Propionic and Methylmalonic Acidemia: Outcome of Nutritional Therapy and Nutritional Status

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INTRODUCTION
Propionic acidemia (PA) and methylmalonic acidemia (MMA) are inherited metabolic diseases due to problems in the metabolism of propiogenic amino acids isoleucine, valine, methionine, threonine, and odd-chain fatty acids. The presence of long-term complications such as growth retardation, intellectual disability, kidney failure, cardiomyopathy, pancreatitis, seizures, and optic nerve atrophy necessitate better therapeutic interventions. It was aimed to evaluate nutritional status as an outcome of nutritional therapy in PA and MMA patients.

METHOD
- Demographic characteristics,
- Clinical and biochemical features,
- Anthropometric data, and
- Recommended dietary treatments following the initial evaluation up to the age of 15 were evaluated retrospectively.

RESULTS
- 28 PA (18F/10M) and 26 MMA (7F/19M) patients were enrolled in the study.
- 35 parents had consanguinity and 23 patients had positive family history.
- Psychomotor retardation was observed in 84.6% of PA patients and 62.5% of MMA patients.
- Chronic malnutrition developed with increasing age both in PA (p<.05) and MMA (p<.05).
- Mean total protein intake (g/kg/day) met or exceeded WHO/FAO/UNU 2007 safe levels in PA and MMA patients.
- Contribution of precursor-free amino acids to total protein was significantly higher in MMA between 0-1 years (40.22±18.96 vs 24.8±10.62, p=.002).
- Plasma essential amino acids except methionine were within the reference range (in PA patients at all ages and between 0-1 years in MMA patients).
- Urine tiglyglycine and methylcitrate levels were significantly higher in PA patients between 2-15 years (p<.05).

CONCLUSION
Although energy and total protein intake meet the recommendations, all patients should be monitored carefully, as growth retardation develops with advancing age.