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The Impact of Rett Syndrome on Caregivers and Family Members: A Multi-Method Study

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INTRODUCTION

- Rett syndrome (RTT) is a childhood-onset, neurodevelopmental disorder causing progressive loss of motor skills and cognitive function.[1]
- Due to their complex needs, individuals with RTT do not develop independence and require continuous care throughout their life.[2,3]
- Caregiving becomes an integral part of parenting, blurring the lines between "typical" parental duties and extensive, sustained medicalized care.

OBJECTIVES

- To explore the impact of caregiving for an individual with RTT on the health-related quality of life (HRQL) of caregivers and their family members, in the UK.

METHODS

- A sequential multi-method study was conducted:
  - **Stage 1:** cross-sectional survey including standardized instruments capturing HRQL (EQ-5D-5L, CarerQol-7D).
  - **Stage 2:** semi-structured online interview exploring the day-to-day experience of caring for an individual with RTT, the HRQL impacts for the primary caregiver being and other household members.
- Participants were recruited by a specialist recruitment agency.
- Interviews were audio-recorded and transcribed. Transcripts were analyzed using content and thematic analysis.
- All participants provided informed consent.

RESULTS

- N=37 caregivers took part in the study. All but one participant was the parent or step-parent of the individual with RTT.
- Median age of care-recipients was 13 years old (range: 4- 41 years). Most could not stand or walk (50%) or could walk only a short distance with support (24%). Majority could not use recognizable words but made some sounds for communication (81%).

Average time spent providing care was 99 hours per week (range: 54-122).

Diagnosis of RTT leads to a sudden and enduring alteration in the life trajectory of caregivers and the wider family unit.

Caregivers' lives centered around their child's needs even as their child had aged and physically grown.

HRQL measures

- **EQ-5D-5L:** Caregivers' mean EQ-5D utility score was 0.621 (SD=0.254); 0.19 points lower than the English population norm (0.811).
  - 48.6% reported moderate-to-severe anxiety/depression.
  - 37.8% reported moderate-to-severe severe problems pain/discomfort.
- **CarerQol-7D:** The total mean CarerQoL-7D utility was 60.8 (SD=18.1), where 0 indicates the worst imaginable caregiving situation and 100 indicates the best imaginable caregiving situation.
  - 15.3 points lower than informal caregivers of people with dementia in the UK (76.1)[4]
  - Estimate is aligned with a previous study examining HRQL in caregivers of children with neuromuscular diseases (range: 0.59-0.81)[5]

Figure 1. Caregiver EQ-5D-5L domain responses (N=37)

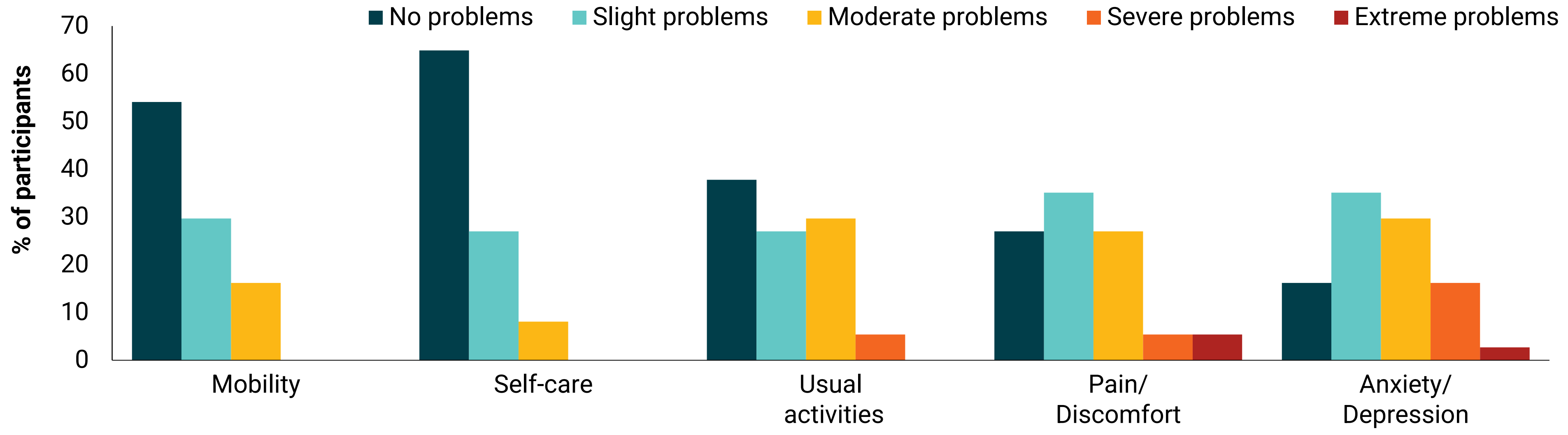
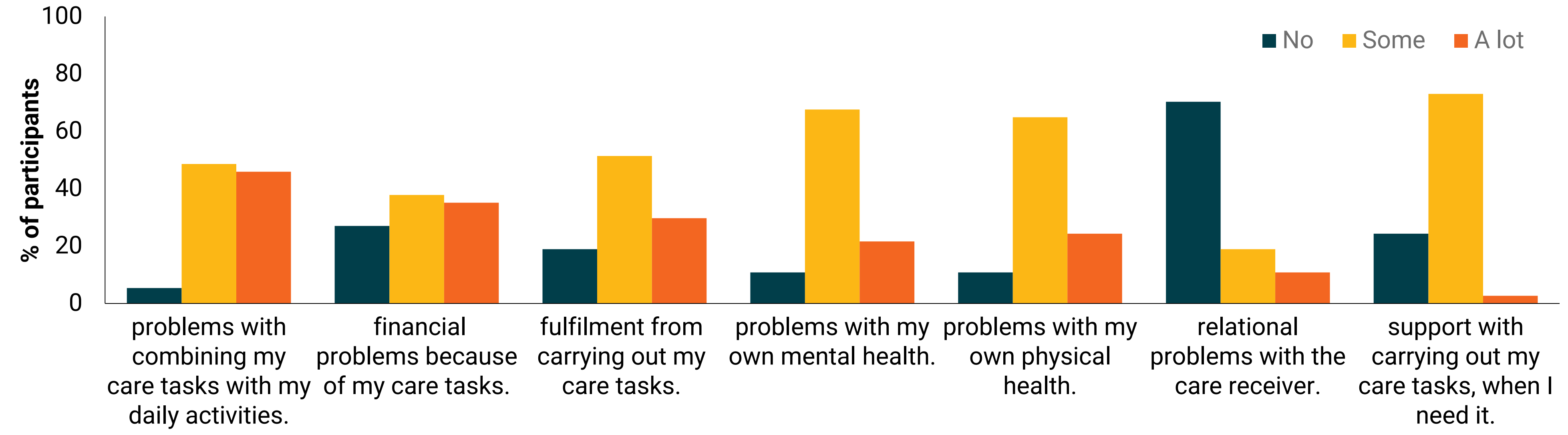


