

Welcome to
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Programme Information & Abstracts

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The National Society for Phenylketonuria (United Kingdom) Ltd.

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The Neuropsychometric Outcome of Adults with PKU On and Off Diet

Shelley Channon
University College London

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

Jane Gick, Paediatric Metabolic Nurse Specialist, Evelina Children's Hospital, Guy's & St. Thomas' NHS Foundation Trust.

R Neil Dalton, Senior Lecturer in Paediatric Biochemistry, The WellChild Trust Laboratory, Evelina Children's Hospital, King's College, London/Guy's & St. Thomas' NHS Foundation Trust

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

mummy and a good daddy or visa versa during these times and can lead to samples not being done. If families feel confident they can perform the test alone following my first demonstration, I suggest the next date for sample and leave them to try sample collection, I will then phone to see how they have managed. In days of old I would return to the house to observe sampling at the next sample date but on the whole most families wanted to try without a professional standing over them. If the sample is managed well, they are on the next step forward to gaining control and independence of their child's condition. If it hasn't gone well, it will the next time, or the one after. There is no rush, just support and reassurance. We haven't used G.Ps or community nurses to perform sampling because it will inevitably time restrict the family to appointments and deadlines that they may not want to be tied to. For families attending the conference today who are not comfortable collecting their child's blood spot sample, I hope some of the points mentioned would reassure.

Sampling devices over the years have made a huge difference to both children and parents alike. The gentler approach of using pens that do not reveal a sharp needle has undoubtedly made life easier for those collecting the sample.

Throughout the country individual service support for parents, children and adults are varied. If you are a family that has help with practice nurses or G.P collecting the sample, ask what support the person collecting the blood spot could offer you, if you wish to learn sampling yourself. Speak to families at conference who routinely collect blood spots from their children. Being self-sufficient is usually much more favourable.

I hope through demonstration of blood spot sample through a short talk with pictorial guide some of these anxieties can be alleviated. As health care professionals, we all appreciate the difficulty parents are faced with when asked to perform blood spot samples. However the benefits of maintaining good phenylalanine control can only be monitored by this method. The best way forward in mastering the skill is to be positive in what you are doing. Being able to do your own or your child's sample allows for greater independence for getting on with life.

The measurement

Collection of the blood spot is the most important step in obtaining an

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.

Research and Developments in Phenylketonuria

Maureen Cleary
Consultant Metabolic Paediatrician
Great Ormond Street NHS Trust Hospital
London

Newborn screening began in the United Kingdom in the 1960's. However, forty years on we are still learning much about the condition. The main developments in the last year will be presented. These can be divided into: **New Technologies in imaging the brain in PKU, New Therapies in PKU and New Ideas in delivering treatment.**

The newest technology in brain imaging is Magnetic Resonance Spectroscopy which shows the chemical peak of phenylalanine in the brain. This measurement can be compared with blood levels and any changes in brain phe levels occurring through dietary alteration can be recorded. The technology is still under development, however, and problems with the interpretation will be discussed.

The new therapies are biopterin and ammonia lyase. Biopterin treatment is being tested this year in an international multi-centre trial. An update on ammonia lyase research will be presented.

It is important to consider how we offer treatment to PKU families and to consider whether different approaches serve some families better. Examples of this are group clinics for PKU where specific topics such as dietary ideas can be presented in detail.

Oh No Not time for Maxamaid

Dr Anna Brazier
Consultant Clinical Psychologist
University Hospital of Wales
Cardiff

Claire Rowlands
Mother of Tesni (4)

'When she was two, my daughter Tesni decided that she wasn't going to 'do' Maxamaid any more. After months of battling and trauma our dietitian referred us to Anna Brazier, a clinical psychologist. My feelings at this point were mixed – I certainly needed help, but the thought of visiting a psychologist was daunting!

I needn't have worried. Anna and I discussed the 'issues' we had been having, not only at Maxamaid time, but also with other aspects of Tesni's behaviour such as the battle of the seatbelt and the toothbrush wars. Through chatting to Anna, I soon came to realise that Tesni was the boss in our house, and that if she refused to do something it wasn't worth fighting about, as we could never win.

Anna suggested that we keep a diary over a couple of weeks of who did what, when and our reaction. When we looked through the notes we soon realised that Tesni was being rewarded (with lots of attention) for *not* drinking her Maxamaid. Together, Anna and I discussed strategies that we could use to turn the situation around.

Through a combination of simple, invariable house rules, rewarding 'good' behaviour with positive consequences and ignoring 'bad' behaviour altogether, we managed to arrive at a point where Tesni now drinks most of her Maxamaid on most days and – mostly – without fuss.

This is how a clinical psychologist helped us. Of course, every child and every situation is different and what worked in our case might not be for everyone, but there are always ways to turn behaviour round if you can take a step back and maybe talk things through with someone else.'

Clinical Child Psychologists take a broad approach to whatever difficulties are brought to them. In practise that means talking about behaviour (who does what when), talking about a child's development, temperament and family relationships, and about the particular challenges of the child's medical condition. Naturally the talking will focus on different aspects of family life depending on the difficulties encountered. Usually some time is spent trying to understand the difficulty and the context in which it happens (and doesn't happen). Very often a discussion together with taking time to think, allows parents to come up with ideas, after all it is they that know the child best. The aim of a session will often be to get the discussion going in a problem solving way; and to take the time to consider the emotional impact of having a child with a medical condition. Sometimes in busy outpatients clinics it can seem as if everyone is just getting on with it without a problem and parents can feel quite isolated.

