Canada-wide Long-chain Fatty Acid Oxidation and Carnitine Metabolism Disorder Research Study: We need your help!

Researchers at the University of Alberta, Department of Medical Genetics and Stollery Children's Hospital are completing a study looking at clinical symptoms and investigations in patients with long- chain fatty acid oxidation (LC-FAO) and carnitine metabolism disorders.

Who can participate:

- All patients diagnosed with
  - Carnitine palmitoyltransferase I (CPTI) deficiency
  - Carnitine palmitoyltransferase II (CPTII) deficiency
  - Carnitine-acylcarnitine translocase (CACT) deficiency
  - Very-long-chain acyl-CoA dehydrogenase (VLCAD) deficiency
  - Long-chain 3-hydroxyacyl-CoA dehydrogenase (LCHAD) deficiency
  - Multiple acyl-CoA dehydrogenation (MAD) deficiency
  - Trifunctional protein (TFP) deficiency
  - Carnitine uptake defect (CUD)

What will we learn:

- Information about the care and outcomes of patients with LC-FAO and carnitine metabolism disorders
- Information about life-threatening events of children and adults affected by LC-FAO and carnitine metabolism disorders.
- About the severity of patients with LC-FAO and carnitine metabolism disorders during different treatments
- Information about the immune functions of patients with LC-FAO and carnitine metabolism disorders
- Information that is meaningful to health care providers and our public health systems

What will you do:

- Your metabolic physician will provide us information about your medical history
- If you are a patient in Edmonton, Alberta, you may be asked to provide a blood sample at your next appointment

Contact: If you have any questions or would like to participate in this study, please contact the study PI by contacting **saadet@ualberta.ca** and **aambrose@ualberta.ca**

Please note communication by e-mail is not absolutely secure. Thus, do not communicate personal sensitive information by e-mail.

The project is titled "*Generation of national real-world evidence for patients with long-chain fatty acid oxidation defects in Canada*" and the principal investigator of this study is Dr. Saadet Andrews, MD, PhD, FCCMG, FRCPC in the Department of Medical Genetics, University of Alberta