4th European Phenylketonuria Group (EPG) Symposium
“Advances and Challenges in PKU”
Rome, Italy - March 23-24, 2012
Aim of the symposium

While dietary restriction of phenylalanine still remains the mainstay of treatment, phenylketonuria (PKU) is an active area of research, and new treatment options are emerging that may reduce the burden of a difficult and limiting diet in these patients and in their families. Basic and clinical research is now focusing on the mechanisms of action of new treatment options, like sapropterin dihydrochloride (also BH₄), on monoamine neurotransmitters metabolism in the brain of PKU patients. Furthermore there’s an emerging need for evidence-based international guidelines to provide standardization in treatment initiation, in evaluation of the response to therapy with sapropterin dihydrochloride, and in blood phenylalanine concentration to target. A number of papers resulting from last year’s symposium reviewed the existing scientific evidences in several aspects of PKU, identifying the current positions in its management. The European Phenylketonuria Group (EPG) and the Serono Symposia International Foundation (SSIF) consider essential to continue their series of successful meetings on PKU, and have organized the 2012 Annual Symposium dedicated to that. The aims of this meeting are to review the most important outcomes of research in the field of PKU and to provide a forum for participants to discuss solutions that can optimize patient management in clinical practice.

Learning objectives

After attending this symposium, the participants will have up-to-date knowledge about:

- Brain pathology of PKU
- Blood-brain barrier and PKU
- CNS neurotransmitters in PKU
- Management of BH₄ deficiency
- Social and financial management of PKU
- Amino acid status in PKU

Target audience

Specialists in pediatric and adult inherited metabolic diseases, dietitians, nutritionists, clinical biochemists, experts in genetics, basic scientists, and professionals within the area of public health will benefit from this symposium.
The world of CME is changing with many different live and online formats, and Serono Symposia International Foundation (SSIF) is continually trying to develop its CME activities. With your participation in a structured series of evaluations, SSIF can provide cutting-edge learning activities designed to give you the greatest value from the time you invest.

SSIF will be running the learning effectiveness project for this meeting. You will be involved in four main steps that will be explained in detail once you register.

Find out more here: 

All Serono Symposia International Foundation programs are organized solely to promote the exchange and dissemination of scientific and medical information. No forms of promotional activities are permitted. All Serono Symposia International Foundation programs are made possible thanks to the unrestricted Educational grant received from: Centre d’Esclerosi Múltiple de Catalunya, ComtecMed, Congrex Sweden, Congrex Switzerland, Cryo-Save, Datanalysis, Esaote, European Society of Endocrinology, Fondazione Humanitas, Fundación IVI, ISFP International Society for Fertility Preservation, ISMF International Society of Men’s Health, K.I.T.E., Merck Serono, Sanofi-Aventis, University of Catania, Vall d’Hebron University Hospital.
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Serono Symposia International Foundation is a Swiss Foundation with headquarters in 14, rue du Rhône, 1204 Geneva, Switzerland

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Margreet van Rijn
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University Medical Hospital Groningen
Groningen, Netherlands
Scientific program
Scientific program  
March 23-24, 2012

Friday - March 23

8.45  Serono Symposia International Foundation (SSIF) Opening

8.50  Welcome to Rome  
Vincenzo Leuzzi (Italy)

8.55  Welcome on behalf of EPG Group and SSIF  
Nenad Blau (Germany)

**Special lecture**

Chair: Harvey Levy (USA)

9.00  PKU in Italy  
Marcello Giovannini (Italy)

**Session I**  
Brain pathology and neurotransmitters metabolism in PKU

Session chairs: Ania Muntau (Germany) - François Feillet (France)

9.30  L1: Mode of action of BH₄ beyond blood Phe  
Nenad Blau (Germany)

10.00  L2: Bioinformatic approaches in PKU research and implications for clinical management  
Søren W. Gersting (Germany)

10.30  Discussion

10.45  Coffee break

**Session II**  
Oral communications

Session chairs: Anita MacDonald (UK) - Mübeccel Demirkol (Turkey)

11.15  Oral communications

13.15  Lunch
14.15 Workshop Sessions 1-4

1. Amino acids supplementation in PKU
   Anita MacDonald (UK) - Margreet van Rijn (Netherlands)

2. BH₄ deficiencies: diagnosis and treatment
   Alberto Burlina (Italy) - Friedrich K. Trefz (Germany)

3. How to improve adherence to PKU treatment
   Francjan J. van Spronsen (Netherlands) - Laurie Bernstein (USA)

4. Crucial aspects in communication to parents at diagnosis
   François Feillet (France) - Ania Muntau (Germany)

15.45 Coffee break and rotation of workshop

16.15 Workshop Sessions 1-4

1. Amino acids supplementation in PKU
   Anita MacDonald (UK) - Margreet van Rijn (Netherlands)

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   Francjan J. van Spronsen (Netherlands) - Laurie Bernstein (USA)

