Remethylation disorders and methylmalonic acidurias

FINAL PROGRAMME

33rd Annual Conference of the Arbeitsgemeinschaft für Pädiatrische Stoffwechselstörungen (APS) (Association for Pediatric Metabolic Medicine) and PMM Workshop from 13 to 15/16 March 2019 Fulda, Germany

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GENERAL INFORMATION

Conference venue
MARITIM-Hotel am Schlossgarten
Pauluspromenade 2
D-36037 Fulda
Phone: +49 (0)661-2820
Fax: +49 (0)661-2824 99
Email: reservierung.ful@maritim.de
www.maritim.de

Arrival and departure [according to own schedule]
By train: very good ICE (high-speed rail) connection from/to Frankfurt airport.
By car: Motorway "Kassel – Würzburg", exit "FULDA NORD", then continue towards the city centre via Leipziger Straße to the "Paulus-Tor". Parking is available in the car park of the hotel.

Conference fee [including dinner and programme]
Registration for the APS Annual Conference by 20/2/2019:
Members of the APS EUR 65 EUR 90
Non-members of the APS EUR 100 EUR 125
Students and PhD candidates do not have to pay this fee on presentation of a corresponding certificate when registering.

Please transfer the conference fee to the APS account when registering:
Deutsche Apotheker- und Ärztebank Düsseldorf
BIC: DAAEDEDD IBAN: DE23 3006 0601 0007 2493 30

Participating in the metabolism workshop is free of charge.

Accommodation
Hotel Maritim, Phone: +49 (0)661-2820
Single room EUR 133/per night Double room EUR 168/per night
Romantik Hotel „Goldener Karpfen“ Fulda, Phone: +49 (0)661-8680-0
Single room EUR 155/per night Double room EUR 175/per night
Hotel Fulda Mitte, Phone: +49 (0)661 8330 907
13/3/19–15/3/19: Single room EUR 115/per night Double room EUR 135/per night
15/3/19–16/3/19: Single room EUR 105/per night Double room EUR 125/per night
Hotel Peterchens Mondfahrt, Phone: +49 (0)661-902350
Single room EUR 98/per night Double room EUR 120/per night
CityHotel Hessischer Hof, Phone: +49 (0)661-78011
Single room EUR 95/per night Double room EUR 110/per night
Hotel am Schloss, Phone: +49 (0)661-250 558 0
Single room EUR 76.90/per night Double room EUR 86.90/per night

Each participant is responsible for their own reservation/cancellation.
It is also possible to individually book a different accommodation through the tourist office of the city of Fulda, phone: +49 (0)661-1021 813

SCHEDULE

Wednesday, 13 March 2019
Opening programme
13.00 hrs PMM examinations Pavilion I
14.00 - 20.00 hrs APS board meeting Pavilion I

Conference programme
20.30 hrs Reception for the participants of the Annual Conference in the MARITIM hotel Foyer (Dinner and informal get-together)

Thursday, 14 March 2019
8.30 - 15.30 hrs APS Annual Conference Großer Saal
12.20 - 14.00 hrs Poster presentation with casual lunch reception Festsaalfoyer
16.00 - 17.30 hrs APS members’ meeting Roter Saal

Social programme
19.30 hrs APS Networking Dinner Großer Saal

Friday, 15 March 2019
8.30 - 12.30 hrs APS Annual Conference Großer Saal
14.00 - 19.15 hrs APS Workshop on Metabolism Roter Saal

Saturday, 16 March 2019
8.45 - 15.30 hrs APS Workshop on Metabolism Roter Saal
PROGRAMME

Thursday, 14 March 2019

8.30 hrs Welcome & introduction
M. Huemer, Zurich/Bregenz
M. Baumgartner, Zurich

Overview of biochemistry and clinical findings
Chair: I. Kern, Geneva & D. Möslinger, Vienna

