Mitochondrial Long Chain Fatty Acid Oxidation and Carnitine Defects in a Single Canadian Metabolic Genetics Clinic

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Background
- LCFAOD and carnitine defects are rare inherited metabolic disorders
- Clinical features:
  - Muscle weakness
  - Exercise-induced muscle pain
  - Cardiomyopathy/cardiac rhythm disturbances
  - Hypoglycemia
  - Coma or death if untreated

Treatment
1. Diet
   - High carbohydrate, low fat diet
   - Restricted long-chain fat diet
2. Carnitine supplementation
3. Illness management
   - High carbohydrate intake during intercurrent illness
   - If decreased oral intake, hospital admission and IV fluid treatment

Materials & Methods
- Retrospective cohort study
- Diagnosis
- Diet
- Variant classification
- Excel database

Results
Participants
- 39 patients included (21 children & 18 adults)
- Mean age: 20.4±17.6 years (3 mo – 55 y)

Investigations
- 50% identified via NMS
- 32 diagnosed via genetic testing
- All confirmed via biochemical methods

Treatment
- 10 patients on the long-chain fat restricted diet
  - CPTII (n=2), LCHAD (n=3), VLCAD (n=4), MAD (n=1) deficiencies
  - Restriction: 10% - 35% VS Intake: 6% - 44%
  - There were 16 patients on supplemented carnitine
  - All patients excluding CUD received illness management

Discussion
- NMS identified 49% of patients
- Prevalence of LCFAOD was 4.75%
- 25.6% of patients had diet management