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Programme Information & Abstracts

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The Neuropsychometric Outcome of Adults with PKU On and Off Diet

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Background

One important question in the management of phenylketonuria (PKU) is how long people should stay on diet. Cognitive outcome including level of intelligence has been linked to dietary control during childhood. Research has shown that starting diet early and remaining on it until adolescence usually leads to a fairly good outcome. Recent guidelines suggest that people should now remain on diet for life, but little work has been done to assess the benefits of this. Our research investigated this by comparing adults who have remained on diet lifelong with those who stopped the diet in adolescence.

Methods

The study groups were adults with PKU who started diet early and remained on diet, adults with PKU who started diet early and stopped in adolescence, and a control group of adults without PKU. All participants were compared on a set of tasks to examine cognitive performance. They also completed questionnaires about quality of life, anxiety and depression.

Findings

Adults with PKU who remained on diet performed slightly better than those who stopped diet in adolescence on some cognitive measures; the groups did not differ on other measures, or on the questionnaire measures of quality of life, anxiety or depression. The adults with PKU who remained on diet performed slightly below the level of the control group on some cognitive measures, but did not differ on others.

Conclusions

The findings suggest that staying on diet beyond adolescence may bring some benefits in terms of cognitive functioning, although these are relatively subtle.

Taking blood samples

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When first told your child has PKU, it can only be imagined by those unaffected what feelings, fears and anxieties a parent experiences. Firstly there are different milks to contemplate and then a diet that is extremely restricted. Literature presented, although not as bleak as in the past, still contains those nasty words "brain damage". To cap it all a discovery that regular blood spot samples are needed must make you feel even more uncomfortable. How can your tiny little bundle be put through all of this? Nevertheless one year on, most of our families are competent and confident in carrying out all of these tasks.

How is this managed?

In our region, when first meeting our families, we talk about diet, very effective treatment and the support our service can offer whilst managing the trials and tribulations of a child on a protein restriction. At this stage a blood sample from the baby's vein is collected, diagnosis confirmed and the family sent home with lots of information and contact numbers. As the nurse specialist, I would then meet families in their own home two or three days later. We will go through all the information given on the first visit and answer any questions that will have been thought of. I would then introduce the family to collecting blood spot samples on their baby. It is a process that is not rushed, we agree it is not the nicest thing to have to do to your baby but is a very positive process that provides us with very important phenylalanine information. We talk through the process and then I would collect the sample with both parents observing. My suggestion is always that both parents learn the process for two reasons; firstly if the person who collects the sample becomes unwell, it is important someone else is able to perform the test. The second is that as the baby becomes a toddler, they will have the odd day that they don't want to have their blood sample collected and it is unfair for a bad

accurate measurement of the level of phenylalanine in the blood. It does not matter how good the laboratory analysis is, if there is a poor blood spot, the answer can be misleading. However, it is very important that when presented with a good blood spot the laboratory measures the phenylalanine and the tyrosine accurately. It is 42 years since the original bacterial inhibition assay, developed by Robert Guthrie, for neonatal screening and monitoring of PKU. The laboratory methods have improved dramatically over the last decade ensuring continued quality in PKU screening and blood phenylalanine monitoring. I will demonstrate how the measurement is now performed in the majority of centres in the UK and emphasise the relationship between the clinical and laboratory teams that can be so successful in achieving the best nutrition for every patient.

