



CIMDRN
Canadian Inherited Metabolic Diseases Research Network

Newsletter 2016

To our participating families,

We cannot thank you enough for your ongoing contribution to our research network. We could not do the work we do without your participation!

There are currently 560 participants enrolled in CIMDRN!

Questionnaires for families

If you indicated that you wished to be invited to participate in our surveys, expect to receive mail from CIMDRN in the coming months!



RECRUITING CENTRES

CIMDRN is currently recruiting participants across Canada at the following 13 treatment centres:



RECENT PUBLICATIONS

“The health system impact of false positive newborn screening results for medium-chain acyl-CoA dehydrogenase deficiency: a cohort study” in *Orphanet Journal of Rare Diseases* (2016)

<http://doi.org/10.1186/s13023-016-0391-5>

“Child and family experiences with inborn errors of metabolism: a qualitative interview study with representatives of patient groups” in *Journal of Inherited Metabolic Disease* (2015)

<http://doi.org/10.1007/s10545-015-9881-1>

“Translating rare-disease therapies into improved care for patients and families: what are the right outcomes, designs, and engagement approaches in health-systems research?” in *Genetics In Medicine* (2015)

<http://go.nature.com/ySvjgQ>

Interviews with guardians

A qualitative interview study was conducted in order to understand caregivers' experiences with the management of their children's inherited metabolic disease (IMD).

Summary of results

We interviewed 21 caregivers receiving care from 4 treatment centres. The interviews provided rich insights into parents'/guardians' personal experiences of having a child with an IMD, with respect to the impact of the disease and its management on child and family life, and their perceptions of interacting with the health care system.

Participants reported that they became well-adjusted to the complex daily management requirements of their child's disease and redefined their 'new normal' through the use of proactive coping strategies. Despite these coping strategies that appeared to ease the management of daily routines, many respondents reported stress related to concerns about the social lives of their children that necessitated parent advocacy, particularly surrounding situations where the child may be excluded because of necessary dietary restrictions.

With regards to experiences with the health care system, most parents interviewed were highly satisfied with care received in the specialist metabolic clinic. However, participants often reported negative experiences with the non IMD-specific components of the health care system that were used frequently, such as the emergency department, pharmacy, or blood laboratory. While these were often single events, they were recounted across many interviews and described as an important source of stress.

The findings from this study point to the potential benefit of a coordinated approach to care that includes the non disease-specific elements of care as well as specialist metabolic care, in order to adequately support the needs of children and their caregivers. Social stressors on parents are important for health care providers to consider with respect to the social and emotional responses associated with disease management. The successful use of a range of proactive coping strategies among study participants suggests the potential value of promoting their use, and this is an important direction for future study. Further research is also needed to better understand the extent to which our findings apply to a broader group of families.

Study results to be published...stay tuned!

Clinical data collection

More than 20 research staff across 13 centres are entering clinical information into the research database for the over 500 participants enrolled in CIMDRN!



STAY CONNECTED!

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