

MANAGING A DELAYED PKU DIAGNOSIS AND T2 DIABETES through the use of a low-carbohydrate, phenylalanine- free medical food/specialized formula

Analyzing past and present symptoms to address a medical enigma

The patient was identified with PKU through newborn screening shortly after birth. He was managed early, incorporating the available PKU formula and following a low-phenylalanine diet. Based on recommendations at that time, he discontinued the restricted-phenylalanine diet at 6 years of age, remaining off both the medical food/specialized formula and the special diet.

In 2002 at 40 years of age, he arrived at the Adult Metabolic Center. He was accompanied by his sister, who stated the patient had a long history of learning difficulties and of issues with concentration and memory. She added that her brother's medical care was now complicated by a diagnosis of Type 2 (T2) diabetes. The patient was also diagnosed with complications from T2 diabetes, including peripheral neuropathy and hyperlipidemia, and was considered to be overweight based on his current body mass index.

Reintroducing the PKU diet after more than three decades

To help the healthcare professionals understand the patient's distinct nutritional needs, baseline tests were administered, including blood phenylalanine and tyrosine levels. His phenylalanine level was 1695 $\mu\text{mol/L}$ (normal range is 36–109 $\mu\text{mol/L}$), and his tyrosine level was 28 $\mu\text{mol/L}$ (normal range is 33–110 $\mu\text{mol/L}$). Additionally, the patient's baseline hemoglobin A1C, a measure of his blood glucose level, was 8.5 percent (normal range is 4–6 percent) over the past three months.

The medical team created a simplified meal plan for easy implementation and management of PKU in a diabetes context. The patient was started on a phenylalanine-free medical formula/specialized formula and a modified meal plan to reduce both his whole protein and carbohydrate intake. This simple change reduced the phenylalanine level to 545 $\mu\text{mol/L}$ and his hemoglobin A1C to 7.5 percent, both of which met the management goals for him at that time. An additional benefit of the streamlined meal plan was that it did not overwhelm the patient or his family. The patient was also able to lose 10 pounds, a benefit in managing his hyperlipidemia.

Revising the dietary approach to PKU management in a diabetes context

After 10 years of successful dietary management, the metabolic clinic received a call from the patient's family physician because of the patient's deteriorating glucose control. This coincided with a deterioration in his PKU control, with blood phenylalanine levels running between 605–726 µmol/L.

To address the deterioration of both glucose and phenylalanine control, a careful reassessment of the patient's diet was conducted. It was determined that the patient's PKU medical food/specialized formula was providing a significant amount of carbohydrates to his overall intake, containing 36 grams of carbohydrates per meal. For successful diabetes management in this patient, the goal was to restrict carbohydrates to no more than 60 grams per meal; the current consumption levels left little room for additional food intake.

To adhere to a more controlled diet, the patient was switched to PKU Lophlex® LQ, which decreased the carbohydrate intake from formula from 36 to 9 grams of total carbohydrates per meal. This allowed him to reduce his overall intake of carbohydrates and afforded for more food choices at each meal. Additionally, My Plate, a resource provided by the U.S. Department of Agriculture (choosemyplate.org), was modified to incorporate his PKU diet principles. This change resulted in improvement in both the patient's blood phenylalanine levels as well as his glycemic control.

Finding success and self-empowerment through a new diet

A simplified plan that encompassed PKU management in a diabetes context empowered the patient to take care of himself more consistently, minimizing family assistance. Key results included:

- Modified PKU medical food/specialized formula to significantly decrease overall carbohydrate intake
- Modified MyPlate meal planning tool to assist in both diabetes and PKU management
- Improved patient adherence to formula intake
- Improved BMI, resulting from a gradual weight loss of 20 pounds over 10 years

In this patient's case, medical food selection was critical in successfully managing multiple conditions. PKU required a diet low in whole protein, and the principles of T2 diabetes management required the implementation of a carbohydrate-controlled meal plan, so striking the right balance necessitated a careful modification of diet programs and their core components. Ultimately, success was found by selecting a medical food/specialized formula that accommodated both conditions, promoted diet ownership and adherence, and offered greater control over food choices.

This case report* is provided by Jill Skrabal, M.S., R.D., L.M.N.T., C.D.E. First published on Nutricia Learning Center in 2015.

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