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Riboflavin non-responsive multiple acyl-CoA dehydrogenase deficiency (MADD) with early severe cardiomyopathy: Favorable long-term outcome on D,L-3-hydroxybutyrate (OHB) supplementation

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Abstract

Background:

MADD results in deficient electron transfer from FAD-dependent dehydrogenases to the mitochondrial respiratory chain. The riboflavin non-responsive phenotype presents as a potential neonatal life-threatening disorder complicated by severe acidosis, hyperammonemia, hypoglycemia and seizures. Early severe cardiomyopathy is frequent and only very few patients are known to have reached adulthood as the treatment of this disorder is very difficult. Orally supplemented OHB is an additional treatment option to be considered in severe cases.

Case study:

We report on 2 cousins (now 18 and 19 years old) with enzymatically and genetically confirmed riboflavin non-responsive MADD. There is a strong family history of severe MADD: 4 siblings died in neonatal period. Both cousins developed a neonatal life-threatening cardiomyopathy unresponsive to conventional treatment. After commencing OHB, the cardiac contractility showed progressive and sustained improvement. The elder cousin presented with a prolonged out-of-hospital cardiac arrest presumed secondary to ventricular arrhythmias in association with cardiomyopathy at the age of 12 years. He was successfully resuscitated. As a preventive measure, both children underwent an implantable cardioverter-defibrillator (ICD) insertion. The younger cousin needed MitraClip placement at the age of 18 years, because of severe mitral valve regurgitation on a background of a longstanding history of left atrial enlargement.

Discussion:

Early and long-term treatment with OHB is a promising lifesaving therapeutic add-on option for patients with severe MADD. It has ameliorated the potentially lethal outcome in our patients. However the risk of long term complications, particularly cardiac life-threatening events including arrhythmias and cardiomyopathy necessitate careful monitoring and management.

Biography:

Friederike Seggewies has earned her medical degree at the Medical University Graz, Austria. She started her work carreer at the University Hospital Eppendorf, Germany in the position of a resident physician at the pediatric cardiology (Head of the department: PD Dr. Kozlik-Feldmann). After 2 years of experience in cardiology she amplified her knowledge in pediatric metabolic medicine at the University Hospital Eppendorf, Germany (Head of the department: Prof. Dr. Ania Muntau). In 2019 she did a metabolic observership at the Great Ormond Street Hospital, London, where she got to know Dr. Grunewald. With her she worked on the above-mentioned project.

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