

Caregiver perspectives on family management of care for children with inherited metabolic diseases: results from a cross-sectional survey

Andrea J. Chow¹, Zobaida Al-Baldawi¹, Ryan Iverson², Monica Lamoureux², Kylie Tingley¹, Isabel Jordan³, Nicole Pallone⁴, Maureen Smith⁵, Pranesh Chakraborty², Jamie Brehaut¹, Alicia K.J. Chan⁶, Eyal Cohen⁷, Sarah Dyack⁸, Cheryl R. Greenberg⁹, Jeremy M. Grimshaw¹⁰, Robin Hayeems⁷, Michal Inbar-Feigenberg⁷, Natalya Karp¹¹, Sara Khangura¹, Jennifer J. MacKenzie¹², Nathalie Major², John J. Mitchell¹³, Stuart G. Nicholls¹⁰, Amy Pender¹², Murray Potter¹², Chitra Prasad¹¹, Andreas Schulze⁷, Komudi Siriwardena⁶, Rebecca Sparkes¹⁴, Kathy N Speechley¹¹, Sylvia Stockler¹⁵, Monica Taljaard¹, Yannis Trakadis¹³, Jagdeep S. Walia¹⁶, Brenda J. Wilson¹⁷, Kumanan Wilson¹⁰ and Beth K. Potter¹

¹University of Ottawa, Ottawa, ON, ²Children's Hospital of Eastern Ontario, Ottawa, ON, ³Patient Partner, Squamish, BC, ⁴Patient Partner, Canadian PKU & Allied Disorders Inc, Toronto, ON, ⁵Patient Partner, Canadian Organization for Rare Disorders, Toronto, ON, ⁶University of Alberta, Edmonton, AB, ⁷Hospital for Sick Children, Toronto, ON, ⁸Dalhousie University, Halifax, NS, ⁹University of Manitoba, Winnipeg, MB, ¹⁰Ottawa Hospital Research Institute, Ottawa, ON, ¹¹Western University, London, ON, ¹²Hamilton Health Sciences, McMaster Children's Hospital, Hamilton, ON, ¹³McGill University Health Centre, Montreal, QC, ¹⁴Alberta Children's Hospital, Calgary, AB, ¹⁵B.C. Children's Hospital, Vancouver, BC, ¹⁶Kingston Health Sciences Centre, ON, ¹⁷Memorial University of Newfoundland, St. John's, NL

Rationale + Objective

Rationale

- Management of pediatric inherited metabolic diseases (IMDs) is complex and often requires significant day-to-day management by families
- Only a few studies have examined specific caregiver contributions to IMD management, none known to be in Canada^{1,2}
- More studies have measured quality of life of caregivers of children with IMD, though none known to be in Canada³⁻⁵

Objective

Describe family experiences with managing healthcare for children with IMDs

Design + Methods

Participants: Parents (“caregivers”) of children ≤12 years with IMD receiving care at & recruited through 1/11 Canadian pediatric metabolic clinics

- We sought a diverse sample on characteristics anticipated to be associated with health care: child age, sex, IMD, treatment centre, and residential proximity to specialty care

Design: Cross-sectional, online survey

Data collection:

- Family time and financial contributions to the management of the child's IMD
- Ease/difficulty in managing & accessing care
- Caregiver quality of life (CarerQOL instrument)⁶
- Family experiences with support services

Analysis: Descriptive statistics (proportions for categorical variables, medians for continuous variables)

Preliminary results

Table 1. Participant characteristics (N=68)

