

Abstract

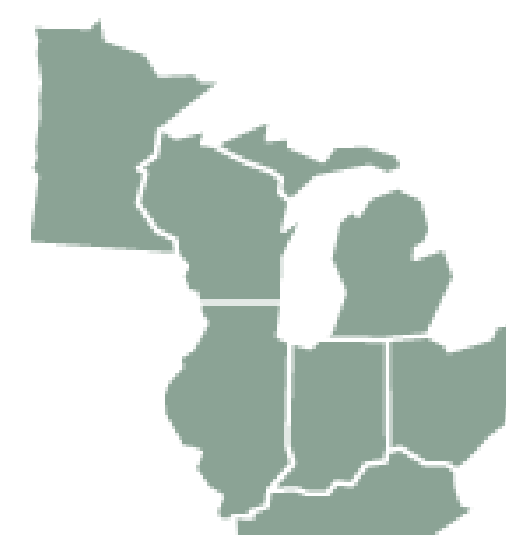
Inborn errors of metabolism (IBEM) are rare genetic conditions in which an enzyme deficiency causes a metabolic error. Treatments need to be condition-specific because each IBEM has different symptoms. Current IBEM treatment methods are based primarily on a metabolic practitioner's personal experience. 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.

Background

Inborn errors of metabolism (IBEM) are rare genetic conditions in which an enzyme deficiency causes a metabolic error. IBEM include disorders of fatty acid oxidation, organic acidemias, and aminoacidopathies. Many IBEM are detectable by newborn bloodspot screening (NBS) tests, especially by the expanded testing using tandem mass spectrometry, which provides opportunity to identify newborns for illness, early diagnosis and intervention. Treatment options for IBEM are condition specific and are currently based primarily on the metabolic practitioner's personal experience. Long-term prognosis of children with IBEM has been limited by differences in clinical practices and unknown best treatment methods.

Methods

Metabolic clinicians and newborn screening specialists from seven state health departments in the Region 4 Genetics Collaborative sought to improve outcomes from NBS. They were interested in monitoring differences in clinical practices and determining the most effective treatment strategies for metabolic disorders diagnosed by NBS. The seven participating states are Illinois, Indiana, Kentucky, Michigan, Minnesota, Ohio and Wisconsin, all of which use tandem mass spectrometry.

