

THE ROLE OF INSTITUTIONS IN SUPPORTING PEOPLE WITH RARE DISEASES AND THEIR CAREGIVERS

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Abstract: *People with rare diseases such as MCT8 deficiency face complex medical and structural challenges. Due to severe developmental disorders, those affected are unable to speak for themselves. Their relatives therefore take on central roles – as caregivers, advocates and liaisons between clinics, research and everyday life. Based on a literature review, this article analyses what care needs exist and to what extent these have been taken into account in previous studies. A total of eleven evidence-based treatment studies and one research initiative on the care situation of those affected were identified. The analysis shows that patient-relevant aspects, in particular quality of life and problems in coping with everyday life, have not been sufficiently taken into account to date. The results underscore the need to systematically integrate patient-relevant objectives – in particular quality of life – into clinical research on MCT8 deficiency and care practice. On this basis, recommendations are derived for needs-oriented, participatory and interdisciplinary support for families with MCT8 deficiency through sustainable institutional structures.*

Keywords: *MCT8 deficiency; rare disease; patient-relevant outcomes; communication processes*

INTRODUCTION

Rare diseases such as MCT8 deficiency not only pose a medical challenge, but also require a comprehensive understanding of the communication processes and the social and institutional conditions under which care is provided to those affected. Due to severe neurocognitive and motor impairments (cf. Allan et al. 1944; Dumitrescu et al. 2004; Friesema et al. 2004; Schwartz et al. 2005), affected children are unable to communicate their needs themselves, have complex requirements and are dependent on lifelong care (cf. Van Geest et al. 2023). Their relatives therefore take on central roles – as carers, advocates and mediators between clinics, research and everyday life.

Needs-based care requires the interaction of various institutions - from specialized medical centers, doctors in private practice and therapy services to care and social insurance providers, educational authorities, self-help organizations and scientific players. While there are established care models and contact points for common diseases, such structures are often lacking for rare diseases or are not tailored to their specific requirements. Parents often find the role of care coordinators overwhelming – a task that should actually be the responsibility of specialist health care agencies (cf. Baumbusch et al. 2018). A global comparison reveals a structural lack of holistic, person-centered and multidisciplinary approaches to the care of rare diseases (cf. Lopez Gousset et al. 2021). In the German healthcare system in particular, the economic focus makes it difficult to provide needs-based care – especially for

children and adolescents with complex chronic diseases (cf. Heinen 2020). The lack of centralized expertise on rare diseases and a lack of understanding of the reality of life for those affected often prevent adequate disease management.

This is particularly evident in the example of MCT8 deficiency, which is characterized by an extremely low number of cases (cf. Schwartz, 2017) and a superregional distribution of those affected (cf. MCT8-Forschung e.V. 2019). Despite isolated studies (cf. Van Geest et al. 2023), only limited knowledge about the reality of care for this patient group is available to date. This gap not only makes it difficult to optimize treatment, but also to support the families of those affected and to implement effective disease management.

The aim of this article is therefore to gain a deeper understanding of the care situation of people with MCT8 deficiency on the basis of a literature review and to critically evaluate existing communication, research and care processes. The analysis provides important insights into treatment needs, gaps in care and everyday needs from the perspective of relatives – information that is indispensable for research, care and policy. Only through a participatory approach can the specific needs of this particularly vulnerable patient group be adequately taken into account and translated into care practice.

RESEARCH METHODOLOGY

The analysis is based on a literature review of treatment and care concepts for MCT8 deficiency. For this purpose, a literature search was conducted in the MEDLINE/PubMed, Google Scholar and ClinicalTrials.gov databases. The search was conducted using the following keyword combinations: ‘[(SLC16A2) OR (MCT8) OR (AHDS) OR (Allan Herndon Dudley syndrome)] AND [(therapy) OR (intervention) OR (treatment)]’ and ‘[(SLC16A2) OR (MCT8) OR (AHDS) OR (Allan Herndon Dudley syndrome)] AND [(patient-reported outcomes) OR (PRO) OR (caregiver) OR (stakeholder)]’ and was completed on 10 February 2024. Evidence-based treatment studies on MCT8 deficiency in humans were considered, as well as studies dealing with the care situation in the context of MCT8 deficiency that were published in peer-reviewed journals and were available as full-text articles. There were no restrictions regarding language. Treatment approaches that have proven to be disadvantageous and are therefore no longer used were excluded. A total of n=11 treatment studies were included (cf. Wémeau et al. 2008; Zung et al. 2011; Tonduti et al. 2012; Verge et al. 2012; Visser et al. 2012; Groeneweg et al. 2019; Remerand et al. 2019; Refetoff et al. 2020; Van Geest et al. 2021; Gazek et al. 2023; Ünsal/Hayran 2023) and n=1 research initiative on the care situation (cf. Van Geest et al. 2023) in the context of MCT8 deficiency that met the requirements.

