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LETTER TO THE EDITORS

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Successful dialysis in a boy with methylmalonic acidemia

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Sirs,

We would like to describe a 7-year-old boy with methylmalonic acidemia (*mut*^{0} phenotype) who has been managed for 4 years by hemodialysis and continuous ambulatory peritoneal dialysis (CAPD) without any serious neurological complications.

In extreme cases of methylmalonic acidemia (MMA, McKusick 251000), methylmalonyl CoA mutase activity is completely absent (*mut*^{0}). Most patients die within 2 months of diagnosis and those who survive have a complicated clinical course, with frequent hospitalizations and both growth and mental retardation [1, 2, 3]. Cardiomyopathy and chronic progressive loss of renal function are frequent and serious complications of long-term survivors [4]. In the literature, there is only one reported case of methylmalonic acidemia treated with CAPD [5].

A 2.72-kg male infant was born by cesarean section at 38 weeks as the second baby of a twin birth. The elder twin brother died during the 1st week of life because of severe acidosis. Laboratory tests revealed metabolic acidosis and hyperammonemia (449 μg/dl). The urine was positive for methylmalonic acid (MMA). A low-protein diet with special formula feeding was started. Complementation analysis of cultured fibroblasts (performed in Dr. Rosenblatt's laboratory in Canada) showed that he had *mut*^{0} type apoenzyme deficiency.

The clinical course was complicated, and was characterized by severe metabolic acidosis, recurrent vomiting, and dehydration. Vitamin B_{12} therapy did not result in a decrease in urinary MMA and did not ameliorate the clinical course. Over the next 33 months, he had 14 hospitalizations because of metabolic decompensation and was treated with sodium bicarbonate, hydration, and a protein-restricted diet.

At 34 months of age, he commenced thrice-weekly hemodialysis for 2-h sessions. His weight was 11.7 kg (3th–10th percentile) and height 88.6 cm (10th–25th percentile). For the next 10 months, he had only 2 hospitalizations due to metabolic decompensation. However, thereafter he was frequently admitted nearly every month for 1 year, despite daily hemodialysis. We recommended liver transplantation, but the parents preferred dialysis.

At 5 years of age, he commenced CAPD with the aim of increasing the excretion of MMA and discontinuing hemodialysis. MMA was measured by isotope dilution gas chromatography-mass spectrometry [6]. Before CAPD, the MMA level of a 24-h urine sample was 482.02 μmol/day and serum MMA was 116.12 μmol/l. Dialysis involved four changes of a standard solution of 500 ml of 1.5% dextrose. After 1 week of dialysis, 24-h urine MMA was 263.56μmol/day and 24-h dialysis fluid MMA was 2,290.44 μmol/day. Serum MMA was 46.11 μmol/l. We confirmed that this regimen could eliminate 2,554 μmol of MMA in urine and dialysate per day. However, serum MMA concentrations (46.11–196.61 μmol/l) were not normalized.

In the 24 months after CAPD was started, his improvement has been dramatic. He had only 2 hospitalizations due to metabolic crises, despite progressive relaxation of the restriction of protein. He is currently 7 years old. His present weight is 25 kg (75th percentile) and his height is 118 cm (25th–50th percentile). The percentiles of weight and height have increased. His renal function is normal.

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His mental capacity was examined by a pediatric psychiatrist and the patient was shown not to be retarded except for his language. His language development is delayed by 1.5 years, but he has no serious neurological complications. Complications of CAPD included three episodes of severe scrotal swelling, and development of an inguinal hernia, which necessitated surgical treatment.

We advocate that CAPD should be an option for the management of the *mut*^{0} phenotype of MMA to prevent serious neurological complications, to improve physical growth, when more-specific forms of therapy are not effective, and to prevent the development of a catabolic state due to glucose supplementation.

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