

Adults with PKU

Back to diet – and new therapies

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- One of 12 adult centres in the UK
- Referrals accepted from all over South of England

★ Adult IEM centre

★ Outreach clinic (held at a different hospital 3-4 times per year)



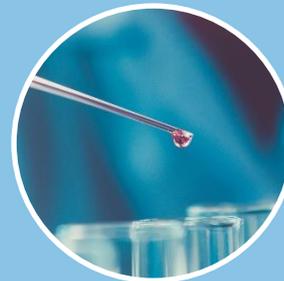
What does being 'on diet' mean?



Restricting protein intake



Taking protein substitutes



Maintaining phenylalanine levels in range





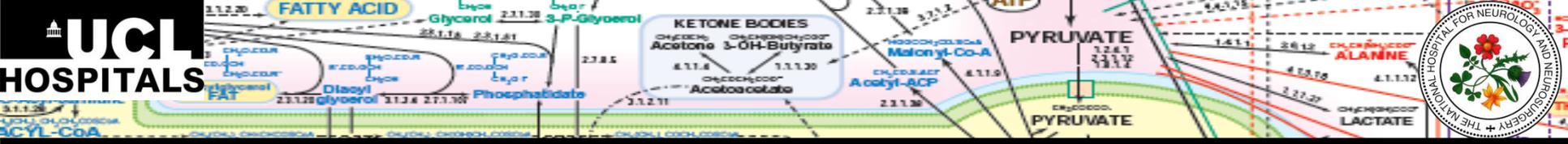
Doing diet “yourself”

- Can be difficult
- Some people struggle with organisation and planning meals
- Confidence with cooking is important
- May need to prepare separate meals for partner/family and own children
- Some people may have little support
- Need reasonable literacy and numeracy skills to read labels and calculate grams of protein/fat etc
- Not everyone will have a beautifully balanced, varied Instagram-able diet (and that’s OK!)



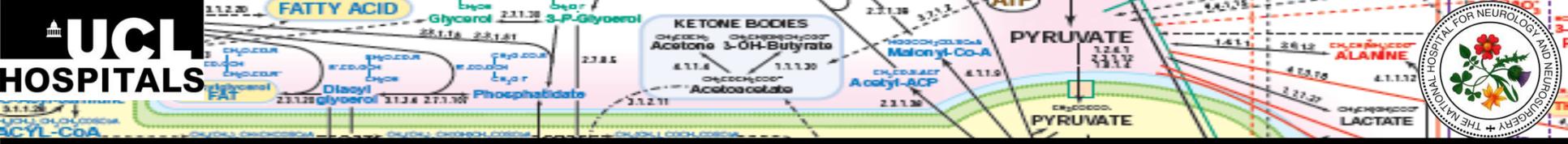
Comorbidities

- Expected as people age
- Consistent data regarding overweight and obesity amongst PKU population – especially in females
- Recent studies seem to suggest increased cardiovascular risk
- Insurance database studies indicate increased risk of multiple comorbidities amongst adults with PKU
- Important to ensure the diet is balanced and doesn't worsen health
- No published PKU-specific health programmes or strategies



Managing comorbidities

- Discuss with patients
- Weight loss when returning to diet is possible
- Identify and address concerns from childhood e.g. avoiding artificial sweeteners, promoting calories
- Offer lower calorie protein substitute
- Encourage dietary protein from nutrient dense sources e.g. wholegrain carbohydrates, vegetables
- Healthy eating principles – reduce saturated fat, increase fibre, limit added sugars
- Importance of exercise



Potential difficulties in returning to diet

Lack of motivation

Uncertainty surrounding perceived benefit(s)

