CR57 Mild carnitine uptake defect due to a novel homozygous mutation in the SLC22A5 gene detected by newborn screening

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INTRODUCTION/OBJECTIVES: Carnitine uptake defect (CUD) is a rare autosomal recessive disorder caused by pathogenic variants in the SLC22A5 gene, resulting in primary carnitine transporter (OCTN2) deficiency and disturbed fatty acid oxidation. Patients may present in infancy with hypoketotic hypoglycemia, metabolic crisis, muscle weakness, cardiomyopathy, or sudden death, while some may remain asymptomatic even if not treated. We present a patient detected by newborn screening (NBS) who harbored a previously unreported homozygous variant in the SLC22A5 gene. OCTN2 activity testing showed mild deficiency.
CASE PRESENTATION: Third child of healthy and unrelated parents was screened positive for CUD by NBS (free carnitine 4.5 µmol/L (cutt-off 8.8)). Follow-up testing showed low free carnitine in plasma and normal carnitine in maternal blood. The patient was supplemented with L- carnitine and the parents were advised about the feeding plan and crisis management. Genetic analysis identified a homozygous variant c.820_821delTGinsGA (p.Trp274Glu) in the SLC22A5 gene, predicted to be damaging but unreported earlier, thus classified as a variant of unknown significance. OCTN2 activity in fibroblasts was 25% of the control value. The treatment and patient monitoring plan were adjusted accordingly.
CONCLUSION: CUD can cause serious medical complications if not treated. Patients diagnosed presymptomatically by NBS have an excellent prognosis. Phenotypic variability poses challenges for individualized treatment and patient management plans, especially in patients with novel variants. Confirmation of significant residual activity by functional testing in the presented patient assured tailored treatment and patient management plan, and reduced disease burden.

CR58 ModuLifeTM- a novel dietary approach in management of a patient with moderate Crohn's disease

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KEYWORDS: Crohn's disease; Crohn's disease exclusion diet; dietary therapy

INTRODUCTION/OBJECTIVES: microbiota, host barrier function, innate immunity, and the Crohn's disease (CD) is a chronic inflammatory bowel disease of unknown aetiology. Evidence demonstrate that a person's diet could be implicated in the pathogenesis of CD. Therefore, there is a growing interest in the use of diet as treatment or adjuvant therapy for CD. ModuLife is a dietary approach based on the Crohn's Disease Exclusion Diet (CDED) combined with a specific Partial Enteral Nutrition (PEN) formula, that aims to reduce exposure to dietary components hypothesized to negatively affect the microbiome, innate immunity and intestinal barrier.
CASE PRESENTATION: We present a case of a 34-year-male patient who was diagnosed with CD at the age of 31, after resection of 40 cm of jejunum and jejuno-jejunal anastomosis was performed due to small bowel stenoses. At that time, the patient was severely malnourished (BMI 17,4 kg/m^2). After stabilization, he started with the second phase of Modulife diet in which 75% of his daily caloric and nutritional needs were met from specific foods and the other 25% from a specific PEN formula. During 6 months of ModuLife diet, patient nutritional status improved significantly (BMI 21.5 kg/m^2) and he achieved clinical and biochemical remission. Currently, he is in deep remission and is still on the ModuLife diet, PEN intake is also reduced.
CONCLUSION: Modulife is a promising dietary approach for inducing and maintaining remission in mild to moderate CD. Due to the restrictive nature of this diet, it is important to work with a qualified health professional to ensure that the diet meets patients nutritional and caloric needs.