



European Society for Phenylketonuria (ESPKU)
and Allied Disorders treated like Phenylketonuria



30th ANNUAL ESPKU CONFERENCE 2016

3-6 NOVEMBER 2016 | DUBLIN, IRELAND

PROGRAMME

Gold sponsors

B••OMARIN®



NUTRICIA
Metabolics
Inspiring Futures

Silver sponsor



Innovation in Nutrition

CONTENTS

PAGE

Welcome	4
Organisational remarks	7
Exhibition floor plan	8
Crèche and childcare	9
Programme	10
Abstracts from invited speakers	12
Oral abstracts	18
Notes	22

Disclaimer

Participation and travelling is at your own risk! ESPKU and the hosting Association are not liable for any loss, damage or injury during travelling to or from the conference or during the event itself.

The programme is provisional. Any change due to unforeseen reasons are up to the organisers, and will not cause any refund of registration fee or travel costs.

ESPKU and the hosting Association are not liable for the content and quality of any information and materials offered during the event by exhibitors, advertisers or other third parties.

WELCOME

Foreword of the President of the ESPKU

It is with great pleasure that I welcome you all to the 30th Annual ESPKU Conference here in Dublin, Ireland. This is the first time we have been here and we really appreciate their enthusiasm in going all out to hold the Conference. They have worked tremendously hard in such a short space of time to get things ready.

For the Irish people involved with PKU, this is a great opportunity for you to come together and use the Conference as a springboard to build PKU Ireland; learn about other PKU Associations and what they do; engage with Professionals at an informal level; make friends and help give PKU Ireland a stronger voice. Carpe Diem!

This year we continue the theme of bringing stories behind the famous people. Let us remember the first person to be treated with a low protein diet was of Irish descent – Sheila Jones. To this end we welcome Anne Green – former director of the metabolic unit in Birmingham who will present the Sheila Jones story in the patients programme.

There has been a lot of discussion about the Guidelines. They are now ready and a preliminary paper has been written for the Lancet. Until this is published we cannot release them publically. This will be explained to you by Professor Francjan van Spronsen during the Common Programme.

Those who were engaged in the process in producing the Guidelines can now take a well earned (short) break before the first review. Studies on PKU come out all the time and undoubtedly there will be some that will add efficacy and credence to what has already been written. This will increase accuracy and increase the scoring of some of the statements as time progresses.

As at all Conferences we shall be welcoming friends from all over the globe. Please make them feel welcome and share your stories.

We acknowledge the great and continued support from our sponsors and, this year from Failte Ireland (the Irish Tourist Board) and help from the Hotel in putting the Conference together.

While this foreword is not equal to the wonderful style of that great son of Ireland – Oscar Wilde – I'll finish with one of his famous quotes

“I have the simplest tastes. I am always satisfied with the best”

And this is what we should all be striving for in the identification, treatment and follow up care of PKU.

I wish you all a successful Conference.

Eric Lange

President, European Society for Phenylketonuria & Allied Disorders

Foreword of the Chairman of the PKU Association of Ireland

On behalf of the PKU Association of Ireland, I would like to welcome you all to Dublin or as we would say in Irish “Céad Mille Fáilte go Baile Átha Cliath”.

It is a great pleasure for us to have so many people from all over Europe and from as far afield as Iran, Australia and America. We hope that you all have a great Conference and based on the range of speakers for all three programmes within the conference and the exhibition stands, I am sure that it will be a great success.

I have been involved in the PKU Association of Ireland for over 10 years and I know from that time, it is at the ESP KU Conference that so much valuable information is made available to so many in such a short space of time. I hope that the delegates here today will go back to their own countries and share what they have learnt with their own PKU associations and with the PKU families they know.