4. Crucial aspects in communication to parents at diagnosis
   François Feillet (France) - Ania Muntau (Germany)

17.45 Closing Day 1
Saturday - March 24

**Session III**  Day to day experiences and treatment aspects in PKU

**Session chairs:** Francjan J. van Spronsen (Netherlands) - Alberto Burlina (Italy)

- **08.30** L3: Use of BH₄ in PKU patients below 4 years  
  François Labarthe (France)

- **09.00** L4: Diet and health in adult PKU: case observations  
  Berthold V. Koletzko (Germany)

- **09.30** L5: NIH PKU Conference report: State of the science and future research needs  
  Harvey Levy (USA)

10.00 Discussion
10.15 Coffee break

**Session IV**  Workshops summaries

**Session chairs:** Maureen Clearly (UK) - Friedrich K. Trefz (Germany)

10.40 Workshop summaries presentations  
Workshops chairmen

**Session V**  Asbjørn Følling lecture and Award

- **12.00** Introduction to Asbjørn Følling Lecture  
  Nenad Blau (Germany)

- **12.10** Asbjørn Følling Lecture  
  Harvey Levy (USA)

**Session VI**  SSIF PKU Award and PKU Academy fellowships

**Chair:** Ania Muntau (Germany)

- **12.40** SSIF Award for the best oral communication  
  **Chair:** Ania Muntau (Germany)

- **12.50** PKU Academy fellowships presentation  
  **Chair:** Francjan J. van Spronsen (Netherlands)

13.00 Meeting closure  
Anita MacDonald (UK)  
Francjan J. van Spronsen (Netherlands)

13.10 Lunch
Call for abstracts

The Scientific Committee invites the submission of abstracts to be considered for oral/poster presentation. Authors are requested to send their abstract by e-mail by February 19, 2012 at the following address: flaminia.masprone@seronosymposia.org

Topic choice – “New aspects related to PKU or BH₄ disorders”.

Submission procedure

Abstracts must be written in English. Abstracts should be 250 words (excluded spaces) in length and include the title of the presentation, the author’s names, the name of the institution, the city and country.

The abstract should be structured with:

- “Introduction”: informative and short
- “Material and Methods”: indicating number and type of human subjects, non-human species or material being studied, the principal procedures, assays, tests or treatments performed
- “Results”: confirming or refuting the hypothesis, supported by statistics if appropriate
- “Conclusions”: stating the major new findings of the study and specifying what these findings add to what is known already

Abbreviations can be used after defining them first. Abstract title: do not use capital letters and do not indent. The title should reflect the contents and should be no longer than 150 characters. List the authors with family name first, followed by initials (i.e. Jones, B.; Smith, J.; and Simpson, A.) The name of the first author is also the presenting author. Do not include professional titles or degrees. Use font size 12, Times New Roman font type and line spacing 1.5.

Authors are requested to disclose any financial support received and/or any potential conflict of interest at the end of the abstract. When referring to pharmaceutical products, authors should not use brand or commercial names; the generic name (active biochemical compound) should be used instead.

The Scientific Committee will review the submitted abstracts choosing the best abstracts for oral or poster presentation. The selected abstracts will be published in the congress abstract book to be distributed on-site to all participants. Authors of the eight abstracts selected for oral presentation will have their registration fee, travel and hotel accommodation expenses reimbursed after the conference.

Notification of accepted abstracts will be sent by by February 29, 2012.

SSIF PKU Award

Serono Symposia International Foundation (SSIF) will reward the two best oral presentations in Clinical practice and Basic research with two awards of Euro 2,000,00 each. The value of these awards will be given as voucher to be spent through the participation in CME (Continuing Medical Education) courses and congresses by September 30, 2013 on the basis of a programme defined with the winners in agreement with SSIF.
General information

Venue
The Symposium will take place at the:
Rome Marriott Park Hotel
Via Colonnello Tommaso Masala, 54
00148 Rome, Italy

Language
The official language of this symposium will be English.

Registration fee
The registration fee is € 400.00.
For students, trainees and young fellows (post-doctoral and PhD fellows) the registration fee is € 250.00.

Note: for students, trainees and young fellows, a letter from the institution head certifying status must accompany the registration.

The registration fee includes:
• Symposium sessions
• Symposium coffee breaks and luncheons
• Symposium dinner on March 23, 2012
• Symposium material
• Certificate of attendance

* If you require special assistance (wheelchair, auxiliary aids) or special dietary requirements, please, contact us at info@seronosymposia.org

Please, note that the deadline to send the registration form is March 16, 2012. Name changes will be accepted within this date.

Hotel accommodation
See the enclosed hotel accommodation form.