RESULTS

People with MCT8 deficiency and their caregivers have complex needs that require a wide range of therapeutic support. However, previous clinical studies have largely ignored the everyday stresses and perspectives of those affected. This defi-

ciency is particularly evident with regard to the practical care problems faced by families. Although individual therapeutic measures, such as TRIAC for peripheral thyrotoxicosis or botulinum toxin for spasticity, have shown positive effects, it is clear that the existing therapeutic options do not adequately cover the multiple, complex needs of AHDS patients and their caregiving relatives.

An evaluation of patient registry data from 2018 to 2022 shows that feeding and sleeping disorders as well as limited motor skills are among the most common challenges in everyday care (cf. Van Geest et al. 2023). Despite these burdens, the majority of patients did not receive professional nutritional counselling. Cardiac comorbidities, which frequently occur in MCT8 deficiency, were also rarely assessed by a cardiologist. In addition, 58% of the parents surveyed (18 out of 31) rated the medical expertise in dealing with MCT8 deficiency as ‘poor’ or ‘very poor’ (cf. Van Geest et al. 2023). This assessment indicates a significant discrepancy between parental expectations and actual care practices. The analysis also makes it clear that only a few studies have systematically recorded patient-reported outcomes to date. As a result, key aspects relevant to everyday life, such as sleep quality, nutrition and motor independence, remain largely underrepresented from the perspective of relatives.

Given the severity of the disease, the complexity of the needs and the high level of care required, there is a clear need to systematically incorporate the perspectives of patients and patient representatives into research and clinical decision-making processes. At the same time, the central importance of structured, interdisciplinary care is evident. Only through a needs-oriented, coordinated and participatory approach – supported by reliable institutional structures – can long-term effective care be guaranteed.

DISCUSSION

The results of the literature analysis show that patient-relevant aspects – especially quality of life and everyday challenges – have been insufficiently addressed in previous research on MCT8 deficiency. Interdisciplinary care areas such as nutritional counseling or cardiological care are rarely used despite clear indications (cf. Van Geest et al. 2023). The assessment of parental satisfaction with professional competence in the field of MCT8 also points to a general knowledge deficit within the healthcare system. In order to tailor care for people with MCT8 deficiency to their needs, research and care practice must be more strongly focused on patient-relevant objectives. The consistent involvement of patients and their representatives is just as essential for this as the expansion of interdisciplinary, cross-sector care structures.

Particularly in the case of rare diseases such as MCT8 deficiency, the development of institutional competencies should be promoted – for example, through specialized treatment centers or central contact points that coordinate care. At the same time, low-threshold, family-oriented services are needed to relieve and support family caregivers. The role of self-help organizations should also be empha-

sized in this context: they make a key contribution to education, networking, and advocacy and should be structurally strengthened in order to further expand their potential as a bridge between science, practice, and those affected.

CONCLUSION

This analysis illustrates the significant discrepancy between the reality of care and the actual support needs for rare diseases such as MCT8 deficiency. The needs of those affected, particularly with regard to quality of life and symptom-oriented support, are not adequately addressed. Disease management for patients with rare diseases requires a reorientation of institutional structures towards participatory, needs-oriented and multidisciplinary forms of support. Family members should be recognized as key cooperation partners and receive targeted support. Only in this way can patient needs be effectively incorporated into care and research and the quality of life of those affected be improved.

Acknowledgements & Conflict of Interest

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РОЛЯТА НА ИНСТИТУЦИИТЕ В ПОДКРЕПА НА ХОРАТА С РЕДКИ БОЛЕСТИ И ТЕХНИТЕ НАСТОЙНИЦИ

Резюме: Хората с редки заболявания, като например дефицит на МСТ8, са изправени пред сложни медицински, социални и структурни предизвикателства. Поради тежките нарушения в развитието засегнатите не могат да говорят сами за себе си. Поради това техните роднини заемат централни роли – като болногледачи, застъпници и връзки между клиниката, научните изследвания и ежедневието. Въз основа на литературен преглед и проучване сред 21 международни представители на пациенти, основано на ангажираност на заинтересованите страни, в настоящия текст се анализират съществуващите нужди и начинът, по който различните институции са се справили с тях до момента. Подобряването на качеството на живот е посочено като основна цел – по-специално чрез по-добър сън, повече удоволствие от живота и намаляване на стресовите симптоми. Удовлетвореността от настоящата терапия и услуги за подкрепа е оценена като умерена във всички области. Резултатите подчертават необходимостта от систематично интегриране на целите, свързани с пациентите – особено качеството на живот – в клиничните изследвания на дефицита на МСТ8 и в практиката на грижите. Проучването извежда препоръки за ориентирана към нуждите, участваща и интердисциплинарна подкрепа на семействата с дефицит на МСТ8 чрез устойчиви институционални структури.

Ключови думи: дефицит на МСТ8; рядко заболяване; резултати, свързани с пациентите; комуникационни процеси

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