

Management of Phenylketonuria

A consensus document for the diagnosis and management
of children, adolescents and adults with phenylketonuria (PKU)

July 2014

Management of PKU

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This revision has been approved by the NSPKU Medical Advisory Panel (MAP)

The following comments have been received on 'Management of Phenylketonuria'

"The Paediatric Group of the British Dietetic Association support its production and distribution and are happy to endorse the contents without amendment."

Paediatric Group of the British Dietetic Association

"It is an impressively produced job and should help to raise the profile of the disease and its treatment."

R.J. Pollitt

Retired Professor, Newborn Screening Laboratory, Sheffield

"I do like the document which has just the right amount of detail to appeal to colleagues and Trust Executives."

Dr. Godfrey Gillett

Consultant Chemical Pathologist

The NSPKU has received enquiries about where it stands in relation to new treatments. Below is the NSPKU's policy statement which includes BH4:

The NSPKU believes, without reservation, that people with Phenylketonuria (PKU) should have access to clinically-proven treatments that help them cope with this hereditary condition.

In the UK, people with PKU follow a strict 'PKU diet'. Whilst this works well for many families and individuals, others have difficulties.

BH4 is a clinically-proven treatment for PKU, available in most European Union countries but not in the UK. The NSPKU welcomes every treatment with proven efficacy that may help people with PKU and their families to manage the condition and will advocate accordingly.

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2 Executive summary

The early diagnosis and treatment of PKU, the most common inborn error of protein metabolism, is one of the great medical success stories of the last forty years.

Prior to the introduction of an effective national screening programme in 1969, most affected children sustained irreversible brain damage before diagnosis. Today, babies born with the disorder can expect to have the same career and social ambitions as unaffected individuals.

As knowledge about early treatment in PKU has grown, and the care of patients has improved, providing a national minimum standard of care has become imperative. This document outlines the views of the Medical Advisory Panel of the National Society for Phenylketonuria (United Kingdom) Limited (NSPKU) and provides a framework for the provision of this minimum standard.

This document firstly lists the aims of treatment. The screening, testing and reporting of results, followed by sensitive handling of positive results by a professional with current knowledge, is highlighted. The biochemical monitoring from first diagnosis to adulthood follows, with the acceptable phenylalanine concentrations for given ages. The management of PKU in childhood requires a multidisciplinary approach across the hospital-community interface.

The success of treatment for PKU means that there is now a generation of young adults who have grown up without significant intellectual impairment. However, there is also an increasing knowledge and awareness of the potential later effects of PKU on the adolescent and adult brain, and, consequently recognition of the importance of continued care during adulthood through dedicated transition and adult PKU services.

The needs of female patients are of particular importance because of the associated risks of pregnancy in the untreated mother with PKU. Untreated PKU will almost invariably lead to miscarriage or the maternal PKU syndrome with severe foetal damage should the pregnancy proceed. Strict control of the diet prior to conception and throughout pregnancy is essential to prevent foetal damage and to ensure that the benefits to one generation are not lost to the next.

Finally, the needs of late-diagnosed patients born before effective national screening was established continue to provide a challenge. Evidence suggests that some of these patients may benefit from dietary intervention or treatment. Although their brain damage is irreversible, improvements in behaviour have been observed, enabling these patients to lead a more fulfilling and happier life whilst also easing the burden of carers.

3 Aims

This document has been compiled by the Medical Advisory Panel of the National Society for PKU (NSPKU) and sets out the standards of practice and management which are considered to be reasonable and attainable in the management of PKU. A broad-based consensus approach has been taken as it is recognised that there is national variation in aspects of management. The Medical Advisory Panel recognises that there are constraints upon the resources of purchasers and providers, but, nonetheless, we wish to focus on the ideal standards of care. Specific aims of this document are...