8.40 hrs Cobalamin: pathways & players
S. Froese, Zurich

9.10 hrs Remethylation disorders: clinical presentations and pathophysiological considerations
M. Huemer, Zurich/Bregenz

9.40 hrs MMA: clinical presentations and pathophysiological considerations
M. Baumgartner, Zurich

10.10 - 10.30 hrs Coffee break / poster / industrial exhibition

Remethylation disorders: Diagnostics and pathophysiology
Chair: A. Das, Hannover & D. Ballhausen, Lausanne

10.30 hrs Diagnostic approaches and newborn screening in remethylation defects
V. Kozich, Prague

11.00 hrs Homocysteine and disease: causal association or epiphenomenon?
L. Hannibal, Freiburg

11.30 hrs Molecular mechanisms underlying the manifestations of impaired remethylation pathway
J.-L. Gueant, Nancy

12.00 hrs Oxidative stress and antioxidant treatment in the cblC defect
D. Martinelli, Rome

12.20 - 14.00 hrs Poster lunch

14.00 hrs MMA-Guidelines for clinical management
F. Hörster, Heidelberg

14.30 hrs Remethylation defects: Guidelines for clinical management
C. Dionisi-Vici, Rome

Free communications I
Chair: M. Lindner, Frankfurt & J. Hennermann, Mainz

15.00 hrs Lessons learned from combined and comparative data analysis of over 1,000 patients with urea cycle disorders
R. Posset, Heidelberg

15.10 hrs Natural history and genotype-phenotype correlation in NBAS deficiency
B. Peters, Heidelberg

15.20 hrs A systematic review and meta-analysis of published cases improves the description of clinical features and natural disease history in multiple sulfatase deficiency
L. Schlotawa, Göttingen

15.30 hrs End of the scientific programme of the 1st day

15.30 - 16.00 hrs Coffee break / poster / industrial exhibition

16.00 hrs APS members’ meeting
Roter Saal

19.30 hrs APS Networking Dinner
Großer Saal
### Free communications II

**Chair:** M. Rohrbach, Zurich & C. Mühlhausen, Göttingen

<table>
<thead>
<tr>
<th>Time</th>
<th>Title</th>
<th>Speaker</th>
<th>Location</th>
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<tbody>
<tr>
<td>8.30 hrs</td>
<td>Delineating the phenotype of COQ4 deficiency with cystic cerebellar degeneration</td>
<td>L. Laugwitz</td>
<td>Tübingen</td>
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<tr>
<td>8.40 hrs</td>
<td>Mitochondriopathy in pediatric patients with unspecific neuropediatric disease</td>
<td>A. van der Ven</td>
<td>Hamburg</td>
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<tr>
<td>8.50 hrs</td>
<td>Evidence for a genotype-phenotype correlation in patients carrying pathogenic GLUT2 (SLC2A2) variants</td>
<td>R. Santer</td>
<td>Hamburg</td>
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<tr>
<td>9.00 hrs</td>
<td>Transferrin glycosylation - analysis from dried blood spots and capillary blood samples</td>
<td>A.B. Wolking</td>
<td>Münster</td>
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<tr>
<td>9.10 hrs</td>
<td>ENPP1-Fc-replacement prevents arterial stenoses through generation of adenosine monophosphate in generalized arterial calcification of infancy</td>
<td>K. Kintzinger</td>
<td>Münster</td>
</tr>
<tr>
<td>9.20 hrs</td>
<td>The use of patient-derived induced pluripotent stem cells (iPSCs) and iPSC-derived cerebral organoids to explore pathomechanisms of succinic semialdehyde dehydrogenase deficiency</td>
<td>H.B. Brennenstuhl</td>
<td>Heidelberg</td>
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<tr>
<td>9.30 hrs</td>
<td>Pharmacologic rescue of hyperammonemia-induced neurotoxicity by inhibition of ornithine aminotransferase in a zebrafish model of acute hyperammonemic decompensation</td>
<td>M. Zielonka</td>
<td>Heidelberg</td>
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<tr>
<td>9.40 hrs</td>
<td>Combined malonic and methylmalonic aciduria (CMAMMA): an inborn defect of the mitochondrial fatty acid biosynthesis (MFASII)</td>
<td>Z.W. Wehbe</td>
<td>Freiburg</td>
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<tr>
<td>9.50 hrs</td>
<td>New insights into human lysine degradation pathways with relevance to pyridoxine dependent epilepsy due to antiquitin deficiency</td>
<td>B. Plecko</td>
<td>Graz</td>
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**10.00 - 10.30 hrs**  
Coffee break / poster / industrial exhibition

