HOUSE OF COMMONS LIBRARY

DEBATE PACK

Number CDP-2018-0152, 18 June 2018

Access to treatment, support and innovative new medicines for Phenylketonuria patients

By Alex Bate
Sarah Barber
Andrew Mackley

Contents

Summary

This House of Commons Library Debate Pack briefing has been prepared in advance of a debate entitled "Access to treatment, support and innovative new medicines for Phenylketonuria patients". This will be led by Liz Twist MP and will take place in Westminster Hall on Tuesday 26th June 2018 at 9.30am. This Pack contains background information, parliamentary and press material, and further reading suggestions which Members may find useful when preparing for this debate.

Phenylketonuria (PKU) is a rare inherited disorder, affecting about 1 in 10,000 babies. Individuals with PKU cannot break down an amino acid called phenylalanine, and when the condition is untreated this can build up in the blood and the brain and cause damage to the brain and nervous system, which can lead to learning disabilities. PKU is managed with a low protein diet, and individuals are monitored with regular blood tests.

There has been some recent debate with regard to access to Sapropterin, marketed as Kuvan, on the NHS. This is a treatment to help improve metabolism of protein and therefore decrease levels in the body among patients that respond to it. NHS England is the responsible commissioner for treatment of PKU, and has produced two commissioning policies with regards to access to Kuvan on the NHS for children and pregnant women, and says that should not be routinely funded, except for some at risk groups of pregnant women. NHS England has recently received a policy proposal for the use of Sapropterin in the management of PKU in adults and children, and has referred the issue to the National Institute for Health and Care Excellence (NICE) to reassess whether the treatment should be routinely provided on the NHS.

1. Background

Phenylketonuria (PKU) is a rare inherited disorder, affecting about 1 in 10,000 babies. Individuals with PKU cannot break down an amino acid called phenylalanine, and when the condition is untreated this can build up in the blood and the brain and cause damage to the brain and nervous system, which can lead to learning disabilities. Other symptoms of untreated PKU include:

- behavioural difficulties – such as frequent temper tantrums and episodes of self-harm
- fairer skin, hair and eyes than siblings without the condition (phenylalanine is involved in the body's production of melanin, the pigment responsible for skin and hair color)
- eczema
- recurrent vomiting
- jerking movements in arms and legs
- tremors
- epilepsy
- musty smell on the breath, skin and urine[1]

Phenylketonuria is one of the nine conditions that is tested for in the newborn heel prick test.

1.1 Treatment and management

PKU is managed with a low protein diet, and individuals are monitored with regular blood tests. NHS Choices recommends blood tests at the following frequencies for children:

> It's recommended that children who are:
>
> - 6 months of age or younger should have their blood tested once a week
> - between 6 months and 4 years of age should have their blood tested once every 2 weeks
> - over 4 years of age should have their blood tested once a month.

A low protein diet will often need to include phenylalanine-free protein supplements. In a March 2018 adjournment debate on PKU, Vicky Ford gave some of the challenges in maintaining a low-protein diet:

> My constituent Cait is 10 years old. She can metabolise only 11 grams of protein a day. She is restricted in every eating experience of her life. Her day is ruled by limited food and constant protein supplements—those drinks taste foul and smell unpleasant. When other children are sharing a meal, or perhaps a birthday cake or chocolate, Cait can only have her protein drink.

---

1 NHS Choices, Phenylketonuria

> Her parents tell me that she is permanently hungry. They say that every day since she was born has been filled with the joy that she brings, but also the misery associated with the daily management of her lifetime condition.
>
> The severe restrictions of a PKU diet place a great burden on patients and their families. The phenylalanine content of all food needs to be carefully restricted, including with vegetables such as potatoes and cauliflower. Cait's grandmother has given up work to care for her. In fact, research shows that more than half of the carers of a child with PKU have stopped working, reduced their hours or changed their job so that they can help to manage the child's diet. Unsurprisingly, the constant worry about what their children are eating, and whether brain damage may be caused by everyday food, puts a huge emotional strain on families. A recent study found that 59% of mothers caring for PKU children had clinical levels of psychological distress themselves.[2]

A number of low-protein foods are available on prescription on the NHS in England, in order to help manage the level of protein in the body. However, the National Society for Phenylketonuria (NSPKU) has highlighted difficulties faced by some individuals with getting sufficient food items prescribed by their GP:

