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Dietary therapy for a patient affected with 3-hydroxy-3-methylglutaric aciduria

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3-Hydroxy-3-methylglutaric aciduria (OMIM 246450) is an extremely rare genetic metabolic disorder of leucine caused by deficiency of 3-hydroxy-3-methylglutaryl-CoA lyase (HMG-CoA lyase). Affected patients present with life-threatening metabolic acidosis, hyperammonemia, and nonketotic hypoglycemia. Optimal outcome for patients depends on early diagnosis and treatment, prompt management of acute metabolic decompensation during the chronic stage, and long-term interprofessional health care programs. Nutritionists play a pivotal role in the care of metabolic disorders throughout the rest of a patient's life.

A 4-year-old intubated Bunun tribe female patient was admitted to the pediatric intensive care unit of the Buddhist Tzu Chi General Hospital (Hualien, Taiwan) due to status epilepticus, hypoglycemia, and severe metabolic acidosis. Results of urine gas chromatography with mass spectrometry revealed markedly elevated 3-methylglutaric acid, 3-hydroxy-3-methylglutaryl-CoA, and 3-hydroxyisovaleric acid, a finding consistent with a diagnosis of HMG-CoA lyase deficiency. She received an interdisciplinary health care program after acute management. Dietary therapy was the major part of the transdisciplinary health delivery process.

Total daily calories were increased from 1400 kcal/day (targeted 60–80%) to 2100–2200 kcal/day (targeted 100%). The ratio of protein lipid and carbohydrate was 13%:24%:63% as recommended for healthy children. Lifelong protein restriction was essential. Total daily protein was given at a dose of 1.5–2 g/kg/day (target: above 50 g/day after the age of 4 years) and was administered at different growth and developmental stages. The daily leucine dose was 1000 mg (suggested dose = 700–900 mg/day), 1.5% of the total protein amount was designed for her. Because leucine is an essential amino acid, close monitoring of optimal serum levels through blood sampling should be reached for better growth and development. Food avoidance was a major focus on restriction of protein from milk, meat, cheese, fish, poultry (including egg), beans, and nutlets, and even any foods containing milk and egg. Partial limited foods included sprouts (alfalfa, clover), radish, red bean, and murgbean. Unlimited fruits included vegetables and low protein starch such as green bean noodles, rice flour, pearl sago, lotus root powder, cornstarch, and corn flour, and low leucine foods such as potato powder were advised. The patient's major protein source was from a special formula (I-Valex I and II; Abbott, Chicago, USA). Other nutrients such as multivitamins and minerals were prescribed. Daily oral L-carnitine was also prescribed to increase excretion of organic acid. The ability to recognize food labeling was taught to the parents, the patient, and school nurses. Servings for daily dietary content (Table 1) and sample meal plans were designed (Table 2).

Table 1
Servings for daily dietary content (11–15 years).

Food category	Services	Protein (g)	Leucine (mg)	Fat (g)	Carbohydrate (g)	Calories (kcal)
Vegetable	3 EX[a]	3	90	0	15	75
Fruit	2 EX	0.8	30	0	30	120
Rice, white (cooked)	6 EX	9	692.3	0	90	420
Oil	6 EX	0	0	30	0	270
Low nitrogen starch	130 g	0	0	0	119	482
I-Valex II	180 g	55	0	23.4	63	738
Total		67.8	825.3	53.4	317	2105
%		13		24	63	

[a] EX = exchange, each exchange list contains foods that are alike; each food choice on a list contains about the same amount of carbohydrate, protein, fat, and calories as the other choices on that list.

Conflicts of interest: none.

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Table 2
One day's meal plan for patient affected with leucine metabolic disorder.[a]

	Solid food	Soup and drink
Breakfast	Vegetable sandwich with egg-free mayonnaise sauce (two pieces of grilled toast packed with 50 g filamentous cabbage, sliced tomato, and cucumber in small pieces)	Formula: 350 mL (special formula I-Valex II 60 g mixed with maltodextrin 30 g in 350 mL water)
Snack	Fruit: an apple, weight = 130 g	
Lunch	1. Fried rice (half bowl of cooked rice 100 g, fried with 50 g carrots, onion, and mushroom) 2. Vegetable (celery 50 g with 20 g konnyaku strips)	100 mL carrot and white radish (30 g) soup with parsley
Snack	Pearl milk tea (special formula I-Valex II 60 g mixed with maltodextrin 30 g in 350 mL water and 40 g tapioca ball)	
Dinner	1. White noodles with vegetable (40 g dry white noodles cooked with 30 g fresh mushroom, 50 g cabbage, and 20 g golden mushrooms) 2. Fruit: cantaloupe (mino) 165 g	
Snack	350 mL formula milk (special formula I-Valex II 60 g mixed with maltodextrin 30 g in 350 mL water)	

[a] This meal plan does not include total daily fluid requirement.

Avoidance of starvation and skipping meals to prevent hypoglycemia was also addressed to parents, school teachers, school nurses, and the patient. No episodes of metabolic decompensation or hypoglycemia were noted during the past 9 years of follow-up. Her growth and development were initially at the 25th percentile and she grew steadily to the 50th–75th percentiles as compared to her peers.

The goal of dietary therapy is to maintain normal plasma amino acid levels, avoid metabolic decompensation, and promote normal growth and development. Dietary meal plans should be designed and combined with special infant formula for treating patients affected with congenital metabolic disorders.

Further reading

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