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THE FOLLOWING IS THE ABSTRACT OF A PLATFORM PRESENTATION AT THE INTERNATIONAL CONGRESS ON INBORN ERRORS OF METABOLISM (ICIEM), HOSTED BY THE SOCIETY OF INHERITED METABOLIC DISORDERS (SIMD).

MCT OIL BASED DIET REVERSES HYPERTROPHIC CARDIOMYOPATHY IN A PATIENT WITH VLCADD

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Fatty acids are oxidized as a major source of fuel by some organs of the body especially during fasting, sustained aerobic exercise and stress. Very long-chain acyl-CoA dehydrogenase deficiency (VLCADD) is one of the genetic defects of mitochondrial fatty acid beta-oxidation presenting in early infancy or childhood manifesting with episodes of hypoketotic hypoglycemia and cardiomyopathy and may also be associated with hepatomegaly, hyperammonemia and high CPK because of rhabdomyolysis. If undiagnosed and untreated, VLCADD may be fatal secondary to the cardiac involvement. Treatment of patients with VLCADD is based on nutrition therapy and involves avoiding fasting, frequent feeding with a diet rich in carbohydrate and MCT and treating episodes of hypoglycemia with IV glucose and possibly L-carnitine. MCT does not require VLCAD for metabolism and has a primary therapeutic effect as a source of energy in patients with VLCADD. Some data suggest that MCT may help prevent hypoglycemia. We assessed the effect of replacing part of the fat in the diet of a 2.5 month old male who presented with vomiting and dehydration with MCT and essential fats. The patient had been found to have persistent elevation of liver function tests, hepatomegaly and pleural effusion. A chest x-ray revealed cardiomegaly and an EKG noted sinus rhythm with a right bundle branch block. Biventricular hypertrophy alongwith pericardial effusion was confirmed on an echocardiogram. Because of the cardiomyopathy, hepatomegaly and abnormal acylcarnitine profile and urine organic acids, he was diagnosed as having VLCADD. This was confirmed via both enzymatic and molecular studies. He was begun on an MCT based formula with added essential fatty acids, uncooked cornstarch (around 1 year of age) and frequent feeds. Due to the aversion to the taste of the formula, G-tube was placed for improved compliance. By 7 months of age, the cardiomyopathy had reversed and by 14 months of age all cardiac medications were discontinued and hypotonia had improved such that physical therapy was no longer required. At 5 years of age he is at the 50th centile for height and weight and has normal development. Further clinical studies with a larger sample size are needed to determine optimal nutrition care for patients with these genetic metabolic disorders.

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