Development of requirements and a pilot registry for long-term follow-up of children with heritable conditions

Reid Holbrook, MD MPH, Catherine Staes, BSN MPH PhD, Nicola Longo, MD PhD
Jeffrey Botkin, MD MPH, Rebecca Anderson, BS MSc RN, Joyce Mitchell, PhD
University of Utah, Salt Lake City, Utah

ABSTRACT
Advances in newborn screening (NBS) have led to earlier detection of heritable conditions. Little is known about the natural history of these conditions or the long-term benefits of NBS. This study will examine the user and data requirements necessary to develop a long-term follow-up registry for these patients. The system will subsequently be analyzed to determine its usefulness for research and reporting outcomes after long-term follow-up of patients.

BACKGROUND
Infants born in the United States are screened for a number of genetic conditions through mandated newborn screening. These disorders are extremely rare and little is known about the natural history of these conditions and whether early identification by newborn screening is beneficial. The Mountain States Genetic Collaboration (MSGC) has proposed the development of a long-term follow-up registry for conditions identified by NBS. There is a need to describe the natural history of conditions, medical complications, specific therapies used, and cost of care. The purpose of this project is to gather system requirements and develop a working prototype registry of patients with heritable conditions.

METHODS
Requirements for the pilot system will be defined using the following procedures. First, we will define the analyses and outputs required for the registry using care process guidelines recently developed by metabolic clinicians with the MSGC, knowledge and experience of local metabolic specialists, and considering the long-term outcomes defined by MSGC. Second, a system analysis of work and data flow in the University of Utah Metabolic Clinic will be performed. Third, medical records of 40 patients from the 4 major groups of metabolic disorders detectable by NBS (Fatty Acid Oxidation, Amino Acid, Organic Acid, and other metabolic disorders) will be reviewed retrospectively.

Source data will be evaluated for structure, availability, as well as the extent of information already stored electronically in the EMR, including laboratory and clinical records. The various attributes will be ranked by priority for fulfilling the performance indicators and outcome measures defined in the care process guidelines.

Data entry, coding, interface, and storage requirements, and finally the data model will be defined before building a database to support needed data and analyses required by the registry. Data for each patient will be entered by hand or electronically transferred from the EMR. We plan to collect demographic, clinical, laboratory data and documented type of diagnosis for each study subject. The data elements will be limited to those that are required to report or derive the performance indicators and outcome measures outlined in the care process guidelines.

ANALYSIS PLAN
We will evaluate the frequency of the availability of different types of data (paper vs electronic; coded, vs free-text) for the 40 patients. All data will be analyzed for its ability to be coded and transferred electronically from the source to the registry. Data entered into the registry will be analyzed for its completeness as compared to performance indicators developed for the metabolic conditions. Overall system effectiveness will be based on the ability of the data model to support the appropriate acquisition, storage, analysis, and reporting of clinically important data to assess outcomes.

CONCLUSION
We will present results of our investigation. The information generated from this project will inform the development of a region-wide registry to be developed in the future.

REFERENCES