In the years that I have been involved with PKU Ireland, we have seen great change in the speed that information can be spread. It is through greater access to information that people with PKU's lives can be made better and awareness and treatment of the condition can be improved. With the aide of social media such as Facebook, Twitter and Instagram we can all reach more people more quickly and I would ask everyone here to share and post online everything interesting that they learn this weekend. Our local PKU Facebook groups are so effective at sharing news throughout the PKU community across Europe and the world. Share links, take pictures, offer advice – we can all do our bit to raise the profile of PKU amongst patients, healthcare professionals and the public. If you do not have a Facebook page I know there are people here willing and able to help you start one – it can be a powerful tool to get information around the PKU community.

Like all of you, I am looking forward to seeing all the food products exhibited here and to hearing of the new developments and scientific research taking place on the treatment of PKU. We can ensure that our own PKU communities benefit from the latest and best care by taking the positivity and increased knowledge gained from this conference back home.

Dublin welcomes the chance to host the 30th Annual Conference, which was switched from Turkey due to the political unrest there earlier this year. We all look forward to a time when the political situation has stabilised and the Turkish PKU Association host a future conference.

Once of the criteria for picking a hotel had to be its close proximity to the airport and we couldn't get much closer. But the hotel is only 9km from the city centre and I hope that you will get the chance either on the organised trips or after the conference to visit Dublin and the surrounding areas.

Finally I hope you all enjoy your visit to Ireland.

Fergus Woodcock

Chairman of the PKU Association of Ireland

Foreword of the Chairman Scientific Advisory Board

The ESPKU story goes on...

Last year in Berlin we were almost ready with the European guidelines and we only needed to write them down.... We already told you there, that could take some time...

It took more or less half a year. In May, we were able to submit the paper on the Key statements to a journal that only aims to have short clear-cut articles. Our problem was how to put our 80 pages of statements and considerations into less than 5000 words. It appears to be possible. The Lancet accepted our manuscript for publication on September 28.

Are we happy now? Yes we are, we are extremely happy that this paper will be published at some stage. It was a very busy period. There was lots of critical remarks both written to us and by us. Writing rebuttals to answer all the questions put to us by the journal, and we are really happy that it is willing to publish the results of more than 3 years of hard work of some 20 colleagues, 14 external reviewers, plus financial support of the ESPKU.

The very important thing is that our work has not been influenced by you as a patient organisation, or anybody from industry. Guidelines need to be seen as neutral as can be, and as much as possible – evidence based.

And now, will the world of PKU change by this document? That is the most important question. When it has been published, we can tell you about its content, not before! These are the rules we have to go by. When we tell you about its content, we can translate the publication and put it on websites referring to the original paper.

We will also publish the whole document of some 80 pages. This again will take some time. That journey can start now!

And what can you do to harmonise our treatment strategies on PKU in Europe?

Discuss the guidelines with your friends, with your families, with your professionals and see what is possible to implement and where the hurdles are and how these should be overcome.

Have a good meeting

Prof Francjan van Spronsen

Chair of the Scientific Advisory Board (SAC) of the ESPKU and Allied Disorders

ORGANISATIONAL REMARKS

BREAKFAST will take place from 06.30 until 10:00 in the Restaurant. Special LP food will be served in the Rose Room adjacent to the Restaurant. Please look for the signs.

LUNCH will be served from 12:30 until 14:00 in the Restaurant and LP food served in the Rose Room

THURSDAY: BUFFET DINNER

19:00 in the Restaurant and Rose Room (LP food)

FRIDAY: BUFFET DINNER

19:00 in the Restaurant and Rose Room (LP food)

SATURDAY: GALA DINNER

Call at 19:00 for surprise pre dinner entertainment

Dinner at 19:45 in the Baskin Suite

Please take care to pay for your extras to the waiters and at the bar.

Participants to wear nametags/lanyards **AT ALL TIMES**, otherwise the hotel staff will ask you to pay for your food. Name tags will be differently coloured...

