

About PKU

Phenylketonuria, or PKU, is a rare genetic disorder that affects a person's metabolism. Screening for PKU takes place for all newborn babies in the Republic of Ireland, and across the UK and Europe.

In Ireland approximately 1 in every 4,500 babies are diagnosed with PKU - a relatively high incidence rate, compared to the UK where it is believed to be closer to 1 in every 12,000 babies.

Living with PKU

Living with PKU is a constant daily challenge. Since its natural progression is so severe, patients must take the condition seriously. The only current widely available approach is to stick to a low-protein diet and to take a protein substitute.

PKU severely affects a person's quality of life

Living with PKU means being constantly surrounded by something that is tempting to eat but that could be harmful. Buying a sandwich, eating at fast food chains or at birthday parties – these all require careful planning with a PKU diet.

Some PKU people manage their diet effectively and live well. While other people will find their diet leaves them hungry, unsatisfied and poorly nourished.

Finding ways to eat well by making the diet as easy as possible is so important to a person's quality of life and long term health outcomes.



The life-long low protein PKU diet is very restrictive

Foods with protein are not only the ones we ordinarily think of as protein such as meat, fish, chicken, eggs, but most normal foodstuffs including bread, cheese, pasta, rice, potatoes and chocolate all contain protein.



The PKU low-protein diet has been a major success. It has prevented hundreds of people across Ireland from developing acute mental and physical disabilities as a result of their condition.

However, it is a punishing and sometimes very difficult diet to stick to and may have long-term health consequences.

A range of low-protein food products are available, which together with foods that contain zero protein, such as some fruits and vegetables, make up the PKU diet. However, patients in Ireland have access to only a small number of the products available to patients elsewhere in Europe. No new foodstuffs have been approved for state funding in many years.

The PKU diet is expensive

A low protein bread supplement is at least 4 times the cost of a normal loaf of bread. In addition the protein substitute drink, which is usually paid for by the state, costs approximately €280 for 10 days supply. Many children struggle with their protein drink due to taste and side effects such as stomach reflux.

PKU can have a lasting impact on your health with widespread physical, emotional and behavioural consequences

With such a restricted diet, people with PKU are believed to be at higher risk of suffering from attention deficit disorders, poor concentration and low energy levels. These in turn can affect performance at school or work.

In the longer term, it is believed that even amongst those who have maintained a strict PKU diet, there may be significant health impacts such as neurocognitive deficits or neurological side effects.

More research should be carried out into PKU in Ireland to understand these health risks and improve treatment.

In Ireland, a diagnosis of PKU entitles people to a small range of supplementary foods paid for by the state. However, the list of products this applies to has not been updated for many years and includes some that are now discontinued and excludes all of the newer products.

Only those able to afford it will be able to access a wider choice. However, some food producers do not make their products available at all in Ireland if they are not included on the approved list of subsidised foods.

It is widely recognised amongst the medical profession that the wider the choice of foodstuffs available the greater the compliance with diet. The Irish health service advocates diet for life but are not offering patients access to the full range of goods that make the diet easier to comply with.

Nothing can replace a normal balanced healthy diet, which PKU people can't have. We should be doing all we can to get as close to that as possible.

More proactive holistic care of PKU throughout childhood and into adulthood is required.

Though blood levels are monitored and dietary advice is available through the specialist hospital, and psychological, educational and neurological assessment is available, there is no strategic outreach to identify where PKU patients would benefit from this.

The burden of caring for this rare genetic disease is placed almost entirely on parents, whose awareness of its socio-psychological effects may be minimal.



Medicines can be effective

Some PKU patients would benefit from taking a medicine called Kuvan (sapropterin) as this can enable them to eat greater quantities of protein, and have a more normal healthy balanced diet, than would otherwise be safe. An improvement in diet would likely benefit the patient's quality of life now, but also affect health outcomes in adulthood. Kuvan is available across USA, Australia and parts of Europe.

KUVAN and other medicines that offer the possibility of a better diet should be a treatment option for PKU children in Ireland.

Adults with PKU

Adults should have the same level of care as paediatric patients.

Compared to the care of paediatric patients, adults with PKU have little in the way of clinical support.

Teenage and early adult years can be the most challenging time for people with PKU and support requirements at this stage are different.



As levels of care change, many people will lose contact with healthcare professionals, despite in some cases this being where they need most support.

It is essential for PKU women considering pregnancy to receive appropriate care.

It is in adulthood that consequences of a poorly managed PKU lifestyle can manifest themselves in quite considerable ways, potentially presenting significant burden on the health system.

The resources at the adult metabolic unit need to be significantly improved and a full time integrated service in place.

The Phenylketonuria Association of Ireland (PKUAI) aims to raise awareness of PKU and support the PKU community in Ireland. Please visit our website or facebook page for information about where to find more support. We always need more volunteers to help run the PKUAI.

We also need your assistance for our campaigns and can help you to contact your TD if you feel your PKU care falls short.

As a patient group we offer support where we can, but strongly advise people with PKU to seek advice from their doctor or dietician.

Principles & Recommendations

- The lifelong PKU diet is very restrictive and severely affects a person's quality of life. More choice in food products could mean all those with PKU having access to a better and more easy to follow diet.
- PKU can have a lasting impact on health, with widespread physical, emotional and behavioural consequences. More PKU research should be carried out in Ireland.
- KUVAN should be a treatment option for PKU children in Ireland.
- More proactive, holistic care of PKU throughout childhood and into adulthood is required.
- The adults' clinic in the Mater hospital needs to be properly resourced and expanded.

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