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

Annual Report 2018

United for a better future

The future depends on what you do today.

Mahatma Ghandi

Words of Gratitude

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

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Preface

Our PKU world is changing fast, but turning in different speeds: While there are still too many patients without access to basic care, the course for new developments of future therapies is set surprisingly fast. Too many are fighting for the chance of a timely diagnosis through neonatal screening, while others are pursuing new therapies through modern genetic medicine. None of this is wrong or unethical, and everything deserves recognition and support. However, we must recognize that the birth-dependent gaps between good, adequate and inadequate care become deeper and more difficult to overcome.

The ESPKU has devoted itself to the task of uniting patients and advocacy groups across Europe and around the world and to build a strong and supporting network to strengthen patients' voices and efforts to close these gaps. This annual report provides a non-exhaustive overview of some of the key activities of the ESPKU in the last 12 months.

ESPKU Conferences and Executive Board Meetings

The ESPKU Annual Conference 2017 took place in Hell, close to the city of Trondheim in Norway, the homeland of Prof. Asbjørn Følling. For the second time in the history of ESPKU, the hosting Norwegian Society for Phenylketonuria pulled together an outstanding event for the about 450 attending patients and families, patients' advocates and delegates, researchers and healthcare providers, industrial representatives and guests from Europe and beyond.

The conference program covered a wide range of different topics: The practical experiences of the patients were reflected as well as current scientific and political questions. Because of its relatively high incidence in Norway, the scientific and common programme also looked at Tyrosinemia Type 1 and its similarities and differences to PKU.

At the Annual General Meeting, the delegates of ESPKU members re-elected the ESPKU Executive Board for another three years term in office.

Only a few months later, ESPKU members' delegates gathered again to their spring meeting in Venice (Italy). The agenda covered three main topics:

- Kate Hall, council member of the ISNS, gave a lecture on challenges and opportunities of neonatal screening and helped us mapping and understanding the current situation in Europe.
- Claudia Rubin, Director of Decideum Ltd., focussed on the increased opportunities for patients associations to influence health politics and successfully advocate for access to a better quality of care.
- Kirsten Ahring, Secretary of the ESPKU Scientific Advisory Committee, discussed how the European PKU Guidelines have been implemented across Europe, and introduced the idea of a "cook book" to provide answers on how to use them in daily life.

After the conference in Hell, the Executive Board and Scientific Advisory Board gathered for another meeting in early 2018. In this meeting as well as in a number of regular skype conferences, current topics as well as the basic politics of the ESPKU were discussed, including the preparation of the first update of the European PKU Guidelines. To look at all topics from different perspectives and to meet the multifaceted tasks of ESPKU more efficiently, the Board has actively involved some volunteers from ESPKU member associations in its work.

Prof. Anita MacDonald appointed as fourth member of the ESPKU Scientific Advisory Board

We proudly announce: On October 6th, 2017 during a Board meeting in Hell, Professor Anita MacDonald was appointed as member of the Scientific Advisory Board of ESPKU.

Professor MacDonald has conducted over 40 studies and published 90 articles and research papers on PKU. She chairs the European Nutritionist Expert Panel in PKU (ENEP) and networks with PKU researchers around the world. Her commitment to PKU patients goes well beyond a purely professional interest, and is much more of a passion. Together with Professor Francjan van Spronsen, Professor Maria Gizewska and Kirsten Ahring, she supports the work of the ESPKU with valuable advice.

Networking with ESPKU member associations and other Patient Groups

Over the past 12 months, ESPKU has paid close attention to promoting PKU within and beyond Europe, and to presenting ESPKU as a strong partner in the global patient network. This included active participation in many events inside and outside the ESPKU and Europe.

The 2017 BioMarin European Patient Advocacy Forum in November was headlined “Advocacy Powered through Data Generation”. Participants from nine European countries attended the meeting in Amsterdam, including Deniz Atakay from the Turkish PKU Family Foundation. There was broad agreement on the value of patient focussed data in health technology assessment (HTA), as well as for approval and access decisions. However, there was disagreement as to whether establishing and maintaining patient databases is a task of patient advocacy groups, clinical research, or other independent institutions. ESPKU President Eric Lange, ESPKU Secretary Tobias S. Hagedorn and ESPKU past President David Abeln presented the ESPKU Closing Gaps in Care Programme. They explained the contribution of benchmarking within the European healthcare economy to the development of new evidence-based treatment guidelines and thus to a potential improvement in the patients outcome.

