

**DEVELOPING A CALL TO ACTION
ON UNMET NEEDS IN
PHENYLKETONURIA (PKU)**



**ONLINE MEETING
OF THE EUROPEAN PARLIAMENT
CROSS-PARTY ALLIANCE ON PKU**

HOSTED BY MEP TOMAS ZDECHOVSKY

WEDNESDAY 8 DECEMBER 2020

EXECUTIVE SUMMARY

MEETING OBJECTIVES:

- **To critically reflect on, discuss, and provide feedback on a draft Call to Action on unmet needs in PKU**
- **To explore the best ways of making use of such a Call to Action**

UNMET NEEDS AND REQUIRED ACTIONS ADDRESSED:

- **Acknowledge PKU as a life-long disorder**
- **Take account of the neurological and mental health impact of PKU**
- **Make new-born screening a reality across Europe**
- **Support the implementation of good practice in PKU treatment: European guidelines**
- **Ensure access and adherence to appropriate dietary therapy**
- **Establish clear and consistent protein food labelling**
- **Facilitate access to innovative treatment**

MAIN CONCLUSIONS

- **Agreement on the identified unmet needs**
- **Strong support for a Call to Action**
- **Agreement on making use of the Call to Action as an advocacy tool to raise awareness of unmet needs in PKU as well as of the activities of the EP Cross-Party Alliance on PKU**
- **During 2021, concerted efforts will be made to broaden the support base for the Cross-Party Alliance in the European Parliament and beyond**

MEETING REPORT

WELCOME AND INTRODUCTIONS

Sue Saville (former Medical Correspondent at ITV News, UK) welcomed delegates from the European Society for Phenylketonuria and Allied Disorders Treated as Phenylketonuria (ESPKU) and MEPs and underlined the aim of the meeting, i.e., to build consensus on the most important and compelling actions that could be taken, at both national and EU level, to address the unmet needs in PKU.

She then gave the floor to **host MEP Tomas Zdechovsky**, who expressed his support for the Cross-Party Alliance as well as for the event. He stated that PKU is a highly complex and lifelong condition, which has many consequences for day to day living and quality of life. One of his primary concerns is PKU’s impact on mental health.

Given that this is the third meeting of the Cross-Party Alliance in 2020, despite the COVID-19 pandemic, it is clear that this Alliance is not to be defeated by adverse circumstances; thus, underlining its dynamic nature.

Tomas Zdechovsky MEP welcomed the Alliance’s move from discussing and sharing the issues faced by those living with PKU to a more active approach; the time has come to work towards concrete advocacy activities and the development of an advocacy tool, i.e., a concrete Call to Action. He expressed his support for future advocacy activities and expressed his hope for the Call to contribute to improving health systems and ensuring lifelong access to the best treatment and care in PKU.

Liam O’Connell, speaking on behalf of **Sean Kelly MEP**, also welcomed the meeting and it aims, as the Alliance is moving to a more action-oriented mode; a Call to Action, specifying what MEPs can do to raise awareness of PKU as well as the policy action that could be taken, will be very helpful. Sean Kelly MEP offered his support to ensure the widest possible dissemination of the Call.

Sue Saville then conveyed a message of support from **Clare Daly MEP**, who also welcomed the dynamic and action-oriented nature of the Cross-Party Alliance; the aim to develop a concrete Call to Action is proof of this. Clare Daly underlined the need for this initiative to guide policymakers towards doing what is right and what is required in this area.

MEPs Barry Andrews and Victor Negrescu sent their apologies for not being able to attend; however, they also expressed their support for the meeting, its outcomes and next steps.

SETTING THE SCENE

Eric Lange (President, ESPKU), setting the scene for the discussion, thanked Tomas Zdechovsky MEP for hosting this event and Sue Saville for moderating. He said that ESPKU is proud to coordinate this Alliance, which was launched in March 2020 with the aim *‘to bring together MEPs to raise awareness of and propose and advocate specific (policy) initiatives to better address unmet needs in PKU across the EU in*

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order to improve the day-to-day situation and quality of life of those affected by PKU and their families'. It is currently supported by 13 MEPs; and this number could and should be increased; PKU currently is an invisible disease which should be made more visible. PKU needs to be brought into the mainstream. He stated that, to date, the Alliance's meetings have explored the various PKU-related issues that need to be addressed as a matter of urgency; it is now time to move into a more action and advocacy-oriented approach. The longer-term aim is for the European Parliament to take a more formal initiative to recognise PKU as a condition which merits more attention, e.g., a Resolution, a report from the Parliament's internal research Unit, etc. However, in recent conversations with the MEPs supporting the Alliance, it became clear that to do so, awareness of PKU will need to be raised across the European Parliament.