- to specify the services required for the management of PKU from diagnosis by neonatal screening to adulthood including pregnancy
- to outline the general rather than the specific principles of management
- to provide evidence-based advice where possible while recognising that much of current treatment is based upon clinical experience
- to clarify the disciplines and professions which care for patients with PKU and to define their respective roles and responsibilities
- to increase the awareness of purchasers and providers about PKU, and to produce guidelines to assist health authorities to define standards of care which the child or adult with PKU can reasonably expect to receive
- to ensure equity of access to services for PKU according to needs assessment
- to ensure that specialist expertise is provided for children and adults and to recognise the multiple clinical interfaces, eg. between general practitioner and paediatrician and adult physician
- to reduce the risk of neurological damage by the effective control of plasma phenylalanine concentrations by dietary means and to ensure that children with PKU achieve their maximal developmental potential
- to encourage the maintenance of dietary and clinical supervision throughout life
- to ensure that all girls and women with PKU are aware of the need for good dietary control when planning pregnancy and throughout pregnancy
- to ensure full recognition of the prescribing implications now that continuing a PKU diet lifelong is recommended
- to ensure that parents and patients are fully informed and have access throughout life to clearly written and regularly updated information on PKU, provided by health professionals, specialist centres and parent support groups

4 What is PKU?

4.1 Background

PKU is the most common inborn error of protein metabolism with an incidence of approximately 1 in 10,000 births (1 in 4,500 in Northern Ireland). If not correctly treated from soon after birth it results in severe brain damage. Treatment by restricted dietary intake of phenylalanine (PKU diet) has been shown to be very effective.

In the relatively short history of PKU, increasing knowledge, coupled with continual improvements in treatment, has resulted in the possibility of an excellent outcome for today's patient. However, it is now also recognised that there is a risk of late sequelae such as impairment of cognitive and / or neuropsychological function and psychiatric symptoms in adult patients who no longer follow a PKU diet. In addition, foetal damage in untreated, or sub-optimally treated pregnant women is well documented. Therefore, whilst recognising that there is individual variation, current advice is to continue a PKU diet throughout life.

In the UK, the provision of services for the treatment of PKU varies widely from centralised clinics and specialist teams to management by a local family practitioner working in isolation.

The Medical Advisory Panel believes that services should be provided or coordinated from a specialist multidisciplinary team based upon a regional centre, but recognise that geographical considerations may make it difficult for patients to attend specialist centres regularly.

4.2 Multi-disciplinary team

In the lifelong management and supervision of PKU the following health care professionals make an important contribution. The responsibilities and roles of team members will inevitably overlap.

The specialist team should include:

- consultant paediatrician/adult physician with knowledge and experience in metabolic disease
- paediatric/adult dietitian with experience of PKU and metabolic disease
- biochemist/chemical pathologist with responsibility for neonatal screening and biochemical monitoring
- clinical nurse specialist
- clinical psychologist
- social worker
- obstetrician/gynaecologist

Specialist teams based at regional centres should be able to provide a multidisciplinary approach, ideally linked with screening centres, and, in order to maintain expertise, should manage a minimum number of 20 patients. A paediatric dietitian and a metabolic biochemical monitoring service are essential elements of the team approach offered by regional centres. The team should work closely with local practitioners, general paediatricians and other agencies, and produce joint protocols for collaborative management which should be regularly reviewed and updated according to audit principles.

5 Neonatal screening

5.1 Requirements

The fundamental aim of screening is the early detection of PKU to allow the initiation of dietary treatment and the prevention of brain damage and later morbidity. The framework and guiding principles of the present neonatal screening programme for PKU were contained in the guidance note issued by the Department of Health in 1969 which specified that:

- Laboratory testing of blood specimens should be centralised and co-ordinated with facilities for confirmation of diagnosis and treatment
- There should be a recording and tracing system to ensure that all babies are tested and results made known

Although the organisation of the NHS has altered significantly since screening began, the basic principles of screening remain unchanged. A series of audit standards for the various steps in the process have been defined and should ideally be incorporated into service contracts for NHS providers. The UK Newborn Screening Programme Centre was established in 2002 (now known as NHS Newborn Blood Spot Screening Programme) with the overall objective of assuring high quality screening services for babies and their parents.

Audit standards may include those listed below:

5.1.1 Collection of specimen

Parents should be provided with written information about the nature of the test and offered a verbal explanation. All babies born in the UK should be sampled on day 5, and in exceptional circumstances between day 5 and day 8 of life. Blood samples must be dispatched promptly to the screening laboratory. There must be a process for the collection of repeat samples. A low proportion of babies may require a repeat sample for a variety of reasons, such as insufficient blood on the first collection.

5.1.2 Laboratory testing

The screening laboratory should comply with national standards for analytical performance and be validated by external quality control schemes, and be accredited. Babies with abnormal results must be referred rapidly for further investigation and treatment.