Personal data treatment - Informative document
In compliance with the article 13 of Italian Legislative Decree n. 196, 30 June 2003, we inform you that your personal data will be processed, with or without the use of electronic or automated means, via suitable tools that will guarantee security and confidentiality. Personal data from you supplied will be communicated to Continuing Medical Education Accreditation Authorities, to agencies that will take care of the whole accreditation process and agencies that will take care of the logistics of the event; should we not receive the below requested data, we will not be able to process your registration to the event. At anytime you may exercise your rights as laid down in the art. 7 of Italian Legislative Decree n. 196, 30 June 2003 of the Italian Law, by contacting the Data Controller Serono Symposia International Foundation (Salita di San Nicola da Tolentino, 1/b - 00187 Rome - Italy) in the person of Data Processor, Michèle Piraux.
Rome is served by the Leonardo da Vinci Fiumicino International Airport (FCO), which is located around 30 km north-west of central Rome. This international airport is connected to the city center by a fast train, “Leonardo Express”, which takes about 30 minutes to reach Rome Termini Railway Station even connected with the metro. As an alternative, it is possible to take the Sabina-Fiumicino line (FR1), which stops in some of the main Roman railway stations (Tiburtina, Tuscolana, Ostiense, Trastevere).

Termini central railway station is the point of arrival and departure for almost all trains from Italy and abroad.

To reach Rome by car, from Northern Italy on the motorway A1, leave the motorway at "Roma Nord" tollgate. Arriving from the South on the same motorway, leave at "Roma Est" tollgate and you will find yourself at the "Grande Raccordo Anulare", major beltway linking the motorways and state roads leading to Rome.
PKU Academy and Masters website

Serono Symposia International Foundation (SSIF) has launched a website dedicated to medical education on Phenylketonuria (PKU):
www.pkuacademy.org

This website is directed at a wide audience of healthcare professionals working in the field, providing a full range of educational tools from the glossary and basic online courses to the advanced e-learning modules. The website has two main areas:

PKU Academy, which provides user-friendly educational tools enabling all healthcare professionals to quickly acquire a basic, up-to-date knowledge of PKU. Healthcare professionals seeking an in-depth understanding of PKU can undertake basic online Courses addressing topics from pathophysiology and diagnosis to management of PKU. Completing these courses delivered by world-renowned PKU experts, will entitle users to CME credits.

The Glossary is an evolving tool and a core reference of key terms aimed at easing users entering the PKU field. The PKU glossary provides rapid updates including explanations of all terms throughout the website.

The Frequently Asked Questions section provides authoritative answers to common questions about the disorder. In these FAQs, top PKU experts share their ‘pearls of wisdom’, providing, in a few lines, key information that users need to know about etiology, pathogenesis and treatment.

Additional information and services provided by the website include a selected Bibliography and a Calendar of congresses in the field.
PKU Masters, a dedicated area for specialists in PKU offering an advanced educational pathway consisting of CME accredited advanced online Courses. The e-learning modules are advanced courses providing PKU specialists with the most up-to-date knowledge from top worldwide experts on each aspect of the disease including genetics, diagnosis and treatment. Completing the modules and passing the learning test entitles users to CME credits.

The Editorial Board of the PKU Academy and Masters is composed of following academic experts:

**Nenad Blau**
Division of Inborn Metabolic Diseases  
University Children’s Hospital  
Department of General Pediatrics  
Heidelberg, Germany

**Anita MacDonald**
Dietetic Department  
The Children’s Hospital  
Birmingham, UK

**Francjan J. van Spronsen**
Beatrix Children’s Hospital  
University Medical Center of Groningen  
Groningen, The Netherlands

To find out more visit:

[www.pkuacademy.org](http://www.pkuacademy.org)  
Registration & Hotel accommodation forms
### Registration Form

#### 4th European Phenylketonuria Group (EPG) Symposium “Advances and Challenges in PKU”
Rome, Italy - March 23-24, 2012

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#### Registration Fee
- The registration fee is € 400.00.
- The registration fee for students, trainees, and young fellows (post-doctoral and PhD fellows) is € 250.00.

*For students, trainees, and young fellows, a letter from the institution head certifying status must accompany the registration.*

#### Friday - March 23, 2012 - Workshops’ Sessions
Each workshop has a limited number of available places. Participants will have the possibility to attend two (2) workshops. Please, mark your preference, in order of interest, for all the 4 workshops (1 most interested, 4 less interested).