- **GREEN:** Patients/Families
 - **RED:** Delegates of ESPKU Member Associations
 - **BLUE:** Scientists/Professionals
 - **YELLOW:** Industrial Representatives
-

During lectures and presentations, please switch off your mobile phones. For the same reason we request that you be in time for each programme you wish to attend.

Photos and videos may be taken during the Conference for public use by the ESPKU & PKU Ireland (print, internet and social media). By attending the Conference, all participants renounce their portrait rights. In case of disagreement, please inform the photographer/cameraman immediately.

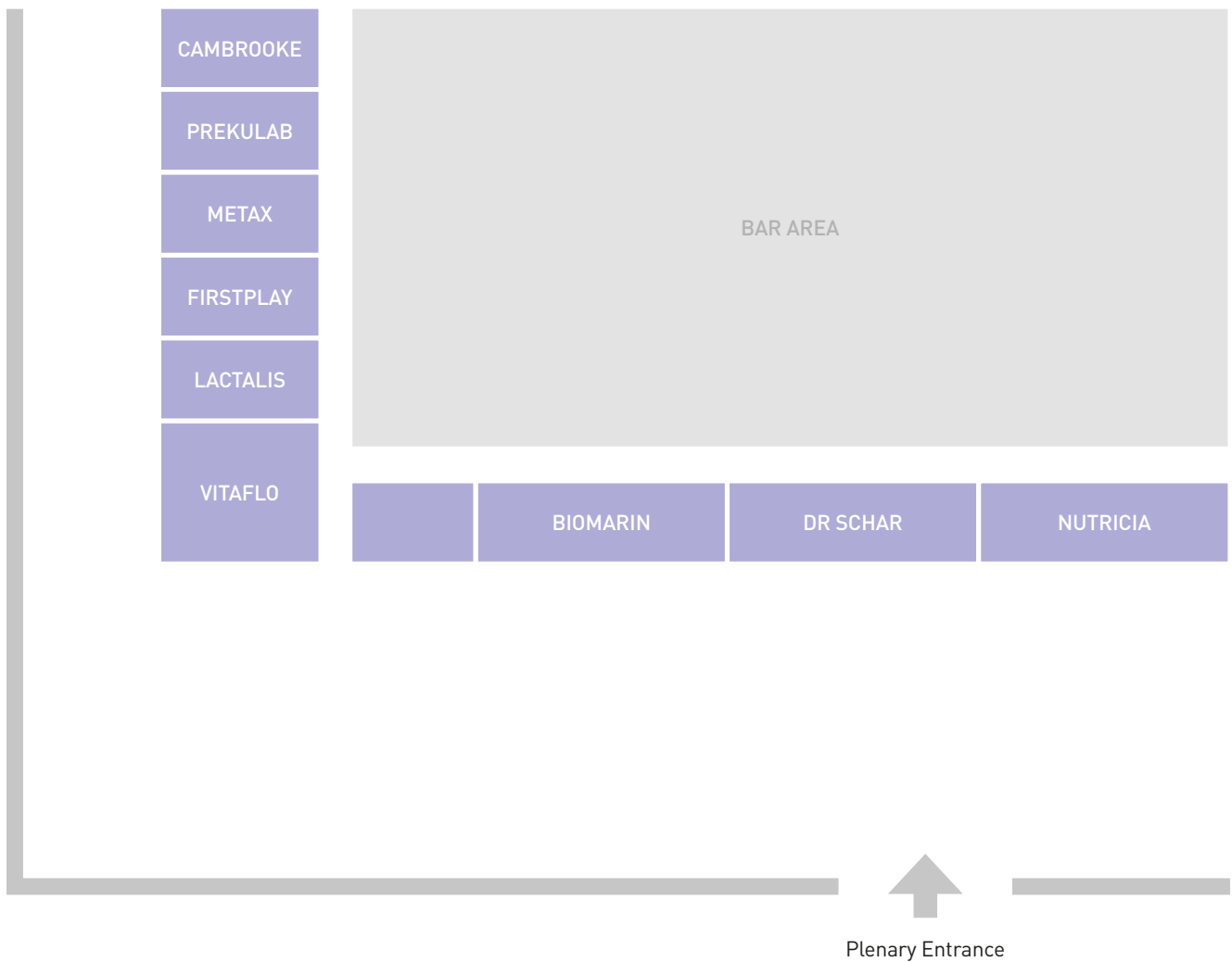
The Conference organisers aim for a peaceful and relaxed Conference atmosphere and expect appropriate behaviour from all participants. The ESPKU; PKU Ireland and the Hotel execute domestic authority. Please attend their orders at any time?

