Carnitine Transporter Deficiency

How to Cite This Article: Alaee MR. Carnitine Transporter Deficiency. Iran J Child Neurol Autumn 2013; 7:4(suppl. 1):14.

Mohammad Reza ALAEE MD

Primary carnitine deficiency or carnitine transporter deficiency is an inborn error of fatty acid transportation caused by a defect in carnitine transport across plasma membrane and impairs the entry of long-chain fatty acids into the mitochondrial matrix. Leading to a variety of symptoms such as, cardiomyopathy, muscle weakness, non ketotic hypoglycemia and hepatic involvement. Acute episodes may be induced by metabolic stress such as prolong fasting, infections or vomiting.

Pathophysiology: Primary carnitine deficiency is caused by a defect in the plasma membrane carnitine transporter, with urinary loss of carnitine causing systemic Intracellular carnitine deficiency that impairs the entry of long-chain fatty acids into the mitochondrial matrix. Consequently, impairs the production of ketone bodies due to defect of beta-oxidation and energy production.

Signs and symptoms: The presentation of patient is varied, from asymptomatic to lethal cardiac manifestations. May be present with non ketotic hypoglycemia encephalopathy, seizure, apnea, myopathy, liver dysfunction and cardiomyopathy

Diagnosis: Primary carnitine deficiency must be considered in every child with emergency situation such as seizures, apnea, coma, cardiomyopathy, or episode of hypoketotic hypoglycemia, encephalopathy and Reye syndrome presentation. Hyperuricemia and Elevated Ammonia and liver transaminases and serum CK levels may be observed in primary carnitine deficiency. On the other hand several patients with primary carnitine deficiency have been detected by newborn screening programs.

Enzyme and Carnitine transport assay in cultured fibroblasts and Mutation analysis are methods for confirmation of diagnosis.

Also in imaging study chest x Ray reveals cardiac enlargement and in the echocardiogram cardiac enlargement and increased thickness of the left ventricular wall. ECG reveals left ventricular hypertrophy and peaked T waves in primary carnitine deficiency.

Treatment: In acute attack with hypoketotic hypoglycemic encephalopathy, start with 4cc/kg D/W 10% and then infusion of serum D/W 10%.

IV carnitine 100 to 200 mg/kg/day in acute attack and continue with oral carnitine.

Management of cardiomyopathy

Oral carnitine therapy in primary carnitine deficiency improves, cardiac function, fasting ketogenesis , growth, and cognitive performance.

Keywords: Carnitine transporter deficiency; Cardiomyopathy; Non ketotic hypoglycemia

Pediatric Endocrinology and Metabolism Department, Mofid Children Hospital, Faculty of Medicine, Shahid Beheshti University of Medical Sciences, Tehran, Iran

Corresponding Author:
ALAEE MR .md
Mofid Children Hospital, Shariati Ave, Tehran, Iran
Tel: +98 21 22227021
Email: alaee2003@yahoo.com