Ad for Fatty Oxidation Disorders Family Support Group website

Volunteers Needed for Research Study on Carnitine Palmitoyltransferase II Deficiency

The Robert Guthrie Biochemical Genetics Laboratory at the State University of New York at Buffalo is currently seeking participants for a research study.

The purpose of the study is to determine if a relationship can be made between the clinical presentation of CPT II deficiency, the confirmatory biochemical and molecular test results, and a test for elevated acylcarnitine in blood. If the relationship can be made, then the investigators hope to predict the expected onset and severity of symptoms in newborns detected with CPT II deficiency in the newborn period. Parents of newborns with an early diagnosis are concerned about whether their baby will become symptomatic, when they will become symptomatic and the extent to which they will be affected by the disorder.

Participants should be adults or children who have:

(1) a complete diagnosis of carnitine palmityoltransferase (CPT) II deficiency by clinical and laboratory testing including but not limited to enzyme analysis in cells or tissues such as muscle biopsy or culture skin fibroblasts and specific mutation detection.

(2) an incomplete diagnosis of CPT II deficiency, that is, the symptoms of CPT II deficiency are present and some laboratory work has been performed that is presumed to be diagnostic but the patient may not have had mutation analysis to identify the specific disease-causing mutations in their case.

Both groups of participants will be asked to collect a few drops of blood from a finger prick onto filter paper cards that will be provided. Testing will be performed to measure a substance called acylcarnitine which has been shown to be abnormally elevated in some cases of CPT II deficiency as well as other fatty acid oxidation disorders, however, it has never been studied in a large group of patients of all ages with the disease to attempt to correlate its concentration in blood with the severity of the disease.

Patients with an incomplete diagnosis will also be asked to collect a small sample of inner cheek cells, called buccal cells, using a soft brush that will be provided in a kit by the investigators. This sample will be used for isolation of genetic material (DNA) to search for specific mutations causing the disorder. There will be no cost to those who participate in the study.

Parents of newborns who have been diagnosed with CPT II deficiency or at increased risk for a diagnosis of CPT II deficiency should contact the Principal Investigator, Dr. Vladutiu, directly to consider confirmatory testing through the Guthrie's Laboratory's clinical testing service before participation in the research study.

For additional information about participation in the study, please call Cathy Kern at 716-829-2695 Tuesday through Thursday between 8AM and 4PM Eastern standard time or by email at ckern@buffalo.edu .

The study protocol has been approved by the Health Sciences Institutional Review Board of The University at Buffalo. The Principal Investigator is Dr. Georgirene D. Vladutiu, Professor of Pediatrics, Neurology and Pathology at the School of Medicine and Biomedical Sciences, The University at Buffalo. Contact information: Room A-767, The Buffalo General Hospital, 100 High Street, Buffalo, NY 14203. Phone: 716-859-7741.