Feedback

At the end of the Conference you will be asked for your feedback about the Conference. Please take a few minutes to answer our online request which will be available on www.espku.org or with this qr code:



EXHIBITION AREA FLOOR PLAN



INDUSTRIAL EXHIBITION

The following companies (in alphabetical order) present their products and services at the industrial exhibition:

Biomarin	Lactalis
Cambrooke	MetaX
Dr Schar	Nutricia
Firtsplay	Vitaflo

The organisers of the ESPKU Annual Conference are grateful for their support.

The exhibition will be open on Friday from 10:30 until 18:00 hrs and Saturday 10:30 until 14:00 hrs

CRÈCHE AND CHILDCARE

Crèche and childcare is offered for children between 3 –11 years old. An experienced and educated team of social workers and nurses will offer a bouquet of games and activities, such as indoor hockey, chess, painting and many more.

For registering your children please contact the staff at the registration desk on Friday morning, 8:00 – 10:00 or Saturday morning, 8:00 – 10:00.

There will be an outing on the Friday afternoon for those 12 years old and above to GoQuest Indoor Challenge Park. At the time of going to press the venue has been provisionally booked.

The children's participation in the program of the ESPKU conference is at their parents' risk and responsibility. The European Society for Phenylketonuria and allied disorders treated as Phenylketonuria (ESPKU) is in no way responsible or liable for any direct or indirect damages of any kind (including any physical or mental injury) incurred in relation to the participation or organization of activities and child care during the conference.

The parents and the guardians of the children and any other persons related to the organization or the child care of the event, hereby irrevocably and unconditionally waive any claims against the ESPKU to the extent permissible by applicable law. Representatives of ESPKU acting as guardians, custodians or in any other function for children who are involved in the program of the ESPKU conference are not responsible or liable for any kind of damages which incur during the event and no claims may be raised against any of these persons.

In addition, the parents of the attending children are required to confirm that they are not aware of any health problem or medical condition of their child which could make their participation risky or questionable. In case of a child's necessity of medical assistance, the parents are obligated to provide this assistance and to inform the guardians about the respective health problem or medical disorder, so that the safety of the child can be granted at each point in time during the conference.

In accordance to provide their children's participation in the ESPKU conference, the parents must sign an exclusion of liability and accept the conditions of participation when allowing their children to take part in the creche and childcare programme. Please contact the people at the registration desk.

PROGRAMME*

Friday – COMMON PROGRAMME	
Time	Topic/Speaker
08:45	WELCOME Fergus Woodcock – Chairman, PKU Ireland Eric Lange – President, ESPKU Prof Francjan van Spronson – Chairman, ESPKU Scientific Advisory Board
	Chairs: Prof Maria Gizewska and Prof Eileen Treacy
09:00	Dr Ahmad Monavari: Care for PKU in Ireland
09:25	Prof Dr Francjan van Spronson: do we know enough about Pathophysiology of PKU to know our treatment?
09:50	Prof Maria Gizewska: Late diagnosed PKU: a need for treatment?
10:15	COFFEE BREAK

Please note that from 10:45 on Friday the Programme will split into three separate programmes that will run concurrently for the rest of the day:

- Scientific Programme
- Patients’ Programme
- Delegates’ Programme

Dinner is at 19:00

The ESPKU Annual General Meeting:
The AGM will be held on Friday at 21:00

Friday – SCIENTIFIC PROGRAMME		
Time	Topic/Speaker	
Chairs: Prof Francjan van Spronson and Prof Eileen Treacy		
10:45	Dr Andrea Pillot: towards elderly PKU	
11:20	Danique van Vliet: Moving beyond phenylalanine levels to further optimise PKU management	
11:40	Free Orals 1. Dr. Denise Ney: Bone health in PKU: a comparison of amino acid and glycomacropeptide medical foods 2. Dr. Hani Alfheaid: Diet-Induced Thermogenesis is Diminished Following Consumption of Phenylketonuria Type Meals 3. Christine Brown, Executive Director NPKUA of America: Patient Input on New Treatments 4. Dr. Aoife Fitzgerald, Metabolic Dietitian: The incidence of overweight & obesity in Irish children with PKU compared with the GUI Study	
	12:45	LUNCH
	Chairs: Prof Maria Gizewska and Dr Amaya Belanger	
	14:00	Free Orals 5. Dr Denise Ney: Glycomacropeptide for nutritional management of PKU: randomised, controlled, crossover trial 6. Danique van Vliet, MD/PhD student: Moving beyond phenylalanine levels to further optimise PKU management 7. Dr. Naser Harzandi: Screening of phenylalanine ammonia lyase in cyanobacteria (<i>Anabaena</i> 90 & 88) isolated from IRAN. 8. Kirsten Ahring, Msc: Absorption of respectively Casein glycomacropeptide (CGMP-20) and free synthetic amino acids (AA) in phenylketonuria (PKU) patients in connection with standardised meal. 9. Orla Purcell, Rd: Dietary management of PKU in premature infants.
15:30		COFFEE BREAK
Nutrition in PKU – Chairs: Kirsten Ahring and Prof Anita MacDonald		
16:00		Dr S Friso: Epigenetics and its influence on outcome in PKU
16:30	Dr Amaya Belanger: Growth and weight gain in dietary treated PKU patients: a multicenter study	
16:55	Prof Anita MacDonald: Glycomacropeptide in PKU treatment	
17:20	Discussion	
17:30	End of the day	

* This Programme is correct at the time of going to press and is subject to change.

Friday – PATIENTS’ PROGRAMME

Time	Topic/Speaker
Chair of all sessions: Fergus Woodcock, PKU Ireland Chairman	
10:45	Anne Green – former director of Metabolic unit in Birmingham who will present the Sheila Jones story
11:30	Debbie Colyer – late diagnosed PKU from Australia awarded a medal of honour by the Australian Government for her activism and pioneering work in PKU
12:00	Karen Farrell – a mother of a child with Homocystinuria (HCU)
12:30	LUNCH
14:00	Leonie Tully – mother of triplets with Phenylketonuria
14:20	Julie Cardy – my life with PKU and being a PKU Mother
14:40	Mandy Macedo – mother of a child with PKU child Don't take no for an answer – one Mother's fight
15:00	Debbie Colyer – how the Australian Association fought the Government and won
15:30	COFFEE BREAK
16:00	Laurie Bernstein – Assoc Prof of Pediatrics at the University of Colorado – Letting Go: How and When to Teach Self-Management Skills.
16:20	John Adams – President of CanPKU and Allied Disorders since 2008: Global Perspectives
16:50	Peter Lavery – young PKU: Climbing Kilimanjaro with PKU