EURORDIS is the largest umbrella organisation that unites more than 800 Rare Disease Patient Organisations from 70 countries. This years’ European Conference on Rare Diseases & Orphan Products took place in May in Vienna and was entitled “Rare Diseases 360° - Collaborative Strategies to leave no one behind”. The agenda covered six different thematic areas on rare diseases, including quality of life, economical perspectives and structuring the research landscape. The ESPKU was represented by President Eric Lange, who enjoyed the exchange of views and experiences with some of the 900 participants.

Patients are taking on ever increasing roles in advocating for their unmet needs. The EURORDIS Summer School is an empowering training programme for patients and researchers to sharpen their advocacy skills and gain an understanding of the regulatory process of therapeutic development. In June 2018, ESPKU President Eric Lange, Florentina Peric from the Croatian PKU Society and Agata Bak from Metabolicos (Spain) joined the Summer School in Barcelona and made contact with approximately 50 attending patient advocates representing different diseases and countries.

In July 2018, ESPKU President Eric Lange and ESPKU Secretary Tobias S. Hagedorn attended the US National PKU Alliance Conference and collected first-hand information about the current state of research for new treatments options. At this occasion, they exchanged views with patient representatives and affected families from the US and other countries of the world.

In September 2018, ESPKU President Eric Lange visited the conference of Metabolicos Spain. In his welcome address during the opening session, he provided information on current European and global topics and encouraged the Spanish ESPKU member organisation to apply for hosting a future ESPKU Conference. With its linguistic proximity, the Spanish member association can be an important transatlantic connection to the South- and Central-American PKU community.

As guest speaker of a meeting of the Russian Association for Rare Diseases, ESPKU President Eric Lange spoke about the legal aspects of providing medical care to patients with rare diseases such as PKU, and about the importance of involving patients' organisations in medical research. He discussed potential collaboration on PKU issues with the Russian Rare Disease Organisation and took the opportunity to strengthen the ESPKU's relations with other attendees from Eastern European countries.

ESPKU launched Sheila Jones Award for voluntary patients' representatives

In 2018, the ESPKU launched the Sheila Jones Award to recognise Sheila Jones and her family's contribution to the PKU community. After numbers of scientific or industrial awards, this award is the first ever distinction particular for patient advocates.

The award is named after Sheila Jones, the first ever successfully dietary treated Patient with Phenylketonuria. In 1951 - 1954, her mother insistently urged Professor Bickel at the Birmingham Children's Hospital to develop a therapy for retarded Sheila. Since then, her persistence made the significant difference between a life with disabilities and a normal life for countless patients.

This award should not only remember the legacy of Sheila Jones, it should be a public recognition for outstanding projects or services of voluntary patient representatives, whose work is often not sufficiently appreciated. It should motivate individuals to volunteer in their patients association. It will prove: With patience, dedication and perseverance, patients can make a difference.

Every year, a jury at Birmingham Children's Hospital, where Sheila was treated, reviews all applications and proposals and selects a worthy award winner. ESPKU received 15 outstanding proposals for the first Sheila Jones Award. The winner will be announced and receive a key-like sculpture in a solemn ceremony during the ESPKU annual meeting 2018.

Joint patients' advocacy can make a difference

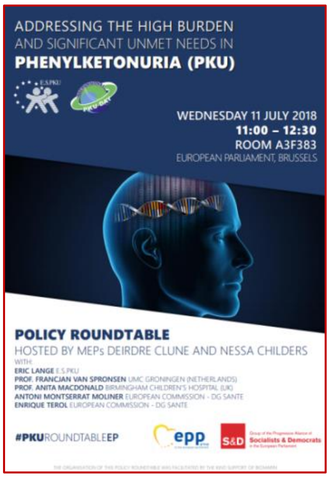
Joint efforts of the ESPKU and its member association "Aproteica" have a potential lasting impact on the reorganization of PKU care in Romania.

In January 2018, our Romanian member association Aproteica informed the ESPKU on the imminent closing of the Screening Centre in Cluj-Napoka. The screening of about 30.000 newborns per year and the follow up of some hundred PKU patients were at stake. As an immediate response, the ESPKU wrote a letter of concern to the Romanian Health Ministry. In a television interview, ESPKU President Eric Lange expressed his dismay at the situation and stressed the importance of neonatal screening as an inalienable human right. Additionally, the ESPKU requested advice and support from the International Society for Neonatal Screening (ISNS). All this has been closely aligned with the activities carried out by Aproteica in Romania.

