

# CALL TO ACTION

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Ensure that people living with **PKU** have lifelong access to the services, therapeutic solutions and support required to best manage the disorder and live to their full potential



## UNMET NEEDS IN PKU:

- Acknowledgement of PKU as a disorder requiring **life-long treatment**
- Recognition of the **neurological and mental health impact** of PKU
- Universal access to **new born screening**
- Broad implementation of **European guidelines** on minimum standards of care
- Universal access (and adherence) to **appropriate dietary therapy**
- Establishment of clear and accurate **protein food labelling**
- Equal access to **innovative treatment**

## What is Phenylketonuria (PKU)?

There are currently approximately 45,000 people in Europe living with PKU - a rare metabolic condition diagnosed through newborn screening. PKU affects a person's ability to break down phenylalanine, an amino acid present in protein. When phenylalanine builds up in the bloodstream, it can lead to intellectual and physical disability. Treatment consists of a protein restricted diet - for life; however, research has demonstrated that adherence to dietary therapy as well as access to the foods required is a challenge for many people living with PKU. Pharmacological therapies with proven efficacy are available - but not to all individuals with PKU.

PKU can have lasting neurological, emotional, mental health and behavioural consequences. Moreover, it affects all dimensions of an individual's life as well as that of their families and carers. Co-morbidities (osteoporosis, high blood pressure, obesity, asthma, chronic ischemic heart disease) are common.

### The European PKU community and the European Parliament Cross-Party Alliance on PKU invite MEPs to support THIS CALL and **HELP US MEET the unmet needs in PKU by**

- Supporting any action that aims to raise awareness of PKU and its unmet needs
- Calling for European-wide implementation of the European PKU treatment guidelines (targeting MetabERN);
- Supporting the call from the 2019 Finnish Presidency and EPSCO Council to the Commission for a comprehensive EU Mental Health Strategy;
- Calling for the EU4Health Programme and EU actions on rare diseases to address issues related to transition of care and the appropriate care infrastructure required;
- Ensuring that the future Action Programme on the implementation of the European Pillar of Social Rights has an appropriate focus on the right to health and long-term care;
- Supporting the European People's Party initiative for a European Care Strategy;
- Calling for universal access to newborn screening across the EU, in line with the Commission's current interest in prevention of ill health;
- Calling for appropriate EU measures on accurate protein food labelling; and ensure that food policy measures that are beneficial to address some specific health issues do not disadvantage people living with PKU (for instance, replacing sugar by aspartame);
- Calling for the role of the European Reference Network on Metabolic Disorders (MetabERN) to include the development of appropriate patient pathways as well as a standard outcome registry to record evidence generated on treatment and care, also in PKU;
- Ensuring that the EU Cancer Strategy functions as a pilot to for coordination in more areas of health and health services, bringing knowledge together in dedicated centres of excellence;
- Ensuring a clinical trials framework that enables patients and clinicians in countries across Europe to participate in trials, thereby increasing their understanding of available treatments. '
- Ensuring that the new Pharmaceutical Strategy maintains a pro-innovation environment to enable the development of better, accessible and affordable treatments with a positive impact on quality of life; the same applies to the ongoing revision of the Orphan Medicines Regulation.

# CALL TO ACTION

The European **PKU** Community and the European Parliament Cross-Party Alliance on **PKU** call on policy makers, relevant authorities and stakeholders at EU and national levels to:

## 1 Recognise PKU as a disorder requiring life-long treatment

PKU requires optimal lifelong treatment. However, structured transition regimes (from child to adult care) are lacking across the EU. This results in growing numbers of patients considered as “lost to follow-up”, with poor physical and mental health outcomes. While the treatment of children with PKU is usually well organised, adult patients should be able to expect similar levels of organisation and quality of care. Moreover, there seems to be a profound lack of awareness and knowledge amongst health professionals of rare metabolic and complex conditions, including PKU.

### NATIONAL LEVEL:

#### HEALTH AUTHORITIES:

- Collect data and monitor the current situation in relation to adult care in PKU, in order to identify gaps and to address these.
- Put in place adequate transition regimes and appropriate care infrastructures to ensure that health-care systems cater for PKU treatment and care at all ages.
- Ensure that health systems plan for the growing number of adults living with PKU and for the uncertainties related to the impact of PKU in old age.
- Collect information on maternal PKU and put in place appropriate care programmes.
- Put in place a registry on adult patients lost to follow-up.

#### POLICYMAKERS:

- Address ‘transition of care’ as an important theme in existing national rare disease plans.

#### CLINICIANS:

- Establish valid medical goals for adult PKU treatment.
- Take account of the specific issues that accompany PKU in adulthood such as multiple morbidities and adherence to diet as well as the challenge of controlling PKU during pregnancy.
- Address ‘transition of care’ as an important theme in existing national rare disease plans.
- Ensure appropriate medical training on PKU, especially for dietitians/nutritionists and young clinicians and enhance recognition of their crucial role in PKU-related care; also, put in place optimal PKU care, ensure the availability of multidisciplinary teams.

## **EU LEVEL:**

### EUROPEAN COMMISSION:

- Include ‘transition of care’ as a theme for the EU4Health Programme as well as in the upcoming Pillar of Social Rights Action Plan.
- Ensure that EU level action on rare disease includes ‘transition of care’ as an important theme.
- Establish mechanisms for the exchange of good practice on health and care services transformation, as referred to in the Strategic Plan for Horizon Europe.
- Explore the role of the European Reference Network on Metabolic Disorders (MetabERN) or another centre of excellence in developing/proposing appropriate patient pathways as well as a standard outcome registry to record evidence generated on treatment and care.
- Explore the possibility of European Social Fund funding for PKU training of medical staff in general (e.g., GPs) as well as for training specialists– e.g., dietitians, nutritionists, psychologists.

