



The Children's Hospital of Philadelphia

34th Street and Civic Center Boulevard, Philadelphia, PA 19104-4399

RESEARCH STUDY ADVERTISEMENT

Mitochondrial disease consists of hundreds of different energy-related, genetic-based disorders. No single laboratory test can accurately diagnose mitochondrial disease in most patients. Furthermore, no effective treatments exist for many mitochondrial disease patients. An NIH-sponsored study at the Children's Hospital of Philadelphia entitled, "**Metabolic Consequences of Primary Mitochondrial Disease**", is recruiting subjects with *definite* biochemical and/or genetic causes of mitochondrial disease to identify a "signature" of metabolic changes that may offer the opportunity for improved mitochondrial disease diagnosis and treatment.

This study is designed to primarily evaluate *previously collected* muscle, skin, and blood from individuals with definite mitochondrial disease. New blood and/or skin specimens will only be requested if no prior specimens are available. Tissue sample(s) will be studied to detect patterns of secondary biochemical and/or genetic alterations that occur in mitochondrial disease.

To learn more about participation in this study, please contact the Research Study Coordinator, Emily Place, MS, directly at (267) 426-9650 and/or placee@email.chop.edu.