

ABSTRACT

Homocystinuria, maple syrup urine disease (MSUD), glutaric aciduria type I (GA I), and tyrosinemia type I are inherited disorders of amino acid metabolism (IMD), the cornerstone of their treatment being a diet restricted in protein or limiting specific amino acids in the diet. The theoretical part of the thesis summarizes general information about inherited metabolic disorders, newborn screening, and nutritional therapy in the Czech Republic and abroad.

The main objective of the practical part was to calculate the intake of limiting amino acids in patients with selected IMDs, compare it with international recommendations, and assess whether Czech patients with IMDs have metabolic compensation in line with recommendations. The thesis also focused on evaluating the potential positive and negative changes in nutritional therapy from a low-protein diet to a diet restricting specific amino acids.

For 5 patients with GA I, 6 patients with MSUD, 3 patients with tyrosinemia type I, and 5 patients with homocystinuria, the calculation of the content of selected amino acids, macronutrients, and energy in the diet was performed, and metabolic compensation and anthropometric data were assessed.

Results: In all patients with GA I, the intake of lysine was significantly lower than recommended by guidelines. On average, patients only consumed 62.5% of the recommended dose for their age category and current weight (with intake ranging from 36.2% to 84.8% of the recommended dose). Nevertheless, only some patients had good metabolic compensation. For patients with MSUD, the intake of leucine ranged from too low to significantly exceeding recommendations; however, we did not find a relationship between their metabolic compensation and leucine intake. Compared to international guidelines, the intake of methionine for most patients with homocystinuria was at the lower limit of recommendations for their age category, and their metabolic compensation was variable. For patients with tyrosinemia type I, most had good metabolic compensation, even though their intake of natural protein was often lower than recommended.

A change in the approach to nutritional management could lead to a partial relaxation of the diet for many patients, but at the same time, it would currently represent a significant time and organizational burden and could temporarily worsen patient compliance. The change would also be demanding for professionals. To make the change feasible, it is necessary to first create a standardized calculation methodology, conduct an analysis of a wider range of foods, and create new food tables, ideally in the form of simple software.

Keywords: maple syrup urine disease, glutaric aciduria, tyrosinemia, homocystinuria, metabolic compensation, low-protein diet, inherited disorders of amino acid metabolism