> One of the most common problems is the confusion with the gluten free prescribable foods. Pharmacists and GP's may be under the impression that the foods and the amount needed are similar. The amount of gluten free foods allowed each month is based on recommendations by the National Prescribing Guidelines, but is a lot less than for a low protein diet. Consequently, many GP's and pharmacists think that people on a low protein diet are getting too much when they ask for their prescriptions. In addition, many of the "luxury" gluten free foods not allowed on prescription can be bought over the counter in supermarkets, but for low protein diets this is not possible and the foods must come on prescription, thereby patients have prescriptions questioned as to why someone with PKU needs cakes and biscuits on prescription and these may be refused or adjusted.[3]

People with PKU are also advised to avoid food and drink containing the artificial sweeter aspartame, as this is converted to phenylalanine in the body. As a result, products that contain aspartame as required to be labelled as such. NSPKU has recently raised concerns that the introduction of the Soft Drinks Industry Levy may unduly affect people with PKU, as drinks may be reformulated to replace sugar with aspartame.[4]

---

2 HC Deb 22 March 2018, c482

3 NSPKU, Low protein foods: how to get what you need, when you need it!, 2016

4 'The sugar tax discriminates against people with the rare disease PKU, says national charity NSPKU', *NSPKU press release*, 8 June 2018

Access to Sapropterin (Kuvan)

Sapropterin, marketed as Kuvan, is a treatment to help improve metabolism of protein and therefore decrease levels in the body, among patients that respond to it. Kuvan is not intended as a replacement for a low-protein diet, which will still continue to be required for people with PKU.

NHS England is the responsible commissioner for treatment of PKU, and has produced two commissioning policies with regards to access to Kuvan on the NHS.

- The use of Sapropterin in children with Phenylketonuria (2015)

This recommends that Saproprterin should not be routinely commissioned for children on the NHS, as a result of a limited evidence base:

> Sapropterin has been considered by NHS England who concluded that there was not sufficient evidence to support the routine commissioning of this treatment. The evidence review provided an assessment of effectiveness and safety of Sapropterin in the short term (up to 10 weeks) and could not demonstrate the benefits of treatment on nutritional status and cognitive development.

- Sapropterin (Kuvan®) For Phenylketonuria: Use in Pregnancy (2013)

This recommends that Sapropterin should not be routinely commissioned for pregnant women with PKU on the NHS, but does identify some at risk groups who are eligible for treatment.

Although NHS England can have a policy not to routinely commission a particular treatment, it cannot impose a 'blanket ban' and must consider exceptional individual cases where funding should be provided, known as Individual Funding Requests (IFRs). Where a patient's GP or consultant feels that a patient has exceptional circumstances that mean they should be able to access a treatment that is not routinely commissioned on the NHS, they can submit an IFR to NHS England on the patient's behalf.

Since the publication of these commissioning policies, NHS England has received a policy proposal for the use of Sapropterin in the management of PKU in adults and children, and has referred the issue to the National Institute for Health and Care Excellence (NICE) to reassess whether the treatment should be routinely provided on the NHS. No further details are available on the timescales for this, as set out in the following PQ:

> **Asked by Layla Moran (Oxford West and Abingdon) 29 March 2018**
>
> To ask the Secretary of State for Health and Social Care, when he expects NHS England and NICE to make a decision on licensing Kuvan for the treatment of phenylketonuria

> **Answered by: Steve Brine 17 April 2018**
>
> NHS England has received a Preliminary Policy Proposal for the use of Sapropterin in the management of phenylketonuria for adults and children, as it is considered by the Clinical Lead that new evidence has now been published to support its use.
>
> This was considered by the Specialised Services Clinical Panel where it was agreed that, as this is a licensed medicine, it should be referred to National Institute for Health and Care Excellence (NICE) for consideration through its technology appraisal process. NICE has confirmed receipt of referral and will be advising on the decision making process and timetable in due course.[5]

An Early Day Motion from March 2018 noted that Kuvan was already provided by health services in several European countries, and called for the appraisal of the treatment to be "concluded swiftly."[6]

Kuvan is not routinely available in Wales or Northern Ireland. NICE technology appraisals also apply to Wales, whilst the Northern Irish Health and Social Care Board has formal links with NICE and will consider implementing any recommendations it makes.