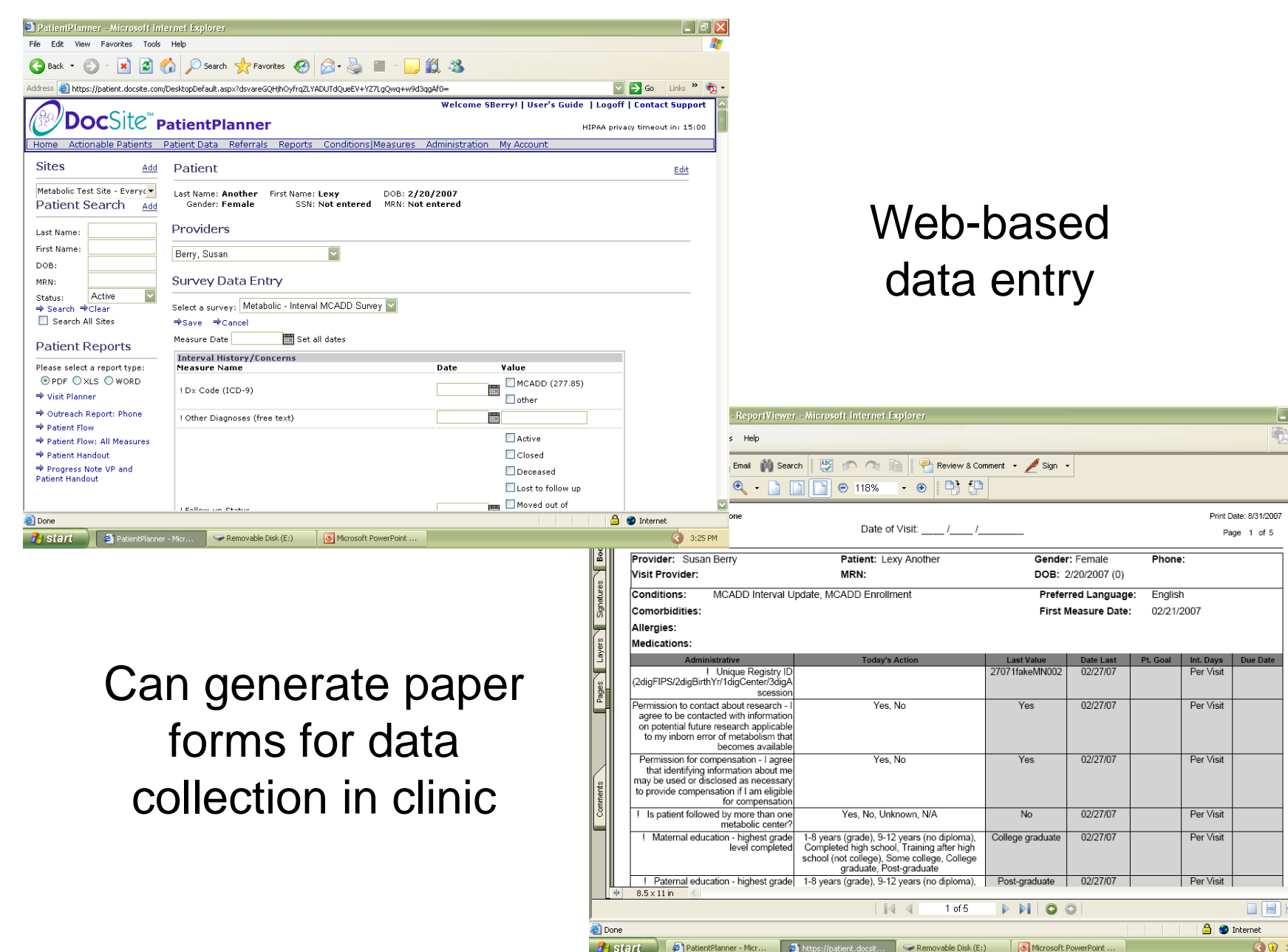


To gather information on patient outcomes, the IBEM-Information System (IBEM-IS) was created by this workgroup to gather uniform data and assess clinical practice differences. The registry began with one condition, medium-chain acyl-CoA dehydrogenase (MCAD) deficiency, because it is one of the more common of the rare IBEM.

Methods Continued

Registry development was guided by the literature and existing database sources, the Mountain States collaborative and Oregon database. Our workgroup wanted to collect information that was already available in the existing databases. We met to agree on essential data elements and general treatment strategies for MCAD deficiency that would be included in the registry. Four main data sections were developed: enrollment, demography, short and long-term follow-up interval surveys, and neuropsychological survey.

Our ultimate goal was to track the health care of children with many genetic disorders. Therefore, to expand the registry we applied for and received special priority funding from the Health Resources and Services Administration (HRSA) to develop and implement a comprehensive IBEM-IS. We met as a workgroup to discuss selection of additional conditions and a strategy for adding new disorders to the registry. This web-based 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. It is expected that the IBEM-IS will result in evidence-based care protocols for kids with metabolic disorders and improve health outcomes.

