

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

Annual Report 2015

Translate visions into reality



**A dream you dream alone is only a dream.
A dream you dream together is reality.**

John Winston Ono Lennon, MBE
1940 - 1980

Words of gratitude:

Thanks to all who supported the work of ESPKU this year and are not mentioned in this report.

Content

- 1 Meetings and Conferences
- 2 ESPKU Guidelines
- 3 Political work of the ESPKU
- 4 Eurordis Meeting 2015, Madrid
- 5 Expansion of the ESPKU
- 6 International PKU Day
- 7 The ESPKU Internet and Social Media Activities
- 8 A look into future
- 9 Impressum

1 Meetings and Conferences

The 2014 ESPKU Annual Conference in Zagreb, Croatia

The 28th ESPKU Annual Conference took place in October 2014 in Zagreb, Croatia. Organised by the Croatian PKU Society, it was one of the best conferences ever – following the high standard set by BOKS.



With a live debate to independently find out what the mindset is, the ESPKU put further emphasis in the development of the guidelines and the implementation of patient perspectives.



With the conference, Croatia has become a link between western and central European countries and south eastern European countries where the gaps in PKU care are huge. The chairman of the hosting Croatian PKU Society Dr. Sanja Peric recapped: „The knowledge we gained in the conference, the experience we exchanged, and the friendships that were made will help PKU patients in their every-day-life.“

The ESPKU General Meeting elected a new executive board. Former ESPKU Vice-President Dinah Lier and Assessor Thomas Møller-Nielson resigned from office. Eric Lange (President), Maarten Criem (Treasurer) and Tobias Hagedorn (Secretary) have been re-elected. Gregor Hammerschmidt was elected as new Vice-President. Paul van Berkel completes the team as project manager.

Furthermore the General Meeting has adopted the „Agenda 2020“. With this policy plan ESPKU will focus on the implementation of best practice care for PKU and allied disorders.

The 2015 Spring Meeting of ESPKU member delegates

On March 28th, 2105 the Spring Meeting of the European Society for Phenylketonuria and Allied Disorders took place in Berlin, Germany. 24 delegates from 13 national PKU Associations took part and discussed the latest political developments.

Resulting from a presentation by Norbert Pahne from Diätverband (Germany) the attending delegates were informed that the on-going revision process of European legislation on dietary food may impact reimbursement and labelling of low protein products. For further details, please read chapter 3 of this annual report.

Another key topic was a presentation of Kirsten Ahring (Secretary of the ESPKU Scientific Advisory Committee) on the development of the ESPKU guidelines (see next chapter). All attending delegates were convinced of the direction of the presented statements, however it was agreed to include the ESPKU Consensus Paper Working Group into the review of the draft Guidelines.

The 2015 ESPKU Annual Conference in Berlin, Germany



The 2015 ESPKU Annual Conference was organised by the German PKU Association at the occasion of its 40th anniversary. In attendance were the descendants of Prof. Følling, Prof. Bickel and Prof. Guthrie. The ESPKU Guidelines were presented to public.



With a documentary film on late diagnosed PKU the importance of neonatal screening, low phenylalanine diet and amino acid supplementation was highlighted.

2 The ESPKU Guidelines

One of the major results of the ESPKU benchmark report „Closing Gaps in Care“, was that PKU management differs considerably across Europe. Development of European guidelines was urged both by health professionals and the ESPKU. Within 3 years agreed guidelines based on the highest quality available evidence have been developed by a multidisciplinary group of 17 professional experts, organised by the ESPKU scientific advisory committee, and presented to the public at the 29th ESPKU Annual Conference in Berlin, Germany.

The goal was to suggest standardized treatment for PKU across Europe that would lead to optimal neuropsychological outcome without overtreatment and unnecessary costs. These guidelines are intended to be used by metabolic physicians, dietitians, gynecologists, psychologists, social workers, biochemists and other professionals involved in the treatment of patients with PKU.

These guidelines are the first ever pan-European guidelines in the treatment of PKU. They also focus on different treatment aspects -nutritional treatment and biochemical / nutritional follow up, neurocognitive outcome including imaging, psychosocial outcome and adherence, adult and maternal PKU, late diagnosed and untreated PKU, diagnosis of PKU including treatment initiation, drugs in PKU. Once implemented into national practice, these guidelines will significantly improve treatment quality and help closing gaps in care.