As a result of joint efforts and national and international public pressure generated, the Romanian Ministry of Health has not only promised to ensure screening will continue for all newborns, but also will look at a better model for the treatment and follow up for all PKU patients in Romania. On request of the Romanian Health Ministry the ESPKU has provided further advice on how to reorganize and secure the care of patients with PKU and allied disorders. The ESPKU will continue the cooperation with Aproteica and ISNS and constructively support their activities.

Political Advocacy for PKU on EU level

On July 11th, 2018 ESPKU organised a roundtable in the European Parliament to address the high burden and significant unmet needs in the management of Phenylketonuria. The meeting was hosted by MEP's Deirdre Clune (EPP, IE) and Nessa Childers (S&D, IE), and generated concrete proposals for action and collaboration.



ADDRESSING THE HIGH BURDEN
AND SIGNIFICANT UNMET NEEDS IN
PHENYLKETONURIA (PKU)

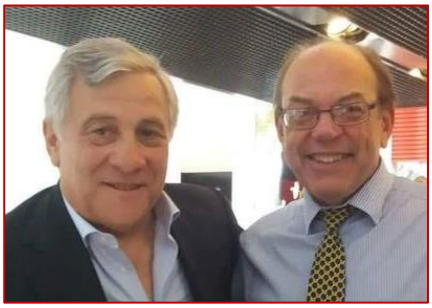
WEDNESDAY 11 JULY 2018
11:00 – 12:30
ROOM A3F383
EUROPEAN PARLIAMENT, BRUSSELS

POLICY ROUNDTABLE
HOSTED BY MEPs DEIRDRE CLUNE AND NESSA CHILDERS
WITH:
ERIC LANGE E.S.PKU
PROF. FRANCJAN VAN SPRONSEN UMC GRONINGEN (NETHERLANDS)
PROF. ANITA MACDONALD BIRMINGHAM CHILDREN'S HOSPITAL (UK)
ANTONI MONTSERRAT MOLINER EUROPEAN COMMISSION - DG SANTE
ENRIQUE TEROL EUROPEAN COMMISSION - DG SANTE

#PKUROUNDTABLE**EP**

In his welcome address, ESPKU President Eric Lange called for audits to ensure that every baby born in Europe is screened for PKU. ESPKU Scientific Advisor Prof. Francjan van Spronsen complained that there is no dedicated body or institution to endorse international treatment guidelines. He called on the EU Parliament and EU Commission to take this up and invited the Reference Network for Metabolic Disorders (MetabERN) to play a role. ESPKU Scientific Advisor Prof. Anita MacDonald underlined the need to harmonise the nutritional composition of FSMP's for PKU, to improve protein labelling and to conduct more work on aspartame labelling. Agata Bak (Spanish ESPKU Member FEEMH) shared her personal experiences as a PKU patient: Being early diagnosed, she can pursue her life goals. However, too many patients still do not have access to low protein food. She explained her concerns about the future and called for a more holistic approach to improve PKU care.

In the Panel Response, Antoni Montesrrat Moliner (EU Commission, DG Sanco) reminded that despite the 2009 EU Council Recommendation on Rare Diseases, there is no European framework for newborn screening in place. Enrique Terol (EU Commisson, DG Santé) explained that healthcare systems remain first and foremost the competence of the Member States. However, he agreed that MetabERN could well be in a position to endorse guidelines as the gold treatment standard. Kate Hall (International Society for Neonatal Screening ISNS) told the audience about a recent visit to Romania. She expressed her shock upon meeting young PKU patients born in the 21st century who are brain damaged due to late diagnosis. She reminded that neonatal screening is not available in several European Countries and has been introduced slowly and imperfectly in others. Prof. Maurizio Scarpa (MetabERN Coordinator) announced his willingness to start a close collaboration with ESPKU.



After the roundtable, Eric Lange spontaneously took the opportunity to introduce Phenylketonuria and the ESPKU to the President of the European Parliament Antonio Tajani in a very short elevator pitch.

Following the success of the meeting, the ESPKU is considering the possibility of initiating an all-party parliamentary group to look after the concerns of PKU patients in Europe.

