Passionate about partnerships –
Diet therapy, psychology, nursing and medical: Optimising partnerships to provide quality care to clients with PKU in their later life

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1. What is PKU?
2. Why promote treatment in later life if the person has not had the recommended early intervention?
PKU – Integrated care

Dr Michel Tchan
Westmead Hospital
Phenylketonuria

- Inborn error of metabolism which, if untreated causes:
  - Mental retardation
  - Microcephaly
  - Decreased pigmentation
  - Eczema
- Intellectual impairment
  - IQ < 35
    - 47%
  - IQ 36 - 67
    - 47%
  - IQ > 68
    - 6%

- Epilepsy
  - 25%

- Other problems: behavioural disturbance with hyperactivity, destructiveness, self-injury, psychiatric problems, tremor,

http://ecast.euclid.k12.oh.us/sproat/sites/fourth/Phillip/phenlketonuria/Phenlketonuria.htm
PKU

- Baby is normal at birth
- Now diagnosed by newborn screening (since 1968)
  - Heel-prick bloodspot at day 3 of life
  - Sample tested for elevated Phe levels by tandem mass spectrometry
  - Further testing to differentiate from other diseases with high Phe
- Strict dietary treatment gives normal outcomes
Dietary treatment

- Low protein diet
  - Minimal amino acid intake
- Supplement all amino acids except for Phe
- Supplement vitamins and minerals
Dietary Treatment

- Previous medical advice was that the diet could be normalised from early teenage years

- BUT

- Complications of high Phe levels in adults
  - Anxiety
  - Increased rates of depression
  - Poor executive function
    - Planning, working memory, attention, problem solving, verbal reasoning, inhibition, mental flexibility, task switching, and initiation and monitoring of actions
Untreated Adult PKU

- There are many adults who missed out on this treatment
  - Intellectual disability
    - Mild to severe (mostly)
  - Behavioural difficulties
  - Seizures

- Restarting diet and lowering Phe levels improves behaviours significantly, and the patients seem happier
Case Study

- 72 year old woman with untreated PKU
- Behaviours
  - Aggressive
  - Uncooperative
- Dermatitis
- Phe levels around 2000 (normal 75-150)
- Introduced diet – average compliance
  - Levels down to 1200
  - Behaviours improved, but still a little aggressive
  - No self harm
  - Dermatitis improved
Case Study

- Poor dietary compliance at group home
  - Levels again 2000
  - Behaviours escalated
  - Self harm

- Moved accommodation – strict diet control
  - Levels down to 600
  - Behaviour very good
  - Seems happy much of the time
Care needs

- Feeding
- Dressing
- Toileting
- Mobility
- Diet
- Behaviours
- Medical oversight
  - Complications of diet
  - Medication to help control behaviours

- Integrated care is vital!
  - Success is dependant on nursing and dietician care
What is the Psychologist’s role in the team helping clients with PKU?

What are the benefits for the client even at this later stage in life?
PKU and Challenging behaviour

- Psychologists work as part of a team supporting clients at Hunter Residences.
- Our focus is generally on challenging behaviours.
- We consider the function of behaviours, triggers, prevention strategies and responses. We look at what changes can be made to improve a person’s life so that challenging behaviours are less likely.
As noted in Dr Tchan’s case study,

- Untreated PKU can lead to challenging behaviours.
- Not adhering to the diet can have similar effects for the person.
- Beginning the PKU diet, even late in life, has been shown to have benefits for the person.
Data

- Before people start on the PKU diet it can be helpful to gather information about any existing challenging behaviours.
- Data sheets are developed by Psychology.
- We rely on the nursing staff to collect the data.
Once someone has been on the diet for a time we can review challenging behaviours by again collecting data to see if the diet has lead to a reduction. This is reviewed in light of follow up pathology Phe levels.

It can be easy to lose sight of the changes for a person. Being able to compare data from before and after diet can remind us of what changes have occurred.
Monitoring of other changes

- Psychology can also help track other changes for a person to look at benefits of being on the diet.
- Health, skin, bowels and irritability can all show improvement with going on the PKU diet, even later in life.
- Again we rely on nursing staff to complete data sheets.
Going on the PKU diet later in life can be seen as a major change for a person. It appears the person is being deprived of things (food) they might value. Any changes of behaviour or mood are sometimes automatically attributed to going on the PKU diet whether this is true or not.
Psychology can help track other events in the person's life. E.g. health, accommodation changes, loss of friends and other major life events.

This helps clarify other issues that the person may be responding to with mood or behaviour changes.
Benefits for the client

- The client has the benefit of everyone's professional expertise.
- Having a variety of disciplines involved with a client makes exchange of information easier.
- Concerns raised can be addressed from different perspectives.
Benefits of Partnerships

- We all learn from other disciplines.
- We can support each other when clients are having difficulties.
- We don’t feel isolated. We know everyone involved with the person is working to give them the best life possible.
1. How a PKU diet is developed and
2. What are the implications for the client in keeping the diet going?
What are we measuring?