This is the background to the draft Call to Action on unmet needs in PKU; once finalised it can be used as an advocacy and awareness raising tool, in the European Parliament and beyond; this can help gather the critical mass required to obtain the support for more formal action afterwards. In order to start the discussion, Eric reminded participants of the 7 main areas addressed in the Call:

- Acknowledge PKU as a life-long disorder
- Take account of the neurological and mental health impact of PKU
- Make new-born screening a reality across Europe
- Support the implementation of good practice in PKU treatment: European guidelines
- Ensure access and adherence to appropriate dietary therapy
- Establish clear and consistent protein food labelling
- Facilitate access to innovative treatment

STATEMENTS FROM PARTICIPATING MEPS

Deirdre Clune MEP stated that there seems to be a different mindset in relation to EU action in the field of healthcare because of the COVID-19 pandemic. The interest to move forward jointly and to share information and resources is stronger. The EU4Health programme is a demonstration of this new spirit. Until recently, there has been less coordination in the area of healthcare and that needs to be changed. In terms of the Cross-Party Alliance, Deirdre Clune MEP suggested that its supporting MEPs can play a role to create more awareness and bring more colleagues on board; she proposed that supporting MEPs directly contact colleagues, outlining the relevance of the Alliance and bringing it into the mainstream.

In relation to the Call to Action, Deirdre Clune MEP emphasised the importance of keeping the focus on the concrete and practical measures that can be taken by the European Parliament. For instance, the Parliament could call for coordination in areas of relevance to PKU, similar to what is currently being proposed for the EU Beating Cancer Plan, which could function as an example for other areas. Research, information and good practice should be brought together and shared in 'centres of excellence', rather than each individual Member State working in silo.

She agreed that efforts to collect online signatures for certain issues can be very helpful as the Commission needs to see that there is substantial support for that particular issue. She also pointed out the possibility of MEPs signing Written Questions to the Commission, which it is obliged to answer.

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Having read the Call to Action with great interest, **Frances Fitzgerald MEP** welcomed the document and expressed her support for wider dissemination once finalised. She raised a number of points which could be taken into account when finalising the document:

- The issues of care – and carers in particular - could have a stronger emphasis. The EPP group is in the process of developing and advocating a 'European Care Strategy', looking at care provision throughout the life cycle. The Strategy would focus on data collection across the EU (e.g., disaggregated data to identify what the care issues are at the various stages of the life cycle) and best practice networks. As a lifelong disorder, this is relevant to PKU; and the topic will feature in many relevant policy discussions in the (near) future.
- A second issue relates to the current pandemic; the EU's resilience and recovery fund, providing 675 billion Euro for the implementation of Member States' national recovery plans, refers to care infrastructure. Relevant groups will have to talk to governments to ensure that this money is well spent; ESPKU members could talk to their governments about this spending.
- A third issue relates to mental health. While recognising that mental health and related services delivery is a national competency, the mental health perspective should function as a lens to focus all our actions.
- New-born screening is also an important topic; much more can be done in the EU and Frances Fitzgerald MEP has recently tabled a Written Question to the Commission to this effect.
- Innovative treatment is another important topic. Today's advances in medicine development are amazing – but equal access is by no means a given. There must be systems in place to ensure equitable access to innovative technologies and medicines. It is said that the price of innovation is high; but the price of not having access to innovation is even higher, in terms of mortality and the negative impact on individuals and their families. Patient involvement is critical to ensure access, as the personal story is the most powerful advocacy for clinicians, policy makers and researchers.
- A high-level working group, bringing together the pharmaceutical industry, advocacy groups, policy makers and other stakeholders is needed in relation to medicines for rare diseases like PKU; Frances Fitzgerald MEP has written, to the Commission proposing such a working group at EU level. The Beating Cancer Plan could function as a pilot for this.