5.1.3 Child Health Information System

There must be routine checking that all babies have been screened and have a conclusive test result recorded on the Child Health Information System by 17 days. Normal screening results should be reported to the parents by post or via the health visitor or midwife. Abnormal results following confirmation should also be notified to the NHS Newborn Blood Spot Screening Programme.

5.2 Diagnosis of PKU

All positive results should be communicated by personal contact with the family from a designated health professional who must have current knowledge of PKU. This task may be undertaken by a clinical nurse specialist who has been trained and is part of the multidisciplinary team. In the event of a positive test arrangements need to be made for immediate follow up within twenty-four hours. This should include referral to a consultant paediatrician in a specialist centre who has an expert knowledge of PKU and immediate access to supporting health professionals and laboratory services.

All positive screening tests must be confirmed with a quantitative measure of phenylalanine on a repeat specimen of blood and investigations undertaken to exclude the possibility of a defect in bipterin metabolism. Dietary treatment should commence as soon as the diagnosis is confirmed, certainly by day 17 (core standard) but preferably by day 14 (development standard).

Following confirmation of the diagnosis of PKU, the extent of shared care should be agreed between the specialist centre, the district general hospital and the local family doctor. Centres undertaking screening should ideally be responsible for the monitoring of phenylalanine during treatment. However, in all monitoring laboratories suitable analytical methods should be employed to ensure appropriate accuracy of testing and laboratories should participate in a national quality assurance control scheme. Results should be available in a timely fashion, i.e. within three working days of sample receipt in the laboratory. Reporting mechanisms should ensure that the results are available for the paediatrician, dietitian or a locally identified co-ordinator within each centre.

5.3 Biochemical monitoring of PKU

The following recommendations are based upon the report of the Medical Research Council (MRC) Working Party (1993) on PKU. The MRC publication "Recommendation on Dietary Management of PKU" is a valuable document which attempts to set out standards for the clinical management of PKU.

5.3.1 Frequency of monitoring

- 0 - School entry – weekly
- School age – weekly to fortnightly
- Adolescents/Adults – fortnightly to monthly

More frequent blood samples may be analysed from older children if the parents wish or if the phenylalanine concentrations are high or abnormally low.

5.3.2 Type of specimen

Phenylalanine concentrations can be measured on liquid blood or dried blood spots. In order to make frequent monitoring practicable, carers should be trained to take blood samples at home which can then be posted to a laboratory with expertise in phenylalanine measurement.

5.3.3 Desired blood phenylalanine levels in treated patients

The current acceptable range of phenylalanine is based upon the MRC report. Please note: Evidence based European Guidelines for PKU are under development and these target phenylalanine ranges may need to be amended.

- 0 - 5 years - 120 - 360 $\mu\text{mol/L}$
- Greater than 5 years - 120 - 480 $\mu\text{mol/L}$
- Greater than 10 years - 120 - 480 $\mu\text{mol/L}$
- Adults - 120 - 700 $\mu\text{mol/L}$

However, dietary compliance may become more difficult in adolescence. Parents and adolescents should be aware that performance and general well-being may be impaired at higher phenylalanine concentrations.

5.3.4 Additional biochemical monitoring of nutritional status

This complements the clinical assessment of nutrition and anthropometry. Periodic testing is indicated if there are concerns about nutritional inadequacies. It may be appropriate to measure plasma vitamin and mineral concentrations. Vitamin B12 concentration should be measured annually in adolescents and adults on a diet in which protein intake is low, but who are not taking a specific PKU protein substitute (also known as an amino acid supplement) with vitamin supplements.

6 General management of PKU

This requires a multidisciplinary approach across the hospital-community interface. Professional roles and responsibilities vary throughout the country and local practices have evolved according to the availability and expertise of health care professionals. All those involved in the care and management of patients with PKU are expected to take part in research as necessary.

6.1 Role of the paediatrician

The fundamental and principal role of the paediatrician is to liaise and co-ordinate the activities of the other health care professionals. The paediatrician should have specialist knowledge of metabolic disease in childhood, and work in a specialist centre where there is expertise in paediatric dietetics and laboratory support for the monitoring of phenylalanine. In some areas of the country shared care with a local paediatrician would be appropriate. Roles of the paediatrician are...

