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

Annual Report 2014

Simplicity, Patience and Compassion

Annual Report 2014

”I just have three things to teach: Simplicity, patience and compassion.

These three are your greatest treasures!”

Lao-Tzu
Chinese philosopher, about 600 B.C.

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

Annual Report 2014

Content

Meetings and Conferences

The 2013 ESPKU Annual Conference in Antwerp

Logo of BOKS

The 2013 ESPKU Annual Conference was hosted by the Belgium Association for Children and Adults suffering from a metabolc diseases "BOKS", and took place in Antwerp from October 31st until November 3rd, 2013. It was opened in attendance of his royal highness Prince Laurent of Belgium. In a video message, the Belgium Vice-Prime Mininstre and Ministre of social affairs and health, Laurette Onkelinx underlined the importance of rare diseases in health politics. Herman van Rompuy, President of the European Council, highlighted the meaning of cooperation between medical experts, patients representatives and politics. The high political attention of the ESPKU Conference is the consequence of ESPKU and its members politicking and purposive approaching of selected members of the European parliament.

Herman van Rompuy

More than 300 patients and their families, delegates of national PKU associations and professional health care providers attended the conference.

In the delegates programme, the executive director of EPPOSI (European Platform for Patiens Organisations, Science and Industry) talked about the ethical cost factors of systematic neo-natal screening programmes in Europe. "There is need for improvements of the rare disease infrastructure and clinical guidelines", she concluded her presentation.

In the General Meeting of ESPKU, the attending delegates and representatives of 16 PKU Associations from all over Europe appointed June 28th as "International PKU Day".

Ida Vanessa D. Schwarz (PhD) introduced the challenging task of treating PKU in Brazil. As in many European countries, economic resources are scarcer in South America.

The professional's programme offered several presentations, such as obesity in PKU, neurocognitive functioning, maternal PKU, quality of life issues and many more. It became visible, that many aspects have to be considered in the first ever pan-european guidelines on the management of PKU. That was also proven by a patients and families workshop on statements in guidelines, that really affect patients and parents.

Annual Report 2014

The 2014 ESPKU Spring Meeting for Delegates in Zagreb

For the 4th time, on march 29th, 2014, the delegates and representatives of 8 ESPKU member associations and the ESPKU Executive Board came together for a working conference to exchange information and experiences and to appoint common activites and political directions with each other.

The future ESPKU policy plan

The main topic on the agenda of the 2014 ESPKU Spring Meeting was the ESPKU future policy plan. Since the ”Closing Gaps in Care” campaign will be completed with the publication of the European Guidelines on the management of PKU most likely in 2015, the delegates agreed to focus on the implementation of these guidelines in national practice until 2020.

Notwithstanding the recently increased attention of rare diseases in European health politics, treatment costs still place a burden on PKU families and represent a significant barrier to the delivery of adequate care in many countries and regions. Subsequently, overcoming the discrepancies in reimbursement policies has been identified as another important issue for the future ESPKU policy.

The future ESPKU policy plan will finally be appointed and presented to public at the 2014 ESPKU Annual Conference in Zagreb, Croatia.

Publication of the ESPKU Consensus Paper on optimal PKU care

At the end of 2013, after 2 years of dedicated work, the first ever pan-European patient/carer perspective on optimal care of PKU has been published in the Orphanet Journal of Rare Diseases.

The paper was developed by consensus of a working group, following a multi-national stakeholder workshop involving representatives from 15 different countries and of the findings from the ESPKU Benchmark Report. On behalf of ESPKU, the authors Tobias S. Hagedorn, Paul van Berkel, Gregor Hammerschmidt, Marketa Lhotakova and Rosalia Pasqual Saludes fleshed out significant deficiencies and inequalities in medical and social care and psychological support of those with PKU. The paper also makes the case for action to be taken to improve the detection and management of the condition on a number of fronts, including:

- Mandatory, universal screening for PKU in newborns
- Equal access to treatment and monitoring of the condition throughout Europe
- Agreeing new standards of best practice
- Establishing European Centres of Expertise
- Reimbursing all aspects of treatment
- Specific interventions for high-risk groups
- Increasing the number of available healthcare professionals where required and elevating the status of dietitians
- Access for patients to multi-disciplinary teams
- Training for carers to provide more comprehensive social support for people with PKU

Hagedorn et al. Orphanet Journal of Rare Diseases 2013, 8:191
http://www.ojrd.com/content/8/1/191

ORPHANET JOURNAL OF RARE DISEASES

REVIEW Open Access

Requirements for a minimum standard of care for phenylketonuria: the patients' perspective

Tobias S Hagedorn[1,3*], Paul van Berkel[2], Gregor Hammerschmidt[1,4], Markéta Lhoták[5] and Rosalia Pasqual Saludes[6]

