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A case study outlining the dietary management of a planned pregnancy for an individual with hyperphenylalaninemia.

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Patient Details

Age:
29 years

Gender:

Diagnosis:
Hyperphenylalaninemia (hyperphe) diagnosed on newborn screening.

Relevant history:
No previous pregnancies.
Works full time as a teaching assistant in a primary school.
Contraception method: intrauterine device (IUD).

The patient contacted the metabolic unit to express her wish to plan a pregnancy.

Relevant Medical History

Dietary restriction of phenylalanine (Phe) not required during childhood. The patient had not attended a metabolic clinic for at least 2 years.

Currently eating a diet containing moderate amounts of natural protein (pulses, legumes, dairy and occasional meat or fish). Not regularly taking any multivitamin or mineral supplements.

Anthropometry prior to commencing dietary intervention:
Weight: 64kg Height: 1.56m
BMI 26.3kg/m^2

Last Plasma Phe:
441µmol/L

Protein requirements at initial assessment:
64 x 0.83 x 1.2 + 1g = 65g/day (WHO, 2007)
51 x 1.15 = 59g/day (Maritz, 2010)

Target Phe for preconception and pregnancy at managing centre:
120-250µmol/L

Action	Rationale
Patient completed a 3-day food diary prior to attending clinic.	To estimate typical dietary protein and micronutrient intake. Helps to facilitate discussion of usual dietary choices.

Findings:
Food diary indicated typical dietary protein intake to be approximately 55g/day. This dietary protein intake is adequate but lower than average intakes for adults in the United Kingdom (Public Health England, 2016). The patient's protein choices were of reasonable biological quality including; small portions or milk, cheese, yoghurt, beans and lentils. Her intake of meat and fish was limited to 1 bacon sandwich per week and occasional fish fingers.

She routinely skipped breakfast due to a busy morning schedule, preferring to have a larger mid-morning snack at her workplace supplemented by milk-containing hot drinks.

Action	Rationale
Patient and her partner received dietary education.	5 hours of dietary education was provided by a specialist dietetic assistant. Education included: protein counting, low protein diet, menu planning and trialling protein substitutes.
Advised to consume 40g protein equivalent (PE) from a protein substitute.	40g PE required to meet protein requirements in a slight excess of requirements to ensure essential amino acids. The patient tried and liked PKU express, as she preferred that the product could be made up to a smaller volume, and that it contained less than 100 calories per 20g PE. She felt able to take it mixed with permitted low Phe drinks twice daily, one dose with her mid-morning snack and one dose with her evening meal.
Advised to restrict to approximately 45g dietary protein.	The patient's Phe level whilst eating a moderate intake of 55g/day dietary protein was 441µmol/L. This indicated that some further dietary protein restriction would be required to achieve target levels for preconception and pregnancy in addition to introduction of protein substitutes.
Twice weekly bloodspot monitoring commenced.	Bloodspots requested twice weekly during preconception at managing centre. In the centre's experience, this facilitates more regular feedback to the patient by providing motivation and encouragement to consistently adhere to their prescribed diet throughout the week, reducing possible variability in plasma phenylalanine.
Additional supplementation of DHA, vitamin D and folic acid recommended.	Intake from diet and protein substitutes would not be sufficient to meet requirements during pregnancy.

After two weeks, Phe levels reached **307µmol/L** but stabilised.
Current practice at this centre is to aim for stable Phe levels within the middle of the target range (150-250µmol/L) for 2-3 consecutive weeks before stopping contraception. Therefore, the patient's metabolic control was not acceptable and it was recommended she continue to use contraception at this time.

