

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

Booking code: **ASMM 2018**

Hotel Alpha-Palmiers by Fassbind

Reservation:

Phone: +41 21 55 55 999

E-Mail: ap@byf.ch

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

Local Organizer

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

diana.ballhausen@chuv.ch

christel.tran@chuv.ch

olivier.braissant@chuv.ch

ilse.kern@hcuge.ch

Symposium Support Service

ZweiPlan

DAS PRODUKTIONSBURO GmbH

Infanteriestrasse 11

D-80797 München

Phone: +49 (0) 89 41 41 445-51

Fax: +49 (0) 89 41 41 445-99

E-Mail: asmm@zweiplan.de

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

Plenum

Bambou Room
978/979

13:30 – 15:45 h Scientific Lectures

Coffee Break

16:10 – 18:10 h Scientific Lectures

19:00 h *Networking Dinner* Caveau St. Vincent
Montreux

Friday, February 2, 2018

8:30 – 10:30 h Scientific Lectures Bambou Room
978/979

Coffee Break

11:00 – 13:15 h Scientific Lectures

13:15 h *Farewell Lunch* Foyer
Bambou Room

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.

Scientific Lectures

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

Scientific Lectures

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. Möslinger, 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

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

13:15/13:30 h *L u n c h and end of meeting*

Departure according to your own arrangement.

Meeting and accommodation

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>

Travel directions

From Geneva Airport:

➡ After the custom, take a left and walk through the whole building. When you go through the revolving door, you will enter the railway station. Buy your ticket to Lausanne at the counter on the opposite side of the hall. For Lausanne, check on the departure panel above each track. Journey to Lausanne lasts approx. 40 minutes.

From Zurich Airport:

➡ After the custom, follow the sign with a train (train station) to the 2nd basement of the airport. Buy your ticket from the counter and check on the blue departure panel when the next train is. Journey to Lausanne approx. 2h30. You will find the schedule of the swiss railway on <http://www.cff.ch>

Once at the Lausanne railway station:

➡ To reach the Hotel Alpha-Palmiers take the road «Rue du Petit-Chêne» that goes upwards. After a 300-meter walk you will find the hotel on the left, next to Mc Donald.

Evening Location

ORCHIS Sàrl
Caveau St.Vincent
Place des Planches 1
Ch-1820 Montreux vieille Ville
Phone: +41 79 833 27 79

ASMM 2018 sponsored by:

in alphabetic order