- The PKU diet is a diet to manage the intake of one of the amino acids – phenylalanine, and monitoring that intake against serum Phe levels.
- Amino acids are the “building blocks” of protein, so we are not just measuring protein as per grams of protein in a food, but mg of phenylalanine in the protein in the food.
- The dietitian must take into account that phenylalanine is an essential amino acid. When the phenylalanine is restricted, the next amino acid in the pathway (tyrosine) should be monitored for adequacy when major changes are made to the diet. Tyrosine has no effect on Phe pathology, but it is important for nutrition.
15mg of Phenylalanine is counted in Australia as 1 Unit Phe, so the client may be given, for example, a 40Unit diet and he/she or the carer adds the units over the day plus supplies the required dose of supplement.

It is important to understand that “1 Unit Phe” is a definitional term and different countries have potentially used a different definition. For example USA uses 10mg phenylalanine as a measure of 1 Unit.

The problem this raises is that the source of your information needs to be known.

There is a caution here about looking something up on the internet and applying it without advice.
What does a PKU diet look like?

- The lower the protein content of any food item, the less phenylalanine is present.
- The PKU diet is therefore a “vegan” diet with nuts and legumes limited or excluded.
- PKU specific supplements are then added based on client weight and protein needs. These contain vitamins, minerals and all amino acids except phenylalanine so that more low protein foods can be included for the client.
- The foods of the PKU diet are therefore low protein, but when the supplements are added the nutrition intake can be actually high protein. This is why the supplement dose is important.
A typical PKU diet would contain

- Limited cereal which can be increased with specialised low protein cereal products such as low protein bread and pastas.
- Phenylalanine free Supplements that are either protein free (such as a Phe free milk-style supplement that can be used to in place of milk) or suppliers of other amino acids (such as is the case with PKU supplements)
- Fruit and vegetables counted as per their individual level of protein
- Fats and oils including cream and margarine are counted as per their protein contribution
LP treats (such as chocolate, cake mixes etc) can be added from specialty food companies.

The dietitian will show the U value of Phe in each food if the menu is supplied, or indicate how to calculate average Phe according to average protein. At Hunter Residences, the actual Units Phe are shown for each PKU menu item.
Eating out

- Clients may still eat out, but they do need to order basic vegan foods as described above and adjustments made if necessary to adding or excluding other items at home if needed.
- The PKU protein supplement needs to be taken either when they are out if a tetra pack option is available or if a powder form supplement then this can be mixed into a glass of water at the restaurant, or doubled up at home if one has been missed.
Cost

- The PKU diet can be drawn from basic foods within the food groups noted above, but these are limited, so options of low protein specialty foods increase the variety and palatability of the diet so that it is more pleasant as a lifelong regime.

- The supplements, both PKU supplement and protein free milk substitute supplement are available on PBS but not the specialty low protein food options.
Alerts for us

- The nurturing side of our natures can tempt us to let the client have a “little bit” of something additional.
- For clients who are intellectually disabled due to this condition, we need to remember that they are not in a position to understand what the diet means, or to make an informed decision about poor compliance. They need us to make those decisions for them. Dr Tchan and Robyn have made reference to this difficulty previously.
- Our clients need us to help them
What is the Inborn Errors of Metabolism (IEM) grant and how clients can apply for it?

How do you take readings for PKU levels for an adult?
To qualify for the IEM grant there are a number of requirements:

- Australian resident with a Medicare card
- >6mths age
- Require a medically prescribed diet
- Be assessed by a metabolic specialist
- Be re-assessed annually with advice that the diet continues.
To maintain eligibility the person must:

- Comply with the diet
- Current personal/banking details
- Current care/custody arrangements for minors
- Permanent Australian residency
Re-application must occur every 12 months to maintain the grant.

- Application is simply done by having an appointment with the metabolic specialist (recognised by the Dept Health and Ageing) who can complete the appropriate form certifying the diet is required.

- At Hunter Residences we were fortunate that Dr Tchan was willing to come to Stockton and do a clinic with the dietitians on site.
Dr Tchan has referred to a newborn baby having a routine heel prick test approx 48hrs after birth.

In adults there are 2 common ways to do the tests:

- A regular blood test that can be uncomfortable and invasive and will cost $100.
- For any person with known PKU this type of testing is classed as inappropriate and sent back not tested.
The appropriate test is similar to the heel prick and is attended in a similar way.

The same piece of paper or card for collecting the baby test is used and a simple finger prick is performed to get the blood for testing. There has to be enough blood in the designated spots on the card to soak through to the back.

This test is free for people with PKU.
The process for using cards

- The cards have to be left to dry for 4 hrs before I wrap them individually in a sheet of paper and then snail mailed to Westmead Children’s Hospital – Newborn Screening Programme.
- Within approx 1 week a result is sent to the MO and I will often ring for the results so I can notify the dietitians.
The requirement for a level to be checked is usually indicated by the dietitian or the GP/MO.

Clients would routinely have a level done annually and more often if there was a health reason for it to be checked more regularly.
To Summarise

- When the person has been diagnosed at birth and treated, they have achieved their cognitive potential. In adult life he/she has the ability to make informed decisions about his/her own dietary compliance and has the government assistance to be able to purchase some of the additional foods.

- These very adults are the ones informing health professionals of their choice to maintain the diet for quality of life reasons.

- For clients with intellectual disability those choices are made for them by others who (you and us) care for them.
Part of the team working together for the client includes the managers and front line workers without whose help and positive attitude with the clients this option would not be available to them.

It’s a pleasure to work together for these clients, we hope you will enjoy joining us in this journey.
On behalf of the panel here - thank you for your attention

If you have Questions please start by identifying the panel member you wish to ask