Respondent	n (%)
Relationship to child, mother	60 (88.2)
Paid employment, full-time	33 (48.5)
Highest education completed	
Secondary school diploma	14 (20.6)
College / vocational degree	15 (22.1)
University degree	39 (57.4)
Residence	
Ontario	37 (54.4)
Alberta	11 (16.2)
New Brunswick / Nova Scotia	12 (17.6)
British Columbia	4 (5.9)
Other	4 (5.9)
Child	
IMD	
Amino acid disorders	16 (23.5)
Urea cycle disorders	8 (11.8)
Organic acid disorders	4 (5.9)
Fatty acid oxidation disorders	21 (30.9)
Mucopolysaccharidoses	10 (14.7)
Other	9 (13.2)
Sex assigned at birth, female	39 (57.4)
Age, years	
0 – 3	27 (39.7)
4 – 6	20 (29.4)
7 – 9	14 (20.6)
10 – 12	7 (10.3)
Other special needs/chronic illness, yes ^a	16 (25.4)

^aI don't know or I prefer not to answer = 5

- Most participants were the mother of a child with an IMD (Table 1); children of participants had IMD diagnoses that were heterogeneous across the sample.
- The most common at-home activities to manage the child's IMD were reported as: medication management (n=37), meal management (n=31), and diet procurement (n=29) (Fig. 1).
- Fewer participants spent time on an exercise regimen or on therapy and device management.

Family experiences managing IMDs

Figure 1. Family time contributions

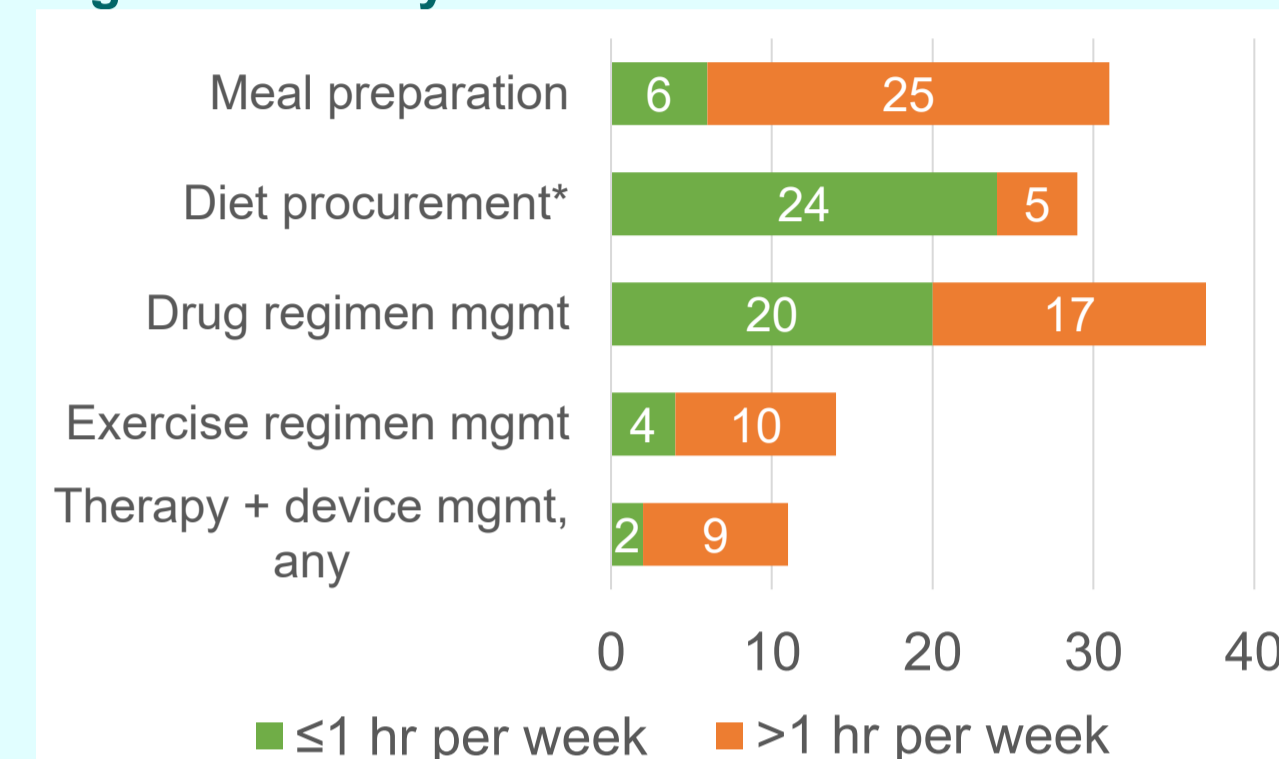
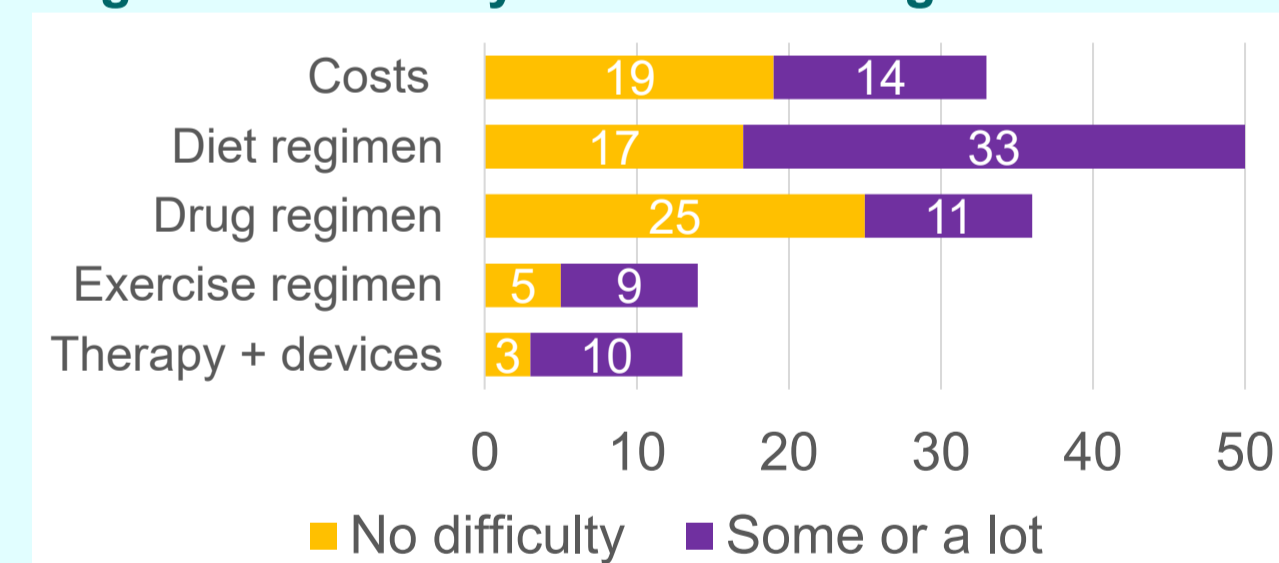


