

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. Treatment options 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 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 to gather uniform data and assess clinical practice differences. The IBEM-IS registry will track long-term newborn screening follow up and evaluation of clinical outcomes to be used as a platform for research. We discuss the registry, expansion to other genetics regions, and integration of data with other electronic information systems, which will ultimately improve care for children with rare genetic disorders.

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