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Programme:

Friday, 15 March 2019

14.00 hrs Welcome
R. Santer, Hamburg

14.05 hrs International clinical guideline for the management of classical galactosemia: diagnosis, treatment, and follow-up
A. Bosch, Amsterdam

14.50 hrs Dietetic treatment of galactosemia
M. Pauli, Erfurt

15.15 - 16.00 hrs Coffee break

16.00 hrs Case workshop “The interesting metabolic case”

17.15 hrs Group work POL case 1
Group leader: S. Wortmann, Munich/Salzburg
C. Mühlhausen, Göttingen
R. Santer, Hamburg
T. Opladen, Heidelberg

18.30 hrs Humour in medical communication
R. Szeliga, Vienna

19.15 hrs End of day 1

Saturday, 16 March 2019

8.45 hrs New guideline: Diagnostic follow-up for suspected congenital metabolic diseases from newborn screening
M. Lindner, Frankfurt

9.25 hrs Metabolic diagnostics, metabolome, exome, genome: When What?
S. Wortmann, Munich/Salzburg

10.05 - 10.30 hrs Coffee break

10.30 hrs Group work POL case 2
Group leader: S. Wortmann, Munich/Salzburg
C. Mühlhausen, Göttingen
R. Santer, Hamburg
T. Opladen, Heidelberg

11.45 hrs Update MMAs and remethylation disorders
M. Baumgartner, Zurich

12.30 - 13.30 hrs Lunch

13.30 hrs Patient workshop MMAs and remethylation disorders

14.30 hrs Group work
Group leader: S. Wortmann, Munich/Salzburg
C. Mühlhausen, Göttingen
R. Santer, Hamburg
T. Opladen, Heidelberg

15.30 hrs Closing remarks and end of the workshop
R. Santer, Hamburg
P-017 Normal development of a 7-month old boy after early start of treatment in severe methylenetetrahydrofolate reductase (MTHFR) deficiency identified by newborn screening

H. Blessing
Erlangen

P-018 Hematopoietic stem cell transplantation in mucolipidosis type II

L.S. Ammer
Hamburg

P-019 Longitudinal cholesterol metabolite profiles in 20 patients with Smith-Lemli-Opitz-syndrome

K. Palm
Magdeburg

P-020 ALG3-CDG- clinical, biochemical and genetic abnormalities of four new patients and identification of a new ALG3 hybrid protein

N. Himmelreich
Heidelberg

P-021 Infantile Pompe disease presents with leukodystrophy after long term ERT

A. Zuehlsdorf
Münster

P-022 Analysis of transition of patients with PKU under real-life conditions

R.-S. Allo
Düsseldorf

P-023 Accidental diagnosis of molybdenum cofactor deficiency type A in a neonate and cPMP rescue prior to irreversible brain injury

S.J. Mayr
Cologne

P-024 A severe neonatal phenotype associated with a pathogenic C12orf65 variant: a disorder of mitochondrial translation and protein synthesis

F. Seggewies
Hamburg

P-025 An infant with milky blood presenting as a sepsis

M. Wachendörfer
Bremen

P-026 A novel thiamine-responsive TPK1 variant in a patient with white matter disease

L.M. Marten
Hamburg

P-027 Life-saving biotin-thiamine treatment in a patient with SLC19A3-associated early-infantile Leigh-like syndrome

