CIMDRN
Canadian Inherited Metabolic Diseases Research Network

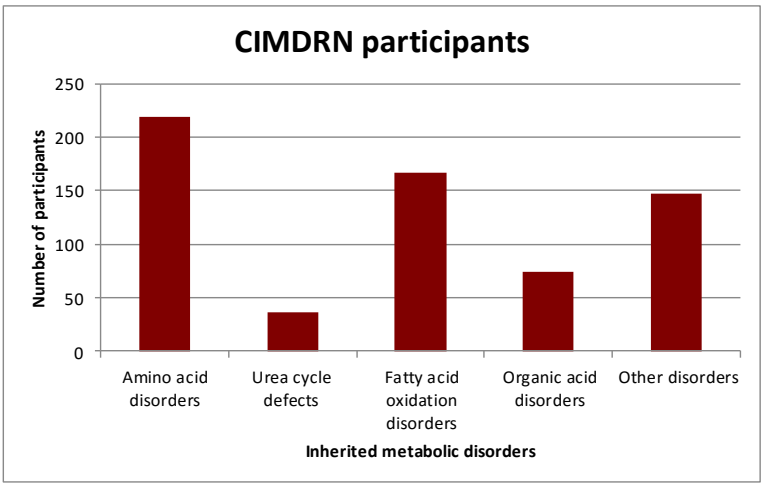
Newsletter

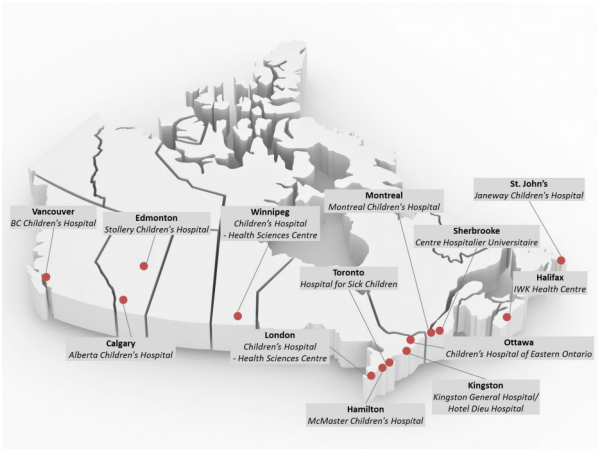
Summer 2018

To our participating families,

Your participation in our research network is what makes our work possible! Thank you for your ongoing contribution to rare disease research!

There are currently 759 participants enrolled in CIMDRN from across the country





- Participants are 51% male, 49% female
- ~80% of participants are between the ages of 2 and 10 years old

RECENT PUBLICATIONS

"Experiences of caregivers of children with inherited metabolic diseases: a qualitative study" in Orphanet Journal of Rare Diseases *(2016)* doi: 10.1186/s13023-016-0548-2

Conferences presented at:

2018 Garrod Symposium, Edmonton, Alberta

39th Annual North American Meeting - Society for Medical Decision Making, Pittsburg, Pennsylvania

2017 Garrod Symposium, Montreal, Quebec

2017 American College of Medical Genetics & Genomics Annual Clinical Genetics Meeting, Phoenix, Arizona

FUTURE PROJECTS

Our funding supporting our original research project is coming to an end. We are happy to announce that CIMDRN has been awarded two grants to continue our research. Our projects will continue to focus on improving care for children diagnosed with inherited metabolic diseases, based on experiences of currently affected families. We are excited to continue our research!

Core Outcomes Set: Surveying Canadian patients and families, health professionals, policy makers, and researchers, we will be creating a core set of outcomes that should be measured in all studies evaluating treatments for children diagnosed with phenylketonuria (PKU) or with medium-chain acyl-CoA dehydrogenase (MCAD) deficiency.

Family-Centered Care: Using surveys and diaries, we are studying families' experiences with care for children with inherited metabolic diseases. We will then work with families and health care providers to design new interventions that are more responsible to the needs of children and families.

Questionnaires for families

Many of you have already participated in a survey of families whose children are participating in CIMDRN. The goal of this study is to describe caregivers' experiences with health care for their children with inherited metabolic disease to identify areas where experience with care can be improved.

Summary of results

The questionnaire is currently being sent to families and data analysis is going. These results summarize findings from 113 respondents who have completed the questionnaire thus far. The majority of respondents have children diagnosed with phenylalanine hydroxylase deficiency or medium-chain acyl-CoA dehydrogenase deficiency.

The role of the metabolic clinic

• 54% of caregivers identified a physician or dietitian at the metabolic clinic as their child's main provider.

• Among those children requiring multiple health services (57%), 56% received care coordination support, most often (68%) from the main provider's office.

• 86% of caregivers were satisfied with care at the metabolic clinic and needs for family-centred care were perceived as being met.

• Needs that were reported as unmet were regarding connections to community services and other resources.

• 55% of respondents travelled at least 1-2 hours to the metabolic clinic, which is a potential barrier to accessing care.

Additional services and factors

• A majority of respondents (>70%) reported that they were satisfied with care within each service outside of the metabolic clinic, including ED care, the pharmacy, and the blood laboratory.

• Across all services, receiving appropriate care and high quality communication from providers were most often rated as essential to satisfaction.

• More than half of families reported that they had out-of-pocket expenses related to their child's care, most often medications and medical foods or supplements.

Initial conclusions

• These findings indicate that caregivers consider the metabolic clinic as central to their child's care and perceive care received as family-centred. Generally, caregivers are satisfied with the care their children receive and most value appropriate treatment and high quality communication with providers.

We look forward to receiving more questionnaires and updating our results!

STAY CONNECTED!

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