

Region 4 Natural History Proposal

Aims and Objectives (DRAFT)

Aim 1- To better understand the relationship between NBS values, genotype and early manifestations and complications of inborn errors of metabolism

- a. To better understand the relationship between NBS values and acuity
- b. To examine the utility of genotype in determining treatment and management decisions
- c. To increase knowledge about the impact of early intervention and treatment on metabolic conditions including secondary targets

Aim 2 – To establish evidence based approaches for managing and treating children with metabolic disorders.

- a. To describe current interventions (nutritional and therapeutic) for treating children with metabolic disorders
- b. To evaluate current interventions (nutritional and therapeutic) for effectiveness
- c. To determine the effectiveness of early intervention and treatment for specific metabolic conditions, including secondary targets

Aim 3 - To overcome barriers to the collection and analysis of long term follow-up data for children with metabolic disorders.

- a. To establish an environment where clinicians engaged in the care of persons affected with inborn errors of metabolism collaborate efficiently and effectively to improve outcomes for those individuals
- b. To provide incentives for data collection
- c. To institute a process that encourages data analysis

Children's Memorial Hospital
Cincinnati Children's Hospital
Nationwide Children's Hospital
Riley Hospital for Children
St. Francis Hospital
Sanford Children's Hospital
University of Illinois
University of Michigan
University of Minnesota
University of Missouri – Columbia
University of Pittsburgh
Waisman Center
Wayne State University