A Case of Pyruvate Dehydrogenase Complex Deficiency Disease with Food Protein-Induced Enterocolitis Syndrome

Yugo NAGAYAMA [1)], Ayako GOTO [1)], Takashi SETOUE [1)],
Tatsuro KODERA [1)], Atsushi ISHII [1)], Eiji OHTA [1)],
Kei KUBOTA [1)], Takashi SAKAGUCHI [1)], Makoto TSUTSUMI [2)],
Shinichi HIROSE [1)]

[1)] *Department of Pediatrics, Faculty of Medicine, Fukuoka University*
[2)] *Department of Pediatrics, Chikushi Hospital, Fukuoka University*

Abstract

A 0-day-old girl had been found to have enlarged ventricles as a fetus and was admitted to the neonatal intensive-care unit (NICU) after birth. Lactic acidosis was observed after birth, and a diagnosis of pyruvate dehydrogenase complex (PDHC) deficiency was made based on mutations in the *PDHA1* gene. When Ketonformula® was started at day 7, vomiting appeared six hours after ingestion. She continued to vomit, but there was no worsening of lactic acidosis, so the attack was determined not to be due to PDHC deficiency. After switching from regular milk to breast milk and medium-chain triglyceride (MCT) oil, the symptoms improved. Eosinophils were positive in the stool, and lactoferrin was positive on the allergen-specific lymphocyte stimulation test (ALST). She was diagnosed with food protein-induced enterocolitis syndrome because an oral food challenge with regular milk and Ketonformula® was positive. As an alternative treatment, nutrition was started with MCT oil and Elental® P. However, she developed intractable diarrhea and growth retardation due to high amounts of MCT oil. While confirming the results of a gastrointestinal mucosal biopsy and the increase/decrease in the peripheral blood eosinophil count, we performed an oral food challenge with long-term titration and were able to induce remission and switch to therapeutic milk at an early stage, with consequent good weight gain.

Key words: Food protein-induced enterocolitis syndrome, pyruvate dehydrogenase complex deficiency, Milk allergy, Ketonformula®