**A.** Please, number (1-4) the Workshops you are interested in:

- Choice n. _____ Workshop 1: Amino acids supplementation in PKU
- Choice n. _____ Workshop 2: BH₄ deficiencies: diagnosis and treatment
- Choice n. _____ Workshop 3: How to improve adherence to PKU treatment
- Choice n. _____ Workshop 4: Crucial aspects in communication to parents at diagnosis

**B.** Please take into consideration that workshop session will be organized with the contribution of participants willing to present/send a case report (2/3 minutes discussion with max. 2 slides will be requested) on a specific topic of one of the above workshops. Participants who wish to present a short contribution are requested to agree on the below statement. They will be contacted by the Workshop Chairmen.
☐ I agree to be contacted by the Workshop Chairmen and contribute by presenting a short case report in the Workshop (tick the box):

☐ Workshop 1: Amino acids supplementation in PKU
☐ Workshop 2: BH₄ deficiencies: diagnosis and treatment
☐ Workshop 3: How to improve adherence to PKU treatment
☐ Workshop 4: Crucial aspects in communication to parents at diagnosis

You consent to provide the Chairmen with your contact details:

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Please, note that the deadline to send the registration form is **March 16, 2012**. Name changes will be accepted within this date.

☐ I require special assistance (wheelchair, auxiliary aids) or special dietary requirements.

**Payment**

☐ **Payment by Bank Transfer** headed to: “Meridiano viaggi e turismo S.p.A.”
BANCA INTERMOBILIARE S.p.A
FILIALE DI ROMA - VIA DONIZETTI, 14 - 00198 ROME - ITALY
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☐ **Payment by Credit Card**
Charge to the following Credit Card *(Credit Card commission Fee 3.5%):*
☐ MasterCard/EuroCard ☐ Visa

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**ONLY REGISTRATION FORMS ACCOMPANIED WITH PAYMENT CAN BE PROCESSED**

**Mail to:** Sara Guglielmini c/o Meridiano Congress International
Via Mentana, 2/B | 00185 Rome, Italy | T +39 06 88 595 211 | F +39 06 88595 234
E-mail: s.guglielmini@meridiano.it

All completed Registration forms will be acknowledged by e-mail or fax.

Cancellations must be received in writing before March 16, 2012. No refund will be provided.
Hotel accommodation form

4th European Phenylketonuria Group (EPG) Symposium “Advances and Challenges in PKU”
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For hotel reservation, please return the completed form with full prepayment of the requested nights. Hotel Reservation forms without prepayment cannot be processed.

DEADLINE FOR RESERVATIONS: March 16, 2012
Reservations after this date are not guaranteed and will be made on a space-available basis.

Rome Marriot Park Hotel

- Double room for single use . . . . . . € 195,00 per night
- Double room . . . . . . . . . . . . . € 218,00 per night

Check in date: ___________  Check out date: ___________  [N° of nights: _____]

Rates include breakfast and city taxes.

Payment

- Payment by Bank Transfer headed to: “Meridiano viaggi e turismo S.p.A.”
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  FILIALE DI ROMA - VIA DONIZETTI, 14 - 00198 ROME - ITALY
  Account number: 000000003808
  IBAN: IT11 C030 4303 2000 0000 0003 808 - SWIFT: BIMITTTTXX

- Payment by Credit Card:
  Charge to the following Credit Card (Credit Card commission Fee 3,5%):
  - MasterCard/EuroCard
  - Visa

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Mail to: Sara Guglielmini c/o Meridiano Congress International
Via Mentana, 2/B | 00185 Rome, Italy | T +39 06 88 595 211 | F +39 06 88595 234
E-mail: s.guglielmini@meridiano.it

Once you confirm rooms to Meridiano, full penalty will be applied in case of cancellation.
Mail to: Meridiano Congress International

Italy
00185 Rome
Via Mentana, 2/B
Attn: Sara Guglielmini
Serono Symposia International Foundation is an independent, non-profit organization dedicated to the Continuing Medical Education (CME) of scientists, physicians, nurses, pharmacists and other healthcare professionals through state-of-the-art clinical and scientific activities designed to enhance patient care and improve healthcare outcomes.

Serono Symposia International Foundation conferences and workshops provide unique opportunities where physicians and healthcare professionals can meet with the world leading experts to learn about the latest scientific developments as well as exchange ideas in an open and stimulating environment.

Serono Symposia International Foundation offers different formats of live educational events:

- **International Conferences**: CME accredited International Conferences updating and evaluating the newest scientific topics.
- **Academia**: CME accredited International Training Courses mainly addressed to junior scientists/physicians who want to specialize in a specific field.
- **Preceptorship**: CME International Workshops based on theory and practice sessions. Tutors and participants are able to directly share practice and knowledge.

If you wish to learn more about Serono Symposia International Foundation’s educational activities, please visit:  

[www.seronosymposia.org](http://www.seronosymposia.org)

The website will provide you with:

- A calendar of upcoming Serono Symposia International Foundation events
- Online CME courses
- Symposia reviews
- Presentations from past events
- Abstracts
- Literature reviews
- Glossaries
- ... and a lot more!