- to organise regular clinical review and support. The outpatient schedule of attendance will vary, but the following options are considered to be a minimal requirement. More frequent review may be helpful during the early years
 - less than 2 years – 3 monthly review
 - less than 5 years – 4 monthly review
 - greater than 5 years – 6 monthly review
 - adults – annually
- to provide basic genetic counselling
- to educate the parent and child by the provision (at relevant times) of written information and advice
- to ensure that girls with PKU and hyperphenylalaninaemia have awareness from an early age of the need for treatment during the preconception period and pregnancy
- to monitor biochemical control by ensuring the regular measurement of phenylalanine concentrations at home or in the clinic
- to provide sample cards, blood sampling equipment, and initial training in the technique of sample collection and to establish a communications system in order that the results may reach the patient / carer in a timely manner
- to ensure access to other specialist services according to need
- to consider and advise on issues of education, careers and employment
- to monitor growth and development
- to liaise with the general practitioner regarding prescriptions

6.2 Role of the dietitian

It is essential that a senior dietitian with experience in paediatrics and PKU cares for children with PKU and provides ongoing continuity of care. Ideally, the dietitian should care for a minimum of 20 patients. Roles of the dietitian are...

- to provide initial teaching of parents (possibly in conjunction with a dietetic assistant where available) in the dietary principles of the management of PKU – parents need to be aware from an early stage that life long dietary treatment is probable
- to ensure nutritional adequacy of the diet, monitoring growth and supporting the family at key stages, e.g. early feeding, weaning and school entry
- to provide education for the family and school and ultimately for the transfer of diet responsibility to the child (this should include awareness of the need for preconception / pregnancy treatment for girls)
- to liaise with the family on results of biochemical monitoring, etc
- to liaise with the school on packed lunches, menu, etc; the general practitioner and community pharmacist on special products, food lists and prescriptions; the paediatrician with an interest in metabolic disease and the health visitor
- to offer patient home visits as needed. Although resource intensive, this enables a more accurate assessment of patients in their own environment
- to regularly adjust the diet according to biochemical control and to provide advice on low protein products, phenylalanine-free foods, phenylalanine exchanges, and protein substitutes, etc
- to develop, revise and disseminate written dietetic resources to families and relevant health professionals
- to potentially provide treatment during pregnancy in the absence of an established adult service – however, this should now rarely be required in the UK and a referral to an appropriate adult service is always preferable

6.3 Role of the dietetic assistant

- to support the role of the dietitian
- to educate patients, their families and carers on the practical aspects of the PKU diet
- to develop recipes and menu plans with patients, taking into account different cultural food needs

6.4 Role of the biochemist

- to organise biochemical testing as part of the definitive diagnosis of PKU, eg. quantitative phenylalanine bipterins
- to provide timely biochemical monitoring for clinical management and appropriate advice
- to provide input into the multidisciplinary team providing patient management
- to provide support for a home monitoring blood collection service
- to ensure that patients are registered with the NBSPC

6.5 Role of the clinical nurse specialist

A specialist nurse with experience and knowledge of all aspects of PKU is available in certain centres. Such a post should be established in each specialist centre to provide an outreach service from the hospital to the community. Roles of the nurse specialist are...

- to liaise with other health professionals about initial diagnosis and ongoing care
- to provide initial contact with parents (if possible by a joint visit with GP) following a positive screening result
- to teach the technique of blood sampling for home monitoring
- to provide a supportive role in early childhood
- to provide sex education, family planning advice and pregnancy counselling in collaboration with others
- to provide families with information regarding national and local support groups
- to support patients not attending a specialist centre, in collaboration with their consultant and dietitian
- to support pregnant women

6.6 Role of the clinical psychologist

Children with classical PKU represent a high risk group for impairment of intellectual function. As the ultimate goal of treatment is the prevention of mental retardation, it is essential that the developmental course of the child is closely monitored. Children with PKU should be assessed as indicated by means of serial psychometric assessments and evidence sought for results that might be attributable to high / fluctuating phenylalanine levels. Treatment by dietary restriction involves considerable modification of normal eating behaviour by the child and cooking behaviour by the carer. Motivation to comply with diet depends on psychological factors such as knowledge of the condition, understanding of the treatment goals, reassurance that the diet is working successfully and resistance to peer and other pressures to deviate from the strict regimen. Roles of the clinical psychologist are...

