Fatty Acid Oxidation Disorders

• Fatty acid oxidation disorders are a group of inherited (passed from parent to child) conditions that occur when the body is unable to turn fat from food into energy.

• Babies with fatty acid oxidation disorders may have vomiting, muscle weakness, and other health problems. Special diets and medications are available to prevent these health problems.

• Indiana’s newborn screen tests for the following fatty acid oxidation disorders:
  – 2,3-Dienoyl-CoA reductase deficiency
  – Carnitine-acylcarnitine transferase deficiency (CAT)
  – Carnitine palmitoyltransferase deficiency I (CPT Type I)
  – Carnitine palmitoyltransferase deficiency II (CPT Type II)
  – Electron transfer flavoprotein (ETF) deficiency (Multiple acyl-CoA dehydrogenase deficiency)
  – Electron transfer flavoprotein:QO deficiency
  – Medium chain acyl-CoA dehydrogenase deficiency (MCAD)
  – Primary carnitine deficiency (Carnitine uptake defect)
  – Short chain acyl-CoA dehydrogenase deficiency (SCAD)
  – Short chain hydroxyacyl-CoA dehydrogenase deficiency (SCHAD)
  – Trifunctional enzyme deficiency
  – Very long chain acyl-CoA dehydrogenase deficiency (VLCAD)
  – Very long chain hydroxyacyl-CoA dehydrogenase deficiency (VLCHAD)

• For more information about fatty acid oxidation disorders, please click on one of the links below.
  – Medline Plus Medical Encyclopedia
  – National Library of Medicine Genetics Home Reference
  – STAR-G (Screening, Technology, and Research in Genetics)