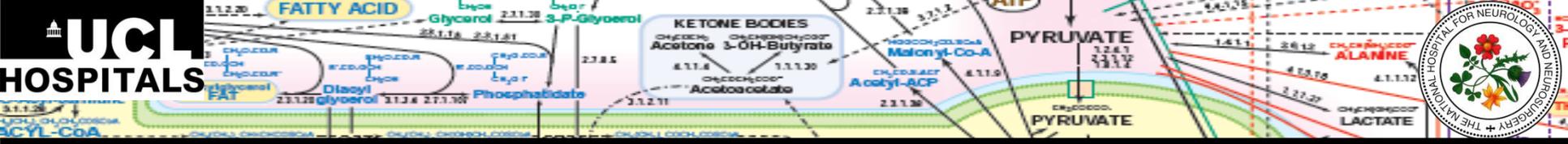
Memories of diet as a child – products/foods

Limited self efficacy

Financial barriers

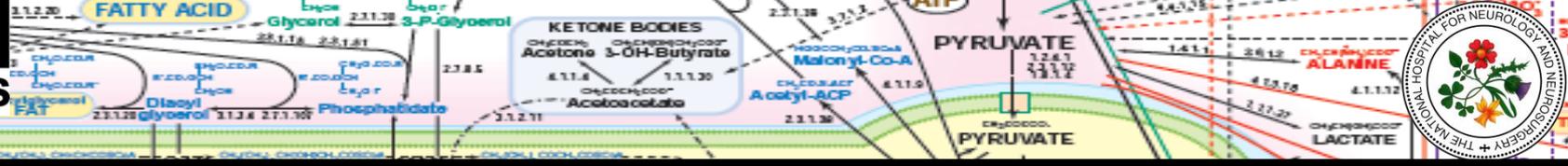
Other commitments – work/families

Fear or anxiety about lack of success



“So, what do you think about returning to diet?”

- Important to have structured assessment
- Need to establish motivation
- Consider social context
- Identify potential barriers
- Manage expectations
- Recognise feelings of guilt or fear
- Explore efforts to return to diet in the past



- Patients may feel pressure to be “on diet”
- Not for healthcare professionals to decide if it is right for them
- Recognition that relapse is very likely – in any behaviour change approach
- Supporting patient to decide if it is the right time
- Do they feel willing and able to commit right now?
- What other support can be offered?

- **Information-related:** understanding of condition, of treatment, of managing difficulties
- **Ability-related:** managing complexity of treatment, managing practical aspects, negative effects, lifestyle factors
- **Motivation-related:** beliefs, fears, attitude towards HCPs/services



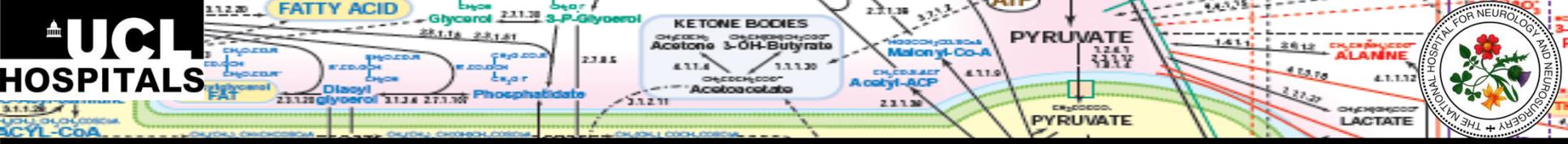
- Recognise that setbacks will happen
- Try to use as a tool for reflection and learning
- Accessing support to become more resilient
- Encourage patients to be kind to themselves
- Challenging unhelpful thoughts:

Pattern	Example
Perfectionism, "shoulds and musts"	"I must do what is best for my health at all times"
All or nothing thinking	"Either I am following the diet 100% or I am failing at diet completely"
Discounting the positive	"I cheated on Tuesday, so the whole week is ruined"
Catastrophising	"If I eat one high protein food I'll go completely off diet"
Overgeneralising	"There's no point, I always fail at everything anyway"
Personalisation	"I'm weak and that's why I can't stick to the diet"



Returning to diet – Charles Dent Metabolic Unit





Key topics addressed during dietary education sessions

Explanation of principles of phenylalanine restricted diet

Provision of phenylalanine exchange list

Information regarding exchange free foods

Education regarding label readings and calculation of exchanges from information on food labels

