

Dietary Treatment of the Untreated Adult PKU



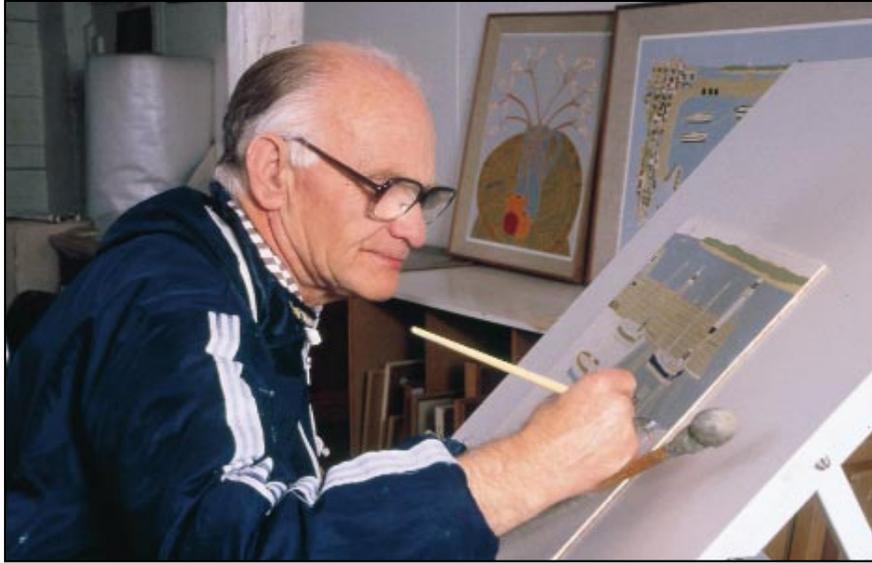
Bryan Pearce – St. Ives Bay with Cargo Ship 1995

Written by Rosemary Hoskin, SRD
Senior Dietitian, Horizon NHS Trust, Harperbury
Radlett, Herts in conjunction
with the Medical Advisory Panel of the
National Society for Phenylketonuria



The National Society for Phenylketonuria (United Kingdom) Ltd.

7, Lingey Lane, Wardley, Gateshead, NE0 8BR



Bryan Pearce working in his studio.

Bryan Pearce is a famous primitive artist. He was born in 1929 and is affected by phenylketonuria, never having been on a diet. However, he found fulfilment in painting at the age of 24 when his mother bought a child's colouring book and paints in a desperate effort to find something useful for him to do and to develop his motor skills. Bryan took to painting as though a lifeline had been thrown to him.

Some of his paintings hang in The Tate Gallery in St. Ives.

Excerpts taken from Marion Whybrow's book on two naïve painters of St Ives to be published Summer 1998 used with kind permission

The painting on the cover is Bryan Pearce's St. Ives Bay with Cargo Ship 1995. Oil on Board 22" by 28". Reproduced by kind permission of the artist ©.

DIETARY TREATMENT OF THE UNTREATED ADULT PKU

INTRODUCTION

This leaflet aims to provide advice for those who want to try dietary treatment for the untreated adult with PKU and describes the potential benefits which may result. The information in this leaflet has been collected from both published and unpublished sources. The principles of the diet are outlined and practical advice is given on important aspects such as baseline measures before dietary changes are made.

A General Practitioner or Consultant and a State Registered Dietitian should always be involved and agreement to treatment should be obtained from the family. Anyone contemplating further work is advised to contact the NSPKU who can provide contact details of other workers in the field.

WHAT IS PHENYLKETONURIA?

Phenylketonuria (PKU) is a rare, inherited disorder which prevents the normal metabolism of the amino acid phenylalanine found in all protein containing foods. Protein, one of the main components of our diet, is made up of different substances called amino acids. The body digests protein and breaks it down into its constituent amino acids and di-peptides which are then absorbed and used for growth and tissue repair of the body. Phenylalanine is one of the amino acids which make up protein and normally any excess phenylalanine is converted into another amino acid called tyrosine. Some or all of this conversion does not take place in PKU and the phenylalanine accumulates in the blood. Untreated, this condition leads to mental retardation.