- to provide serial developmental assessments from infancy to adulthood. This should include assessments of dyslexia and dyscalculia
- to determine the extent to which indices of development reflect phenylalanine control as opposed to psychosocial or other factors
- to assist patient and parent understanding of the need for dietary treatment
- to foster patient and parent motivation to comply with treatment
- to liaise with educational psychology services regarding aspects of learning and behaviour that might be related to PKU or its treatment

6.7 Role of the social worker (sometimes performed by the clinical nurse specialist)

- to inform families of national and local support groups
- to review benefits to which the family may be entitled

Other professional roles

6.8 Role of the general practitioner

The family doctor is responsible for the co-ordination of care within the community and the overall management of the child and family. Roles of the GP are...

- to ensure the prescription of protein substitutes and low protein foods in sufficient quantities to minimise the need for frequent attendances and repeat prescriptions. This includes the prescription of low protein foods and protein substitutes for adults who wish to return to a PKU diet at a later stage in life.
- to ensure effective liaison with all health care professionals
- to ensure that adolescents and adults with PKU have access to specialist services; to advise on family planning and refer for preconception and pregnancy management

6.9 Role of the health visitor and midwife

The responsibility of health visitors and midwives for screening is subject to regional variation. Roles of the health visitor are...

- to undertake the collection of initial and/or repeat blood samples for neonatal screening and to despatch these expeditiously
- to provide information to the mother and family at the time of initial sample collection as to the nature of the test (leaflets are available)
- to confirm that all babies have had a neonatal screening test and that the result is recorded within the child's health record according to agreed procedures
- to act as liaison between the hospital and community

6.10 Role of genetics services

PKU is inherited as an autosomal recessive disorder. Couples who have an affected child have a one in four recurrence risk for each further pregnancy. Following the birth of an affected child with PKU, options for future pregnancies and possible impact on other family members should be discussed. All families should be offered genetic counselling and should be referred to regional genetic counselling services if appropriate.

6.11 Role of support groups

These groups provide an opportunity for families to meet others with PKU. They offer social and practical support through the exchange of ideas and information on diet, etc., via newsletters, chat forums, conferences and educational holidays. They provide a forum to lobby nationally on behalf of patients for an equitable share of health care resources for the management of PKU.

7 Adults and adolescents: special considerations

7.1 Clinics for adults and adolescents

These exist in only a few centres in the UK and provide for the transfer of care of children with PKU to adult services. An adolescent/adult transition clinic is the ideal. This should be run jointly by a paediatrician and an adult physician, both with an interest in metabolic disease.

Paediatric specialist centres should consider, in collaboration with purchasers, a strategy for the provision of long term services for patients with PKU. Where it is current practice to care for adults and adolescents in paediatric centres this should continue until effective alternatives have been established.

7.2 Frequency of attendance at adult clinics

This will depend upon the degree of dietary restriction. Stable patients on a phenylalanine restricted diet should be reviewed at least annually. In view of reports of cognitive and/or neuropsychological impairment and psychiatric symptoms in a minority of patients, adult patients not following a PKU diet still require clinical review and should also be seen annually. Vitamin B12 concentrations should be measured annually in those on a protein restricted diet but who are not taking specific PKU protein substitute with vitamin supplements.

7.3 Prescription charges for adults

These become payable after the eighteenth birthday with exemptions for those unemployed or pregnant. Prescription charges can create difficulties for many families particularly for treatment during the pre-conception period. Adults will require advice about the most cost effective way to meet prescription charges.

7.4 Lifelong treatment for PKU

Currently lifelong dietary treatment with phenylalanine restriction is recommended for the majority of patients as it is recognised that some patients who have discontinued PKU dietary treatment after childhood have subtle but significant neurological and psychological problems which limit function and performance.

However, the NSPKU recognises that some adults, for various reasons, choose not to maintain lifelong dietary phenylalanine restriction and will return to a normal unrestricted diet. Adult services should continue to provide support to these patients and to offer regular annual follow-up appointments.

Similarly, individuals with hyperphenylalaninaemia, who are not on a PKU diet, should be offered ongoing outpatient follow-up eg. every two years. This is particularly important for women with hyperphenylalaninaemia who may require dietary intervention when planning a pregnancy.

7.5 Preconception counselling

Counselling of women before pregnancy on the foetal risks associated with a high plasma phenylalanine concentration is essential. Ideally family planning advice should be given by the paediatrician and clinical nurse specialist before transfer to an adult clinic. Reiteration of this advice and the provision of written information is important throughout adolescence. Later, at the adult clinic, specific risks of the maternal PKU syndrome (ie. congenital malformation, intellectual impairment, and behavioural problems in the children of untreated/sub-optimally treated pregnant women) should be fully explained.

