P-129 - GLUTARIC ACIDURIA TYPE I: NUTRITIONAL MANAGEMENT EXPERIENCE IN PEDIATRIC PATIENTS IN AN ARGENTINE POPULATION.

Crespo D¹, Becerra A², Guelbert G², Guelbert N²

1 Department of Nutrition-Hospital de Niños de la Santísima Trinidad; 2 Section of Metabolic Diseases-Hospital de Niños de la Santísima Trinidad; Cordoba-Argentina decrespo05@hotmail.com

INTRODUCTION: Glutaric aciduria type 1(GA-1) is an inherited autosomal recessive metabolic error of lysine, hydroxylysine and tryptophan, due to deficiency of the enzyme glutaryl-CoA dehydrogenase. The clinical manifestations vary broadly, from classical forms (macrocephaly, dystonic encephalopathy and frontoparietal atrophy), to subacute, adult and/or asymptomatic forms. Nutritional management is the most important general therapy in these patients to avoid encephalopathic crises, neurological deterioration and inadequate nutrition.

GOALS: - To show the casuistry and nutritional management in patients with GA-1. - To provide family members and/or care givers with education in the nutritional management of patients with GA-1.

PATIENTS AND METHODS: Ten patients diagnosed with GA-1 with different phenotypes were followed nutritionally and clinically, all under a lysine, hydroxylysine and tryptophan restricted diet. Tables of specific amino acid and anthropometric requirements were used to evaluate nutritional status. General biochemical indices and determination of amino acids were measured. All families were trained with special diets and food selection. RESULTS: The gastrostomy performed on 3/10 patients allowed for improved nutritional management, adherence to diet and growth; 2/10 patients died due to intercurrent infections; 2 other patients evolved very well neurologically, one thanks to neonatal diagnosis and early treatment and the other despite receiving a partial diet. The rest of the casuistry presents severe dystonic symptoms with nutritional compromise. Food education for family members helped to improve the nutritional management of children by exchanging knowledge and motivations. CONCLUSIONS: Nutrition is the most useful tool in inborn errors of intermediate metabolism. GA-1 with a close interaction between nutrients and disease requires specific dietary management. Interdisciplinary management, education and the integration of family members as part of the team are the key elements for good nutritional management.