AUDIENCE DISCUSSION

Sue Saville then opened the discussion, reminding participants of the two main questions that needed to be addressed:

- what do you see as missing from the draft Call to Action?
- what do you think the most important messages are?

She also underlined the importance of linking the Call to some relevant items currently on the EU agenda, such as Horizon Europe (research), the EU4Health programme, or the Sustainable Development Goals; there are a number of other relevant 'hooks' as well, such as:

- The need for better implementation of treatment guidelines (targeting MetabERN)
- The increasing policy interest in mental health and actions to advocate for an EU Mental Health Strategy
- Actions on rare diseases (including national action plans on rare diseases)
- The European Pillar of Social Rights (articles 16 and 18, on health and long-term care, for instance)

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- The current PPE interest in long term care as one of their main communication platforms
- The increasing focus on prevention of illness
- The increasing advocacy efforts on equal access to treatment across the EU
- Earlier EU initiatives on new-born screening
- Ongoing work (and competency on) food labelling)

Bernadette Gilroy (PKU Association for Ireland, IRL) welcomed the comprehensive nature of the Call to Action and underlined the importance of the European PKU treatment guidelines. These are very good; ESPKU's Scientific Advisory Committee has done a tremendous job. The Call should emphasise that these guidelines address minimum standards of care for PKU as opposed to 'good practice' only.

She also suggested adding the word 'accurate' to the section on 'clear and consistent food labelling'. She also stated the importance of including a reference to increasing knowledge on ongoing clinical trials within countries so that patients (and their advocates) are more familiar with what is available (and what they may be missing out on).

Laura Petreus (Asociatia Aroteica din Romania, RO) underlined the importance of the European guidelines becoming the standard for PKU. In Romania, some doctors had proposed a different set of guidelines; the Aroteica Association took this matter up with the Ministry of Health; it recently argued for these guidelines to be removed from the Ministry's website, asking for them to use the EU guidelines and consult with EU specialists. Laura Petreus asked the ESPKU to send a letter of recommendation about the use of the EU guidelines to the Health Ministry. ESPKU kindly supported this action. In general, doctors in Romania should be better trained in PKU; the standards of medical care seem to be higher outside Romania.

Júlio César Rocha (NOVA Medical School Lisbon & Reference Centre of IEM-CHULC, PT) stated that the draft Call is concise and complete. He made some additional suggestions:

- Dietary treatment is the basic treatment in PKU. However, in some countries the crucial involvement of dieticians and nutritionists is still not at the desired level in order to truly meet patients' need.
- The importance of ensuring a multidisciplinary team in the care of PKU cannot be emphasised enough. In some countries, nutritionists are not allowed the work at the hospitals as they are not training to work with patients living with PKU and other metabolic conditions. In his work, Julio sees (undiagnosed, untreated) people affected with severe mental retardation as well as (diagnosed, treated) people who can live their lives to their full potential. Having access to treatment determines whether a person is either lost or can play a role in society.
- New born screening should be available everywhere. Likewise, dietary treatment should be available for every single patient. This is not yet the case; in some cases, innovative treatments are available before dietary therapy. While pharmaceutical innovation is important, the foundation of treatment – i.e., dietary treatment - should already be in place.

Tobias Hagedorn (DIG PKU, DE) supported what had been said by his colleagues and shared a personal story to demonstrate the importance of mental health and PKU. People living with PKU do not feel recognised as an individual: lack of access to treatment – dietary or innovative - has an effect on mental health. Inadequate (or no access to appropriate transition regimes has an impact on mental health. There needs to be broad understanding and recognition that the PKU diet is a medical need. The lack of

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recognition in the areas above has a huge effect on individuals living with PKU as well as on the PKU community. Mental health plays a role in all points made in the draft Call to Action.