Figure 2. Out of pocket financial contributions



Figure 3. Difficulty with IMD management



- A majority of participants reported out-of-pocket financial costs associated with a child's IMD management (n=35, Fig. 2).
- Among 47 caregivers working full/part-time, 18 (38.3%) missed >15 days of work due to child care over 12 months. In lifetime, 42.7% respondents (n=29) had quit a job or reduced work hours due to child's IMD (data not shown).
- The most common challenges participants reported in IMD management related to diet regimens (n=33) and costs (n=14) (Fig. 3).

Caregiver experiences accessing care needs

Table 2. Difficulty experienced in accessing care needs

	No. using item	Access	
		Easy	Hard
		n (%)	
Child's IMD management	N = 68		
Prescription drugs or medication	37	36 (97.3)	1 (2.7)
Medical foods or formulations	31	26 (83.9)	5 (16.1)
Dietary supplements ^a	27	17 (65.4)	9 (34.6)
Dietary prescription or support services	44	39 (88.6)	5 (11.4)
Exercise prescription or support services	14	8 (57.1)	6 (42.9)
School assistance	21	10 (47.6)	11 (52.4)
Home, respite or social services	14	11 (78.6)	3 (21.4)
Developmental support ^b	14	9 (64.3)	5 (35.7)
Therapies or devices	13	10 (76.9)	3 (23.1)
Family supports	N = 65		
Counselling or support groups	17	11 (64.7)	6 (35.3)
Financial support	12	7 (58.3)	5 (41.7)
Respite services	10	5 (50.0)	5 (50.0)
Home visits	6	6 (100.0)	0 (0.0)
Care coordination / case management, system navigation ^c or advocacy support	15	8 (53.3)	7 (46.7)
Education, skill building or coping strategies	9	8 (88.9)	1 (11.1)
Paid leave from work	4	4 (100.0)	0 (0.0)

^aMissing, n=1; ^bdevelopment support = neurological or cognitive assessment or early childhood interventions; ^chealthcare or other systems

- The most common supports used in their child's IMD management were reported as: medication (n=37), diet-related (n=27-44) & school assistance (n=21) (Table 2).
- Among those reporting the use of specific supports, difficulties with access were reported by ≥50% for school assistance (11/21) and respite services (5/10).

Discussion

- Caregivers of children with IMDs reported supporting their children's care at home, with management of special diets and medications being most common.
- A significant proportion of participants faced at least some difficulty managing their child's diet (n=33, 66.0%), exercise regimens (n=9, 64.3%), and various therapies/devices (n=10, 76.9%); and nearly half had quit a job or reduced their work hours to support their child's care.

Caregiver well-being

Figure 4. Participant well-being related to caregiving (N = 68)

