

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

Annual Report 2016

We understand the patient's needs best



**Change is the law of life,
and those who look only to the past or present are certain to miss the future.**

John F. Kennedy (1917 – 1963)

President of the United States of America 1961 - 1963

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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Review of the 2015 ESPKU Annual Conference in Berlin, Germany



475 Patients and their family members, delegates of ESPKU member associations, Scientists, Healthcare Providers and other professionals and industrial representatives from about 40 countries attended the 29th ESPKU Annual Conference from October 22nd until October 25th 2016 in Berlin, Germany. The German PKU Association DIG PKU organised this event as the final highlight of their 40th anniversary celebration year.

The overarching theme of the conference was „building bridges“. It was more than symbolic, that at the ESPKU

General Meeting, the delegates accepted the application for ESPKU membership of both, Israel and Palestine / The Gaza Strip.

The conference programme offered information and education on past and future of PKU treatment and connected scientists and patients from all over the world. The descendants of the Pioneers of PKU Følling, Bickel and Guthrie offered intimate insight in their fathers and grandfathers life and earned standing ovations for their presentations. A touching documentary edited by the hosting DIG PKU and adapted into English by NSPKU, reminded the fate of late diagnosed patients and the importance of screening, dietary treatment and amino acid mixtures for PKU patients.

Harvey Levy looked into his crystal ball and talked about future treatment options for PKU and allied disorders. Francjan van Spronsen and Annemiek van Wegberg presented the key statements of the first European Guidelines for PKU, which have just been finalized by an independent group of Experts two days before.

In her presentation during the conference dinner, ESPKU and DIG PKU honorary member Dr. Ursula Wachtel remembered the past decades of PKU and reminded the audience that only dedicated and devoted hard work will help closing gaps in care and improving the lives of future patient generations.

Having heard this message, guests and organisers celebrated the future of PKU, dancing their feet off in an outstanding conference party.



The 2016 ESPKU Member's Delegates Meeting (Spring Meeting)

About 15 delegates from only 7 countries met at the Clayton Hotel in Dublin (Ireland) from April 15th until April 17th, to exchange information on their current agendas and initiatives and to discuss latest developments.

One of the topics on the agenda was the implementation of Allied Disorders in the ESPKU policy. Some ESPKU member organisations are purely for PKU, others include PKU and allied disorders in their membership and offer related programmes and activities. A few ESPKU members are Rare Disease Organisations. Due to these different structures of our national and regional member organisations it was and still is difficult to integrate programmes and activities dedicated to other allied disorders in the ESPKU policy.

However, since „Allied disorders treated like Phenylketonuria“ are included in the Associations name, the attending delegates agreed that ESPKU shall at least offer membership to other rare disease organisations and organisations for allied disorders of protein metabolism, because they potentially will benefit from any ESPKU achievements for PKU patients. Furthermore, future ESPKU Conferences shall include information and presentations on allied disorders, just like the presentation on MSUD in the patients programme at the 2015 ESPKU Annual Conference in Berlin.

Other topics on the Spring Meeting Agenda, such as European Reference Network for Metabolic Disorders, International PKU Day, the European Guidelines on PKU and the 2016 ESPKU Annual Conference will be discussed in other chapters of this ESPKU Annual Report.

The 2016 ESPKU Annual Conference in Dublin, Ireland



Primarily the 30th ESPKU Annual Conference was intended to take place in Istanbul, Turkey. There was no doubt that the Turkish PKU Foundation with Deniz Atakay and her team would organise an outstanding and unforgettable Conference and be good hosts for all guests. However, the political developments in Turkey and the region had an impact on security issues and the ESPKU could not ignore this.

For that reason only, the ESPKU Executive Board was asked to find an alternative location. Encouragingly there were a number of countries interested, but the Board chose Dublin, Ireland. Asked by the ESPKU Executive Board, the PKU

Association of Ireland did not hesitate to stand in and accepted the challenge to host this event at the Clayton Hotel in Dublin. Since there was less than one year to organise this event, the ESPKU offered massive support to the Irish PKU Association. The experiences with the organisation of the ESPKU Conferences 2015 and 2016 will help to rewrite the guidelines and recommendations for future conference hosting ESPKU members.

The 2016 Conference Programme will cover a wide range of topics, e.g. Homocystinuria, late diagnosed PKU, maternal PKU, the parents perspective, the European Guidelines, elderly patients with PKU, Pathophysiology and many more.

At the moment when this ESPKU Annual Report was edited, the conference was sold out without a disclosed programme.



International PKU Day 2016



International ESPKU Day is an initiative of ESPKU. Since 2013, 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. In 2016, a new website was opened for International PKU Day: www.pkuday.org.



For International PKU Day 2016, ESPKU organised the Robert Guthrie Memorial Dinner: To identify PKU babies affected by Phenylketonuria immediately after birth, Robert Guthrie invented the possibility for newborn screening, also known as the "Guthrie test". Uncounted patients owe him the chance of early treatment and a normal life without the devastating symptoms of untreated PKU. On June 28th, 2016 was the 100th anniversary of Robert Guthrie's birth. On this occasion, ESPKU invited people from all

over the world to commemorate him and his lifetime achievements. No matter if dining alone at home or out with family, friends, colleagues or teammates: People from around the world dedicated their dinner on June 28th to Robert Guthrie and his work, and uploaded a photo to a picture wall at www.pkuday.org.

Also at International PKU Day 2016, a touching documentary by the German Association for Phenylketonuria and allied inherited disorders (DIG PKU) calling attention to the fate of late diagnosed and untreated patients was published online by ESPKU. This film is available in German, English and German. Up to the date of this ESPKU Annual Report, more than 6000 people watched this film online.