Abstract

Phenylketonuria (PKU, ORPHA716) is an inherited disorder that affects about one in every 10,000 children born in Europe. Early and continuous application of a modified diet is largely successful in preventing the devastating brain damage associated with untreated PKU. The management of PKU is inconsistent: there are few national guidelines, and these tend to be incomplete and implemented sporadically. In this article, the first-ever pan- European patient/carer perspective on optimal PKU care, the European Society for Phenylketonuria and Allied Disorders (E.S.PKU) proposes recommendations for a minimum standard of care for PKU, to underpin the development of new pan-European guideline for the management of PKU. New standards of best practice should guarantee equal access to screening, treatment and monitoring throughout Europe. Screening protocols and interpretation of screening results should be standardised. Experienced Centres of Expertise are required, in line with current European Union policy, to guarantee a defined standard of multidisciplinary treatment and care for all medical and social aspects of PKU. Women of childbearing age require especially intensive management, due to the risk of severe risks to the foetus conferred by uncontrolled PKU. All aspects of treatment should be reimbursed to ensure uniform access across Europe to guideline-driven, evidence-based care. The E.S.PKU urges PKU healthcare professionals caring for people with PKU to take the lead in developing evidence based guidelines on PKU, while continuing to play an active role in serving as the voice of patients and their families, whose lives are affected by the condition.

Keywords: Phenylketonuria, Standards of care, Screening, Guidelines, Europe, Centres of Expertise, Healthcare agenda, Patient advocacy, Patient group, Patient voice

Introduction

Phenylketonuria (PKU, ORPHA716) is a rare inherited disorder that affects around one in every 10,000 children born in Europe [1]. The metabolic defect underlying PKU is a mutation in the gene coding for the enzyme, phenylalanine hydroxylase (PAH), which is responsible for the transformation of phenylalanine into tyrosine [2-4]. Impairment of PAH activity in PKU causes increased levels of phenylalanine that if untreated cause devastating damage to the brain, with severe mental disability, reduced IQ, seizures and tremors, impaired executive function, psychological and behavioural issues and social difficulties [4-6].

Most patients with PKU are identified during neonatal screening [6] and all patients then require lifelong treatment [7]. The mainstay of the therapeutic management of PKU is a modified diet that includes specially manufactured foods low in protein, and phenylalanine-free amino acid supplements [3,4]. Maintaining adequate adherence to this diet is challenging, but effective in preventing the severe brain damage associated with uncontrolled blood phenylalanine, and allowing individuals with PKU to lead full and successful lives [5,7-9]. A pharmacologic treatment option, sapropterin, is available for prescription in a growing number of counties [10,11]. A number of other potential treatments that may contribute increasingly to the management of PKU in the future include better and more palatable phenylalanine-free foods, glycomacropeptide (a natural protein free of phenylalanine), large, neutral amino acids, phenylalanine-ammonia lyase (an injectable enzyme that metabolises phenylalanine) and – for the

* Correspondence: tobias-hagedorn@t-online.de
[1]European Society for Phenylketonuria and Allied Disorders (E.S.PKU), Melsele, Belgium
[3]Deutsche Interessengemeinschaft Phenylketonurie, Hiddenhausen, Germany
Full list of author information is available at the end of the article

BioMed Central

The publication of this consensus paper is a significant milestone in the collective effort to improve the treatment and management of PKU in Europe. It represents the most far-reaching attempt to date to capture the views of patients, professionals and advocates, providing a bluprint for universally supported guidance for clincal practice.

The paper is an open access publication and freely available at http://www.ojrd.com/content/8/1/191. The development of the paper was supported by an unrestricted grant form Merck Serono, a division of Merck KgaA, Darmstadt, Germany, with no influence on any content.

Development of the 1st Pan-European Guidelines for the Management of PKU

Parallel to the ESPKU consensus group, since 2012 a group of experts is working to find consensus on guidelines on the treatment of PKU for every age thoughout life. The expert group consists of 18 well established PKU researchers, who are able to step down from their national politics and therefore can reach for the guidelines on many PKU issues on European level. The key chapters of these guidelines will be:

- Neurocognitive outcome including imaging
- Psychosocial outcome and adherance
- Nutritional treatment and biochemical / nutritional follow up
- Adult and maternal PKU, late treated PKU
- Diagnosis of PKU, drugs in PKU (when to start)

The publication of the first ever pan-European PKU guidelines is expected for 2015. However, even today we recognise scientific activities on absolutely essential revision of existing national guidelines and agreement on new guidelines in some European countries.

The ESPKU Expert group is working independent from any industrial influence. The reimbusements of their expenditures are covered by a specific ESPKU budget, collected from several grants and support.

International PKU Day

Logo of the International PKU Day

Like many rare diseases, it is a characteristic of Phenylketonuria that the general public is not sufficiently informed on the disorder and the needs of patients.

At the ESPKU Delegates Meeting 2013 in Antwerp, it was decided to initiate the International PKU Day, taking place annually on June 28th. This is quite a special day because, not only is it nicely in the middle of the year but also, it is the birthday of two stalwarts of PKU, Robert Guthrie (1916-1995) and Horst Bickel (1918-2000).

Regional, national or international associations, individuals like patients and their relatives, health care providers, public bodies such as insurance companies, food producers and other industries in the field of PKU – everybody is invited to organise events or activities that appeal to the general public and raise awareness of PKU. A start was done in 2014: PKU picnics, press releases, concerts and individual activities followed by twitter and other social media have been organised to put PKU and the needs of patients into a public spotlight.

