



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

Annual Report 2013

Setting the future direction of PKU care





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**The world stands aside to let anyone pass
who knows where he is going.**

David Starr Jordan

US biologist, educator, & ichthyologist (1851 - 1931)
1st president of Stanford University

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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New Members

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Impressum

French Les Feux Follets rejoins ESPKU after 7 years of absence.

According to the ESPKU Statutes, the General Meeting 2013 has to decide on the applications for legal membership of France and Galicia.

Les Feux Follets is the «Association Nationale de Parents d'enfants atteints de Maladies Metaboliques Hereditaires" (the national association of parents whose children are affected by a inherited metabolic disorder) from France. It has already been member of ESPKU from 1996 until 2006.

Its foundation was declared to the Journal Officiel de la Republique Francaise on June 26, 1995. In these days the association was regionally based, but within one year it became recognised as national association. In the 1990th, it was Les Feux Follets who initiated the European PKU Day taking place annually on June 28th.

The contact between Les Feux Follets and ESPKU remained even after their first membership. ESPKU is happy to welcome back this important association as legal member.

New membership application from ASFEFA (Galicia)

The second application for ESPKU membership is from Fenilcetonúrica and OTM Association of Galicia (ASFEFA). ASFEFA is one of fifteen regional associations that make up the Spanish Federation of Inherited Metabolic Disorders. Founded in 1990, ASFEFA today counts 280 members.

Besides being very active on the political level in Galicia and Spain, they also offer their members a daily life support. As an example, they run a local office and warehouse property for special foods that functions as a distribution center for Galicia.

Meetings and Conferences

General Meeting 2012 took place in Liverpool, United Kingdom

The General Meeting 2012 took place in Liverpool on October 19th, 2012. At this formal occasion, the attending delegates of ESPKU member association received the latest information on the ESPKU activities, such as the ESPKU guidelines on PKU management and the ESPKU consensus paper. The supplementing financial data proved accurate accounting and firm financial basis of ESPKU. The executive board members have been discharged.



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learn about the 10 steps to build and implement a successful engagement campaign.

For the third time, the ESPKU Spring Meeting for delegates took place on March 23, 2013. The venue was the Radisson Blu Astrid Hotel in Antwerp, Belgium. ESPKU member's delegates and Executive Board members from 11 countries exchanged information on actual topics in a very productive and informal atmosphere. The discussions have been valuable also to future ESPKU politics and projects.

The development of social support for PKU patients in Europe was one key topic of the meeting. Still, there are remarkable inconsistencies in social support for PKU and rare disease patients across Europe. And still the ESPKU member organisations have to put much effort in defending the status quo from any impact of the socio-economic change. In some specific areas, their advocacy campaigns have been already successful, and in some cases they could even reach an improvement of some details in social support for PKU and RD patients.

To support a more dedicated and targeted advocacy, ESPKU members also considered a European or even international PKU day. Compared to the existing rare disease day, it would focus on the demands and needs of PKU patients, instead of all rare diseases in general.

As a new project, the delegates considered to produce some video clips to address major PKU issues, such as benefit of treatment, newborn screening and nutritional treatment. These clips shall serve as a supportive tool for political lobbying and raise awareness on PKU.

During two intense Executive Board Meetings in Liverpool and Antwerp, the ESPKU board members and representatives of its scientific committee coordinated their work and action and appointed further strategies to achieve the goals of ESPKU.

Spring meeting highlights need for social support

International PKU day considered to focus on the needs of PKU patients

Video clips to address major PKU issues appointed

ESPku Consensus Paper

In 2011, the delegates of ESPKU members agreed on the most important cornerstones of optimal PKU care from patient's perspective, and appointed to publish an ESPKU consensus paper on

ESPKU consensus paper submitted for publication in Orphanet Journal of Rare Diseases

results of the working group have been presented to the ESPKU delegates for further discussion.

After having finalized the draft of an ESPKU consensus paper in spring 2013, the authors have submitted the final paper for a peer reviewed publication to the Orphanet Journal of Rare Diseases, which is one of the most important journals in this field. The final title of the manuscript is "Requirements for a minimum standard of care for phenylketonuria: The patient's perspective".

In October 2013, the publication was awaiting the decision of the editor or OJRD. The peer reviewers testified the manuscript being "an article of outstanding merit, importance and interest in its field".