The ESPKU is thankful to all who contributed and gave support to this important project, including the professionals in the guideline group, the numerous sponsors and donators, the reviewers and last but not least Francjan J. van Spronsen (chairman of the ESPKU Scientific Advisory Committee and guidelines working group) and Annemiek van Wegberg (ESPKU Guidelines project manager).

3 The political work of ESPKU

The continued consolidation of European legislation on dietary food may have an impact on reimbursement and labeling of low protein food products. Currently low protein foods as required for the PKU diet may be marketed as “foods for particular nutritional purposes sui generis” according to Article 11 of Directive 2009/39/EC – the current so-called framework directive for dietary foods. The concept of dietary foods and Directive 2009/39/EC including its Article 11 is repealed by Regulation 609/2013 which introduces a new legal framework for foods for special consumer groups (FSG) instead. This new Regulation will enter into force in July 2016 and does not foresee any rules for low protein foods as required for the PKU diet.

As a consequence the affected food products like pasta, bread, flour and other low protein food will have to be switched to foods for general consumption and the relevant legislation will apply. In this case the quantitative labeling of Phenylalanine will not be possible from July 2016 onwards. Alternatively low protein products could to be switched to medical food in the sense of the legislation for foods for special medical purposes (FSMP). But the European Commission and the Member States are not supposed to accept low protein convenience foods as part of the FSMP category. This development may also have an implication on reimbursement of low protein food.

The ESPKU has presented these concerns to MEP Francesco Carlucci, who promised that the new framework of Regulation (EU) No 609/2013 will in no way lower the level of protection of PKU patients. Furthermore he reassured that the Commission will take into account existing rules applicable to these products. With regard to the expected impact of the EU legislation, ESPKU President Eric Lange has again explained in a personal meeting that the issue not applies to Amino Acid Supplements (FSMP), but to the special low protein phenylalanine food. ESPKU will continue advocacy for a sufficient labelling of low protein food.

4 Eurordis Meeting 2015, Madrid

The ESPKU was represented by President Eric Lange, who used the opportunity to meet with several stakeholders and to establish and improve the ESPKU networks.

The most important topic of the meeting was “European Reference Networks for rare diseases (ERN)”. An ERN is a framework for healthcare pathways for rare-disease patients through a high level of integrated expertise. ERN’s provide the structure that facilitates a doctor’s ability to access specific knowledge across borders. Following the EUCERD recommendations, Centers of Expertise shall collaborate with patient organisations to bring in the patient perspective. Patient organisations are not legally required to participate in the governance and evaluation of ERN’s, but ERN’s are required to demonstrate patient-centric care and patient empowerment. The involvement options of patients associations such as ESPKU is yet to be formalised.

5 The Expansion of ESPKU

In 2015, PKU Associations from Ireland, Belarus, Israel and Gaza Strip applied for legal ESPKU membership. At the time of writing, it is assumed that the ESPKU General Meeting 2015 will accept all applications, the ESPKU membership will increase to 34 countries. As many members are not from EU member states, the ESPKU must decide upon its non EU activities and in close cooperation with the new members.

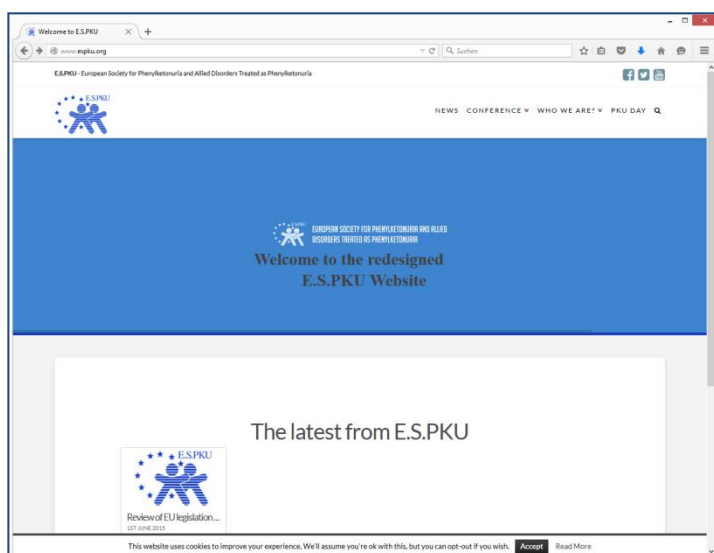


6 International PKU Day



The International PKU Day was celebrated for the second time with increasing activities of ESPKU members, such as meetings, press releases, picnics and camps. Not only ESPKU members, but also other PKU and rare disease associations, industries, institutions and individuals have been active to raise awareness for PKU and promote the demands of patients all over the world.