K. Talakas
Hamburg

P-028 Disorders of ketone body transport and utilization as a cause of recurrent metabolic acidosis: succinyl-coenzyme A: 3-oxoacyl coenzyme A transferase (SCOT) deficiency (mutations in the OXCT1 gene) and monocarboxylate transporter 1 (MCT1) deficiency

S. Roloff
Berlin

P-029 Concentrations of (iso)valerylcarnitine (C5) in dried blood spots (DBS) are higher in premature infants

S. Murko
Hamburg

P-030 Effect of newborn screening and adherence to recommended treatment on clinical outcome in glutaric aciduria type 1: a meta-analysis

N. Boy
Heidelberg

P-031 Twelve-year experience with a rapid and simple fluorometric tripeptidyl peptidase 1 (TPP1) assay using dried blood specimens (DBS) to diagnose CLN2 disease

Z. Lukacs
Hamburg

P-032 Sirtuins in patients with glycogen storage disease type 1: possible biomarkers and role in pathophysiology

A.B. Potthast
Hannover

P-033 Impact of nutritional habits on sirtuins in blood from recreational runners

A.B. Potthast
Hannover

P-034 Ketogenic diet as a treatment option in malignancies: effect of C8 and C10 on sirtuins at cellular level

K. Stühnwohlt
Hannover

P-035 Challenges facing patients in PKU therapy and measures to support patients

J.E. Thiele
Mannheim

P-036 Fatal outcome after heart surgery in PMM2-CDG due to exon 5 skipping of the PMM2 gene

M. Hutter
Heidelberg

P-037 Ketogenic diet in pharmacoresistant epilepsy: impact of beta-hydroxybutyrate on sirtuins and respiratory chain enzymes in human and mouse neuronal cells

P. Dabke
Köln

P-038 Interference of carnitine supplementation and physiological adaptation mechanisms in OCTN2 deficient (OCTN2-/-) mice

N.M. Mingirulli
Freiburg

P-039 Sex specific metabolic phenotype in very-long chain acyl-coA dehydrogenase deficient (VLCAD-/-) mice

K. Alatibi
Freiburg

P-040 A severe case of DHDDS-CDG due to a homozygous variant in the dehydrodolichol diphosphate synthase

A. Hülle
Heidelberg
SPECIAL REMARKS

APS Annual Conference:
Deadline for submitting abstracts is 14 January 2019.
Abstracts for oral presentations (7 min. plus 3 min. discussion) and posters can be submitted again this year and can be uploaded on the APS website (www.aps-med.de/abstracts).
Please pay attention to the standard structure (introduction, patients/methods, results, conclusion). Abstracts must not be longer than 1400 characters (including spaces).
Accepted abstracts will be published in the monthly journal “Monatsschrift Kinderheilkunde”. Abstracts must be written in English.

The best oral presentation will be awarded EUR 1000 by the APS. In addition, Dr. Ursula Wachtel, honorary member of the APS, has donated an additional prize of EUR 500. Each year, this prize is awarded to a European scientist (under 35 years of age) who presents the best poster on a pediatric metabolic disorder at the APS Annual Conference.

The prizes will be awarded by a jury from the APS board at the end of the Annual Conference. The winners are asked to be present in person at the award ceremony.

The Annual Conference has been certified as an advanced training event by the Landesärztekammer Hessen (State Chamber of Physicians of Hesse).

APS workshop participants:
Abstracts for “The interesting metabolic case”
Workshop participants are requested to submit their own clinical case from the field of congenital metabolic diseases. Please send an informal abstract (maximum half a A4 page) to chris.muehlhausen@med.uni-goettingen.de when registering. From the abstracts submitted, cases will be selected which will be presented by the participants as a short case presentation (5–10 min) in the context of the workshop part “The interesting metabolic case”. Workshop participants are asked to organise their travel schedule in a way that they can stay until the end of the workshop and do not have to leave early. Out of respect for the patients and their families, who come specifically for the patient workshop, we ask everyone to participate until the end of the program.