Access to Kuvan in Scotland is currently under review, and a decision is expected in August, as set out in the following Scottish Parliament written answer:

> **Question S5W-16945: Miles Briggs, Lothian, Scottish Conservative and Unionist Party, Date Lodged: 29/05/2018**
>
> To ask the Scottish Government, in light of the findings of the report, Review of Access to New Medicines, what progress has been made in ensuring access to treatments for (a) phenylketonuria and (b) other rare diseases.
>
> **Answered by Shona Robison (06/06/2018):**
>
> I wrote to the Health and Sport Committee on 17 May to provide a further update on the Scottish Government's progress in delivering the recommendations from the Review of Access to New Medicines.
>
> I can confirm that the pharmaceutical company, BioMarin, has made a submission to the Scottish Medicines Consortium (SMC) for sapropterin (Kuvan®) for the treatment of phenylketonuria. The SMC will publish their advice in August.
>
> Decisions made by the SMC are independent of Ministers and the Parliament.[7]

---

5 PQ 135194, 17 April 2018

6 Early Day Motion 1044, 8 March 2018

7 Question S5W-16945, 6 June 2018

2. Parliamentary material

2.1 Written Parliamentary Questions

- Sapropterin

Asked by: Moran, Layla | **Party:** Liberal Democrats

To ask the Secretary of State for Health and Social Care, when he expects NHS England and NICE to make a decision on licensing Kuvan for the treatment of phenylketonuria

Answering member: Steve Brine | **Party:** Conservative Party | **Department:** Department of Health and Social Care

NHS England has received a Preliminary Policy Proposal for the use of Sapropterin in the management of phenylketonuria for adults and children, as it is considered by the Clinical Lead that new evidence has now been published to support its use.

This was considered by the Specialised Services Clinical Panel where it was agreed that, as this is a licensed medicine, it should be referred to National Institute for Health and Care Excellence (NICE) for consideration through its technology appraisal process. NICE has confirmed receipt of referral and will be advising on the decision making process and timetable in due course.

17 Apr 2018 | Written questions | Answered | House of Commons | 135194

Date tabled: 29 Mar 2018 | **Date for answer:** 16 Apr 2018 | **Date answered:** 17 Apr 2018

- Sapropterin

Asked by: Thomas-Symonds, Nick | **Party:** Labour Party

To ask the Secretary of State for Health and Social Care, what his Department's policy is on the administration of Kuvan to women with Phenylketonuria throughout their pregnancy.

Answering member: Steve Brine | **Party:** Conservative Party | **Department:** Department of Health and Social Care

NHS England has a published policy on access to "Kuvan" (drug name Sapropterin) for phenylketonuria during pregnancy. Further information is available at:

https://www.england.nhs.uk/wp-content/uploads/2013/04/e12-p-a.pdf

28 Feb 2018 | Written questions | Answered | House of Commons | 128680

Date tabled: 20 Feb 2018 | Date for answer: 22 Feb 2018 |

Date answered: 28 Feb 2018

- Sapropterin

Asked by: Hodgson, Mrs Sharon | **Party:** Labour Party

To ask the Secretary of State for Health and Social Care, how many pregnant women with phenylketonuria have received Kuvan (sapropterin) treatment since that drug has been licenced for use in the UK; and what is the average duration of the course of that treatment.

Answering member: Jackie Doyle-Price | **Party:** Conservative Party | **Department:** Department of Health and Social Care

The information requested is not held centrally.

In 2013, NHS England published a policy on the use of Sapropterin for the management of Phenylketonuria (PKU) during pregnancy:

https://www.england.nhs.uk/wp-content/uploads/2013/04/e12-p-a.pdf

NHS England has now received a Preliminary Policy Proposal for the use of Sapropterin in the management of PKU for adults and children, as new evidence has now been published to support its use. This was considered by the Clinical Panel this month where it was agreed that NHS England will need to further review the evidence. NHS England will be working with the National Institute for Health and Care Excellence to agree the best approach to this and whether the policy should subsequently be reviewed. The NHS England process for development of Clinical Policies can be found here:

https://www.england.nhs.uk/publication/specialised-commissioning-service-development-policy-and-process/

https://www.england.nhs.uk/publication/methods-national-clinical-policies/

31 Jan 2018 | Written questions | Answered | House of Commons | 124254

- Phenylketonuria: Medical Treatments

Asked by: Harman, Ms Harriet | **Party:** Labour Party

To ask the Secretary of State for Health, whether he plans to allow NHS England to negotiate directly with pharmaceutical companies to improve access to treatments for phenylketonuria.