Marketa Lhotokova (NSPKU, CZ) fully supported the Call to Action and underlined access to dietary treatment as the most important issue. Access to treatment determines whether a person with PKU lives a disabled or a non-disabled life. The diet is difficult to adhere to – patients, carers and families - but it is effective. Access to this treatment is not easy as the dietary foods required are expensive but not reimbursed – they are not seen as a medical treatment. The financial burden is difficult for many families. Marketa Lhotokova's association has applied for reimbursement many times, but these efforts have not yet born fruit. It would be helpful if these advocacy efforts could be supported across the EU.

Malgorzata Henek (PL/E.S.PKU) highlighted the Call's section related to PKU being a lifelong disorder as one of the most important messages; this is crucial as the number of adult and ageing patients is increasing. PKU care provision varies across EU countries; access to and quality of care is not the same. Another important point relates to reimbursement. While PKU formulas currently are being reimbursed in Poland, this may change if the government needs to save money and make budget cuts – like in times of COVID-19. Lastly, Malgorzata Henek underlined mental health as a very important and overriding issue.

Anita MacDonald (Birmingham Children's Hospital, UK) made a number of additions and comments:

- The Call should speak of lifelong treatment rather than lifelong condition; every congenital condition is lifelong. Treatment should be optimal and lifelong and meet the standards as outlined in the EU guidelines. In this respect, the guidelines should specify the minimum requirements rather than the optimal ones.
- Universal screening is of course essential, but care should be taken to ensure that immigrants and those people moving from country to country are not missed out.
- 'Transition' is a trendy topic at the moment; but it should not be forgotten that transition also means moving into a new clinical setting. It needs to be specified that transition should go hand in hand with the establishment of fully established clinics with trained professionals.
- A register of how many patients are lost to follow up would be useful as this number seems to be on the increase – and in many cases, it is the most vulnerable patients that are lost to follow up.
- Maternal PKU is an issue that needs attention. At this point in time, it is not known how many maternal PKU cases there are in EU; and there is a lack of information on maternal PKU care.
- There is a huge number of problems with accuracy of protein labelling and lack of monitoring of this; it is simply not seen as a problem by the relevant authorities. Moreover, government policies addressing obesity and reducing sugar content in certain food will often lead to increased proteins (or sweeteners providing a source of phenylalanine like aspartame) in these foods - and that will further impact that food that people with PKU can eat. It needs to be acknowledged that government policies designed for the majority will negatively affect a minority.
- In terms of access to protein substitutes and special low protein foods, people should have access to the best products that are developed and made with the best technological advances.
- Equal and immediate access to innovative medicines once approved is essential as adherence to dietary treatment is difficult. Pharmaceutical companies have a responsibility to ensure that their treatments are accepted by governments across Europe by providing evidence about cost effectiveness and clinical effectiveness, thereby facilitating patient access to these medicines.

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Loïc Lalin (Les Feux Follets, F) focused on the issue of patients being lost to follow up. Until 2 years ago, PKU was not seen as a lifelong disorder. As a result, many patients are lost to follow up. Many did not have access to screening as this was either not available or not effective. This is an important issue.

A second point related to the fact that medical treatment does indeed make a difference. Dietary treatment is the minimum standard of care; all those living with PKU should have access to this. Innovative pharmacological treatments can really support patients as the diet is difficult to adhere to - but the dietary treatment should be ensured for all. Therefore, innovative dietary solutions should be supported as a factor improving the quality of life for those living with PKU.

Florentina Peric (Croatian PKU Association, CT) told the story of her experience with PKU care in two different countries, Croatia and The Netherlands. In the Netherlands, dieticians are much more involved; Florentina sees her dietician every three months and that level of involvement is crucial as well as reassuring. This does not exist in Croatia; the dietician can be called in case of questions, but active involvement is much lower.

In the Netherlands, every patient has a dietician and this dietician is strongly involved. Mandatory involvement of a dietician should become the norm across the EU. Florentina welcomed the Call as very complete and highlighted the importance of the implementation of the guidelines as well as the focus on mental health.

Karen Willetts (PKU Association for Ireland, IRL) welcomed the Call as complete and comprehensive. As a priority she listed lifelong treatment, as adult PKU care and transition from child to adult services need to be improved. She also called on the ESPKU delegates to build personal relationships with MEPs. If ESPKU wants to raise awareness and improve services, they should get involved with MEPs as the best awareness raising can be done by those directly affected by PKU (patients, carers) themselves.