## **2 Take account of the neurological and mental health impact of PKU**

**PKU has a profound neurological, intellectual and mental health impact: people affected by PKU generally have lower than average IQ and have problems with brain development and functioning. Intellectual disability, ADHD and other mental health issues (e.g., depression) are also common.**

## **NATIONAL LEVEL:**

### CLINICIANS:

- Take account of the ‘hidden’ neurological and mental health aspects of PKU and make these part of diagnosis, treatment and services plans.
- Address the neurological and mental health aspects in the existing national rare disease plans.

## **EU LEVEL:**

### EUROPEAN COMMISSION:

- Develop a comprehensive EU Mental Health Strategy, as called for by the 2019 Finnish Presidency conclusions and Employment, Social Policy, Health and Consumer Affairs (EPSCO) Council, taking into account the issues listed above.

### **3 Make new born screening a reality across Europe**

PKU is diagnosed through new born screening. If not detected early and managed, PKU can lead to profound and irreversible intellectual and physical disability later in life. The benefits of new born screening – for the quality of life of those affected, for health budgets and for economies as a whole – are self-evident. However, while screening policy frameworks are in place in most European countries, gaps in access to screening continue to exist.

#### **NATIONAL LEVEL:**

HEALTH AUTHORITIES:

- Carry out reviews and audits at national level to ensure that every baby born in Europe is screened for PKU, including babies from migrants and people moving between countries.
- Ensure that every hospital has appropriate and accessible screening programmes in place.
- Carry out robust direct cost/benefit analyses of new born screening, also considering all indirect potential costs, e.g., health, psychological, economic – and the fact that it will be cost-saving in the longer term.

#### **EU LEVEL:**

EUROPEAN COMMISSION:

- Exchange of good practice in the area of new born screening.

### **4 Support the implementation of good practice in PKU treatment: European guidelines**

European PKU treatment guidelines, aimed to specify minimum treatment standards were developed in 2016 and are currently being revised and updated. However, some European countries do not implement these.

#### **NATIONAL LEVEL:**

HEALTH AUTHORITIES:

- Monitor and ensure implementation of European PKU treatment guidelines.

#### **EU LEVEL:**

EUROPEAN COMMISSION:

- Explore the potential role of MetabERN as a dedicated body to ensure dissemination and implementation.

## **5 Ensure access and adherence to appropriate dietary therapy**

In some countries, it is difficult to obtain the specific low protein foods required for dietary therapy. These foods are also expensive, and in many cases not reimbursed. Moreover, adhering to this highly restrictive diet requires lifelong discipline and vigilance; it can be a permanent source of pressure and stress, as well as an obstacle to having a normal social life.

### **NATIONAL LEVEL:**

POLICYMAKERS:

- Ensure easy access to affordable, high-quality low protein foods for everyone living with PKU.
- Put in place reimbursement regimes for dietary therapy.

### **EU LEVEL:**

EUROPEAN COMMISSION:

- Facilitate exchange on good practice in this area.

## **6 Ensure clear and accurate protein food labelling**

There is a lack of harmonised EU guidance and control on protein substitutes and low protein special foods. While the EU regulates for gluten, allergens, lactose, weight control foods, infant formula and baby food, this is not the case for people depending on low protein diets. This adds to the burden of living with PKU and adherence to required low protein dietary therapy.

### **EU LEVEL:**

EUROPEAN COMMISSION:

Address and put right the current lack of guidance and control with respect to protein substitutes.

- Ensure that imported foods comply with EU protein labelling rules.

### **NATIONAL LEVEL:**

POLICYMAKERS:

- Ensure implementation of future EU regulations in this field.

## 7 Facilitate access to innovative treatment

Equal and easy access to innovative treatment should become a reality.

### NATIONAL/EU LEVEL:

#### POLICYMAKERS :

- Foster a pro-innovation environment to enable the development of better treatments with a positive impact on quality of life of patients as well as carers.
- Ensure implementation of the 2020 Pharmaceutical Strategy, in particular in the proposed areas of 'delivering for patients: fulfilling unmet medical needs and ensuring accessibility and affordability of medicines', 'initiatives on access to medicines' and 'initiatives on innovation'.

#### CLINICIANS:

- Provide people living with PKU with information about available treatment options, empowering them to make informed treatment choices.

#### HEALTH AUTHORITIES/RESEARCHERS:

- Involve patients when developing and approving new treatments for PKU, reflecting what matters most to them.
- Governments need evidence about effectiveness and cost-effectiveness of these new medicines. Pharmaceutical companies have a responsibility to facilitate patient access to these medicines. Therefore, these companies should also do research in order to get treatments accepted, rather than research to obtain marketing authorisation only.

For **all of the above**, the EU level could play an important part in collecting relevant data and generating evidence, facilitating the exchange of good practice and learning between Member States, and stimulating discussion on these topics.



# CONCLUSIONS

Sustained efforts are required to ensure PKU has an equal footing in public health on EU and national policy agendas.

As well-being and long-term health being are increasingly recognised as important drivers for an equitable and prosperous Europe, the European PKU community and the European Parliament Cross-Party Alliance on PKU call on policymakers and relevant authorities to work with them to explore all avenues for policy action to allow people with PKU to live well and participate fully in society.

# MORE INFORMATION

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