Every child born in the United Kingdom is offered screening for PKU by a blood test, usually at about 6 days. This service began in the late 1960s. Prior to that, screening using urine was carried out in some areas of the UK. The prevalence of PKU is approximately one in 10,000 live births. Following diagnosis affected babies are immediately put on to a low phenylalanine diet, provided there is good dietary compliance during the critical period of early development, the prognosis for normal development is excellent.

This means that many individuals with PKU born prior to nationwide screening were not diagnosed until mental retardation was evident. Consequently, many individuals with PKU have spent their lives in long stay hospitals for people with learning disabilities, group homes or at home needing constant supervision. In addition to learning difficulties, high blood phenylalanine levels may cause other problems. Some individuals exhibit behavioural problems, for example – aggressiveness, hyperactivity, self-injurious behaviour, sleep problems and they may also suffer from skin conditions such as eczema.

CAN DIETARY RESTRICTION OF PHENYLALANINE BE BENEFICIAL TO THE UNTREATED PKU?

On the present information available (see references) some untreated PKUs may benefit from late dietary intervention, yet it is not clear how to predict who will benefit from the diet and who will not.

Starting the restricted diet after brain damage has occurred will not reverse the damage, however current research indicates that the diet may be beneficial in other ways. Case studies on untreated adult PKUs have shown some or all of the following improvements:

- 1) physical
 - improvement in eczema and other skin conditions
 - decreased body odour
- 2) behavioural
 - reduced aggressive and self-injurious behaviour
 - reduced hyperactivity
 - more positive moods
 - increased social awareness
 - increased attention span thus allowing the individual to gain from behavioural intervention
- 3) medical
 - e.g. reduction of psychotropic medication

WHAT ARE THE BASIC PRINCIPLES OF THE DIET?

To lower the blood phenylalanine levels the amount of phenylalanine in the diet has to be reduced. To achieve this the quantity of protein from normal foods is severely restricted.

- a) Meat, fish, eggs and cheese are rich in protein and therefore phenylalanine, so they are not allowed. Milk is also rich in protein but is sometimes included as an exchange to make the diet more acceptable see, b) below. These foods are replaced with an amino acid supplement from which the phenylalanine has been removed e.g. XP Maxamum (which includes vitamins and minerals), Phlexy 10 (which requires supplemental vitamins and minerals) or Aminogram (which requires supplemental vitamins and minerals).

The amino acid supplement is an essential part of the diet for anyone with PKU and must be taken regularly with meals and evenly spread throughout the day.

- b) Other foods which contain some protein, such as potato, milk and cereals, are given in small measured quantities (exchanges). A system of exchanges has been found to be a practical approach to the dietary management – one exchange is equal to 1g of protein or 50mg of phenylalanine. These exchanges are spread out as part of the day's meals and/or snacks. Examples of one exchange are 15g Cornflakes, 80g of boiled potato or 45g of boiled rice. The exact number of exchanges used long term will depend upon the level of blood phenylalanine which maintains improvements in the quality of life.
- c) Most fruit and some vegetables can be taken in normal quantities. Sugar, jam and fats, such as butter, lard and cooking oil can be used quite freely. There are some manufactured foods which are low in protein and available for the person with PKU on prescription. These include low-protein bread, biscuits, flour, low protein milk substitute, spaghetti and other pasta. These are important to provide sufficient energy in the diet and to add variety.

All the normal ways of cooking (frying, baking etc) can be used and herbs, spices and flavourings can be used to add interest to the diet.

The diet comprises:

- **Phenylalanine Free Amino-Acid Supplement***
- **Phenylalanine Exchanges**
- **Free Foods**

* **vitamin/mineral supplements may or may not be needed**

From current case studies it appears that starting with 8-11 phenylalanine exchanges will reduce the phenylalanine level in the blood to the level at which general improvements may be seen. Improvements usually occur with phenylalanine levels below 600-700 $\mu\text{mol/l}$.