Annual Report 2014

The ESPKU travel passport

ESPKU Travel Passport

Based on the initiative of some adult PKU-patients at the ESPKU Adults Meeting in St. Triden (Belgium), a draft multilingual travel passport was elaborated, including basic information on seven inherited disorders of protein metabolism and the medicinal and nutritional products the patients have to have during travel. After the final layout, the document will be presented to the ESPKU member associations at the 2014 ESPKU Annual Conference in Zagreb. This ESPKU activity is supported by a grant of Vitaflo® International Ltd.

Meetings with European politicians

Participants of the ESPKU Adults Meeting 2013 in St. Triden (Belgium) visited the European Parliament in Brussels and made contact to MEP Angelika Werthmann from Austria. After being confronted with the stories of the life of 14 PKU-patients from 18 to 44 years of age, Angelika Werthmann offered to present the most important issues of PKU as an example for rare diseases to the EU parliament. As with regard to the European elections 2014 it was not possible to organise an adequate format for a parliamentary debate, this opportunity will be picked up in 2015 again.

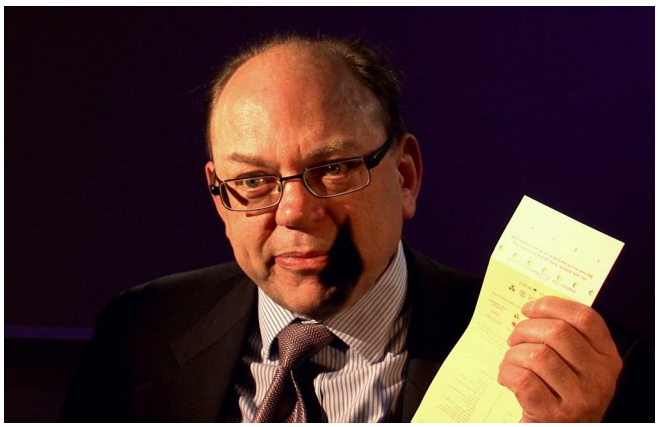
A look into history and future

2014 was a year of transition for ESPKU. Still working on the final step of the “Closing Gaps in Care” campaign we prepared our future policy at the same time. Like in the past, to achieve our future objectives, ESPKU will

- support its member associations on European level in their political activities
- network with member associations, professional healthcare providers, other associations and health politics
- stimulate scientific discussion and development of PKU care
- raise public awareness on PKU and allied disorders

Most important is to act as a community. In a filmed interview with the PKU Academy, ESPKU President Eric Lange said: “Since its inception in 1987 ESPKU has evolved over the years into a particular strong brand in terms of an umbrella body for patients organisations throughout Europe. We believe ESPKU can help its member association close their gaps in care, whereas our member associations can help ESPKU improve knowledge of PKU with policymakers and public. Together, there’s synergy: Two plus two equals five.” (The interview is available online at http://www.pkuacademy.org/en/views/perspectives/eric-lange.html?vid=:1402156192828.)

Implementing new comprehensive treatment guidelines advocating for reimbursement of all treatment aspects must include those who are untreated or late treated. Still, this group of patients tend to get marginalised, but yet can be helped to improve the quality of their life. Even if there is a small minority of consultants who believe that for some patients it is okay to come off diet at a certain age, because there is no evidence that coming off diet will impact these patients, we shall turn it around and ask them for the evidence that coming off diet won’t have any negative outcome.



ESPKU President Eric Lange

At present and in future there is a lot going on. When we prepare our activities to follow our policy plan, it seems to be quite difficult to consider all aspects and facets of health politics in general and PKU care in particular.

Eric Lange: “We all shall remember that most important things are simple. In this wonderful world of computers, mobile phones, tablet computers etc. we need to remember how one simple thing can prevent disability, and that’s a piece of blotting paper.”

E.S.PKU

Annual Report 2014

Annual Report 2014

Impressum

The Annual Report 2014 was presented to the delegates of the attending member associations of ESPKU at the 2014 General Meeting of ESPKU, held on October 25th, 2014 in Zagreb, Croatia. It provides insight into the main working fields of ESPKU from October 2013 until September 2014, 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 (ESPKU)
and Allied Disorders treated as Phenylketonuria

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

Address of correspondence:
Mr. Tobias S. Hagedorn (Secretary)
Wilhelm-Mellies-Strasse 10
D-32120 Hiddenhausen, Germany

www.espku.org

Annual Report 2014

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

Austria	**Hungary**	**Portugal**
Belgium	**Iceland**	**Romania**
Bulgaria	**Ireland**	**Slovakia**
Croatia	**Italy (2)**	**Slovenia**
Czech Republic	**Latvia**	**Spain (2)**
Denmark	**Lithuania**	**Sweden**
Estonia	**Moldova**	**Switzerland (2)**
France	**Serbia / Montenegro**	**The Netherlands**
Georgia	**Norway**	**Turkey**
Germany	**Poland (2)**	**United Kingdom**

Bank Account: **IBAN: BE66 7360 0196 7043** **BIC: KREDBEBB**