Meal planning

Tasting of protein substitutes and discussion of properties (dose, calories, sugar)

Trialling specialist low protein products

Determining monthly prescription requirements for protein substitutes and specialist low protein products

Discussion of home delivery services available

Advice on managing prescription costs or use of a prepayment certificate

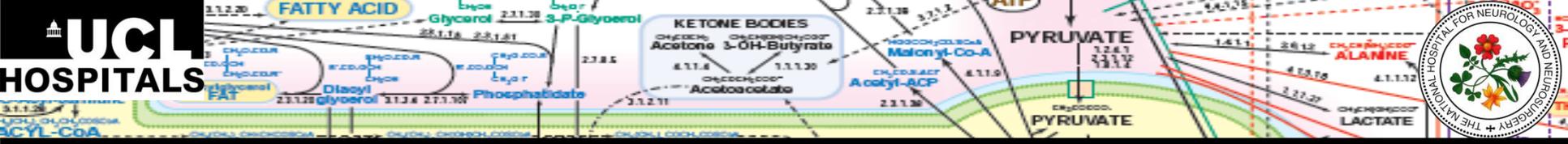
Practical cookery using specialist low protein products

Supermarket visit

Advice and tips for eating out or managing travel and holidays

Tailored education related to clinical need (e.g. advice regarding weight management)

Teaching on performing bloodspots (provided by clinical nurse specialist)



New therapies



(New) therapies:

BH4

Pegvaliase

Gene therapy





BH4 in pregnancy

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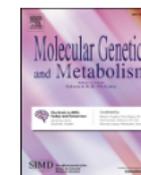


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Review article

International best practice for the evaluation of responsiveness to sapropterin dihydrochloride in patients with phenylketonuria

Ania C. Muntau^a, Darius J. Adams^b, Amaya Bélanger-Quintana^c, Tatiana V. Bushueva^d, Roberto Cerone^e, Yin-Hsiu Chien^f, Ana Chiesa^g, Turgay Coşkun^h, Javier de las Heras^l, François Feillet^l, Rachel Katz^k, Florian Lagler^l, Flavia Piazzon^m, Fran Rohrⁿ, Francjan J. van Spronsen^o, Paula Vargas^p, Gisela Wilcox^q, Kaustuv Bhattacharya^{r,*}



Sapropterin dihydrochloride treatment should be considered in pregnant women with PKU if:

- they are unable to achieve a blood Phe concentration $\leq 360 \mu\text{mol/L}$ with dietary management OR***
- they are already on sapropterin dihydrochloride, which had been started prior to becoming pregnant, and request to continue taking this medication during pregnancy***



Gene therapy

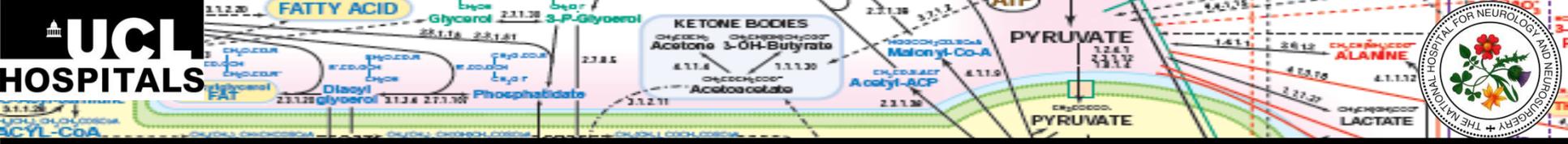
Currently in clinical trials

If effective, would potentially be curative

Can only be offered from adolescence

Safety for pregnancy?

Will require long term dietary changes to maintain nutritional status



All new therapies likely to present dietary challenges

Metabolic control may become secondary priority to overall nutritional status

Treatment adherence issues?



PKU - dynamic field requiring response to new demands

Dietary treatment

Will play a key role in PKU for many years to come

Treatment needs are different in adulthood

Alternative treatments are changing dietetic needs

Full extent of adult needs have not yet emerged

Many opportunities for future