ESPKU European Guidelines on the management of Phenylketonuria

ESPKU launched working group to develop international guidelines for PKU treatment

After 2012 was a significant milestone for ESPKU with the launch of the first PKU benchmark report *Closing the Gaps in Care* at the European Parliament in Brussels on Rare Disease Day, ESPKU built on that momentum through the launch of a new working group dedicated to developing guidelines for the optimal treatment of PKU.

"We need to ensure that PKU patients get an equal chance of living a healthy and productive life, no matter where they live. We cannot accept that some people with PKU are still being left untreated in Europe. The publication of the E.S.PKU consensus paper will contribute to addressing that," said Dr Francjan J van Spronsen, chair of the Scientific Advisory Committee of the E.S.PKU, at the 2012 Annual ESPKU Conference in Liverpool. "The consensus paper asks us as professionals in PKU care to develop European or even world-wide guidelines on the treatment of PKU for every age throughout life."

The expert group consists of 16 well established PKU researchers, who are able to step down from their national politics and therefore can reach for guidelines on many PKU issues on European level. They aim to present the first international PKU guidelines within the next 2 years. In their initial meetings, they have already agreed in the key chapters and formed dedicated small working groups on

- Neurocognitive outcome including imaging
- Psychosocial outcome and adherence
- Nutritional treatment and biochemical / nutritional follow up



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

Political Activities

On behalf of ESPKU, our President travelled to Brussels two times and met with different policy makers on the European level. Most talks were on European Reference Networks (ERN) and the European Union Committee of Experts on Rare Diseases (EUCERD).

In a meeting with Antoni Montserrat from EUCERD, Mr Montserrat explained that there is no controversy with regard to PKU. PKU is included in all Rare Disease Plans that have been submitted to date, and likely to be in one of the earlier European Reference Networks. He appreciated that ESPKU has responded on a questionnaire on European Reference Networks.

Another Meeting with Enrique Terol from the European Commission was the occasion to gauge what the Commission had in mind with regard to the ERNs following the recent publication of a public consultation on setting up criteria ERNs will have to fulfil. He highlighted the fact that the Commission had limited powers. Indeed, organisation of health systems is a national competence in the treaties and the Commission can only play a limited role, usually fostering cooperation between Member States.

Davide Lecchini, the Italian health attache, mentioned Italy is currently identifying potential priorities for their presidency of the European Council in 2014, and patient organisations should advocate to put rare diseases on the agenda. This is particularly true as 2013 will be marked by the implementation report on rare diseases and further strategies on the topic should be fostered in coming years.

On European Reference Networks he explained that the recently launched public consultation aims at gathering stakeholders' input on what Networks should be, their functionality, governance systems, as well as agreeing on general criteria common to all Networks and specific criteria depending on the field of expertise. The Commission will work with the Cross-Border Directive expert group to reach a common understanding of what constitutes an ERN and its organisation. Knowing about the Challenges of inequality across

**Meetings with
several European
policy makers on
European Reference
Networks (ERN) and
the European Union
Committee of
Experts on Rare
Diseases (EUCERD)**

ESPKU applies for membership in EUCERD!

EUCERD to play a key role in the setting up of the ERNs, he noted that EUCERD produced a recommendation on criteria for centres of expertise and is currently working on a Recommendation on ERNs for rare diseases so as to provide concrete guidelines for specific diseases.

In response to the call for new members of the EUCERD, ESPKU recently expressed interest to be appointed as a member representing patient's organisations in the field of rare diseases. At the date of editing this annual report, we were awaiting an answer on our application.

On another occasion, our President met with Amelia Areias, Assistant to Maria Da Graça Carvalho Portuguese MEP and discussed rare diseases.

With Director Generale for Employment Arnaud Senn (policy officer in charge of active ageing, health and long-term care) he discussed accessibility people have with PKU – both to treatment and products.

Our President also had a teleconference with Maurizio Scarpa from Brains for Brain and attended an initial meeting on "The Prospects of Brain Research within Horizon 2020: Responding efficiently to Europe's societal needs". He made contacts with the key players and explained that the older generation of those on the PKU diet in reaching their '50's will need to be monitored for research purposes

Finally, he met the Greek Health Attache Antonias Lanaras who was very interested in PKU. He was most concerned that Greece is not a member of the ESPKU. He has provided a contact of a metabolic consultant in Athens to encourage the setting up of a Greek Society.