However, some patients require levels to be down to 400 $\mu\text{mol/l}$ before improvement is seen and on rare occasions levels need to be as low as 200 – 300 $\mu\text{mol/l}$. Mood swings have been observed when levels have fallen below 200 $\mu\text{mol/l}$.

Blood levels usually start to reduce quickly ie. in the first two weeks on diet, but these lower levels need to be maintained for at least six months before the subject shows consistent improvement in behaviour, though improvement in attention span, eye contact and social awareness may be seen earlier.

If dietary intervention is being considered, then a State Registered Dietitian must be consulted for further details regarding the diet. The NSPKU also produces various leaflets and other literature which are a valuable source of dietary advice.

ADDITIONAL FACTORS TO BE CONSIDERED

- 1) The diet should only be given to subjects under close medical supervision. Detailed information of the PKU diet should be obtained prior to intervention.
- 2) Eating patterns/nutritional intake need to be assessed by a State Registered Dietitian prior to dietary changes.
- 3) Prior to starting the diet the levels of challenging behaviour of each subject need to be defined and quantified for the baseline. This will enable the change, or lack of change, in behaviour to be measured.
- 4) Baseline measures of health e.g. eczema and mental functioning/capability to achieve daily tasks need to be established.
- 5) Initial blood phenylalanine levels need to be established before introduction of the restricted diet. Once the diet is introduced they need to be measured weekly until there is an improvement in behaviour and other parameters, accompanied by a consistently reduced phenylalanine level. On the present information improvements should be seen if the phenylalanine level is below 600 $\mu\text{mol/l}$. However, as previously stated, some patients require levels to be down to 400 $\mu\text{mol/l}$ before improvement is seen and on rare occasions levels need to be as low as 200 – 300 $\mu\text{mol/l}$.

If a consistently low phenylalanine level is achieved over a six month period without clinical or behavioural improvement consider discontinuing the diet.
- 6) Thereafter blood phenylalanine levels need to be monitored monthly for the subsequent six months and then once every three to six months – provided that there are no adverse changes.
- 7) The multi-disciplinary team and carers supporting the subject must be fully committed to collecting the relevant data for evaluation purposes and to the project itself.
- 8) Staff/carers working with the client may need special training in the evaluation procedures.

- 9) Ideally significant changes in the subject's life should not be made, e.g. medication or environment whilst the diet is being introduced and evaluated.
- 10) Regular review meetings need to be held on the subject's progress to maintain a good team approach in support of the subject.

PROBLEMS WHICH MAY BE ENCOUNTERED

- Expectations may not be met as not all subjects will benefit from dietary intervention. Current research suggests that approximately half may benefit (Ref 1).
- Untreated adults who have previously been on a free diet may find the restricted diet difficult to accept, in some cases this is due to –
 - 1) restriction of familiar foods
 - 2) acceptability of low protein products
 - 3) palatability of amino acid supplements
- The diet and amino acid supplement required is expensive – ensure that funds are available from the GP if the subject is in the Community or from the Hospital if a resident.
- Staff/carers will need to be trained to cope with the preparation of the diet and subsequent monitoring to ensure compliance.
- Agreement from the family/close relative should be obtained and may be needed from the local ethics committee if part of a research study.

SUMMARY

- 1) Prior to implementing the diet, establish a baseline in conjunction with a Psychologist, Doctor and state registered Dietitian for –
 - * behaviour
 - * health
 - * mental functioning/capability of the subject
 - * blood phenylalanine levels
 - * dietary intake
- 2) When introducing a low phenylalanine diet the use of a phenylalanine free amino acid supplement is essential – this should be supervised by a State Registered Dietitian. NB A low protein diet without a phenylalanine free amino acid supplement and a vitamin/mineral supplement could lead to serious nutritional inadequacies and may not be effective in maintaining low phenylalanine levels long term.
- 3) Monitor carefully any changes in the original baseline levels, as the phenylalanine levels in the blood fall, including any adverse reactions.

Establishing a low phenylalanine diet in the untreated adult PKU is difficult but in some cases may be very worthwhile. Significant behavioural changes may occur as the blood phenylalanine levels fall, resulting in benefits which can outweigh the difficulties of implementing a low phenylalanine diet. Improvement may also be seen in skin conditions e.g. eczema.

