5th Austrian-Swiss Metabolic Meeting (ASMM)

Lausanne, February 1-2, 2018

Lausanne

Metabolic myopathies
Phenylketonuria – Controversies on therapy

FINAL PROGRAM
**General Information**

**Location**

**Hotel Alpha-Palmiers by Fassbind**
Rue du Petit-Chêne 34
CH-1003 Lausanne

Phone: +41 21 55 55 999
E-Mail: ap@byf.ch
Website: https://byfassbind.com/hotel/alpha-palmiers

**Accommodation**
Reservation according to your own arrangement with a special room rate until **January 5, 2018**

**Hotel Alpha-Palmiers by Fassbind**

*room rate (incl. breakfast)*  
single room  170,– CHF

**Registration**

Your participation is free of charge.
Please register until latest **January 25, 2018** via the online link
http://www.asmm-registration.com/2018

**Organization**

**SGIEM – Swiss Group for Inborn Errors of Metabolism**
Kinderspital Zürich – Eleonorenstiftung
Steinwiesstr. 75
CH-8032 Zürich

**CHUV – Centre Hospitalier Universitaire Vaudois**
PD Dr. Diana Ballhausen  
Médecin adjoint, Pédiatre FMH  
Centre des Maladies Moléculaires  
Service de Médecine Génétique  
Avenue de Beaumont 29  
CH-1011 Lausanne

**Local Scientific Committee**

PD Dr. Diana Ballhausen, CHUV Lausanne  
Dr. Christel Tran, CHUV Lausanne  
PD Dr. Olivier Braissant, CHUV Lausanne  
Dr. Ilse Kern, HUG Geneva

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**Symposium Support Service**

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**Meeting Schedule**

**Thursday, February 1, 2018**

**Pre Symposium Meeting**

10:00 – 12:30 h  
Meeting of the Swiss Group for Inborn Errors of Metabolism  
Room 976

**5th Austrian – Swiss Metabolic Meeting**

12:30 h  
**Welcome Lunch**  
Foyer  
Bambou Room  
978/979

**Plenum**

13:30 – 15:45 h  
Scientific Lectures  
Coffee Break

16:10 – 18:10 h  
Scientific Lectures

19:45 h  
**Networking Dinner**  
Caveau St. Vincent  
Montreux

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**Friday, February 2, 2018**

8:30 – 10:30 h  
Scientific Lectures  
Coffee Break

11:00 – 13:15 h  
Scientific Lectures  

13:15 h  
**Farewell Lunch**  
Foyer  
Bambou Room  
978/979

End of Conference

Departure according to your own arrangement.

13:30 - 15:30 h  
Meeting of the Austrian Group for Inborn Errors of Metabolism  
Room 973

For this symposium credit points have been applied.
Thursday, February 1, 2018

13:30 h  Welcome

**Session I: Metabolic Myopathies**

14:00 h  Key lecture on metabolic myopathies from neonatology to childhood  
M.C. Ørngreen, Copenhagen

14:45 h  Key lecture on metabolic myopathies in adolescence and adulthood  
R. Quinlivan, London

15:30 h  Discussion

15:45 h  Coffee Break

**Session II: Short Communications**

16:10 h  Infantile Pompe disease: decision making, ethical considerations and potential criteria for starting and stopping ERT  
M. Rohrbach, Zurich

16:20 h  When the genotype does not help to predict the clinical course of a patient: a case report of a child with neuronopathic Morbus Gaucher  
N. Faignart, Lausanne

16:30 h  Arts syndrome- a case report and pathophysiological considerations  
M. Huemer, Zurich

16:40 h  Untargeted clinical metabolomics in Pyridoxine-Dependent Epilepsy  
L. Crowther, Zurich

16:50 h  Secondary brain creatine deficiency and neurological impairment in BDL rats, an in vivo model of chronic cholestatic liver disease  
O. Braissant, Lausanne

17:00 h  Severe motor involvement in a non-compliant adult with biotinidase deficiency: The necessity of life-long therapy  
G. van Winckel, Lausanne

17:10 h  Liver failure, lactic acidosis and hyperammonaemia in a newborn  
A. Roscher, Vienna

17:20 h  Defects of ATP synthesis are biochemically heterogeneous with similar clinical manifestation  
J. Mayr, Salzburg