7 ESPKU internet and social media activities



The ESPKU website was completely overhauled with new software, design and content. It is now offering more information about ESPKU projects and more dedicated news on the conference, to gain more interest. An online feedback-questionnaire will help ESPKU and members to improve future conferences to an even higher level.

Aiming to improve communication with and amongst its members and the public, the ESPKU has implemented a newsletter for ESPKU member delegates.

A travel network was launched in cooperation between www.pkuboard.info and ESPKU, possibly to build a contact network on PKU's willing to help while traveling.

The ESPKU of social media has launched on facebook and twitter. Offering news on current topics within ESPKU as well as the conference, we have already approximately 500 likes on facebook and 125 followers on twitter.



8 A look into future

Over the last 4 years the ESPKU has become more and more politically active on an EU level. In the same period we expanded our membership considerably. We are now finishing the first ever pan-European guidelines framework on PKU.

A basis for this is the successful teamwork between ESPKU members, ESPKU board and the Scientific Advisory Committee of ESPKU.

Due to this development ESPKU is no longer recognized just as a patients association. This has enhanced our reputation as the European PKU organization for both patients and professionals. Consequently the ESPKU has to address some important questions on its future direction. This is the challenge ahead, and it offers the opportunity to increase our influence as ESPKU and for our increasing membership base. That includes our non EU activities.

Another future challenge is a more integrative approach to allied disorders. It will be important for ESPKU to be involved in other guideline developments, e.g. for Tyrosinemia Type I and II, Urea Cycle Defects, Homocystinuria, Organic Acidurias such as MSUD, MMA, PA, IVA, Glutaric Aciduria Type I and maybe even Vitamin B6 dependent epilepsy.

Finally the participation of the ESPKU in the governance and evaluation of European Reference Networks offers the opportunity to implement more patient centric care across Europe, even if the participation of patients associations in that process formally remains to be unclear.

9 Impressum

The Annual Report 2015 was presented to the delegates of the attending member associations of ESPKU at the 2014 General Meeting of ESPKU, held on October 23rd, 2014 in Berlin, Germany. It provides insight into the main working fields of ESPKU from October 2014 until September 2015, without laying claim to completeness. This annual report is published at www.espku.org.

On behalf of the ESPKU Executive Board



Eric Lange, President



Tobias S. Hagedorn, Secretary

European Society for Phenylketonuria (ESP KU)
and Allied Disorders treated as Phenylketonuria

A non-profit organisation promoting exchange of information about phenylketonuria (PKU) and allied disorders of amino acid metabolism.

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The European Society for Phenylketonuria and Allied Disorders treated like Phenylketonuria
is a non-profit organisation with members in the following countries:

Austria	Germany	Poland (2)
Belarus (Candidate for membership)	Hungary	Portugal
Belgium	Iceland	Romania
Bulgaria	Ireland (Candidate for membership)	Serbia / Montenegro
Croatia	Israel (Candidate for membership)	Slovakia
Czech Republic	Italy	Slovenia
Denmark	Latvia	Spain (2)
Estonia	Lithuania	Sweden
France	Moldova	Switzerland (2)
Gaza Strip (Candidate for Membership)	Norway	Turkey
Georgia		United Kingdom

Executive Board:

President: Mr. Eric Lange, United Kingdom
 Vice-President: Mr. Gregor Hammerschmidt, Austria
 Treasurer: Mr. Maarten Criem, Belgium
 Secretary: Mr. Tobias S. Hagedorn, Germany
 Project Manager: Mr. Paul van Berkel, The Netherlands

Board of the Scientific Advisory Committee:

Chairman: Dr. Francjan J. van Spronsen, The Netherlands
 Vice Chairman: Ass. Prof. Dr. Maria Gizewska, Poland
 Secretary: Mrs. Kirsten Ahring, Denmark

Bank Account:

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