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Quality of Life Impact of Caregiving for Patients with MCT8 Deficiency: Results From a Cross-Sectional Survey

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Objectives

Monocarboxylate transporter 8 (MCT8) deficiency is a very rare, highly debilitating and life-limiting genetic disease characterized by a serious neurodevelopmental disorder as well as chronic thyrotoxicosis[1]. The burden on caregivers of people affected by MCT8 deficiency is poorly understood[2].

This study aimed to quantify the health-related quality of life impact of caring for a person with MCT8 deficiency.

Methods

Informal caregivers of people with MCT8 deficiency completed a cross-sectional online survey. This would be a person who identifies themselves as a main and unpaid caregiver, typically a close family member or friend.

This survey included the PedsQL Family Impact Module instrument, which measures the impact of pediatric chronic health conditions on parents with multiple domains. Each domain includes questions that are answered on a 5-point Likert scale (Figure 1). Scores range from 0 to 100; higher scores indicate better functioning.

DOMAINS
Physical function
Emotional function
Social function
Cognitive function
Communication
Worry
Daily activities
Family

RESPONSE OPTIONS
Never
Almost never
Sometimes
Often
Almost always

Figure 1. Domains and response options of PedsQL instrument

Results

Participants' mean age was 42.2 years (n=22); 81.8% were female and 95.5% were the parent of the person with MCT8 deficiency. The mean age of the person with MCT8 deficiency was 8.8 years, ranging from 0.8 to 30.4 years.

The mean total PedsQL score was 41.1. Caregivers reported the lowest mean scores for daily activities, social functioning and physical functioning. This is due to the higher proportion of caregivers reporting 'often' and 'almost always' for each item. Within each domain, most participants reported 'often' feeling too tired to do the things they like to do (66.7%), not having enough energy for social activities (57.1%) and family activities taking more time and effort (47.6%) (Figure 2).

The highest scores were reported for family relationships, emotional functioning and cognitive functioning. Here, fewer participants reported 'often' and 'almost always' for each item.

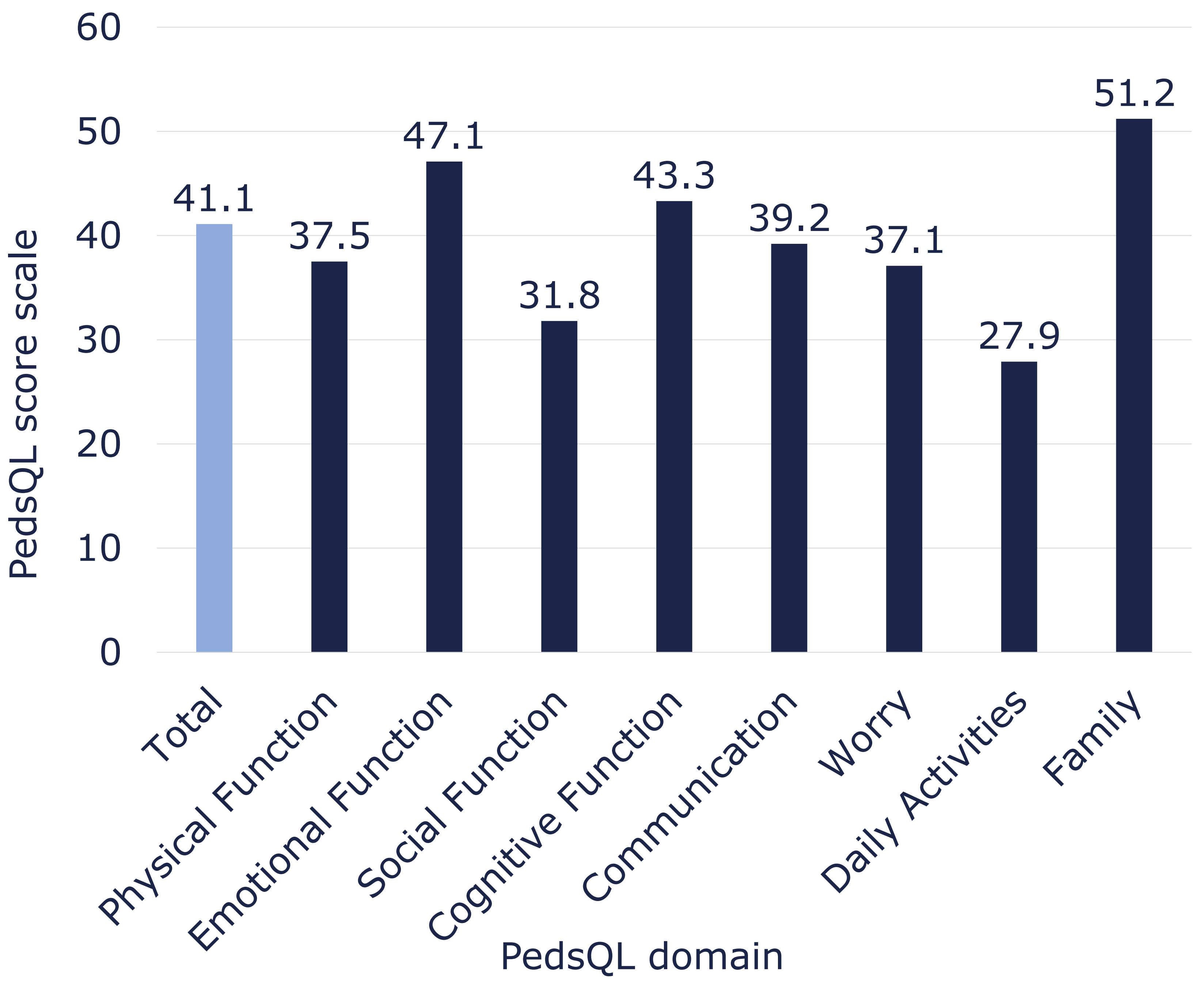

Figure 2. Mean PedsQL scores by domain

Most participants reported 'sometimes' having family conflict and difficulty making decisions as a family (57.1%), finding it hard to think quickly (57.1%), and feeling angry and frustrated (52.4%) (Figure 2).

Caregivers reported 'almost always' worrying about the future of the person with MCT8 deficiency and whether treatments were working. Caregivers also 'almost always' experienced a lack of time and energy to engage in social activities outside of caregiving.

Caregiving impacted participants' level of attention and memory, as well as contributing to experiences of anxiety, sadness, anger, frustration and helplessness. Participants reported frequently experiencing feelings of isolation, a perceived lack of understanding of their family situation and an inability to talk about the person's condition and to open up to healthcare providers about caring for the person with MCT8 deficiency.

Conclusion

Caregivers reported a low to moderate level of functioning across domains of the PedsQL, indicating that caring for someone with MCT8 deficiency has an impact on HRQoL. Caregivers reported lowest scores for daily activities, social functioning and physical functioning.

References

1. van Geest FS et al. Monocarboxylate transporter 8 deficiency: from pathophysiological understanding to therapy development. Front Endocrinol. 2021;12:723750.
2. van Geest FS et al. Parent Perspectives on Complex Needs in Patients With MCT8 Deficiency: An International, Prospective, Registry Study. J Clin Endocrinol Metab. 2024;109(1):e330-e335.