Title

Emerging team in rare diseases: achieving the “triple aim” for inborn errors of metabolism

Project Description

Our team is undertaking a program of research designed to support evidence-informed health care for children with rare diseases, beginning with inborn errors of metabolism (IEM). The team is comprised of experts in health services and policy research, provision of clinical care for children with IEM and is supported by a multidisciplinary advisory board that includes patient and family representatives.

The research program builds on existing partnerships and incorporates a new research framework that highlights the importance of measuring outcomes from a range of perspectives, emphasizing key challenges in developing effective health care for children with rare diseases. To support our research, we are developing a unique information system for Canadian children with IEM. With families’ permission, this information system will enable us to describe children’s health outcomes, their use of health services, and the treatments they receive. Importantly, we will also incorporate patients’ and parents’ self-reported well-being and their experiences with health care. The findings that result from this work will be used to make recommendations for improving care for children with IEM and other rare diseases.

The work of the emerging team will also form part of the establishment of a sustainable research network in the field -- the Canadian Inherited Metabolic Diseases Research Network (CIMDRN). CIMDRN aims to establish and develop collaborative teams, train new researchers and providers and continue to build research synergies in the field of IEM that will ultimately support the improvement of patient health outcomes for children with IEM and their families in Canada.

Principal Investigator

Beth Potter, University of Ottawa