

Inborn Errors of Metabolism Information System, \*A. Jurek (University of Minnesota, Minneapolis, MN 55455).

Inborn errors of metabolism (IBEM) are rare genetic conditions in which an enzyme deficiency causes a metabolic error. Treatments need to be condition-specific because each IBEM has different symptoms. Current IBEM treatment methods are based primarily on a metabolic practitioner's personal experience. To improve this, metabolic clinicians and newborn screening specialists from seven state health departments in the Region 4 Genetics Collaborative sought to monitor differences in clinical practices and to determine the most effective treatment strategies for metabolic disorders diagnosed by newborn screening. As a result, the IBEM-Information System (IBEM-IS), a web-based disease registry, was created by this workgroup to gather uniform data and assess clinical practice differences. The IBEM-IS registry will track long-term follow up of newborn screening and clinical outcomes to be used as a platform for research regarding new strategies for treatment of IBEM. As of January 2009, the registry includes 62 cases: 46 medium chain acyl-CoA dehydrogenase deficiency, 7 maple syrup urine disease, 4 very long chain acyl-CoA dehydrogenase deficiency, 3 short chain acyl-CoA dehydrogenase deficiency, 1 long-chain 3-hydroxy acyl-CoA dehydrogenase deficiency, and 1 trifunctional protein deficiency enrolled by centers in 6 states. We discuss the registry, expansion to other Regional Genetics Collaboratives, and integration of data with other electronic information systems, all endeavors designed to ultimately improve care for children with rare genetic disorders. This project is funded by Cooperative Agreement U22MC03963, the Region 4 Genetics Collaborative, from HRSA/MCHB.

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