Eric Lange (President, ESPKU) reiterated the need 'to make the invisible visible' as the main task of ESPKU and the Cross-party Alliance. As one of his priorities, he listed the need for higher appreciation and better recognition of the role of dieticians and nutritionists across the EU. Training, especially in the EU's eastern Member States, could possibly be fostered by one of the EU's support funds. He noted that while the guidelines are increasingly being implemented, we should make sure that the dieticians and nutritionists are in place to facilitate this implementation.

Sue Saville then invited participants to come forward with their views on how to best take the Call to Action forward. The following issues were raised:

- Signing petitions and making use of social media are useful tools that could be put to use.
- Messaging needs to be kept simple: the KISS ('Keep It Simple, Stupid') principle should be adhered to, ensuring that the Call's messages are concrete and action oriented.
- Once the ESPKU Call to Action is in place, EU level stakeholders should encourage Member States to check their PKU-relevant legislation and work with patients to update these, ensuring the resources and services that are required.
- The potential role of MetabERN (the European Reference Network addressing rare metabolic diseases) could play explored in the dissemination of guidelines or in changing and influencing policies.
- Because of the new-born screening measures, introduced in the 1960s, patients are now becoming first-time older patients and they will need to have their needs addressed. In terms of

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media attention, it was suggested that it might be interesting to find the oldest patient in Europe and listen to their story and authentic experience. On the other hand, it was said that it is difficult for patients to share their stories in the media as their main message may get lost; making use of case studies in the media should be a last resort. It may be better to conduct some qualitative research about the lived experience of PKU (patients, families and carers' perspectives) and share this information without exposing individuals.

- It could be worthwhile to find a celebrity living with PKU to champion the main message.

CONCLUSIONS AND NEXT STEPS

In conclusion, **Eric Lange** outlined the next steps that will be taken: first of all, a second version of the Call to Action will be prepared, based on today's feedback and discussion. Then, the feedback and endorsement of the MEPs supporting the Cross-Party Alliance will be sought, with a view to finalising it before the end of January.

Dissemination and endorsement gathering actions will then be launched, such as outreach in the European Parliament, e.g., an information stand on relevant Days (e.g., International PKU Day, Rare Disease Day...) and organising meetings with MEPs (COVID permitting), letter campaigns involving the ESPKU community, online signatories, social media activity, etc.

Eric Lange underlined that these activities would make a difference to the lives of people living with PKU and their families across Europe.

He then thanked Sue Saville, participants and those working behind the scenes for their contribution to this event.

Sue Saville closed the meeting, also thanking participants, MEPs and organisers.

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ANNEX: PARTICIPANTS

Surname	First name	Organisation	Country
Boëne	Marc	BOKS	Belgium
Clarke	Una	Office of Frances Fitzgerald MEP	Ireland
Clune	Deirdre	MEP	Ireland
Crima Mihaela	Talos	Asociatia Aroteica din Romania	Romania
Deery	Eimear	Office of Maria Walsh MEP	Ireland
Dvorak	Tomas	Office of Tomas Zdechovsky MEP	Czech Republic
Finnegan	Rachel	BioMarin	Ireland
Fitzgerald	Frances	MEP	Ireland
Gilroy	Bernadette	PKU Association of Ireland	Ireland
Hagedorn	Tobias	DIG PKU	Germany
Henek	Gosia	ESPKU	Poland
Lalin	Loic	Les Feux Follets	France
Lange	Eric	ESPKU	
Lhotakova	Marketa	ESPKU	Czech Republic
MacDonald	Anita	Birmingham Children's Hospital	
Marking	Christine	Marking Public Affairs	Netherlands
O'Connor	Liam	Office of Sean Kelly MEP	Ireland
Peric	Florentina	Croatian PKU Association	Croatia
Petreus	Laura	Asociatia Aroteica din Romania	Romania
Rocha	Julio	NOVA Medial School, Lisbon	Portugal
Saville	Sue	former Medical Correspondent at ITV	United Kingdom
Willets	Karen	PKU Association of Ireland	Ireland
Zdechovsky	Tomas	MEP	Czech Republic