Further work is continuing in this field and anyone contemplating dietary treatment should endeavour to obtain the most up to date advice – contact the NSPKU.

REFERENCES

- 1 **YANNICELLI S and RYAN A (1995)**
Improvements in behaviour and physical manifestations in previously untreated adults with phenylketonuria using a phenylalanine restricted diet.
Journal of Inherited Metabolic Disease, National Survey 1995
Vol. 18 No.2 131-134
- 2 **HARPER M and REID A H (1987)**
Use of a restricted protein diet in the treatment of behaviour disorder in a severely mentally retarded adult female phenylketonuria patient.
Journal of Mental Deficiency. Research Vol. 31 209-12
- 3 **MARHOLIN D, POHL R E, STEWART R M, TOUCHETTE P E, TOWNSEND N M and KOLODNY E H (1978)**
Effects of diet and behaviour therapy on social and motor behaviour of retarded phenylketonuria adults :- an experimental analysis.
Paediatric Research Vol. 12. 179-87.
- 4 **HOSKIN R G, SASITHARANT and HOWARD R (1992)**
The use of a low phenylalanine diet with amino acid supplement in the treatment of behavioural problems in a severely mentally retarded adult female with phenylketonuria.
Journal of Intellectual Disability Research Vol. 36 183-191
- 5 **HARVEY E L and KIRK S F (1995)**
The use of a low phenylalanine diet in response to the challenging behaviour of a man with untreated phenylketonuria and profound learning disabilities.
Journal of Intellectual Disability Research Vol. 39 520-526
- 6 **KOCH J H (1997)**
Robert Guthrie The PKU Story pages 155-158
- 7 **DOLAN B.E., KOCH R. et al (1998)**
Diet intervention guidelines for adults with untreated PKU
National PKU News. California U.S.A.

- 8 **BAUMEISTER A.A. AND BAUMEISTER A.A. (1998)**
Dietary treatment of destructive behaviour associated with hyperphenylalaninemia
Clinical Neuropharmacology 21 No1 pp18-27
- 9 **KOCH R. et al (1999)**
Long-term beneficial effects of the phenylalanine restricted diet in late diagnosed individuals with phenylketonuria
Molecular Genetics and Metabolism 67, 148-155
- 10 **LEVITAS A.S. (1999)**
Phenylketonuria (PKU) and the Hyperphenylalaninemias 111
Psychiatric and behavioural aspects
Mental Health Aspects of Developmental Disabilities Vol.2. No 4
- 11 **FITZGERALD B., MORGAN J. et al (2000)**
An investigation into diet treatment for adults with previously untreated phenylketonuria and severe intellectual disability.
Journal of Intellectual Disability Research Vol.44 Part 1 53-59

Produced by the National Society for Phenylketonuria
and its Medical Advisory Panel. With special thanks to Rosemary Hoskin,
Senior Dietitian, Harperbury Hospital Hertfordshire



The National Society for Phenylketonuria (United Kingdom) Ltd.

The Society is a registered charity.
It offers support to PKUs and their families by producing various
publications including a quarterly newsletter, organising formal and
informal meeting and conferences.

SHS
INTERNATIONAL

THE NATIONAL SOCIETY FOR PHENYLKETONURIA IS VERY GRATEFUL TO SCIENTIFIC
HOSPITAL SUPPLIES U.K. FOR THEIR SPONSORSHIP OF THIS BOOKLET.

SPECIALIST CLINICAL NUTRITION

Further information and detail can be obtained by contacting:
The NSPKU Helpline on: 0845 603 9136
Email: nspku@ukonline.co.uk
or writing to:
The National Society for Phenylketonuria (United Kingdom) Ltd.
7, Lingey Lane, Wardley, Gateshead, Tyne & Wear NE10 8BR

Charity No. 273670

© Copyright NSPKU

Company No. 1256124

<http://web.ukonline.co.uk/nspku>

April 2000/2k