General and technical information for speakers

We would like to point out that the speakers are required to present in a product- and service-neutral manner. Possible conflicts of interest must be announced at the beginning of the presentation.

We would like to ask you that both your presentation and your talk are in English.

For your PowerPoint presentations, you can expect the following specifications for the presentation hardware:
Server: Intel i7 / 4x3.4 GHz, 16 GB RAM, SSD
Projector: Panasonic PT-RZ12K, 3-Chip DLP Laser Phosphor Projector, 12,000 ANSI lumen, resolution: 1920 x 1200 Pixel (WUXGA)

For reasons of compatibility, you should not use your own notebook for your presentation.

Please submit your presentation as a PC-compatible file on a data storage device (CD-ROM, DVD or memory stick). After saving it on the data storage device, please make sure the presentation is running smoothly. Speakers are responsible for their presentation’s compatibility.

If you include pictures, videos and audio in your PowerPoint presentation, please note that the data needs to have the following formats:
Pictures: .jpg videos: .avi audios: .wav

Please make sure that all of the pictures, videos or audio in your presentation are fully stored on your data storage device.

Please hand in your data storage device at the “PreView desk” at least 2 hours before your presentation.

You can also send your presentation in advance as an e-mail attachment to aps@zweiplan.de (by 11 March 2019 at the latest).

MAC users are urgently requested to save their presentation on the data storage device in a PC-compatible way and ensure that it will run without issue.
The contents of this event will be independent of any products and services. We confirm that the scientific management and the speakers disclose potential conflicts of interest to the participants. The following companies act as sponsors:

Platinum: (EUR 30,000)

Gold: (EUR 15,000)

Silver: (EUR 10,000)

Bronze: (EUR 5,000)

(Disclosure of sponsorship services in accordance with the FSA Code as part of the expanded transparency requirement for the support of congress events)
Host:

**APS Annual Conference**

Prof. Dr. med. Dipl.-Psych. Martina Huemer
1. Kinderspital Zurich (Zurich Children's Hospital) – Eleonorenstiftung, Abt. für Stoffwechselkrankheiten (Dept. of Metabolic Diseases)
   Steinwiesstr. 75, CH-8032 Zurich
   Email: martina.huemer@kispi.uzh.ch
2. LKH Bregenz, Abt. fur Kinder- und Jugendheilkunde (Dept. of Pediatrics and Adolescent Medicine)
   Carl-Pedenz-Str. 2, A-6900 Bregenz
   Email: martina.huemer@lkhb.at

**APS workshop**

Prof. Dr. med. Chris Mühlhausen
Klinik für Kinder- und Jugendmedizin (Dept of Pediatrics and Adolescent Medicine)
Universitätsmedizin Göttingen
Robert-Koch-Str. 40, D-37075 Göttingen
Phone: +49 (0)551-39 66 210
Email: chris.muehlhausen@med.uni-goettingen.de

Prof. Dr. med. Matthias Baumgartner
Kinderspital Zurich (Zurich Children's Hospital) – Eleonorenstiftung, Abt. für Stoffwechselkrankheiten (Dept. of Metabolic Diseases)
Steinwiesstr. 75, CH-8032 Zurich
Email: matthias.baumgartner@kispi.uzh.ch

**Univ.-Prof. Dr. med. René Santer**
Universitäts-Klinikum Eppendorf
Klinik für Kinder- und Jugendmedizin (Dept of Pediatrics and Adolescent Medicine)
Martinistr. 52, D-20246 Hamburg
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Email: r.santer@uke.de

**Service:**

Symposium Support Service
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Infanteriestr. 11, D-80797 Munich
Phone: +49 (0) 89 41 41 445-51
Fax: +49 (0) 89 41 41 445-99
Email: aps@zweiplan.de

**Registration**

APS website: www.aps-med.de
Direct link
www.Jahrestagung.apstagung.de