E.S.PKU

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

Annual Report 2017

Persistence pays off

A very little key will open a very heavy door.

Charles Dickens (1812 – 1870), British author
Hunted down, Chapter II

Words of gratitude

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

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Preface

A bunch of keys is an important symbol for the success of courage, engagement and science. It was this bunch of keys that drew Sheila Jones attention when she was treated with the first phenylalanine-restricted casein hydrolysate as the main source of protein in her nutrition. The history of PKU is characterized by the cooperation of the patient’s perseverance and scientist’s commitment.

This ESPKU Annual Report presents a non-exhaustive overview on some important activities that our community has taken or has been part of, without claiming to be complete. All these projects and tasks have one in common: Perseverance, commitment and cooperation of patients and their families, scientists, healthcare providers and other stakeholders. Whoever has been, is, or will be involved to improve just one single patients life, we all owe them gratitude.

The work of the ESPKU Executive Board: Key learnings

In three meetings and several phone calls and skype conferences, the Board has discussed and decided many different topics. Apart from care-related topics like the European Guidelines, European Reference Networks and EU legislation issues, questions on the future orientation of ESPKU were also on the agenda, including numerous current and future initiatives.

In addition, contacts have been established and maintained with other PKU- and rare disease associations from all over the world, professional associations, representatives of European health policy, industries and other stakeholders.

ESPKU quickly responded to a current development: Within a few days and in close cooperation with our Spanish member and the Scientific Advisory Board, ESPKU published a supportive position paper on the Situation of Refugee Rare Metabolic Disorder Patients.

The number of tasks and agenda items is growing steadily and requires greater commitment. However, the capacities of a voluntarily working executive committee are limited. In order to meet the topics and duties more efficiently, the Board has begun to outsource administrative tasks to external service providers.

Annual Conference 2016 in Dublin: Key results

Since for safety reasons it was impossible to organise the 2016 ESPKU Annual Conference in Istanbul (Turkey), the PKU Association of Ireland took over to be a perfect host. With no conference organising experience and within just eight months, Karen Willets and her team have successfully prepared and executed the meeting in the Irish capital Dublin.

About 470 patients and relatives, patient representatives, scientists, nutritional experts and sponsors from all across Europe and the entire world showed up in this important event in the global PKU calendar. After a weekend full of outstanding presentations by superb speakers on the need for treatment and on potential improvements of nutritional PKU therapy, Fergus Woodcock as president of the hosting PKU Association of Ireland summarised the most important conference outcomes. As in PKU history, it still needs determined parents and patients. He was optimistic that persistence will pay off: “PKU can’t hold us back. Anything is possible.”

The 2017 ESPKU Spring Meeting for members delegates: Future key projects

The ESPKU Advocacy Toolkit, the ESPKU Benchmark Report, the ESPKU Consensus Paper on Minimum Requirements for PKU Treatment, the initiation of European PKU Guidelines: ESPKU has achieved much in recent years. All these projects reflected issues common to patients across European countries and were the result of ESPKU member’s cooperation.

The 2017 ESPKU Spring Meeting for member’s delegates took place in Hell (Norway). Chaired by Marketa Lhotakova (Czech Republic), the attending delegates identified potential future key projects for ESPKU having a significant benefit to European patients. Taking into account challenges and opportunities, they sketched a draft action plan on each of them, including proposals for related ESPKU and ESPKU member’s activities.

Amongst other ideas, the non-exhaustive list of potential projects included:

- Transition from paediatric to adult care
- Equal access to products, health services and treatment options for PKU patients
- European PKU Database
- The Inconsistency of neonatal screening across Europe

The discussion will be followed up in the upcoming ESPKU member’s delegates meetings.

European Guidelines on Diagnosis and Management of patients with PKU: Key statements

To close some major gaps in PKU care identified in the 2012 ESPKU Benchmark Report, in their consensus paper ESPKU and its members called for comprehensive European Guidelines in 2013. Initiated and funded by ESPKU and coordinated by the chairman of the ESPKU Scientific Advisory Committee Prof. Francjan J. van Spronsen, a multi-disciplinary group of experienced clinical professionals and researchers has scientifically developed 70 recommendations. 10 key statements have been published in the Lancet Diabetes and Endocrinology on January 2017, including recommendations on the treatment initiation and duration as well as target Phe levels:

Review

Key European guidelines for the diagnosis and management of patients with phenylketonuria

We developed European guidelines to optimise phenylketonuria (PKU) care. To develop the guidelines, we did a literature search, critical appraisal, and evidence grading according to the Scottish Intercollegiate Guidelines Network method. We used the Delphi method when little or no evidence was available. From the 70 recommendations formulated, in this Review we describe ten that we deem as having the highest priority. Diet is the cornerstone of treatment, although some patients can benefit from tetrahydrobiopterin (BH4). Untreated blood phenylalanine concentrations determine management of people with PKU. No intervention is required if the blood phenylalanine concentration is less than 360 µmol/L. Treatment is recommended up to the age of 12 years if the phenylalanine blood concentration is between 360 µmol/L and 600 µmol/L, and lifelong treatment is recommended if the concentration is more than 600 µmol/L. For women trying to conceive and during pregnancy (maternal PKU), untreated phenylalanine blood concentrations of more than 360 µmol/L need to be reduced. Treatment target concentrations are as follows: 120–360 µmol/L for individuals aged 0–12 years and for maternal PKU, and 120–600 µmol/L for non-pregnant individuals older than 12 years. Minimum requirements for the management and follow-up of patients with PKU are scheduled according to age, adherence to treatment, and clinical status. Nutritional, clinical, and biochemical follow-up is necessary for all patients, regardless of therapy.

