Phenylketonuria and Personal Independence Payments

What's the Connection?

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Phenylketonuria (PKU) is a rare inherited disorder of phenylalanine metabolism.[1]

The treatment for adults with PKU in the UK is a low phenylalanine diet, which is complex and time consuming – it takes on average 19 hours a week.[2] Without treatment, people with PKU suffer severe and irreversible brain damage. Early treated adults with PKU can experience a range of outcomes, with some experiencing cognitive impairments which can affect daily functioning.

Some adults (over 16 years) with PKU could be eligible for Personal Independence Payments (PIP) if they need supervision, or assistance, with managing their treatment or other aspects of their daily functioning.

Applying for PIP, however, can be a lengthy and challenging process. Support may be needed by metabolic dietitians and other members of the metabolic multidisciplinary team. This article, firstly, outlines the evidence for why PIP is relevant in PKU, and secondly, what PIP is and some of the work done by NSPKU around PIP.

Adults on a low phenylalanine diet – who are they?

PKU occurs due to inherited mutations in the gene encoding phenylalanine hydroxylase. Lifelong phenylalanine restriction and intake of protein substitutes is essential for the best outcome, yet the diet is burdensome, by being both time consuming and often difficult to follow.[3] Consequently, patients may stop following the PKU diet and may also become disengaged from metabolic review. Furthermore, in the past, patients with PKU have been either discharged, or advised to cease treatment because of prevailing beliefs at the time, about brain development being complete. There are patients who are attending specialist clinics following the diet who are early diagnosed, and early and continuously treated, as well as early treated patients, who may have stopped the diet, and subsequently resumed. There is also a group of patients who are classed as late-treated (i.e., not diagnosed during newborn screening, which only started in 1969), and these late-treated patients may have learning difficulties of varying severity.

Low phenylalanine diet – what is involved?

The only treatment, in the UK, for PKU is a complex lifelong dietary regimen, with tighter control needed at different life stages.[1] The diet usually restricts protein to about 10 g per day, and protein containing foods are either counted, or measured down, to 0.3 g protein per serving.[3] The complexity of the diet has evolved over 20-30 years and details have only just been agreed by specialists.[4]

As the diet depends upon restricting the phenylalanine content of foods it is necessary to plan, shop for ingredients, weigh foods and cook/prepare foods with great care. Baking with low protein products – and no egg – takes quite some ability and not everyone acquires these skills. The time commitment needed for a low protein diet is considerable and has been measured. NHS England has accepted (in its policy work on PKU), the peer reviewed evidence showing that the time needed for PKU management is approximately 19 hours per week.[2] Another relevant research finding is that PKU control is better in adult patients who work in part-time jobs rather than full-time or undertake shift work.[5]

The diet is highly artificial which means using prescribable foods low in protein, plus a prescription–only protein substitute either as a drink (e.g., 125 mls x 3 per day) or tablets with meals (e.g., about 80-120 tablets per day).

Managing PKU therapy requires the ability to plan and execute a complex diet. It is necessary to be able to analyse nutrition labels and calculate the phenylalanine content of different foods. It is necessary to use aids like the NSPKU Diet Information book, digital scales, and a calculator. It also necessary to sustain these efforts over the long-term which requires extraordinary commitment and motivation. Evidence shows systems for people with PKU to get prescription items they depend upon as part of their treatment (protein substitutes and low protein food), are broken systems which result in food- and treatment-insecurity, and considerable time spent to acquire the items needed.[6]

Definition of disability

You're disabled if you have a physical or mental impairment that has a 'substantial' and 'long-term' negative effect on your ability to do normal daily activities.

The diet-brain function relationship

Raised blood phenylalanine affects the brain, and specifically disrupts executive functions needed for carrying out tasks in life. Functions such as working memory, planning and inhibitory control can be affected, and this is possibly due to hypomyelination of the brain's white matter in the presence of raised phenylalanine.[7-9] Neurotransmitters are also likely to be disrupted due to excess phenylalanine and insufficient tyrosine, leading to poor mental health such as anxiety and low mood, and there is evidence about PKU patients experiencing tremor more frequently than controls.[10-15]

Common neuropsychological, and neurological features, of symptomatic PKU:

- impaired working memory,
- reduced sustained attention,
- reduced inhibitory control,
- impaired flexible thinking and problem solving,
- tremor,
- anxiety, including panic attacks,
- low mood.