ESP KU Executive Board Meetings: Key Results

The Executive Board met four times in 2016 and discussed different issues regarding ESPKU management, future ESPKU direction, current ESPKU activities and other areas.

The first meeting was attended by the entire board of the ESPKU Scientific Advisory Committee. The meeting focussed on corporate governance issues and topics related to science and treatment of PKU, such as the European Guidelines, European Reference Networks and others. A patients' questionnaire on potential health problems related or not related to PKU was appointed to be conducted as soon as possible.

Another board meeting happened without any SAC participation, and allowed the Executive Board Members to brainstorm on the future policy of ESPKU and to discuss solutions for administrative challenges. As an overarching statement of the future ESPKU agenda, the Executive Board believes that all patients in Europe should have free access to all available amino acid supplements, low protein food and pharmaceutical products necessary for their treatment for life.

To make ESPKU fit for future, the Executive Board considered the organisational structure of ESPKU, the regular operations that need to be executed. There are projects on the way, like the European PKU Guidelines, the International PKU Day, and the ESPKU Conference Organisation. We have to build alliances with other continental patients associations and to re-focus on Europe and patients advocacy together with our members.

At the 2016 European Conference on Rare Diseases and the EURORDIS Membership Meeting in Edinburgh the ESPKU was represented by their President Eric Lange. European Reference Networks was one of the focus topic of the Eurordis Membership Meeting.

European Reference Network for Metabolic Diseases (METAB-ERN)



European
Reference
Networks

The establishment of European Reference Networks (ERN) is based on the EU Directive 2011/24/EU on patients' rights in cross-border healthcare. ERNs are aimed to help professionals and centres of expertise in different countries to share knowledge. They should apply EU criteria to tackle rare diseases requiring specialised care, serve as research and knowledge centres treating patients from other EU countries and ensure the availability of treatment facilities where

necessary. One of these ERN's called METAB-ERN will focus on hereditary metabolic diseases and including PKU and related disorders. In summer 2016, a proposal aimed to establish the METAB-ERN was been submitted to the European Commission, including 69 healthcare providers from 18 EU countries.

Initially there was no provision made for patients' association involvement in ERN's. However, EURORDIS initiated Patient Advocacy Groups (EPAG) to participate in the ERN decision-making process. Early in 2016, we supported the election of Lut de Baere from the Belgish ESPKU member organisation BOKS as EPAG Patient Representative for metabolic diseases.

ESPKU and its member associations have addressed the issue of European Reference Networks being EU centric: Healthcare providers from non-EU-countries have no access to ERN's. Since ESPKU is not EU-centric, this was one of the key reasons why ESPKU refrained from putting forward its own candidate as an EPAG Patient Representative.

In their discussion during the „ESPKU Spring Meeting 2016“, the attending member's delegates were sceptical with regard to the efficiency of ERN's, since the administrative overhead looked very complex.

ESPKU generally welcomes the establishment of ERN's, especially the METAB-ERN, and supports their aims, and offered close cooperation to Maurizio Scarpa, the coordinator of METAB-ERN and of course to Lut de Baere, the EPAG Representative for the METAB-ERN. However, we address the issues of efficiency and restrictions for non-EU-countries.

European Guidelines on diagnosis and management of Phenylketonuria: The current status

Initiated by ESPKU, the guidelines have been finalised by the guideline group of dedicated professional experts, independently from patients and industries. During the recent month a paper on the key statements and submitted to the Lancet Diabetes & Endocrinology Journal, a highly valued scientific journal, who recently accepted it for publication.

The Lancet operates a strict embargo policy. Therefore we are not able to publish any part of manuscript before it is published online.

When this paper is published, we can then go ahead and publish the full Guidelines. ESPKU and Member Associations can go ahead to implement the guidelines.

We have anticipated translation into several languages so that patient organisations of all European countries can see with their professionals what the hurdles are to implement these guidelines. To this end, an ESPKU working group currently has identified suitable professional agencies.

A look into future

The ESPKU Executive Board recognised the challenges ahead and has looked into the issue of finance and administration support. This is somewhat complicated as we are registered in Belgium and have to obey Belgium charity Law. At the time of writing we have identified 3 providers who can meet our needs for the future.

We are increasing our contacts worldwide and we have recognised for a while now, the need to unite PKU Organisations from around the world. PKU needs a politically strong worldwide voice to be able to e.g. ensure research is done with patients in mind and to avoid duplication of scientific research, to influence healthcare and health insurance policies and to produce unified guidelines.

As a result discussions have and are being held to progress the above. There may be an announcement at the Conference.

We understand the patient's needs best. That's the difference between us and other organisations.

Impressum

The Annual Report 2016 was presented to the delegates of the attending member associations of ESPKU at the 2016 General Meeting of ESPKU, held on November 4th, 2016 in Dublin, Ireland. It provides insight into the main working fields of ESPKU from October 2015 until October 2016, 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 (ESP KU)
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	Germany	Portugal
Belarus	Hungary	Romania
Belgium	Iceland	Serbia / Montenegro
Bulgaria	Ireland	Slovakia
Croatia	Israel	Slovenia
Czech Republic	Italy	Spain (2)
Denmark	Latvia	Sweden
Estonia	Lithuania	Switzerland (2)
France	Moldova	Turkey
Palestine / Gaza Strip	Norway	Ukraine
Georgia	Poland (2)	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 Ahning, Denmark

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