

GLUTARIC ACIDEMIA, A PRELIMINARY SURVEY ON IRANIAN CHILDREN

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Abstract

Objective

Glutaric aciduria type 1 (GAL 1) is a cerebral organic academia, which manifests as encephalopathy with long-term neurological handicap. In this study, clinical presentation, neuroimaging, molecular finding of CGDH mutation of our patients were reviewed.

Materials and Methods

This was a descriptive and cross-sectional study. Patients in whom GLA1 were suspected by clinical manifestation, neuroimaging or metabolic study during last 10 years (2001-2011) in pediatric Mofid hospital were tested for CGDH gene mutation.

Results

Patients age range at study times were 15-102 months. Patients' ages at diagnosis time were 5-17 months. All of parents of our patients were relative. Clinical presentation in order were: developmental delay (54%), macrocephaly and seizure (45%), dystonia (36%), neurodevelopmental regression (27%), acute encephalopathy after fever and vaccination (18%). Neuroimaging finding in brain CT scan and MRI in majority of patients included brain atrophy, widely open sylvian fissure and basal ganglia calcification. Enzymatic study was not performed. Molecular testing results of CGDH in all patients were abnormal. A new mutation in CGDH was detected in our patients.

Conclusion

GLA1 has a protean clinical presentation with different neurological sequel. It is essential to detect patients by newborn screening . Molecular testing and enzymatic study of CGDH activity establish the diagnosis of patients and prenatal diagnosis, too.

Keywords: Glutaric aciduria; CGDH gene mutation; diagnosis.

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