For people with PKU, being on, or resuming dietary treatment, is likely to improve cognition and mental health.[16-17] There is radiological evidence which suggests that resuming the PKU diet, improves white matter changes in the brain, if the changes have manifested in adulthood, and thus, neurocognitive deficits in adults may also improve with better phenylalanine control.[18]

The conundrum is that the diet requires meticulous planning, organisation, motivation and restraint – these are the skills which are affected by PKU, especially if there have been any deviations from the target range of blood phenylalanine.

What is the purpose of PIP?

PIP is a benefit payment which is designed to help with the extra costs of a long-term physical, or mental health, condition or disability.

- The weekly rate for the daily living part of PIP is either £60 or £89.60.
- The weekly rate for the mobility part at the standard rate is £23.70.

An individual can get PIP whether they are working or not. The individual must be aged 16 years, or over, to claim and usually have not reached State Pension age. They must have a physical, or mental health, condition or disability which is long term and:

- Have had difficulties with daily living or getting around – or both – for three months.
- Expect these difficulties to continue for at least nine months.

An individual may get the daily living part of PIP if they need help more than half of the time with things like:

- preparing or eating food,
- managing your medicines or treatments,
- engaging with other people,
- washing, bathing and using the toilet,
- dressing and undressing,
- reading and communicating,
- making decisions about money.

An individual may be eligible for the mobility part of PIP if they need help going out of their home environment or otherwise being able to move around. This can include problems related to cognitive issues, or mental health problems, such as an inability to take a journey alone.

The default way in which the Department of Work and Pensions (DWP) makes an assessment about ability to prepare or eat one's own food is usually through whether a claimant can make a simple snack such as a boiled egg and toast. Whereas an applicant with PKU will need to explain that while they *might be able to boil an egg*, they will be very unlikely to safely eat it, and so this standard test question is not relevant. Hence, the full complexity and time-consuming nature of the dietary regimen must be explained in detail.

What is the process of a PIP application?

If a child with PKU is approaching their 16th birthday and they already claim Disability Living Allowance, the patient should be prompted to apply for PIP when they turn 16 – they should receive a letter from the DWP. Adult patients wanting to apply for PIP can do so by contacting the DWP.

All claimants will need to complete a lengthy form called: *'How your Disability Affects You'* and will be prompted to include details of the healthcare professionals that treat them. It is extremely helpful if a healthcare professional can provide information which explains their condition, the treatment regime, and how the patient is impacted in daily life.

The DWP will start processing the claim when they receive the completed form. The claimant will usually be invited to an assessment by a healthcare professional engaged on behalf of DWP. The healthcare professional will not have specialised knowledge of different medical conditions and will question the claimant on the impact that their condition has on their daily living activities and ability to get around.

The DWP will make a decision based on the information in the form, any supporting medical evidence and the assessment. Once a decision is made, the patient is informed by letter. It is possible to challenge the decision made, by requesting a mandatory reconsideration. Patients need to ask for mandatory reconsideration within one month of the date of the PIP decision. If the mandatory reconsideration stage still finds that the patient does not merit PIP, then it is possible to progress to a tribunal. It should be noted that if the PIP is eventually awarded then the monies paid to the patient will be backdated to the date of original application. As of July 2021, 74% of PIP appeals claimants won at tribunal.[19]

How can the NSPKU help?

NSPKU provides guidance to help people apply for PIP that can show people how to better explain their issues to the DWP. We have also directly supported a small number of individuals to apply, for example:

- 16-18-year-olds needing ongoing carer/parent support to implement treatment.
- 18+ year olds going to higher education, needing ongoing support for treatment.
- Women with PKU who have children who need extra time for their self-care needs to be able to perform their parenting to the best of their abilities.
- Women or men who have come off diet early and tried to get back on, who might have extra support needs through difficulties associated with being off diet.
- Women or men who have had late-treated PKU, e.g., treated from a couple of months old rather than 15 days old and who have extra support needs.
- People who have PKU, and other health challenges, which combine to impact on their daily lives.
- People with PKU for whom their health, living conditions, and demands upon them to do their PKU care are too difficult to meet their own needs without support.

