Abstract:

The bachelor thesis deals with rare hereditary metabolic disorders, LCHAD (long chain 3-hydroxyacyl-coenzyme A dehydrogenase) deficiency and MTP (mitochondrial trifunctional protein) or TFP (trifunctional protein) deficiency. Deficiencies of these enzymes cause a disturbance in the cycle of β-oxidation of long-chain fatty acids. The basis of treatment for these disorders are dietary and regimen measures. The main goal of the bachelor´s thesis was to determine the level of compliance in dietary and regimen measures and calcium intake in patients with LCHAD/MTP deficiency.

The theoretical part first describes hereditary metabolic disorders as a large heterogeneous group of serious hereditary disorders, some of which occur rarely. These rare diseases include LCHAD/MTP deficiency. First, general findings of hereditary metabolic disorders, their history, occurrence, heredity, pathogenesis, diagnosis, clinical manifestations and treatment, are briefly summarized. Furthermore, the theoretical part of the thesis deals with fatty acids as a nutrient, which plays a crucial role in the diet of a patient with LCHAD/MTP deficiency, as well as the β-oxidation process, which is disrupted in these deficiencies. The main part of this thesis is devoted to the disease itself, the LCHAD/MTP deficiency. It describes the heredity of the disease, diagnosis, clinical manifestations and treatment of these disorders. The conclusion of the theoretical part of this bachelor thesis is devoted to calcium, a mineral substance that is indispensable for the human organism. The role of calcium in the human body, its homeostasis, the consequences of its deficiency, sources of calcium in food and the recommended daily intake are described.

In the practical part of the thesis, the result of research that included all patients with LCHAD/MTP deficit who were diagnosed in the Czech Republic at this time, i.e. twenty-two patients, are presented and evaluated. All patients were required to record their dietary regimen of at least three days, including the dietetic preparations, supplements and modular dietetics used. Furthermore, the laboratory parameters related to calcium metabolism and anthropometry were evaluated. An analysis of the dietary regimen was carried out and the data were compared with doctor´s orders.

The results of the research showed that patients with LCHAD/MTP deficiency who are predominantly energy receiving from a special infant formula (Lipistart, Nutrilon Allergy Digestive Care) or a combination of a special infant formula and food for special medical purposes (Milupa Basic F) receive sufficient calcium doses in their diet and meet its reference daily intake (RDI). Patients who no longer consume these formulas or consume them in a minimal dose and draw calcium mainly from a complementary food, no longer meet the RDI of calcium. From the data found, it can be stated that calcium supplementation should be an integral part of nutrition therapy for patients with LCHAD/MTP deficiency, who take major part of energy from a complementary food.

The research also showed that the laboratory levels of serum calcium, phosphorus, parathyroid hormone and alkaline phosphatase were within the reference values in all patients. Calcidiol levels (25-OHD concentrations) were deficient, especially in the first and the fourth quarter of the year in all patients over one year of age. It is therefore appropriate to ensure sufficient vitamin D supplementation during these months. Anthropometric data suggest that despite the frequent food serving with polysaccharides content and MCT fats, efficient nutrition management can keep energy intake under control and thus avoid other health complications associated with overweight or obesity. As further research of this bachelor's thesis showed, it is appropriate to regularly monitor the actual intake of particular nutrients, energy intake and a level of a compliance with diet and regimen measures, re-educate patients and their family members and repeatedly obtain feedback on their knowledge of nutrition management.

Key words: LCHAD deficiency, MTP deficiency, TFP deficiency, hereditary metabolic disorders, β-oxidation, fatty acids, calcium, compliance, nutrition therapy