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Glutaric aciduria type 1: Diagnosis, clinical features and long-term outcome in a large cohort of 34 Irish patients

Glutaric Aciduria type 1 (GA1) is a rare neurometabolic disorder that can lead to encephalopathic crises and severe dystonic movement disorders. Adherence to strict dietary restrictions, in particular a diet low in lysine, carnitine supplementation and emergency treatment in pre-symptomatic patients diagnosed by high-risk screen (HRS) or newborn screen (NBS) leads to a favourable outcome.

We present clinical and biochemical characteristics and long-term data of Irish patients up to 40 years of age. Sixteen patients present clinically: median age at diagnosis of one year. Eighteen patients on high-risk screen and newborn screening: median age at diagnosis, four days.

Clinical events occurred after six years of age were unused but happened.

So, we support the recommendation for diet, carnitine supplementation, as well as emergency treatment for life.

Biography

Prof. Monavari has more than 20 years' experience in Inherited Metabolic Disorders in Ireland, the United Kingdom and Canada. He has been a permanent consultant in The National Centre for Inherited Metabolic Disorders (NCIMD) for more than 10 years. He is the Clinical Director of the NCIMD. Prof. Monavari is President and co-founder of the Irish Society of Inherited Metabolic Disorders (ISIMD). Prof. Monavari is interested in all Inherited Metabolic Disorders. His clinical and research interests include Glutaric Aciduria, Urea Cycle Defects, and Maple Syrup Urine Disease (MSUD).

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