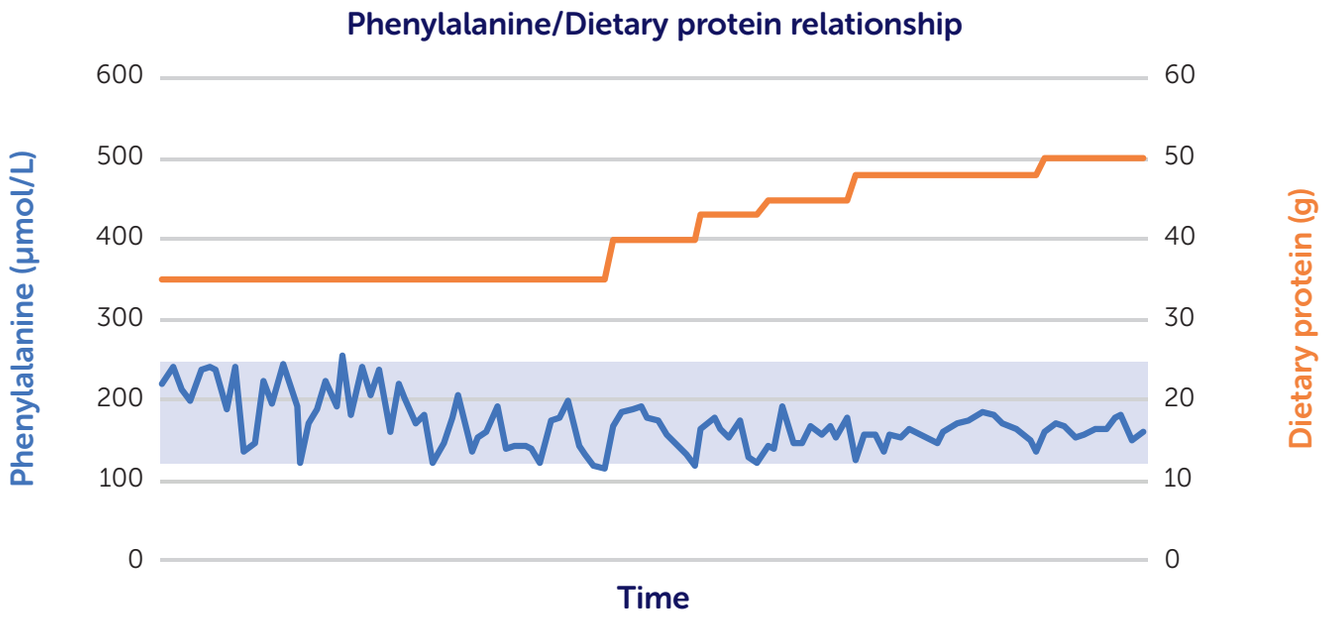
Action	Rationale
Patient completed 3-day food diary.	To establish dietary pattern, energy and dietary Phe intake.
Patient regularly monitored her weight at home.	Weight stability indicates adequate energy intake to prevent catabolism. Weight loss might indicate that an increase in energy intake is required to stabilise Phe levels rather than/in addition to a change in dietary protein or protein substitute.

Findings:
The food diary demonstrated that the patient was taking her protein substitute twice daily with her mid-morning snack and evening meal, as had been advised.
The patient was managing the dietary protein restriction well and consuming 45g dietary protein per day. However, dietary protein was mainly consumed in the evening rather than spaced out during the day.

Her weight remained stable.

Action	Rationale
Reduce dietary protein to 35g per day and distribute intake more evenly during the day.	The patient was finding it difficult to spread their protein intake out during the day and said they felt confident they could reduce their dietary protein intake further.
Increase protein substitute to 50g PE/ day.	To promote reduction in Phe levels. The patient agreed to take 1 x PKU express20 with breakfast, 1 x PKU cooler10 with lunch and 1 x PKU express20 with her evening meal.

Following these changes, metabolic control was achieved and maintained for 3 weeks (average Phe levels between 120-250µmol/L), and the patient was therefore advised that contraception could be discontinued.
The patient had a positive pregnancy test after 4 months on diet, and was advised to increase bloodspot monitoring to three times per week which is routine care at the managing centre.



Metabolic control for the first trimester remained stable with this dietary prescription. In the second trimester (16/40 onwards) Phe levels began to fall consistently below 150µmol/L, indicating increased Phe tolerance. Initially, PKU cooler10 was stopped and the patient continued with 40g PE from 2 PKU express20 to help maintain Phe levels > 120µmol/L, and then dietary protein (Phe exchanges) was increased (often in 5 x 50mg Phe increments) to achieve acceptable metabolic control. As the patient did not have a strong preference for meat or fish, alternative options to increase daily protein intake were suggested - including eggs, small pots of yoghurt, mini cheeses and milky drinks which could be consistently incorporated into her daily eating pattern.

Rationale for dietary change:

1. Protein intake exceeding requirement
2. Patient choice and convenience

By the end of pregnancy total protein intake was 90g (40g protein equivalent from PKU express, plus 50 x 50mg dietary protein (Phe exchanges). Total maternal weight gain during pregnancy was 11.5kg.

A healthy male infant was born at 40+2/40, weighing 3.4kg (25–50th centile) with a head circumference of 34cm (25th centile). He was breastfed. The patient was advised to discontinue PKU express after delivery, and encouraged to return to an unrestricted protein intake to ensure sufficient energy and protein to support lactation. It was advised that she take a calcium and Vitamin D supplement to meet increased requirements for lactation. She was seen in clinic 16 weeks post-partum. At that time, her estimated dietary protein intake was 65g/day and plasma Phe 532µmol/L and her infant received normal paediatric checks. He will be offered developmental follow up at the managing centre at age 2, 4, 8 and 14 years which is routine care for infants born to women with PKU or hyperphenylalaninaemia at this centre.

Reference:
Maritz C, Ellerton C. 2010. Dietary Management of Maternal Phenylketonuria: A Practical Guide.

Public Health England (PHE), F.S.A.F. National Diet and Nutrition Survey (NDS). 2016 3rd May 2018]; The National Diet and Nutrition Survey assesses the diet, nutrient intake and nutritional status of the general population of the UK.]. Available from: https://www.gov.uk/government/collections/national-diet-and-nutrition-survey.

World Health Organization (WHO), Food and Agriculture Organization of the United Nations (FAO), United Nations University (UNU). (2007). Protein and amino acid requirements in human nutrition. WHO Technical Report . Series 935, 1-265

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All information correct at the time of print

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