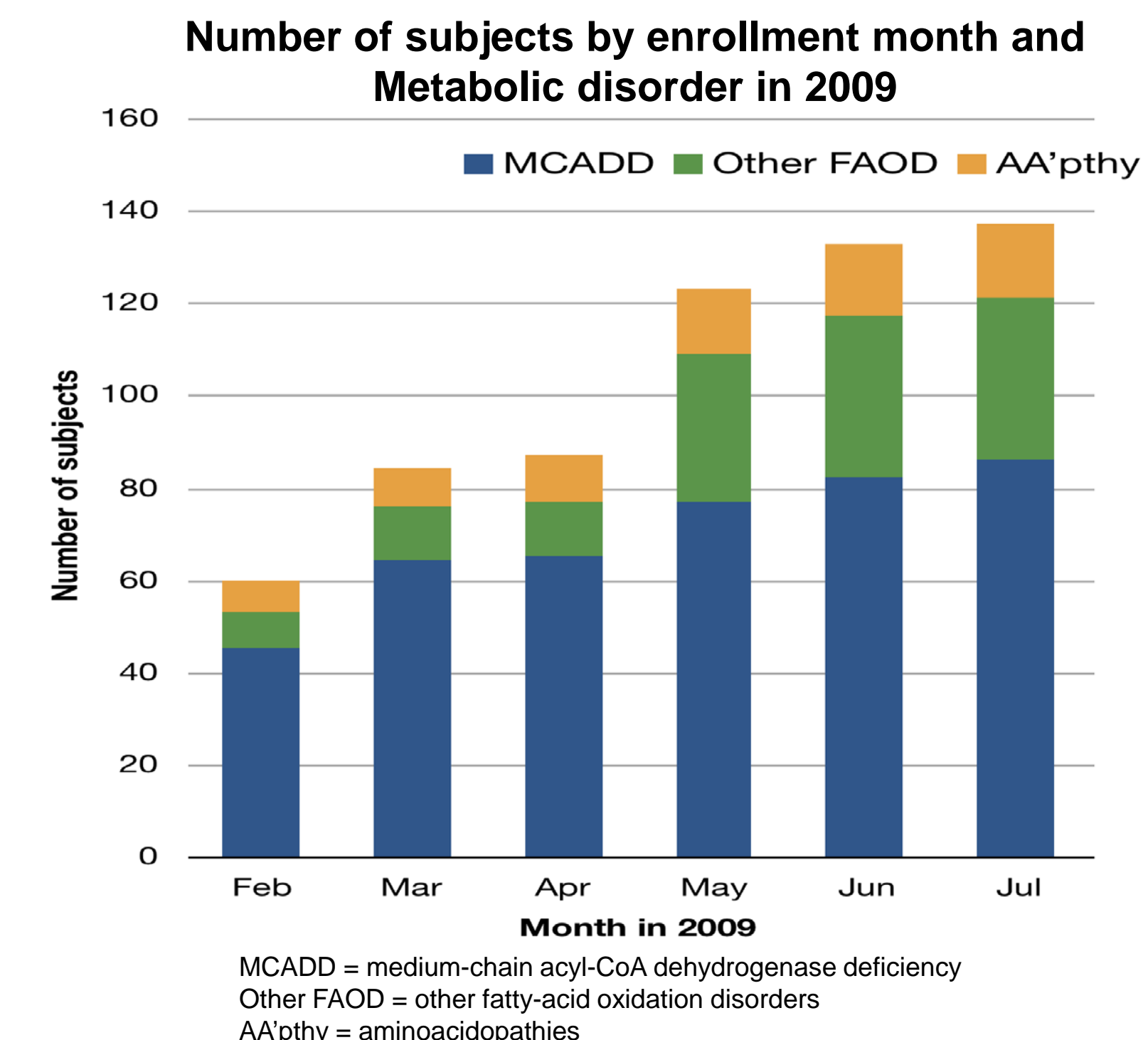


Web-based data entry

Can generate paper forms for data collection in clinic

Future Directions

We are interested in incorporating direct linkages with other electronic records to expand the usefulness of the IBEM-IS. These sources include data from state NBS programs at Departments of Health and the Midwest Emergency Medical Services for Children Information System (MEMSCIS). Additionally, we would like to include collaborations with other regions on this project.



Conclusions

We have:

- Established a collaborative workgroup in IBEM
- Accumulated relatively rapid data collection on patients enrolled in the registry
- Developed a reasonably prompt method to look at natural history and outcomes

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