ESPKU will remain addressing PKU issues at EU level!

ESPKU will remain addressing PKU issues at the EU level, to raise the attention amongst European health politics. Since we started with these talks, we recognise an increasing interest on our positions.

Participation in other conferences and meetings

ESPKU's competence is recognised among professionals

ESPKU's competence is also recognised among professionals. We received invitations to present our messages at different conferences.

At the Serona Symposium 3rd Dietitians workshop in Istanbul, ESPKU highlighted the fact that although much research is being done into



He also was invited to speak at the 2nd Balkan Rare Disease Conference in Sofia. There he made several contacts within the Balkan Community, in particular Macedonia where there is no NBS screening whatsoever.

The ESPKU attended the International Congress of Inborn Errors of Metabolism (ICIM) Conference in Barcelona with the National PKU Alliance (NPKUA) of America and the Canadian PKU Society CanPKU. This was an opportunity to meet with several players in industry. The discussions with the NPKUA & CanPKU revolved around an international PKU day and a world PKU conference.

Summary

Due to the variety of cultural and political traditions across Europe the work of the national PKU associations is essential. The meaning of self help has increased in the first decade of this century: The self care associations have developed from a pure "get-together of patients" to an important stakeholder in the field of health politics.

ESPKU plays this role by consequently following its long-term programme "Closing Gaps in Care", to minimize inconsistencies in treatment and access to treatment of a rare disorder of protein metabolism. With the publication of a consensus paper on the requirements of a minimum standard of PKU care from a patient's perspective, and the work on the first ever international guidelines on the management of PKU, we will set a new benchmark for health care and health politics across Europe.

To support the results of the ESPKU activities, we use the opportunity to get in touch with several policymakers on EU-level and to inform them on the need of political action. On the same importance are our contacts to the scientific world, where we also present our demands and call-to-actions.

All these activities are highly depending on the functioning of ESPKU as a strong community its member associations. Without the ongoing efforts within each single country, all European activities will fail at the end. Therefore ESPKU offers support and education for political lobbying, where needed. And we recognise that more and more of our member associations remarkably increase their political activities.

Self help associations have developed from a pure „get-together of patients“ to an important stakeholder in the field of health politics.

Without efforts within each single country, all European activities will fail!

countries recognise the importance and value of our work and become new members of ESPKU. In return, this will raise the importance and meaning of our common work all across Europe.

A look into future

With the publication of the European guidelines on the management of Phenylketonuria, the recent policy of ESPKU will reach an important stage: We are setting the future directions of PKU care across Europe.

There will be no time to rest on our laurels.

Nevertheless, there will be no time to rest on our laurels. We need to define strategies to implement the results of our recent work into practise in treatment, health politics and social support. Furthermore, it is time to appoint new goals for future, building on what we have achieved so far.

The majority of PKU patients in Europe are left untreated!

Still, the majority of PKU patients in Europe is born before screening, and therefor never had any access to treatment. Untreated and late treated PKU is tended to be sidelined in our society. When once a minimum standard of care is achieved for PKU patients across borders in Europe, it should not be exclusively available to those lucky ones that have been identified as PKU patients at birth. Bringing back late- and untreated PKU into the spotlights of our activities will also proof the benefits of neonatal screening and dietary treatment.

Another issue coming up at the horizon is PKU and maturity.

Another issue coming up at the horizon is PKU and maturity. How will PKU impact the life of a senior? What are the specific demands of an aging society with regard to rare diseases such as PKU and allied disorders? In time we should be prepared to present questions, answers and strategies to the public. We owe this to the first generation of PKU patients, who already have been the guinea pigs for PKU and allied disorders in the first half of their lifes.

A project may be completed, but the topic remains: Access to treatment for all patients. ESPKU is ready to act.



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The Annual Report 2013 was presented to the delegates of the attending member associations of ESPKU at the 2013 General Meeting of ESPKU, held on November 1st, 2013 in Antwerp, Belgium.

It provides insight into the main working fields of ESPKU from November 2012 until October 2013., without laying claim to completeness.

This Annual Report 2013 is published at **www.espku.org**.

On behalf of the ESPKU Executive Board

Tobias S. Hagedorn, Secretary

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

is 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

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)

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