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Case Report

Improvement in Growth and Developmental Milestones with Nutritional Intervention in Methylmalonic Acidemia: A Case Report

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ABSTRACT

Introduction: methylmalonic acidemia (MMA) is a metabolic disorder and especial nutritional support has an important role in improvement of growth and development in these patients.
Case presentation: A 3-month old female infant with known MMA was admitted to emergency department of Dr Sheikh Children Hospital with primary diagnosis of pneumonia and sepsis .This patient was a full term baby; MMA was diagnosed at 3th day of her life after several episodes of tachypnea, lethargia, poor feeding and irritability. After patient was stabilized, she was referred to nutritional support team for specialized MMA medical nutritional therapy. The patient's growth and development improved significantly after 2 months of follow up.

Keywords: Diet, Methylmalonic acidemia, Nutrition.

Introduction

Patients with methylmalonic acidemia can be divided into two subgroups: a severe, early-onset form with a high mortality rate, and a less severe form with better survival. The former is usually associated with defects in methylmalonyl coenzyme (1).

Inadequate oral intake secondary to poor appetite is a common and bothersome complication in long-term management of these patients. Consequently, enteral feeding (through a nasogastric tube or gastrostomy) should be considered early in the course of the treatment (2). Close monitoring of blood pH, amino-acid levels, blood and urinary concentrations of methylmalonate, and growth parameters are necessary to ensure proper balance in the diet and the success of therapy. Prognosis depends on the severity of symptoms and the occurrence of complications. These patients survive with major extrapyramidal (tremor, dystonia) and pyramidal (paraplegia) sequelaes (3). Methylmalonic acidemia presents early in life with a characteristic picture of vomiting, failure to thrive, hepatomegaly, lethargy, and ketoacidosis. And if left untreated, these children usually die early in life (4).

Case Presentation

Our patient was a 3 months old female infant with methylmalonic academia (MMA), with chief complaint of fever and history of frequent hospitalization in pediatrics intensive care unit of Dr. Sheikh hospital. The primary diagnosis of patient in hospital was pneumonia and sepsis. This patient was a full term baby who was born by normal vaginal delivery from a healthy mother. After birth her general condition was apparently normal, but she shortly developed tachypnea, lethargia, poor feeding and irritability. Laboratory examination at the 3th day of life- indicated sever metabolic acidosis and hyperammonemia.

Signs of developmental delay were apparent in the patient including hypotonisity, lack of neck holding, and inability to rolling supine to prone and prone to supine in 3th month of life. This baby had little respiratory distress. On her biochemistry, blood sugar was 103 mg/dl. Other tests showed Hb 8.2 g/dl, Ammonia 83.9, lactate

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49, magnesium 2.3, phosphor 3.5, urea 5, cratinine 0.4, sodium 136 and potassium 4.2. The patient excreted large amounts of methylmalonic acid in the urine and the blood level of MMA was elevated.

About diet history, she had been fed by breast milk in the first 14 days of life and then she was fed with a special formula (MMA/PA). In 3th month of life her weight was 4600 grams that is between 5 and 10th percentile. The expected weight for her was 5600 grams (for 50th percentile).

Nutritional considerations for the patient include feeding by the only available special formula (MMA/PA), supplementation with B_{12} vitamin, vitamin B_1 and carnitin, protein restriction (0.5-1 mg/kg/dl) and avoidance from long term fasting.

The energy intake of baby was 500-600 kilocalories from 250 millilitres dextrose water, 10 modules MMA/PA formula for providing 7.5-8 grams protein, 10 modules Aptamile formula for providing 5-6 grams protein and 7-8 milliliter MCT-oil. She was also fed by porridge with at smallest amount of protein including rice flour, starch and oil at the tolerance.

We followed the status of patient after discharg from hospital for two months (at the 5th month of life). Marked muscular hypotonia, developmental retardation, and neutropenia have decreased with controlling acidosis by protein restriction and treatment . In 5th month of life the weight of baby was 6000 grams, that is between 10 and 25th percentile (increased 1400 grams in 2 months).

The baby psychomotor development was markedly delayed before nutritional intervention. We found a clear improvement in development of hearing and understanding including moving eyes in direction of sounds, responding to changes in tone of voice, noticing toys that make sounds, and paying attention to music. Improvement in speech development includes babbling sounds more speech-like with many different sounds including *p, b,* and *m,* vocalizes excitement and displeasure, makes gurgling sounds when left alone and when playing with the caregivers. Improvement in movement development included holding her head up.

Discussion

Effects of metabolic disease on brain development, especially in the first year of life are irreversible .As observed in this study, the role of nutritional support in improving children's developmental status is important. Thus dietary recommendations should be included in the disease management program. In this patient, we had observed that the developmental delay showed a significant improvement by special nutritional intervention for MMA. MMA is a rare inborn metabolic disease, diet and nutritional advices may have an important role in management of patients. Also since there is no definite treatment for this disease, finding ways to manage the disease, including nutritional interventions, can play an important role in delaying progress to more advanced stages.

Conclusion

Our results showed that early diagnosis of MMA and nutritional intervention have a significant role in improving growth and developmental milestones of these patients.

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