Introduction

Patients with untreated blood phenylalanine concentrations less than 360 µmol/L do not require treatment. With untreated phenylalanine levels between 360 µmol/L and 600 µmol/L, they should be treated until the age of 12 years. Patients with untreated phenylalanine levels more than 600 µmol/L should be treated for life.

In treated patients up to the age of 12 years, target phenylalanine concentrations should be 120–360 µmol/L. In treated patients aged 12 years or older, the target phenylalanine concentrations should be 120–600 µmol/L.

The publication of the 10 key statements has received great attention and broad support in a short time. They have already been implemented in numerous metabolic centres across Europe.

The publication of the entire 70 European guidelines is expected soon.

EU legislation on dietary food: A political key issue

In summer 2016, the EU regulation 609/2013 on food for special consumer groups replaced the old framework directive on dietary food. As a result, low-protein products would no longer have been labelled as such, as well as no quantitative labelling of phenylalanine would have been allowed anymore. This would have affected the applicability of these products and their reimbursement by national health systems. To prevent this, it was agreed with Francesco Carlucci from the EU Commission, that no provision shall be introduced that will make it more difficult for PKU patients to use food for special medical purposes (FSMP) intended for the dietary management of PKU. Dedicated low protein food should be covered by the new FSMP directive in future.

In spring 2017, the ESPKU Executive Board had access to a draft guideline on the classification of FSMP, which gave rise to further concerns about some potential and unacceptable restrictions. Once again, we have successfully addressed our concerns to Francesco Carlucci: The critical points of the draft have been revised.

Currently the internal consultation process on the draft guideline is going on. The ESPKU Executive Board and some ESPKU member association are following the process at various political levels. The release of the final version is not expected before 2018.

Taking care for others: A key duty of any patients association

End of 2016, at the height of the European refugee crisis, the ESPKU took a clear position and called for rapid support for refugee patients with rare metabolic disorders. In a position paper, ESPKU highlighted the need for quick identification of diagnosed and undiagnosed patients and their prioritised relocation. Only in a steady environment, these vulnerable individuals can receive the required multidisciplinary care and dietary treatment to avoid further deterioration of their health status.

Foundation of a Global Association for Phenylketonuria: A key initiative

At the occasion of the ESPKU Annual Conference 2016 in Dublin, patient representatives from Australia, Canada, Europe and the USA appointed to start with a new initiative.

In July 2017, several country-based PKU patient organizations from Argentina, Australia, Brazil, Canada, Chile, Europe, Mexico, Turkey and the United States of agreed to form the first global association for PKU.

This entity will serve as a supportive umbrella organization to regional and national patient organisations and groups in bridging the gap for PKU worldwide.

The vision of the Global Association for PKU is that all people living with PKU, despite their origin, will be able to reach their full potential. The mission is to advance PKU as a global health priority and strengthen PKU associations and groups throughout the world.

The priorities of this global network of patients associations are:

- to create a global platform for PKU advocacy that includes ensuring universal access to neonatal screening and diagnosis, treatments and comprehensive care;
- to mentor like-minded patient associations and groups and offer best practices and support that are sensitive to their experiences, culture, language(s) and laws;
- to increase collaboration among PKU researchers, scientists, clinicians and patient groups to move basic science and research forward to accelerate new knowledge, treatments, and a cure.

The formation of this group is in the early stages. Organisational tasks have been assigned and plans are progressing for other meetings and the final formal foundation of the Global Association for Phenylketonuria.

Summary and outlook

Since its foundation in the late 1980th, ESPKU has grown to a number of 40 member associations representing 33 countries. Beyond the organisation of an annual meeting of international importance, the ESPKU has achieved very much in recent years:

- With the Benchmark Report and the ESPKU Consensus Paper on minimum requirements on PKU treatment, ESPKU has initiated European Guidelines on the management of PKU.
- ESPKU is actively working to influence politics on both EU and national levels and has an impact on EU legislation.
- ESPKU is promoting the foundation and cooperation of local and national PKU patients associations, especially in Eastern European countries.
- ESPKU provides solidly researched information on current issues through various channels like the ESPKU website or social media.
- ESPKU has increased its networking activities with numerous rare-disease associations, healthcare providers and professional associations, political stakeholders, industries and many more.

It becomes more and more evident, that the future ESPKU agenda must balance political efforts with different projects to target both social and healthcare improvements as well as a greater public perception of ESPKU as a patient centric stakeholder with a significant added value for its members and all European PKU patients and other stakeholders.

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The ESPKU Annual Report 2017 was presented to the delegates of the attending member associations of ESPKU at the 2017 General Meeting of ESPKU, held on October 6th, 2017 in Hell, Norway. It provides insight into the main working fields of ESPKU from October 2016 until October 2017, without laying claim to completeness. This annual report will be published at www.espku.org.

On behalf of the ESPKU Executive Board

Eric Lange, President

Tobias S. Hagedorn, Secretary

European Society for Phenylketonuria (ESPKU)
and Allied Disorders treated like 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
- Belarus
- Belgium
- Bulgaria
- Croatia
- Czech Republic
- Denmark
- Estonia
- France
- Palestine / Gaza Strip
- Georgia
- Germany
- Hungary
- Iceland
- Ireland
- Israel
- Italy
- Latvia
- Lithuania
- Moldova
- Norway
- Poland
- Portugal
- Romania
- Serbia / Montenegro
- Slovakia
- Slovenia
- Spain
- Sweden
- Switzerland
- Turkey
- Ukraine
- 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

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