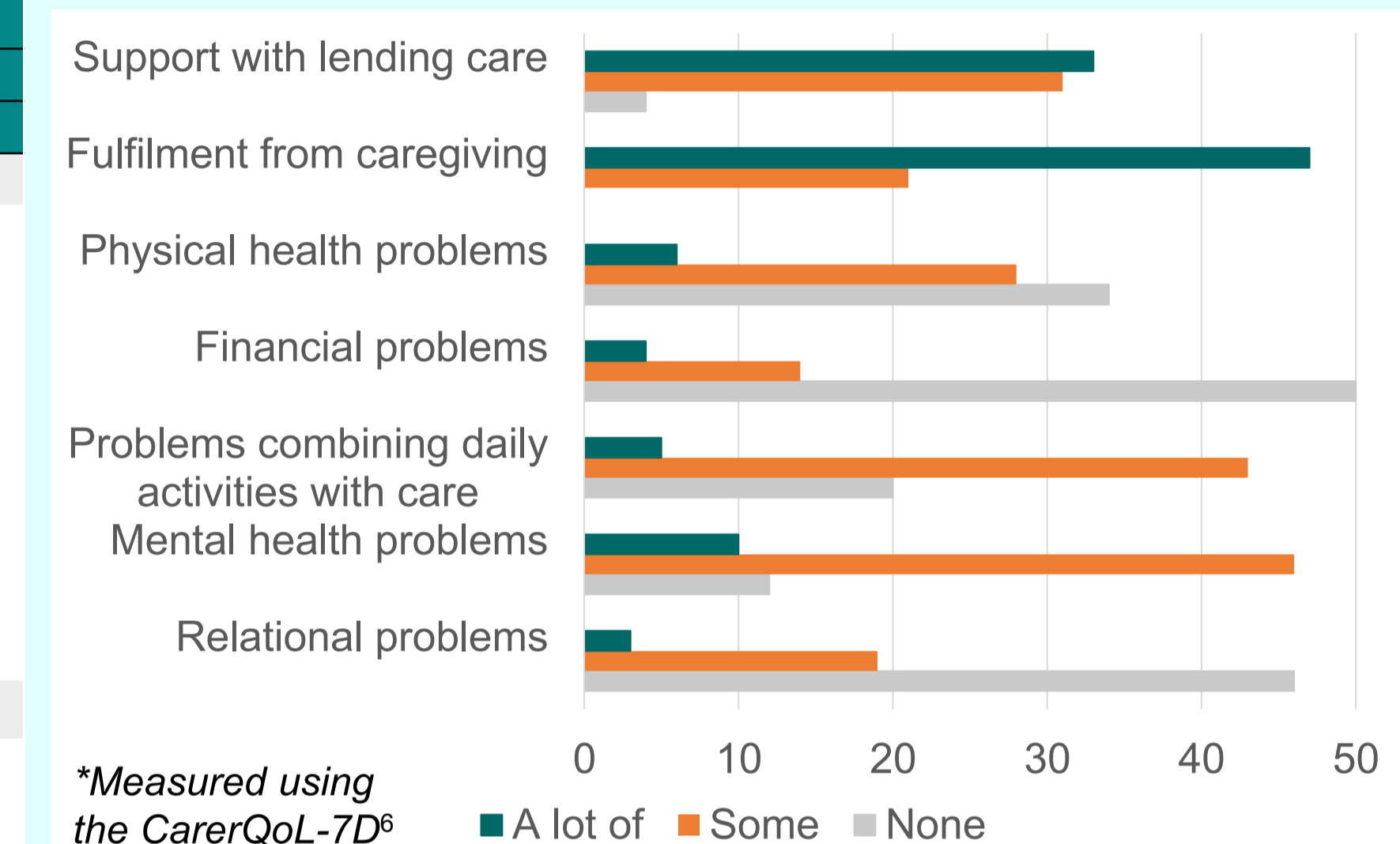
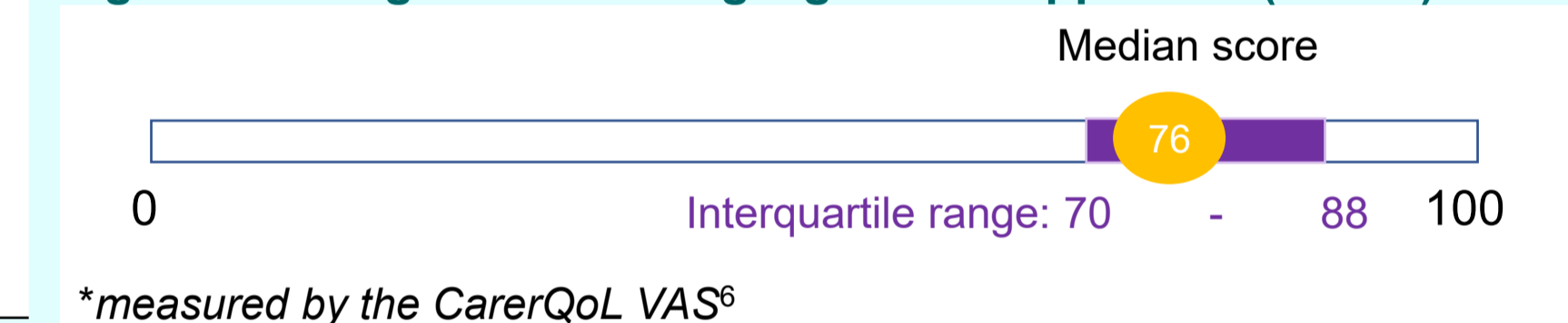


Figure 5. Caregiver well-being – general happiness* (N = 67)



- Caregivers frequently reported “a lot of” fulfillment from caregiving (69%) & support in providing care (49%) (Fig. 4).
- A majority also reported experiencing some or a lot of mental health difficulties (83%) and challenges in combining daily activities with care (70%).
- Caregivers' median happiness rating (scale 0-100) was 76 (interquartile range 70-88) (Fig. 5).

- While caregivers generally found their role fulfilling, many struggled with mental health and with balancing other activities; caregivers' happiness was similar to other studies of parents of children with chronic illness that used the Carer-QOL tool^{7,8}
- Limitation:** We collected data during the COVID-19 pandemic, which may have played a role in care, access & mental health.
- Conclusion:** Improved access to supports for caregivers should be prioritized as part of holistic pediatric IMD management.