Figure 2. Caregiver CarerQoL-7D domain responses (N=37)



Qualitative measures

Table 1. Summary of interview findings regarding HRQL impacts experienced by caregivers (N=37)

Physical impacts (n=35)	• ***Fatigue, back pain, joint pain*** and **exacerbation of existing conditions** due to physical demands of caring (e.g. lifting, hoisting) • Physical problems are compounded by **sleep disruption** due to monitoring and care duties during the night	*"The pain is excruciating. Sometimes I can't even stand up myself.."* *"She's getting heavier every year. I hurt all the time, my back, my hips…"*
Impacts on daily (n=36) & social (n=34) activities	• Daily activities ***center around the needs of their child*** • ***Reduced time*** and ***freedom*** to engage in other activities • Outings and social events are often routine-based and require extensive planning	*"She can't be left alone so it's a real juggle trying to do anything normal day-to-day."* *"I can no longer be spontaneous and drop everything...you know a lot of parents would be in that situation when their child is small but it's always like that now you know, regardless of how old she gets."*
Relationship impacts (n=36)	• Deterioration or breakdown of romantic relationships due to ***lack of quality time*** and caregiving pressures • Strained relationships with family and friends due to ***divergence in life circumstances with peers***, ***limited availability*** and ***last-minute cancellations***	*"I'm no longer with her dad. And I think a lot of that is down to how much care she required…it does definitely impact relationships."* *"You can't promise anything, you can't say 'I'll definitely be there', and even just going to visit family and stuff, they don't get it."*
Emotional and psychological impacts (n=37)	• All caregivers described a negative emotional impact • ***Anxiety*** and ***worry*** due to the unpredictability of RTT • ***Sense of loss*** for the life they had envisioned especially as their life trajectory continues to diverge from others their age • ***Guilt*** about the sacrifices made when balancing caregiving with other wants and needs	*"You've got a level of anxiety always there going, 'Am I going to get a phone call?'"* *"So, like your typical family, the mum, the dad, the two kids like that makes me jealous. Not that I'd be anybody else, but I still struggle with why is that not us?"* *"Guilt, heartache, grief...You're grieving someone who's alive. That's a really, really kind of mentally hard one to manage."*
Financial impacts (n=25)	• Restrictions to work - ***60% of participants had stopped working to care for their child full-time*** • ***Increased household expenditure*** to accommodate child's additional needs (e.g., home adaptations, medical supplies, additional electricity and water bills, specialized dietary requirements)	*"The cost of the washing, the electricity, the water, the equipment we need to buy that isn't funded…there is a significant cost involved."* *"I was just at the start of my career when the diagnosis came."*

CONCLUSIONS

Caring for someone with RTT has a profound and long-lasting impact on the HRQL of primary caregivers, and other family members.

Caregivers described a sudden and permanent shift in lifestyle, relationships, and future expectations.

- Findings echo qualitative studies relating to progressive pediatric conditions.
- Unlike caring for aging relatives, caregiving responsibilities occur unexpectedly during a transitional life phase of family planning when social, financial and career foundations are still being established

These results highlight the spillover effects of the burden of life-limiting, childhood onset conditions beyond the individual living with the condition.

DISCLOSURES

DD and MS are employees of Acadia Pharmaceuticals Inc. who sponsored this study. AP, RH, TO and AL are employees of Acaster Lloyd Consulting Ltd. who conducted the study.

ACKNOWLEDGMENTS

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