Answering member: Steve Brine | **Party:** Conservative Party | **Department:** Department of Health

There are two policies relating to Sapropterin (Kuvan). The first allows for access to sapropterin for women with phenylketonuria (PKU) who are pregnant and cannot maintain appropriate PKU levels with their diet. This can be found at:

https://www.england.nhs.uk/commissioning/spec-services/npc-crg/group-e/e09/

For the remaining patients, sapropterin is not routinely funded. This policy can be found at:

https://www.england.nhs.uk/commissioning/spec-services/npc-crg/group-e/e06/

This second policy is currently in the process of being reviewed, with a decision expected in 2018. This will involve evaluating any new clinical evidence that has been developed since this policy was originally formed.

NHS England has a number of mechanisms available to secure access to medicines in secondary care from pharmaceutical companies. For example, the Commercial Medicines Unit undertakes procurements through framework agreements that enable individual trusts to purchase medicines at the framework price. Where medicines have been approved by the National Institute for Health and Care Excellence, a patient access scheme may be in place to allow the National Health Service to benefit from a discounted price. The mechanism used will depend on the medicine in any given case.

04 Dec 2017 | Written questions | Answered | House of Commons | 116628

Date tabled: 29 Nov 2017 | **Date for answer:** 04 Dec 2017 | **Date answered:** 04 Dec 2017

- Children: Behavioural Disorders

Asked by: Howlett, Ben | **Party:** Conservative Party

To ask the Secretary of State for Education, what guidance her Department makes available to teachers on supporting children who display behavioural problems resulting from phenylketonuria.

Answering member: Edward Timpson | **Party:** Conservative Party | **Department:** Department for Education

We know how important it is that children with medical conditions are supported to enjoy a full education. That is why we introduced a new duty to require governing bodies to make arrangements to support pupils with medical conditions and have provided statutory guidance outlining schools' responsibilities in this area.

The Department has also issued advice on behaviour and discipline for schools. This advice is clear that schools should assess the needs of pupils who present with persistently difficult behaviour. We make clear that schools should consider whether the continuing disruptive behaviour is a result of unmet educational or other needs. At this point, the school should consider whether a multi-agency assessment is necessary.

27 Mar 2017 | Written questions | Answered | House of Commons | 68064

Date tabled: 15 Mar 2017 | **Date for answer:** 20 Mar 2017 | **Date answered:** 27 Mar 2017

- Hereditary Diseases: Screening

Asked by: Glen, John | **Party:** Conservative Party

To ask the Secretary of State for Health, what the annual cost to the NHS is of providing screening for (a) sickle cell disease, (b) cystic fibrosis, (c) congenital hypothyroidism, (d) phenylketonuria, (e) medium-chain acyl-CoA dehydrogenase deficiency, (f) maple syrup urine disease, (g) isovaleric acidaemia, (h) glutaric aciduria type 1, and (i) homocystinuria (pyridoxine unresponsive) (i) in total and (ii) on average per each test delivered; and how many such tests were undertaken in the latest year for which figures are available.

Answering member: Jane Ellison | **Party:** Conservative Party | **Department:** Department of Health

Data on the annual cost to the National Health Service of providing screening for sickle cell disease (SCD), cystic fibrosis (CF), congenital hypothyroidism (CHT), phenylketonuria (PKU), medium-chain acyl-CoA dehydrogenase deficiency (MCADD), maple syrup urine disease (MSUD), isovaleric acidaemia (IVA), glutaric aciduria type 1 (GA1) and homocystinuria (pyridoxine unresponsive)(HCU) are not held centrally.

From Public Health England data, the following number of tests were undertaken in England in 2014/15:

	Number tested for PKU	Number of babies tested for CHT	Number of babies tested for CF	Number of babies tested for MCADD	Number of babies tested for SCD
England	673,328	673,233	671,120	672,107	668,117

No data is available for MSUD, IVA, GA1A and HCU as screening was not routinely offered until January 2015.

30 Jun 2015 | Written questions | Answered | House of Commons | 3515

Date tabled: 22 Jun 2015 | **Date for answer:** 24 Jun 2015 | **Date answered:** 30 Jun 2015

2.2 Oral Parliamentary Questions

- Topical Questions

Asked by: Layla Moran | **Party:** Liberal Democrats

I have a young constituent who has PKU, a rare inherited disorder that requires a strict diet and treatment for life. She had been in receipt of the disability living allowance, but now that she has turned 16, she has scored zero in every personal independence payment category. Will the Minister meet my constituent and me so that we can iron out this clear case of "the computer says no"?