17:30 h  Investigation of bioenergetic adaptations under galactose stress in OXPHOS deficient fibroblasts using a combined metabolic Flux and HR-MAS NMR approach  
D. Hertig, Bern

17:40 h  Difficulties in proving the pathogenicity of a COQ4 missense mutation in a child with progressive encephalomyopathy  
B. Plecko, Graz

17:50 h  To cut a long story short: the specific phenotype of HNF4A mutation R63W  
M. Gautschi, Bern

18:00 h  Kyphoscoliotic Ehlers-Danlos Syndrome caused by biallelic mutations in FKBP14: Expansion of the clinical and mutational spectrum and description of the natural history  
C. Giunta, Zurich

19:00 h  Networking Dinner

Friday, February 2, 2018

**Session III: Phenylketonuria (PKU) – controversies on therapy**

8:30 h  Opinion lecture on treatment in PKU: low cut-offs (new guideline)  
F. van Spronsen, Groningen

9:00 h  Opinion lecture on treatment in PKU: high cut-offs (comment on new guideline)  
P. Burgard, Heidelberg

9:30 h  Treatment in PKU: actual practice in Austria  
D. Mölsinger, Vienna

9:45 h  Treatment in PKU: actual practice in Switzerland  
A. Tsouka, Lausanne

10:00 h  Interactive discussion

10:30 h  Coffee Break
**Session IV: Short Communications**

<table>
<thead>
<tr>
<th>Time</th>
<th>Title</th>
<th>Speaker Location</th>
</tr>
</thead>
<tbody>
<tr>
<td>11:00 h</td>
<td>Transitory episodes of headaches, confusional state and low cognitive efficiency: a case report of late-diagnosed atypical hyperphenylalaninemia</td>
<td>M. Jequier Gygax, Lausanne</td>
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<tr>
<td>11:10 h</td>
<td>DNAJC12 chaperonopathy leads to hyperphenylalaninemia and a neuro-metabolic phenotype</td>
<td>B. Thöny, Zurich</td>
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<tr>
<td>11:20 h</td>
<td>Deleterious effects of sepiapterin on developing reaggregated rat brain cell cultures</td>
<td>N. Remacle, Lausanne</td>
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<tr>
<td>11:30 h</td>
<td>Citrin deficiency – a rare but treatable cause of neonatal intrahepatic cholestasis. First Austrian case</td>
<td>J. Spenger, Salzburg</td>
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<tr>
<td>11:40 h</td>
<td>How to unravel a patient with LPI through anamnesis and clinical examination</td>
<td>D. Rymen, Zurich</td>
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<tr>
<td>11:50 h</td>
<td>Reduction of complexity: Explaining metabolic diseases and treatment to children</td>
<td>N. Zeltner, Zurich</td>
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<tr>
<td>12:00 h</td>
<td>From Biochrom to LC-MS : a methodological shift in the amino acid analysis for inborn errors of metabolism</td>
<td>C. Roux, Lausanne</td>
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<tr>
<td>12:10 h</td>
<td>Decreased concentrations of isovaleryl-carnitine in patients with Maple Syrup Urine Disease (MSUD)</td>
<td>R. Fingerhut, Zurich</td>
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<tr>
<td>12:20 h</td>
<td>Early detection of myocardial diastolic dysfunction in patients with LCHADD – a pilot study</td>
<td>J. Berchtold, Innsbruck</td>
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<tr>
<td>12:30 h</td>
<td>Very long-/ and long Chain-3-Hydroxy Acyl CoA Dehydrogenase Deficiency correlates with deregulation of the mitochondrial fusion/fission machinery</td>
<td>D. Karall, Innsbruck</td>
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<tr>
<td>12:40 h</td>
<td>Adaptive behavior is more impaired than executive function in adult patients with classical galactosaemia – a Swiss observational study</td>
<td>F. Scherer, Bern</td>
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<tr>
<td>12:50 h</td>
<td>Management of diabetes mellitus and severe osteoporosis in an adult patient with glycogen storage disease type III</td>
<td>L. Bosanska, Bern</td>
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<tr>
<td>13:00 h</td>
<td>Closing remarks</td>
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**Departure according to your own arrangement.**
ASMM 2018 sponsored by:
in alphabetic order

ACTELION

ALEXION

BIOMARIN
BioMarin Europe Ltd.

NUTRICIA Metabolics

SANOFI GENZYMEN

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Pioneer in Rare Diseases