Promoting universal Neonatal Screening

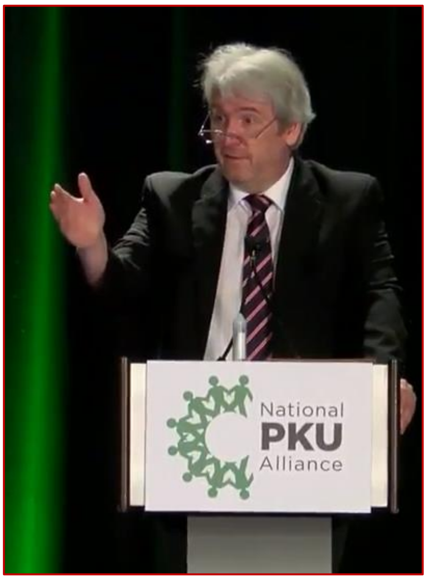
Newborn screening goes back to PKU and still is and will be the most fundamental basis of PKU treatment for today and in future. Once developed by Robert Guthrie to identify babies with PKU before they show symptoms of the disease, today neonatal screening helps to ensure protection of children from many life-quality threatening conditions beside PKU.

However, because its implementation across Europe took more than 40 years and has not been finished yet, there are significant inconsistencies regarding experience, extent and access to newborn screening. The ESPKU has initiated close collaboration with the International Society for Neonatal Screening (ISNS) to provide greater support for implementation of universal newborn screening.

Kate Hall, member of the Council of ISNS, supported an ESPKU video statement to call politicians and health care providers to make available, implement and maintain universal neonatal screening for all newborns around the world. This video was recorded at the ESPKU 2018 Spring Meeting for Members' Delegates. It was published on our facebook account and website on International PKU Day (June 28, 2018), and shared by many ESPKU members and others. https://youtu.be/VC8IttbzUW8

In the past twenty years, many attempts to develop a home monitoring kit have been unsuccessful. In 2018, the ESPKU and its scientific advisory board have both promoted two promising projects with supportive letters. We called on the involved companies to not only develop a reliable tool for improving the self-management of treated patients, but to laser-focus on making neonatal screening accessible to newborns in regions with inadequate medical infrastructure, where it is not available yet.

Global Association for Phenylketonuria unveiled



As already reported in 2017, during the 2016 Annual Conference in Dublin, ESPKU took the initiative to encourage the foundation of a global community of national and regional PKU associations.

After a first founders meeting in summer 2017 in Toronto, the ESPKU hosted another conference in Amsterdam in February 2018. At this meeting the Toronto agreements were converted into a constitution. The founding documents of the Global Association for Phenylketonuria (GAP) were solemnly signed by the cofounders from Europe, Australia, North-, Middle and South-America. Further required regulations were agreed at a final founders meeting in July 2018 in Atlanta (USA).

On July 5th, 2018 ESPKU Secretary Tobias S. Hagedorn proclaimed the foundation of GAP at the occasion of the NPKUA Conference in Atlanta, USA. In his address, he emphasized the responsibility of the international patients' community to advance PKU a global health priority and to help closing the gaps in PKU care worldwide. (See video with his complete speech at https://youtu.be/HtWCLxM2g3U)

GAP has applied for registration as Charitable Incorporated Organisation in England and is waiting for approval from the Charity Commission. ESPKU President Eric Lange and ESPKU Secretary Tobias Hagedorn are members of the provisional executive committee of GAP. Deniz Atakay from the Turkish PKU Family Foundation has been appointed as trustee for the Middle East Section of GAP.

Summary and outlook to future

With its targeted advocacy work for all PKU and allied disorder patients in Europe, ESPKU has received increasing attention amongst all stakeholders in recent years, not only within Europe, but globally. Thanks to this attention, the ESPKU can make a difference and achieve real improvements for patients.

In the future, it will be the duty of the ESPKU to prudently use these opportunities and to support the diverse needs of the patient community without leaving anyone behind. It will be the ESPKU's task to constantly strengthen and nurture the network of local and national patient advocacy groups. And it will be the responsibility of the ESPKU to equally involve all stakeholders in this community and to foster their cooperation.

Annual Report 2018

Imprint

The ESPKU Annual Report 2018 was presented to the delegates of the attending member associations at the 2018 ESPKU General Meeting, held on November 2nd, 2018 in Venice, Italy.

It provides insight into the main working fields of ESPKU from November 2017 until September 2018, 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 Phenylektonuria (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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E.S.PKU

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**
- **Macedonia**
- **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

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

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