Friday – DELEGATES’ PROGRAMME

Time	Topic/Speaker
Chair: Tobias Hagedorn, Secretary ESPKU	
10:45	Eric Lange – Welcome
11:00	T Hagedorn – What's on in... Introduction of attending delegates/Presentation of the hosting Irish PKU Association/Presentation of members activities
12:30	LUNCH
14:00	Prof Tangerass – European Reference Network METAB-ERN Update on latest development/Patient participation/information from inside
14:45	T Hagedorn – Latest news from the EU Latest developments on the review of the EU legislation on dietary food (FSMP) and other EU legislations
15:30	COFFEE BREAK
16:00	Prof F van Spronsen – European Guidelines Status quo of publication process, timeline/preparation of implementation campaign
16:30	Paul van Berkel – ESPKU's future agenda Presentation and discussion of a mindmap explaining ESPKU policies and activities
17:15	T Hagedorn – ESPKU: A Community? Options for bi-lateral and multilateral cross-border cooperation of ESPKU member associations
18:15	Eric Lange – Any other business/Close

18:00 Patients & Parents – Meet the experts (the Plenary room)

Saturday – COMMON PROGRAMME

Time	Topic/Speaker
Chairs: Prof Maria Gizewska and Assoc Prof Laurie Bernstein	
09:15	Dr Tafweg Ben-Omran: Classical Homocystinuria and PKU in Qatar: To Better Meet Family and Patient Needs
09:15	Prof Maria Gizewska: Can a woman with PKU become a happy mother?
09:45	Dr Aria Setoodah: PKU in Iran – what are the challenges to be met?
10:15	COFFEE BREAK

Time	Topic/Speaker
Chairs: Prof Francjan van Spronsen and Kirsten Ahring	
10:45	Annemiek van Wegberg/Prof Francjan J van Spronsen: Guidelines finished: the journey only starts now!
11:45	Laurie Bernstein: How does your PKU play a role in your day?
12:45	CLOSING CEREMONY

ABSTRACTS

Late diagnosed PKU: a need for treatment

Maria Gizewska

Department of Pediatrics, Endocrinology, Diabetology, Metabolic Diseases and Cardiology, Pomeranian Medical University, Szczecin, Poland

Despite many years of neonatal screening for PKU and great success of early treatment offered to vast majority of patients, the problems of late-diagnosed and therefore potentially late-treated or untreated persons with PKU still remain to be solved.

It applies to many countries worldwide and not only to those which have not introduced neonatal screening for PKU yet. Thanks to improvement in medical care of persons with intellectual disabilities, even in regions with long history of neonatal screening, untreated or late-diagnosed patients with PKU may reach an advanced age.

There is no clear answer to the questions if all late-diagnosed patients with PKU should be treated but this option should be always considered. In untreated PKU patients introduction of treatment can be beneficial in several domains, but individual decision of initiation is necessary.

It was proven that in some of late-diagnosed or untreated patients with PKU, dietary treatment can result in positive changes in their challenging behaviours, including decrease in aggressiveness, irritability and hyperactivity, severity of neurological symptoms as well as number and/or severity of epileptic attacks. Effectively treated patients often demonstrate improvement in verbal communication, eye contact, intellectual functioning, mood and social awareness.

As it is impossible to predict who will response to late introduction of treatment and patients have individual sensitivity to dietary intervention – a treatment trial of 6 months should be given before the outcome of treatment is evaluated. Diet should be introduced methodically, enabling the patient to gradually adapt to taking and tolerate a low phenylalanine diet. The PKU team should closely support caregivers, providing practical advice and regular review.

Professor Maria Gizewska MD, PhD graduated from the Pomeranian Medical University in Szczecin, Poland in 1985. Since 1986 she has been working as an academic teacher for medical students at Pomeranian Medical University.

Professor Gizewska started her career at the Department of Biochemistry and soon joined the Department of Children's Diseases. In 1995 she obtained her PhD. She is a specialist in general pediatrics and pediatric metabolism.

Since 1998 she is the Vice-head of the Department of Pediatrics, Endocrinology, Diabetology, Metabolic Diseases and Cardiology of Pomeranian Medical University in Szczecin. From the mid-nineties her research is focused on early diagnosis and treatment of children, adolescents and adults with different types of inborn errors of metabolism, including almost 250 patients with phenylketonuria. She is a consultant on inborn errors of metabolism for the Region of West Pomerania, Poland.