person	€ 200	€ 250

Attendees can register by using the online registration form

Full Delegate (SSIEM Members, non members, students & residents-in-training):

- Admission to all oral and poster sessions and exhibition area
- Scientific programme and Symposium bag
- Coffee, tea and lunch bag during Symposium breaks
- Welcome Reception

Single day ticket:

- Admission to all oral and poster sessions and exhibition area
- Scientific programme and Symposium bag
- Coffee, tea and lunch bag during Symposium breaks of the day/ days of the ticket

Accompanying persons:

- Welcome Reception
- Thursday Networking Evening

Privacy Policy

Personal data will be collected, processed and stored by Congress by design (the "Processor"), on behalf of the SSIEM2019 Congress organizers, in its capacity as data controller, only for the preparation and execution of SSIEM Annual Symposium, September 3-6, 2019. Delegates' personal data will be retained securely and under the applicable law, you may, at any time, request access to your personal data, as well as its rectification, erasure or limited processing, portability of your data, or oppose to the processing by written request addressed to the Data Controller through the email address info@ssiem2019.org.



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Congress by design does not give your personal information to anyone else unless one of the following applies;

- You would reasonably expect that your information will be passed on to the recipient as part of the provision of products or services to you ancillary to your registration for this symposium;
- You have consented;
- It is required or authorized by law;
- It will prevent or lessen a serious and imminent threat to somebody's life or health;
- It is reasonably necessary for the enforcement of the criminal law or of a law imposing a pecuniary penalty, or for the protection of public revenue.

Confirmation

A confirmation email will be sent to you on completion of your registration. In this email you will receive an invoice/receipt and more information about your registration.

Cancellation Policy

Notification of cancellation of registration must be sent in writing to the SSIEM2019 Meeting Secretariat at info@ssiem2019.org.

- For cancellations received up to and including 3 July 2019, a cancellation fee of 25% will be charged.
- For cancellations received on and after 4 July 2019, no refund will be given, full amount required.
- Name changes are not accepted, unless the participant is from the same company.
- A handling fee of EUR 40 per registration will be charged for every modification received after 4 July 2019.
- All refunds will be processed after the event.

Cancellation of congress

It is mutually agreed that in the event of total or partial cancellation of the congress due to fire, strike, natural disaster (either threatened or actual), government regulations or causes which would prevent its scheduled opening or continuance, the contractual relationship between SSIEM2019 and the participant will be terminated immediately and SSIEM2019 shall determine an equitable basis for the eventual refund of a portion of the registration or other fees after due consideration of expenditures and commitments already made. Under no circumstances is SSIEM2019 responsible for any participant expenses (such as travel and lodging) or any other incidental or consequential damages.

CME Accreditation

The SSIEM Annual Symposium 2019 has applied for European CME credits (ECMEC®s). More information will be available soon on the Symposium website.

Aeglea BioTherapeutics-sponsored symposium

THE STORY OF ARGINASE 1 DEFICIENCY: TIME FOR A NEW CHAPTER

Wednesday 4 September 2019, 18:00–19:30

Chaired by Professor Martina Huemer

This interactive Aeglea BioTherapeutics-sponsored symposium will provide an overview of the latest understanding of arginase 1 deficiency, explore the impact of the disease on patients and their families, and review its current management, as well as the evidence for a novel therapeutic approach.

Programme:

Time	Session
18:00–18:05	The prologue
18:05–18:40	Arginase 1 deficiency: From the beginning
18:40–19:10	Understanding the cast of characters
19:10–19:20	The plot thickens: A look ahead to a new treatment approach
19:20–19:30	Can we change the ending? <i>Audience and panel discussion</i>

Date of preparation: May 2019