Through the presentation we will aim to show how stepping outside a situation and thinking about the different factors that contribute to children's behaviour can help parents help their children to manage what can be a demanding medical regime.

Summary Of 'LIFE WITH PKU'

Owen Brown

Introduction:

Hello everyone, it's nice to meet you all. My name is Owen Brown and I have been asked to talk to you about my life with PKU. I am 21 years old and live with my parents in the Scottish Borders. I am currently a HNC modern musicianship student at Jewel & Esk valley college in Edinburgh. My three main hobbies are music, computing (building websites) and cycling particularly in the summer months. I am into playing guitar, keyboard, drums but my main love is singing.

School Years:

I would like to take some time to talk to you about my childhood and school years. Challenging though they were, a lot of support during my childhood and school years was offered. I was also diagnosed with mild hearing difficulties at the age of 6 and because of this, class work was that little bit more difficult. One school pupil who was giving me a hard time had action taken against him, very quickly, by school staff, who were very understanding and aware of my problems. School parties were good but I often felt out of place and different as I often took in my own food, which raised the questions like 'Why have you got that' etc. In my last year at primary School our class travelled to France for a week long excursion, this entailed getting letters from doctors, dietitians etc so that I could get my Aminogran mix through Customs!! It was really good to be treated the same as everyone else and given the opportunity to go.

Secondary School:

Despite being anxious and worried about the change of primary to secondary school, secondary school staff were also very understanding and tried hard to cater for my needs. This is perhaps best expressed when I went on what is known as 1st year exodus (a week long excursion) and staff were proving very helpful not only with PKU, but also building up my levels of confidence because of certain responsibilities and tasks that we had to take part in but again as well as being rewarding, it was indeed very challenging, particularly where PKU was concerned. It entailed supplies being sent from home everyday that I was on the excursion, which was prepared and dealt

with by my parents. Secondary school gave me the chance to expand on the one thing that I had been confident with for a long time, music. This helped my confidence and as a result I learned to play the guitar, a bit of keyboard and drums as well as taking part regularly in high school musical events, including a 'Friday Fringe' event, which featured a regular slot with a pop idol type talent show, which was not only fun to be part of, it was very funny!! In 1999, when I was in 3rd year of high school, I was presented with an Electro Acoustic guitar at the schools annual prize-giving ceremony to encourage musical development and it did exactly that!! Soon after I performed at the primary school, in which I was a pupil three years earlier to raise money for the local youth club, it proved to be quite a success!!

Life As A Student With PKU:

I started college in September of 2002 and yet again it was another huge step that I was very apprehensive about taking. Being so apprehensive about this meant that I got the bus almost 50 miles up the road and then immediately walked back out of grounds of the college and got the next one home!!!. I missed my first week at college but huge persuasion from both of my parents and one particular tutor at college meant that I found that little bit extra courage to get my backside back on the bus and get back to the college and I have been there ever since. College tends to get harder year by year and although 1st year was relatively easy it was challenging for my PKU and meant that packed lunches often had to be prepared because the canteen did not offer a huge choice of meals and what I was allowed to eat, didn't come without the extra cost. My aminogran is taken twice a day, once in the morning, which is normally a bit of a struggle and one in the evening before I go to bed. As the years of college goes on, the workload tends to get heavier but with the right help and facilities balancing the workload with PKU can be overcome. I did find that creating awareness amongst college staff difficult in some cases. Some staff were very supportive and others could be very dismissive of the condition and tend to think that it's nothing more serious than a special diet. I have also noticed that many will believe that it is the choice of the individual to be on the 'special diet' and I feel that there is a huge lack of understanding right across the board, not just with college staff as I have experienced in the past. The college offers study support for people with special needs or requirements, that includes people with PKU but again getting them to understand the condition can be challenging and frustrating at times.

Coping With Diet In General:

Going out for a family meal, although can be done tends to be avoided in my family due to the very fact of PKU because I usually end up with something boring compared to everyone else. Vegetarians are always catered for but frustratingly always involves cheese, pasta or both!! Living in a rural area also does not come without its problems as getting a wider choice of food is harder because the nearest major supermarket is miles away and we try to make do with what we have. Being at home with a PKU diet tends to be a little less stressful than having to be committed to something such as full time education but taking responsibility and having a schedule, which allows you to aim for your goals can be rewarding too. Sticking to the PKU diet will bring you huge benefits, especially when you are involved with a career or a full time education. You will be in more control of your concentration than you would be off the diet. You will feel better, more energetic and happier in yourself. Having lapsed from the diet some time ago, I started to notice that shakiness in my hands and both my knees was hugely noticeable and reflexes were very poor. As if it wasn't enough, eyes would be very bloodshot, feel very heavy, I would have regular mood swings as well as depressive and negative thoughts. Sticking to your diet is hugely important.

Closing Thoughts:

People with PKU very often feel isolated and alone with the condition as they feel like the only one in the world who have it because although it's on the increase (very slowly though), it is still very rare. For those who have internet access, I feel that creating a friendship between two or more people with PKU on websites and in various PKU chat communities is hugely important so that they do not feel as isolated as they once did, and to share their thoughts, experiences and problems and I am glad to see that I have noticed a huge increase in the number of websites now offering this facility. For those who don't have internet access, I feel that publications would help in the forming of friendship between people with PKU, particularly those who live in rural areas and outwith areas, where they are not able to attend PKU groups that have been set-up exactly for these reasons.

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