Answered by: Sarah Newton | **Party:** Conservative Party | **Department:** Work and Pensions

I would be absolutely delighted to meet the hon. Lady and to go through this constituency case with her.

21 May 2018 | Topical questions - 1st Supplementary | Answered | House of Commons | House of Commons chamber | 641 c558

Date answered: 21 May 2018

2.3 Debates

- Phenylketonuria and Kuvan (HC Deb 22 March 2018 cc482-488)

2.4 Early Day Motions

- ACCESS FOR PHENYLKETONURIA PATIENTS TO TREATMENT, SUPPORT AND INNOVATIVE NEW MEDICINES

That this House recognises the need to raise awareness of phenylketonuria (PKU), a rare metabolic disease which inhibits the ability to metabolise phenylalanine within protein; notes that the disease can cause irreversible brain damage or impaired cognitive or neurological function if not correctly treated; is concerned that the only treatment currently provided by the NHS is a severely restricted diet which places a great burden on patients or carers; acknowledges the impact of PKU on patients, carers and family life; further notes the particular need for care and treatment to support women with PKU in pregnancy and prior to conception to ensure safe pregnancies that minimise stress on women with PKU; further recognises the need to ensure consistent access to special dietary foods, protein substitutes, dietetic and psychological support to assist people with PKU to manage their condition; understands that the medicine Kuvan for the treatment of PKU is due to be appraised for use on the NHS; further understands that Kuvan is widely available for patients with PKU in the majority of European countries; and supports the calls of the charity the National Society for Phenylketonuria (UK) Limited for the appraisal to be concluded swiftly.

08 Mar 2018 | Early day motions | Open | House of Commons | 1044 (session 2017-19)

Primary sponsor: Rashid, Faisal | **Party:** Labour Party

Other sponsors: Austin, Ian · Farron, Tim · Williamson, Chris · Aldous, Peter · Twist, Liz

Number of signatures: 28

3. Press articles

A life without meat, dairy or nuts: Meet the 28-year-old woman who can only eat SIX GRAMS of protein a day due to rare genetic disorder that severely restricts her diet

MailOnline, 18 February 2018

Scots schoolgirl with rare genetic disorder forced to ditch Irn-Bru amid fears new recipe could leave her brain damaged

The Scottish Sun, 6th January 2018

NHS agrees to fund 'life-changing' drug for seven-year-old

BBC News, 29 September 2017

PKU funding battle: Family wins High Court challenge over drug

BBC News, 8 August 2017

Parents of autistic boy, 7, with a rare protein allergy say they are 'greatly heartened' as judge rules NHS must reconsider refusal of funding for 'life-changing' drug after a two-year fight

The Daily Mail, 8 August 2017

What is PKU disease, what are the symptoms of Phenylketonuria and what sort of diet do sufferers need to follow?

The Sun, 18th July 2017

Maternal PKU - Why I'm Having A Burrito Delivered To The Labour Ward

The Huffington Post UK, 28 June 2017

4. Press notices

NSPKU Sugar Tax Press Release

National Society for Phenylketonuria, 8 June 2018

Statement to NSPKU members regarding the appraisal of Kuvan by NHS England

National Society for Phenylketonuria, 15 March 2018

5. Further reading

- Commons Library, NHS Commissioning of Specialised Services, June 2017
- Commons Library, The Soft Drinks Industry Levy, 12 April 2017
- National Society for Phenylketonuria, Low protein foods: how to get what you Prescriptions need, when you need it!, August 2016
- NHS England, Clinical Commissioning Policy: The use of Sapropterin in children with Phenylketonuria, July 2015
- NHS England, Clinical Commissioning Policy: Sapropterin (Kuvan®) For Phenylketonuria: Use in Pregnancy, April 2013

DEBATE PACK

Number CDP-2018-0152

25 June 2018

About the Library

The House of Commons Library research service provides MPs and their staff with the impartial briefing and evidence base they need to do their work in scrutinising Government, proposing legislation, and supporting constituents.

As well as providing MPs with a confidential service we publish open briefing papers, which are available on the Parliament website.

Every effort is made to ensure that the information contained in these publically available research briefings is correct at the time of publication. Readers should be aware however that briefings are not necessarily updated or otherwise amended to reflect subsequent changes.

If you have any comments on our briefings please email papers@parliament.uk. Authors are available to discuss the content of this briefing only with Members and their staff.

If you have any general questions about the work of the House of Commons you can email hcinfo@parliament.uk.

Disclaimer