7.6 Maternity care

Women with PKU and their partners who wish to start a family should be seen by clinical and dietetic experts at a specialist centre or adult clinic. Reintroduction of diet or the (re)establishment of strict dietary control is essential before conception.

With some local variation, the aim of dietary treatment is to maintain the woman's blood phenylalanine concentration between 100 and 300 $\mu\text{mol/L}$, prior to conception and throughout pregnancy in order to prevent the maternal PKU syndrome in the foetus.

The dietitian, with support from a dietetic assistant where available, will provide (re)education regarding the principles of dietary protein restriction and provide practical advice on diet preparation. Arrangements will be made for regular blood testing. Blood phenylalanine should be measured at least 2-3 times weekly during pregnancy. Full amino acid profile, vitamins, and full blood count should be measured during each trimester. On occasion, hospital admission may be necessary for dietary stabilisation.

Foetal ultrasound and scanning should be scheduled as per routine antenatal care unless there are clinical concerns about intrauterine growth retardation or other foetal problems. Many obstetricians will not have managed a pregnant woman with PKU previously and it is helpful to provide them with written information on the basic principles of management and potential foetal risks.

7.7 Children born to mothers with PKU

Children born to mothers with PKU should be assessed for PKU as per the routine neonatal screening programme. Initial examination of the infant should be by a neonatologist aware of the clinical features of infants born to mothers with PKU. If resources permit, it is recommended that a clinical psychologist should conduct assessments at 18 months, four, eight and fourteen years to identify any developmental or cognitive problems and suggest additional educational support if required.

7.8 Role of the clinical psychologist in adult life

The roles of the psychologist in adulthood are...

- To conduct formal neuropsychological assessments to document any cognitive deficits in attention or processing information that might be attributable to high/ fluctuating phenylalanine levels. Typically those adults that have been off diet and are re-commencing, are seen by a psychologist before they re-start and again once they have been on the diet for some time to document any change over time
- To help support the adult to remain on the diet if they wish to and are finding it difficult to do so
- To promote psychological and emotional well-being in adulthood by direct work with individuals or by signposting them onto local services
- To assist the individual and their family in promoting independence into adulthood

Adults with PKU may report difficulties with emotional well-being and can struggle with issues relating to dependency on others as they enter adulthood and greater demands are placed upon them. Anxiety and depression may be reported and require support. In general these can be treated in the patient's local area with guidance given to local therapists.

Women who are trying to conceive and pregnant women with PKU may need additional psychological support during this time.

7.9 Dietary treatment of the late-diagnosed (untreated) adult with PKU

Older patients, not identified by newborn screening, who may never have had dietary protein restriction or have only been treated for a brief period, create special practical problems of management as many have learning difficulties and behavioural problems. The role of dietary protein restriction and treatment in these patients is uncertain. There is currently no clear data as to who will benefit from diet. Starting restrictive diet after brain damage has occurred will not reverse damage, but research indicates that the diet may be beneficial in other ways. Case studies have shown that there may be improvement in observed behaviour with reduced aggression, self-injurious behaviour and hyperactivity, with more positive mood and increased social awareness, and an increased attention span.

However, untreated adults who have previously been on a normal diet may find the restrictive nature of the PKU diet difficult to accept, and staff and carers will need to be trained to cope with the preparation of the diet and subsequent monitoring to ensure compliance. Therefore, the diet should only be given to patients under close medical supervision (dietetic involvement is essential). Prior to implementation a baseline for behaviour, cognitive functioning, nutritional intake and phenylalanine concentrations should be established.

7.10 Previously treated adult patients returning to a PKU diet

A number of adults, diagnosed on newborn screening, who may have stopped the PKU diet in adolescence, will opt to return to dietary treatment at a later stage of life – often because they present with symptoms related to their mood, energy levels or concentration. Such patients should have a formal neuropsychometric assessment and be given full support as above to return to a PKU diet.

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The NSPKU exists to help and support people with PKU, their families and carers. The NSPKU actively promotes the care and treatment of individuals with PKU and works closely with medical professionals in the UK. It organises events such as conferences and study days throughout the UK, produces a wide range of publications (including food lists) for parents, patients and medical professionals. Local support groups also hold study days and other events.

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