What are the outcomes so far?

Kate Learoyd, Advocacy & Campaign Manager at NSPKU says:

"We run a helpline for families affected by PKU and are repeatedly confronted with evidence of how PKU can interact with social issues – such as insecure work, inadequate housing, and poverty. People with PKU were telling us that they couldn't afford to buy the fresh fruit and vegetables they needed to manage the diet, or they were unable to store and cook prescription foods in cramped bedsits. There can be a downward spiral effect where uncontrolled PKU can make people more vulnerable to social deprivation [which makes managing PKU even harder]. We were aware that many people were motivated to claim PIP but were struggling to navigate the system. However, we have found that with support the majority of claims are successful after a request for mandatory reconsideration to the DWP or at tribunal."

"Working with people with symptomatic PKU has really opened my eyes to the extent to which people may have invisible issues which really significantly affect everyday life."

"Claimants often score points across several daily living and mobility activities. These claims can help stabilise a person's home life creating circumstances gives them a better chance of living a healthy and independent life. We can't easily separate social vulnerability and health outcomes in PKU. I am very thankful for the NHS dietitians and other health professionals who have supported patients with evidence which explains the impact of this misunderstood disease to the DWP. I have seen for myself how much difference the extra money from PIP can make."

Case study

What does this mean for the people themselves, who receive PIP – what have they said about it?

"I am a mum of two young children and have PKU. I went off the PKU diet in my late teens and twenties and I think this has had a big effect on my health. I now struggle with organisation, my memory and completing tasks and I have poor mental health. This means I rely on family help a lot to cope with my complex PKU diet as I can't remember what to buy or how to weigh out PKU exchanges. My struggle with anxiety also means I don't cope with mixing with people I don't know, and other basic things like catching the bus."

"I claimed PIP benefit because the extra money would help us cope financially. My hospital helped with writing letters that explained my condition and I also had support from the charity NSPKU. The DWP did not seem to understand PKU, and I took the case to a tribunal. The three members of the panel asked me questions about my life and the impact of PKU. They read letters from my hospital. My mum and my sister also explained how they support me with my diet and household jobs."

"The tribunal awarded PIP at the enhanced rate for daily living and lower rate mobility. This extra money of £113 a week has made a massive difference. I have paid off debts that had built up. I can afford the extra foods to help my PKU diet. I am planning to spend some of the money on a carer to help with shopping and household jobs. I feel it has lifted a weight from us. I really hope other people in my position can get the help they need."

How can dietitians support their patients in the future?

The DWP will want to see supporting letters from a clinic treating the patient for their long-term condition or disability, to 'corroborate' what the patient themselves is stating about their daily life. Dietitians can spell out the individual's specific difficulties and challenges that are caused by PKU and will be able to put these in lay terms so that the DWP can understand PKU's impact on daily life. The supporting letter can comment on everything from what the diet involves, which parts are particularly challenging, neurocognition, mental health issues, prescription difficulties and bloodspot monitoring.

NSPKU has formulated some template letters which can be adjusted for different patients – we are happy to supply these and help dietitians help their patients. We know that a letter from a health care professional about a specific individual they know, and treat, is most effective to explain a patient's needs. Our experience is that information from clinics is crucial to success with PIP claims as the DWP assessors may not understand the disease, the treatment and the impact on patients.

Conclusion

PKU can impact the daily life of adults in different ways, such as through impaired mental processes, known as executive functioning, and poor mental health, due to the way PKU can affect brain biochemistry. PKU treatment in the UK for adults is by diet alone. The diet is a very complex and restrictive regimen, and decision makers in the NHS agree that the diet involves a significant time commitment of about 19 hours per week. Thus, it maybe that an individual with PKU can apply for benefits to support them and ease the pressure of their dietary treatment after the age of 16. Personal independence payments can help individuals who need support for different aspects of their daily living, not just the diet.

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