Lessons in diet therapy in long chain 3-hydroxyacyl-CoA dehydrogenase deficiency (LCHADD)

Brooke Pinsent1,2, Erin Mullane1,2, Kristen Fitzell1,2, Maureen Evans1
1 Department of Metabolic Medicine, Royal Children’s Hospital Melbourne.
2 Department of Nutrition and Food Services, Royal Children’s Hospital Melbourne.

Long chain 3-hydroxyacyl-CoA dehydrogenase deficiency (LCHADD) results in impaired β-oxidation of long chain fatty acids. Dietary management includes reduced fasting times, long chain triglyceride (LCT) restriction and supplement of medium chain triglycerides (MCT). We report on dietary changes to stabilise a 3-year-old boy with LCHADD.

Methods:
Data were collected retrospectively from medical records. Dietary manipulations were made to optimise metabolic and/or symptom control. Key outcomes included admission days (per month) for decompensation or gastrointestinal (GIT) symptoms. Decompensation admissions were defined as a creatine kinase (CK) level >1000. GIT admissions were defined as presence of vomiting and/or diarrohoea, and where CK was <1000. Growth trends were noted through analysis of World Health Organization (WHO) growth charts, corrected for prematurity.

Results:
Figure 1: major dietary changes over admissions in first 3.5 years of life

- Change (a) increased fasting time; patient was previously tolerating 4 hours max fasting at timepoint (a), at timepoint (b) (11 months old) fasting overnight was increased to 7 hours which was poorly tolerated evidenced by increased decompensation admissions from 0.63 day/month to 10.5 days/month respectively.
- Change (c) formula change; change to reduced total fat content from 4.53g/100kCal to 2.9g/100kCal and reduced LCT content from 8% total calories as LCT to 4% as LCT resulted in less decompensation admissions 10.5 days/month to 0.3 days/month.

Figure 2: formulas 1 and 2 - macronutrient distribution range

Contact details: metabolic.dietitians@rch.org.au

Results:

- Change (d)(e) increased MCT supplementation; after increased activity at 3 years old, further MCT was provided in addition to formula using an emulsified MCT product. This stabilised CK and reduced decompensation admission but increased GIT admissions from 0.45 days/month to 1.5 days/month.
- Change (f) altered MCT supplement: change from liquid MCT emulsion to a powdered MCT product with protein and carbohydrate. This and altered dose timing, resulted in nil GIT admissions to date.

Figure 3: formulas 3 and 4 - macronutrient distribution range

Figure 4: Growth trends with increased calorie intake

Increased calories were required to stabilize patient, this resulted increased weight percentile from 5%ile CGA at birth to 95%ile CGA by 2-3 years old. Calories when well were reduced to stabilise weight. At 3 years old a reduction to 95%ile with subsequent tracking was achieved.

Discussion:
- Frequent acute illness management with IV Dextrose and increased calories contributed to excessive weight gain.
- Decreased LCT and increased MCT resulted in decreased admissions for metabolic decompensation.
- Reduced fasting tolerance required overnight continuous feeds, with daytime boluses to stabilize our patient.
- MCT related GIT symptoms, including nausea, vomiting and diarrhoea, without metabolic decompensation existed in our patient.
- Altered timing and type of MCT product were successful dietary manipulations to control GIT symptoms and admissions.

Conclusion:
Reduced LCT fat intake and changes to supplemental MCT product and dose timing achieved stabilisation in this patient.